

# International Conference on Birth Defects and Disabilities in the Developing World

"Health for all: Accelerate Efforts for Birth Defects Prevention and Care"

23 to 26 February 2020 Cinnamon Grand Hotel, Colombo, Sri Lanka

**PROGRAMME & ABSTRACT BOOK** 



icbd.marchofdimes.org

# International Conference on Birth Defects and Disabilities in the Developing World

"Health for All: Accelerate Efforts for Birth Defects Prevention and Care"

## Organised by

Sri Lanka Medical Association

**MARCH OF DIMES** 

Family Health Bureau Ministry of Health Sri Lanka U.S. Department of Health and Human Services Centers for Disease Control and Prevention

## **Partner Organizations**



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## ICBD 2001 - 2020

## About the International Conference on Birth Defects and Disabilities in the Developing World

Recognizing the need to build capacity in lower-income countries for the prevention of birth defects and preterm birth and care of those affected, the goal of these biennial conferences has been to provide a platform for sharing tools and approaches that developing country participants can use to implement and strengthen surveillance, health care delivery systems policy and funding for improving birth outcomes in their respective countries.

## **Previous Conferences**

Bogotá D.C., Colombia (2017) Dar-es-Salaam, Tanzania (2015) Cebu, Philippines (2013) Lodz, Poland (2011) New Delhi, India (2009) Rio de Janeiro, Brazil (2007) Beijing, China (2005) Johannesburg, South Africa, (2001)

## **CONFERENCE COMMITTEES**

Conference Chair	Dr. Kapila Jayaratne	
Conference Co-Chairs	Dr. Anula Wijesundare Dr. Salimah Walani	
Scientific Committee Co-Chairs	Prof. <i>Vidya Jyothi</i> Vajira H. W. Dis Dr. Salimah Walani	sanayake
Sustaining Advisory Committee	Dr. Kelle Moley Dr. Salimah Walani Dr. Cynthia Moore Dr. Neena Raina Dr. Pablo Duran Dr. Carmencita Padilla Dr. Ignacio Zarante Dr. Kapila Jayaratne Mr. Federico Nuñez Perea	
Scientific Programme Committee	Dr. Navoda Attapattu Dr. Manjula Danansuriya Ms. Lasanthi Daskon Dr. Boris Groisman Dr. Madhulika Kabra Prof. Dhavendra Kumar Dr. Saqib Mahmood Dr. Rajesh Metha Dr. Rathna Puri Dr. Nirmala Sirisena Dr. Nilam Thakur Dr. Saraji Wijesekara Dr. Piyara Rathnayake Dr. Ornella Lincetto	Dr. Hannah Blencowe Dr. Ernawati Darmawan Dr. Ruwangi Dissanayake Dr. Paula Hurtado Dr. Vijaya Kancheria Dr. Nishani Lucus Dr. Cara Mai Dr. Lahiru Prabodha Prof. Raj Ramesar Prof. Samanmali Sumanasena Dr. Sarah Verbiest Dr. Anuruddha Padeniya Dr. Nathalie Roos
Editorial Committee	Dr. Ruwan Ferdinando Dr. Manjula Danansuriya Dr. Saraji Wijesekara Dr. Buddhika Mahesh Dr. Sameera Senanayake Dr. Samthushya Fernando Dr. Sumudu Awanthi Hewage Prof. Carukshi Arambepola Dr. Anuji Gamage Prof. Sampatha Goonewardena Dr. Nirupa Pallewatte Dr. Thilanga Ruwanpathirana Dr. Priyadarshani Samarasinghe Dr. R. Surenthirakumaran Dr. Dilhani Samarasekera	
Website and Social Media Manager	Dr. Megha Ganewatta	

### **Other Representatives of Local Partner Organizations**

Sri Lanka Medical Association Prof. Indika Karunathilake Dr. Sumithra Tissera

Family Health Bureau, Ministry of Health Dr. Chithramalee de Silva Dr. Gayan Jinadasa Dr. Kshyama Welwagamage

Human Genetics Unit, Faculty of Medicine Dr. Nirmala Sirisena

*Ministry of Health* Dr. Sudath Samaraweera

College of Community Physicians of Sri Lanka Dr. Sapumal Dhanapala Dr. Saveen Gamage Dr. Ruwan Ferdinando

Perinatal Society of Sri Lanka Dr. Kaushalya Kasturiarachchi Dr. Asiri Hewamalage

Sri Lanka College of Paediatricians Prof. Vasantha Devasiri

Sri Lanka Association for Child Development Dr. Saraji Wijesekara

Sri Lanka Heart Association Dr. Duminda Samarasinghe

Sri Lanka Association of Paediatric Surgeons Dr. Ananda Lamahewage

Health Informatics Society of Sri Lanka Dr. Rohana Marasinghe Dr. Roshan Hewapathirana

World Health Organization Dr. Manjula Danansuriya

## MESSAGE FROM THE MINISTER OF HEALTH AND INDIGENOUS MEDICAL SERVICES, WOMEN & CHILD AFFAIRS & SOCIAL SECURITY



It is with great pleasure that I send this message to convey my best wishes to participants of the 9th International Conference on Birth Defects and Disabilities in Developing World.

We are proud, as a nation with excellent indices in maternal and child health, to host this prestigious international conference. I congratulate the Sri Lanka Medical Association and other partnering local professional organizations for coming together in organizing this important conference.

Birth defects are emerging as a leading cause of morbidity and mortality among children nationally and globally. They are a major burden to the healthcare services of developing countries. It is noteworthy that during the past few decades, Sri Lanka has achieved remarkable success in reducing infant deaths by providing quality care at both field and institutional level, but still more has to be done.

I have come to know that there are many best practices that have been developed in various parts of the world to prevent birth defects and care for those with these conditions, based on scientific evidence. I believe it is vital that we have the opportunity through this conference to come together to share the scientific evidence and showcase the best practices so that we could learn from each other and implement these practices to improve lives of people in our respective nations.

I thank March of Dimes, the US Centres for Disease Control and Prevention and the World Health Organization for supporting Sri Lanka in numerous ways to host ICBD2020 in Colombo. I would like to thank them for the faith that they had kept in us in spite of numerous challenges to host the conference in Sri Lanka after the tragic events of 21 April 2019.

I am delighted to see all key players in the field of prevention and treatment of birth defects and disabilities in Colombo for what is set to be an inspirational ICBD Conference. I wish all of you a wonderful conference and, a pleasant stay in Sri Lanka for the Foreign Guests.

### Honourable Pavithra Wanniarachchi

Minister of Health and Indigenous Medical Services, Women & Child Affairs & Social Security

## MESSAGE FROM THE CONFERENCE CHAIR, CO-CHAIRS, & THE CO-CHAIRS OF THE SCIENTIFIC COMMITTEE



We wish Aayubovan (ආයුබෝවත් - *May you live long*) to all the participants of the *9th International Conference on Birth Defects and Disabilities in the Developing World (ICBD)* in Sri Lanka- often referred to as "the pearl of the Indian Ocean". We have the honor to serve as the chairs of this ICBD on behalf of our esteemed organizations, Sri Lanka Medical Association and March of Dimes, USA. We thank all the members of the various conference committees for helping us deliver this conference to you to continue the tradition that started with the first conference, almost 20 years ago.

The theme of this Conference is "Health for All: Accelerate Efforts for Birth Defects Prevention and Care". This theme aligns with the Sustainable Development Goal (SDG) # 3 and its aim to ensure Universal Health Coverage - health for all people, everywhere in the world regardless of their power to pay or speak for themselves. Birth defects affect all nations, but the burden is very high in Asia and Africa where many children with birth defects are often left behind. At the ICBD in Sri Lanka, presenters will discuss issues related to birth defects surveillance, prevention and care in connection with the SDG3 targets.

About 350 attendees from 36 countries are in attendance at this conference - they are healthcare professionals, policy makers, researchers, volunteers and students representing healthcare service organizations, academic institutions, governments, international organizations and civil society organizations. In addition, colleagues from many prominent organizations are here and we hope you will find opportunities to network and learn from each other.

ICBD 2020 offers a high quality mix of sessions and unique networking opportunities. The technical program is rich with 7 plenary sessions with invited speakers, 15 simultaneous symposia including symposia featuring oral presentations and 10 poster sessions. We have secured the highest caliber of speakers who inform and educate on a variety of birth defect-related issues. ICBD 2020 received over 150 abstracts on different topics, mostly in surveillance of birth defects, public health policies, health care systems to ensure prevention and on how to improve care for persons with birth defects. We believe this academic event would be a great opportunity to meet colleagues from all over the world, exchange valuable

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experiences and contribute to prevention and management of patients and families with birth defects at global and national levels.

We hope you enjoy the academic program and the social activities. Colombo offers a unique cultural experience. We encourage you to enjoy the delightful food, colorful landscapes and the spiritual vibe that Colombo offers.

**Dr. Kapila Jayaratne** *Conference Chair* 

**Dr. Anula Wijesundare** *Conference Co-Chair* 

**Dr. Salimah Walani** *Conference Co-Chair & Scientific Committee Co-Chair* 

**Prof.** *Vidya Jyothi* Vajira H. W. Dissanayake Scientific Committee Co-Chair

## **MESSAGE FROM MARCH OF DIMES**



On behalf of March of Dimes, I am delighted to welcome you to the 9th International Conference on Birth Defects and Disabilities in the Developing World. Since our first conference in 2001, March of Dimes has been a proud co-sponsor of this important conference series that brings together health professionals, researchers, policymakers and civil society organizations dedicated to birth defects and premature birth surveillance, prevention and care. Thank you to the Sri Lanka Medical Association for hosting this year's conference in Colombo, Sri Lanka.

Birth defects and preterm birth continue to be major causes of child mortality and lifelong disability all over the world, but the burden is particularly high in Asia and Africa. This biennial conference has helped us increase visibility and understanding of the burden of birth defects and preterm birth worldwide, and collaboration to find solutions to prevent and care for those impacted.

This conference is part of our work at March of Dimes to end the maternal and infant health crisis. This is one crisis, not two, as the health of moms and babies is deeply intertwined. Building on our decades-long legacy, March of Dimes is accelerating knowledge, research and programs and advocating for policies that fight for the health of all moms and babies in the U.S. and globally. We are proud to have you as partners in this critical work.

Thank you for joining us at this year's conference and for your commitment to fighting for healthy moms and strong babies.

Enjoy the conference!

**Stacey D. Stewart** President and CEO March of Dimes

## **MESSAGE FROM US CENTERS FOR DISEASE CONTROL & PREVENTION**



Working toward a day when every child is born with the best health possible and the potential for a full and productive life-that is our mission. However, it can be a challenging one to realize for the many millions affected by birth defects around the world. Birth defects are common, costly, and critical. Affecting about 1 in 33 babies worldwide each year, birth defects can affect babies regardless of where they are born, their ethnicities, or their races.

of life. And babies who survive and live with these conditions can have lifelong challenges. including long-term disabilities. Birth defects not only impact babies born with these conditions, they also have an emotional and financial impact on their families and communities.

The US Centers for Disease Control and Prevention (CDC) applies a public health approach to understand and prevent birth defects. Accurately tracking birth defects and analyzing the collected data is a first step in the prevention of birth defects. CDC uses tracking and research to identify causes of birth defects, find opportunities to prevent them, and improve the health of those living with these conditions. Understanding the causes of birth defects can lead to recommendations, policies, and services to help prevent them and care for those affected.

Birth defects tracking systems are also an essential component of preparedness in our efforts to protect mothers and babies from emerging health threats. As you know, the 2016 Zika virus outbreak served as a stark reminder of the vulnerability of pregnant women and babies to emerging infectious diseases and the destructive consequences that can result from infections during pregnancy. The outbreak demonstrated that birth defects can be the first sign that an emerging infection causes serious harm and that rapid data collection can be critical for public health action. We are now applying lessons learned from the 2016 Zika virus outbreak to address other emerging health threats. Through a strong foundation of birth defects tracking and research, we can identify and monitor known or emerging threats to pregnant women and their babies.

Together, we must continue working to understand birth defects so that we may provide a voice and hope for millions of families all around the world.

### **Margaret A. Honein**

Director, Division of Birth Defects and Infant Disorders National Center on Birth Defects and Developmental Disabilities US Centers for Disease Control and Prevention

## MESSAGE FROM THE WORLD HEALTH ORGANIZATION SOUTH EAST ASIAN REGIONAL OFFICE



Greetings to all the delegates of the conference and the experts from various parts of the world!

Birth defects or congenital anomalies contribute to increasingly higher proportion of fetal loss and child deaths as the infectious cause of child deaths are decreasing in the countries of South-East Asia Region. In addition, birth defects are associated with significant long-term morbidity and disability. Long-term care for children affected with

birth defects puts additional burden on the already over-stretched health system. Potential stigma and discrimination associated with birth defects leads to high social cost on the families.

Member States adopted a Resolution on birth defects in the World Health Assembly (2010) that has requested the WHO to assist countries in setting up surveillance mechanisms for determining the burden of birth defects and implement strategies for prevention and better care of children affected with birth defects.

In response, WHO Regional Office of South-East Asia (WHO-SEARO) initiated a holistic initiative on birth defects in collaboration with CDC-USA in 2011-2012. We have supported countries in the Region to set up hospital-based birth defects surveillance systems that is showing good progress and is providing good quality data. Around three million births have been reported to the SEAR-NBBD database since July 2014. We also prepared a Regional Strategic Framework for prevention and control of birth defects that has guided the countries to develop national plans for birth defects. By now nine of eleven countries of the Region have national plans and Maldives has recently renewed their national plan for the next five years. Regional communication strategy on birth defects has also been developed for adaptation and use in the countries to undertake advocacy, social mobilization and behaviour change

WHO-SEARO has recommended that the countries integrate interventions for prevention of birth defects in the existing national programmes for reproductive, maternal, child and adolescent health, nutrition and immunization. We have strongly advocated for fortification of staple food with folic acid and vitamin B-12 to prevent occurrence of neural tube defects in addition to supplementation approaches. WHO-SEARO is committed to provide technical support to Member States of South-East Asia Region in advancing strategies for prevention, management and rehabilitation of babies born with birth defects for better survival and improved quality of survival.

With best wishes for successful deliberations in the conference.

### Neena Raina

Senior Advisor Reproductive, Maternal, Newborn, Child & Adolescent health and Ageing World Health Organization Regional Office for South-East Asia

## **PROGRAMME AT A GLANCE**

DAY 1: Sunday	DAY 2: Monday	DAY 3: Tuesday	DAY 4: Wednesday
Feb 23, 2020	Feb 24, 2020	Feb 25, 2020	Fe 26, 2020
08:00 AM - 01:00 PM	08:00 AM - 5:00 PM	08:00 AM - 05:00 PM	08.00 AM - 10:00 AM
REGISTRATION	REGISTRATION	REGISTRATION	REGISTRATION
09:00 AM - 03:00 PM	08:30 AM - 10:00 AM	08:30 AM - 10:00 AM	08.30 AM - 10.00AM
WORKSHOPS	PLENARY 1 [Venue: Hall – A]	PLENARY 5 [Venue: Hall – A]	SIMULTANEOUS SYMPOSIA [13-15]
WORKSHOP 1	PARTNERSHIPS AND PROGRAMS FOR	UNADDRESSED CHALLENGES OF BIRTH	SYMPOSIUM 13
	IMPROVING BIRTH DEFECTS	DEFECTS	[Venue: Hall – A]
USING EPIDEMIOLOGIC	SURVEILLANCE		EXPERIENCES
SURVEILLANCE DATA			SYMPOSIUM 14
PREVENTION, CARE			[Venue: Hall – B]
AND POLICY			PRECONCEPTION CARE: ENGAGING CIVIL SOCIETY
WORKSHOP 2			CVMDOCHUM 1 F
DEVELOPMENT OF			[Venue: Hall – C]
EDUCATION			REDUCING RISK FACTORS FOR ADVERSE
			BIRTH OUTCOMES
	10.00 AM - 11.00 AM	10.00 AM - 11.00 AM	10:00 AM- 10:30 AM
	PLENARY 2	PLENARY 6	Poster Sessions 9 & 10
	[Venue: Hall – A]	[Venue: Hall – A]	Networking and Tea
	PREVENTION BEFORE PREGNANCY	ADDRESSING BIRTH DEFECTS FROM	10.00 AM - 12.00 NOON
		THE LENSE OF HUMAN RIGHTS & INCLUSION	PLENARY 7 [Venue: Hall – A]
	11:00 AM - 11:30 AM	11:00 AM - 11:30 AM	THE VOICE OF THE COMMUNITY &
	Poster Sessions 1 & 2	Poster Sessions 5 & 6	PARENT ORGANIZATIONS

DAY 1: Sunday Feb 23, 2020	DAY 2: Monday Feb 24, 2020	DAY 3: Tuesday Feb 25, 2020	DAY 4: Wednesday Fe 26, 2020
	11:30 AM - 01:00 PM SIMULTANEOUS SYMPOSIA [1-3]	11:30 AM - 01:00 PM SIMULTANEOUS SYMPOSIA [7-9]	12.30 PM - 01.00 PM CLOSING CEREMONY
	SYMPOSIUM 1 [Venue: Hall - A]	SYMPOSIUM 7 [Venue: Hall - A]	01:00 PM - 02:00 PM Lunch and Departure
	FOLIC ACID FORTIFICATION FOR IMMEDIATE PREVENTION OF GLOBAL EPIDEMICS OF NEURAL TUBE DEFECTS: GLOBAL EXPERIENCE AND NEXT STEPS IN ASIA	STRENGTHENING BIRTH DEFECTS SURVEILLANCE FOR SAVING AND IMPROVING LIVES.	VISIT TO 'AYATI' - THE NATIONAL CENTER FOR CHILDREN WITH DISSABILITIES, UNIVERSITY OF KELANIYA
	SYMPOSIUM 2 [Venue: Hall - B]	SYMPOSIUM 8 [Venue: Hall - B]	
	LIFE COURSE APPROACHES AND PROGRAMS FOR IMPROVING THE HEALTH OF WOMEN AND GIRLS BEFORE, DURING AND AFTER PREGNANCY	DIAGNOSTIC STRATEGIES: GENETIC TESTING, NEWBORN SCREENING, PATHOLOGICAL EVALUATIONS	
	SYMPOSIUM 3 [Venue: Hall - C]	SYMPOSIUM 9 [Venue: Hall - C]	
	COUNTING BIRTH DEFECTS: INNOVATIONS IN SURVEILLANCE	NETWORKING AND PARTNERSHIPS FOR BIRTH DEFECTS RELATED RESEARCH, PREVENTION AND CARE	
	01:00 PM – 02:00 PM Lunch and Networking	01:00 PM – 02:00 PM Lunch and Networking	
	02:00 PM – 3:00 PM PLENARY 3 [Venue: Hall - A]	02:00 PM – 3:00 PM PLENARY 7 [Venue: Hall - A]	
	LINKING BIRTH DEFECTS TO SUSTAINABLE DEVELOPMENT GOALS (2030)	PREMATURE BIRTH	

DAY 1: Sunday	DAY 2: Monday	DAY 3: Tuesday	DAY 4: Wednesday
Feb 23, 2020	Feb 24, 2020	Feb 25, 2020	Fe 26, 2020
	03:00 PM - 4.00 PM	03:00 PM – 4.00 PM	
	PLENARY 4	PLENARY 8	
	[Venue: Hall - A]	[Venue: Hall - A]	
	DEDUCING EVDOCUDE TO HADMEU	DICITAL HEALTH CTANDADDC AND	
	REDUCING EXPUSURE TO HARMFUL SUDSTANCES AND INFECTIONS FOR	DIGITAL HEALTH STANDARDS AND	
	IMPROVING RIPTH OUTCOMES	RIDTH OUTCOMES	
	IMI KOVING BIKTII OOTCOMES	DIRTH OUTCOMES	
	04:00 PM - 04:30 PM	04:00 PM - 04:30 PM	
	Poster Sessions 3 & 4	Poster Sessions 7 & 8	
	Networking and Tea	Networking and Tea	
	04:30 PM - 06:00 PM	04:30 PM - 06:00 PM	
	SIMULTANEOUS SYMPOSIA [4-6]	SIMULTANEOUS SYMPOSIA [10-12]	
	SYMPUSIUM 4 [Vonue: Hell A]	SYMPOSIUM 10 [Vorue: Holl A]	
	[venue: nan - A]	[venue: han - Aj	
	EVIDENCE GAPS AND NEW KNOWLEDGE TO	COUNTING BIRTH DEFECTS: HOSPITAL	
	SUPPORT NTD PREVENTION ACTIVITIES IN	AND POPULATION BASED SURVEILLANCE	
	LMICS		
	SYMPOSIUM 5	SYMPOSIUM 11	
	[Venue: Hall - B]	[Venue: Hall - B]	
	PEODIENTING HEALTHCADE CYCTEM FOD		
	REURIENTING HEALTHCARE SYSTEM FOR	EPIDEMIOLOGIC APPROACHES IN THE	
	AND NEWBORN HEALTH SERVICES	DEFECTS	
	SYMPOSIUM 6	SYMPOSIUM 12	
	[Venue: Hall - C]	[Venue: Hall - C]	
	OUALITY OF LIFE STRATECIES TO DEDUCE	FOOD FORTIFICATION FOD DDEVENTION	
	MORTALITY AND IMPROVE PHYSICAL AND	OF BIRTH DEFECTS	
	DEVELOPMENTAL OUTCOMES OF		
	CHILDREN WITH BIRTH DEFECTS		
6:00 PM onwards		7.30 PM Onwards	
<b>OPENING CEREMONY</b>		CONFERENCE DINNER	

**PROGRAMME IN DETAIL** 

## **23 FEBRUARY 2020: PRE-CONFERENCE WORKSHOPS**

## **PRE-CONFERENCE WORKSHOP 1**

## Using Epidemiologic Surveillance Data for Birth Defects Prevention, Care and Policy

Organised in collaboration with US Centers for Disease Control and Prevention, March of Dimes, World Health Organization, and International Centre for Birth Defects Surveillance and Research

Purpose	To understand and critically assess quality of birth defects surveillance data and its use to impact prevention, care, and policy		
Objectives	<ul> <li>Understand how birth defects surveillance can be an important tool for driving public health action</li> <li>Improve surveillance data collection and data quality</li> <li>Understand methods for data aggregation and analysis</li> <li>Prepare data for presentations, reports, and high-quality publications</li> </ul>		
Facilitators	CDC: Cindy Moore and Cara Mai ICBDSR: Boris Groisman WHO: Ornella Lincetto, Nathalie Roos, Rajesh Mehta and Pablo Duran		
09:00 AM	Welcome & Introductions		
09:30 AM	<b>Birth Defects Surveillance: A Tool for Driving Public Health Action</b> <i>Overview from data collection to data utilization</i>		
10:15 AM	Quality Control and Quality Improvement Methods in Birth Defects Surveillance		
	Part 1: Critical processes, e.g., improving description, coding and classification		
11:00 AM	Tea Break		
11:15 AM	Quality Control and Quality Improvement Methods in Birth Defects Surveillance		
	Part 2: Tools for the job (if time permits, an exercise on quality improvement tools)		

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12:30 PM **Lunch** 

## 01:15 PM **Data Interpretation, Reporting, and Utilization for Prevention, Care** and Policy Change

## Part 1: Interpret, prepare and present data

How to ensure your data are of high enough quality to use them to drive decision making

- Publish your data in peer-reviewed journals
- Types of data in the published literature
- Inform and drive prevention strategies
- Improve care

Part 2: Tools for the job

- 2:45 PM **Summary of Key Concepts**
- 3:00 PM Conclude Workshop

## **PRE-CONFERENCE WORKSHOP 2**

## **Development of Targeted Genomic Education**

Organised in collaboration with the Global Consortium for Genomic Education (GC4GE) A multi-disciplinary workshop on genetic and genomic education and training for application and translation of genomic advances and new developments in clinical medicine and healthcare.

Co-Chaired by Dhavendra Kumar & Vajira H. W. Dissanayake

## 09:00 AM Welcome/Introduction

09:15 AM Session 1: Status of Genetic and Genomic Education & Training

- Global overview Dhavendra Kumar
- Current strategy for genetic/genomic education in India
   Madhulika Kabra
- Experience and achievements of genetic/genomic education in Sri Lanka - Vajira H. W. Dissanayake
- Genetic/genomic education & training in Philippines - Carmencita Padilla
- Genetic/ genomic education & training in the Latin America - Paula Hurtado

## 10:45 AM **Tea Break**

## 11:15 AM Session 2: Targeted Genetic/Genomic Education & Training

All participants will split into three teams led by one of the Faculty. Each team will find out the current status and draw out conclusions. The Faculty lead will then present to the whole workshop for open discussion. Team A - Genetic and genomic education for 'Doctor of the Future' Team B - Genetic/genomic core knowledge & skills for 'Specialist Doctors' Team C - Genetic/ genomic education and training for career grade 'clinical geneticists, genomic physician, genetic counsellors and specialist genetic nurses'

12:45 PM **Lunch** 

- 1:30 PM Session 3: Structure/ Format of Genomic Education & Training Reports by Teams Discussion & Recommendations
- 3:00PM Close

## 23 FEBRUARY 2020: INAUGURATION

5.45 pm Guests Take Their Seats

6.00 pm Arrival of the Chief Guest

6.05 pm Introduction of Representatives of Partner Organizations to the Chief Guest

6.15 pm Ceremonial Procession

6.20 pm National Anthem

6.25 pm Lighting of the Oil Lamp

**6.30 pm Welcome Address** Dr. Kapila Jayaratne, Conference Chair

**6.35 pm Welcome Remarks by Co-chairs** Dr. Salimah Walani, Conference Co-Chair Dr. Anula Wijesundere, Conference Co-Chair

6.45 pm Welcome Remarks by March of Dimes

6.50 pm Welcome Remarks by the U.S. Centers for Disease Control and Prevention

6.55 pm Address by the World Health Organization South East Asia Regional Office

7.00 pm Welcome Remarks by the President, SLMA

## 7.05 pm

Address by the Chief Guest

Honorable Mrs. Pavithra Wanniarachchi, Minister of Health & Indigenous Medical Services, Women & Child Affairs & Social Security

7.15 pm

Presentation of Mementos

7.30 pm

## **Keynote Address**

"The Continuing Need for Services for People with Congenital Disorders in Low- & Middle-Income Nations: A View from the Past for the Future" Prof. Arnold Christianson

**8.15 pm Vote of Thanks** Prof. *Vidya Jyothi* Vajira H. W. Dissanayake

8.25 pm The Procession Leaves the Hall

8.30 pm Cocktail Reception

## **KEYNOTE ADDRESS**

The Continuing Need for Services for People with Congenital Disorders in Low- & Middle-Income Nations: A View from the Past for the Future



**Prof. Arnold Christianson** University of the Witwatersrand, Centre for Ethics (WiCE), Johannesburg. South Africa

In 1958, the March of Dimes (MoD) became the first organization, national or international, to confront the challenge of what it termed birth defects. Professor Victor McKusick called for the establishment of a specialty of clinical genetics in 1959 and in 1961 the WHO initiated the first international study of the epidemiology of congenital disorders (CDs). Following Bernadette Modell & Anver Kuliev, the WHO in 1985 recognized the need for community-based services for people with CDs in developing countries. Translating this need into action waited until the 1997 Cape Town Declaration calling for an international conference on the topic, and for the WHO to become proactive, on the issue. Two WHO expert meetings occurred in 1999 & the 1st International Conference on Birth Defects & Disabilities in the Developing World was held in 2001. The WHO meetings delineated issues central to developing services, including confirming services for care and prevention should be in primary health care linked to secondary & tertiary care; acquiring epidemiology for development of services through health needs assessment; and resolving issues of definitions to assist the former. The WHO unfortunately withdrew support for these efforts in 2002 to promote 'Genomics & World Health'. Organizations, including MoD, CDC & International Genetic Alliance, and individuals, including local champions, continued to promote the development of services for people with congenital disorders (CDs), and how to integrate these into existing health services. In 2006 the MoD published its Global Report on Birth Defects, including the Modell Database of global CDs epidemiology. Prompting immediate concern & action from WHO, a 2007 MoD/WHO expert report confirmed the acceptability of the modelled Modell Database epidemiology and addressed issues regarding definitions. Eventually, in 2010, the World Health Assembly resolution WHA 63.10 recognized the need for services for people with congenital disorders. That need, in my opinion, has still to be equitably fulfilled in most low- & middle-income countries (LMICs). This paper will discuss the characters, events and issues, including controversies, involved in the development & integration of appropriate care & prevention services for people with congenital disorders in LMICS, and how they relate to today.

## 24 FEBRUARY 2020: CONFERENCE DAY 1

#### 08:30 AM – 10:00 AM: PLENARY 1 - PARTNERSHIPS AND PROGRAMS FOR IMPROVING BIRTH DEFECTS SURVEILLANCE [Venue: Hall - A]

Chairpersons: Anula Wijesundere and Salimah Walani

**Surveillance for Emerging Threats to Pregnant Women and Infants** Peggy Honein, Centers for Disease Control & Prevention, USA

**Regional Surveillance Programs and Burden of Birth Defects in South East Asia** Neena Raina, South East Asian Regional Office, World Health Organization, India

**Expanded Objectives of Birth Defects Surveillance in Developing Countries** Boris Groisman, International Centre for Birth Defects Surveillance and Research, Argentina

**Births Defects Surveillance Efforts in Sri Lanka** Kapila Jayaratne, Ministry of Health, Sri Lanka

### 10.00 AM – 11.00 AM: PLENARY 2 - PREVENTION BEFORE PREGNANCY [Venue: Hall - A]

Chairpersons: Vasantha Devasiri and Neena Raina

**A Life Course Approach to Preconception Health and Pregnancy Planning** Gita Mishra, University of Brisbane, Australia

**Primary Prevention of Birth Defects** Cindy Moore, Centers for Disease Control & Prevention, USA

### 11.00 AM - 11.30 AM: POSTER SESSION 1 & 2, NETWORKING, TEA

#### 11:30 AM – 01:00 PM: SIMULTANEOUS SYMPOSIA [1-3] [Venue: Hall - A]

### SYMPOSIUM 1 - FOLIC ACID FORTIFICATION FOR IMMEDIATE PREVENTION OF GLOBAL EPIDEMICS OF NEURAL TUBE DEFECTS: GLOBAL EXPERIENCE AND NEXT STEPS IN ASIA [Venue: Hall - A]

Organised by the Center for Spina Bifida Prevention & Food Fortification Initiative, Emory University, USA

Chairpersons: Sussie Perera and Rajesh Mehta

## Mandatory Cereal Grain Fortification: Global Status, Best Practices, and Opportunities

Scott Montgomery, Food Fortification Initiative, USA

## 2019 Global Status of Prevention of Folic Acid Preventable Spina Bifida and Anencephaly

Vijaya Kancherla, Center for Spina Bifida Prevention Emory University, USA

Preventing Neural Tube Defects in Haryana, India: A Project to Implement Wheat Flour Fortification in the Public Distribution System and Safety Net Programs Sonia Trikha, State Health Systems Resource Centre Haryana State, India

#### NTD Care and Advocacy, its Role in Fortification

Lieven Bauwens, International Federation for Spina Bifida and Hydrocephalus, Belgium

#### Sri Lanka: Identifying Opportunities and the Path Towards Mandatory Fortification

Renuka Jayatissa, Medical Research Institute, Ministry of Health, Sri Lanka

### SYMPOSIUM 2 - LIFE COURSE APPROACHES AND PROGRAMS FOR IMPROVING THE HEALTH OF WOMEN AND GIRLS BEFORE, DURING AND AFTER PREGNANCY [Venue: Hall - B]

Chairpersons: Iyanthi Abeywickrama and Gita Mishra

#### **OP-01: Workplace Wellness across the Lifespan: A Pilot Program for Health Promotion in a Middle-Income Country**

Khalid Yunis, American University of Beirut Medical Center, Lebanon

### **OP-02: Workplace Wellness Modules across the Lifespan: Utilizing e-Learning** Access to Knowledge

Carmencita Padilla, University of the Philippines Manilla, Philippines

**OP-03: Readiness for Implementation of Preconception Care in Uganda** Nathan Isabirye, Makerere University School of Public Health, Uganda

#### **OP-04: Preconception Health Care Services Delivered in Selected Medical Officer** Areas of Kandy District Sri Lanka Suranga Ubesekara, General Hospital, Matara, Sri Lanka

#### **OP-05: Impact of Antenatal Care on Stillbirth at a Tertiary Care Referral Hospital of** India.

Nishi Choudhary, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi. India

### SYMPOSIUM 3 - COUNTING BIRTH DEFECTS: INNOVATIONS IN SURVEILLANCE [Venue: Hall - C]

Chairpersons: Duminda Samarasinghe and Cara Mai

### **OP-06: Global Birth Defect Inventory of Resources for Surveillance and Research** Helen Dolk, Ulster University, United Kingdom

#### **OP-07: A Reflection of Birth Defect Surveillance in Bangladesh: Challenges and Opportunities**

Shahidullah M.1\*, Mannan M.A.1\*, Dey S.K.1, Moni S.C.1, Shabuj K.H.1, Jahan I.1

#### **OP-08: Integrating Birth Defect Surveillance Midwives into Existing Labor Ward** Structures: A Comparison between Government and Private-Not-For-Profit Hospitals in Kampala, Uganda.

Joyce Namale Matovu, Makerere University - Johns Hopkins University Research Collaboration, Kampala, Uganda

**OP-09: Pattern of Congenital Anomalies Among Women Opting for Elective Termination of Pregnancy for Fetal Anomalies (ETOPFA)** Bharti Sharma, Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India

**OP-10: Comparison of Modelled and Observed Data for Isolated Oral Facial Clefts in South Africa** 

Helen Malherbe, University of KwaZulu Natal, South Africa

**OP-11: Overcoming the Defect: A Perinatal Review of Congenital Anomalies in 6984 Consecutive Deliveries in a Tertiary Center in Western Nepal** Shreyashi Aryal, Lumbini Medical College Teaching Hospital, Nepal

#### 01:00 PM - 02:00 PM: LUNCH

02:00 PM – 03:00 PM: PLENARY 3 - LINKING BIRTH DEFECTS TO SUSTAINABLE DEVELOPMENT GOALS (2030) [Venue: Hall - A]

Chairpersons: Chithramalee de Silva and Cynthia Moore

**An Introduction to the 2030 SDGs in the Context of Child Health and Birth Defects** Nathalie Roos, World Health Organization, Switzerland

Surveillance, Prevention and Care of those with Birth Defects Contribute to Countries Achieving their SDG Child Health Targets Salimah Walani, March of Dimes, USA

**Polling to Understand the Perception of ICBD Conference Attendees about Birth Defects and their Relation To SDGS** Vijava Kancherla, Center for Spina Bifida Prevention Emory University, USA

# 03:00 PM – 4.00 PM: PLENARY 4 - REDUCING EXPOSURE TO HARMFUL SUBSTANCES AND INFECTIONS FOR IMPROVING BIRTH OUTCOMES [Venue: Hall - A]

Chairpersons: Padma Gunarathne and Vijaya Kencherla

**Impact of Opioids and Alcohol on Birth Outcomes** Rahul Gupta, March of Dimes, USA

**Medications During Pregnancy and Birth Defects** Priyadarshani Galappatthy, Department of Pharmacology, University of Colombo, Sri Lanka

**Infectious Diseases and Birth Defects** Pablo Duran, Pan American Health Organization, World Health Organization, USA

#### 04.00 PM - 04.30 PM: POSTER SESSION 3 & 4, NETWORKING, TEA

#### 04:30 PM - 06:00 PM: SIMULTANEOUS SYMPOSIA [4-6]

#### SYMPOSIUM 4 - EVIDENCE GAPS AND NEW KNOWLEDGE TO SUPPORT NTD PREVENTION ACTIVITIES IN LMICS [Venue: Hall - A]

Organized by Nutrition International

Chairpersons: Narada Warnasuriya and Lakmini Magodarathne

#### Introduction: The Folate Task Team Homero Martinez, USA

Folate Status Assessment in Women of Reproductive Age: Capacity Building for Red Blood Cell Microbiological Assay Renuka Jayatissa, Sri Lanka

**Salt as an Alternative Food Vehicle to Provide Folic Acid in Select LMIC** Vijaya Kancherla, USA

New Information for Policy Makers to Support the Adoption of Mandatory Large-Scale Food Fortification Programs Homero Martinez, USA

#### SYMPOSIUM 5 - REORIENTING HEALTHCARE SYSTEM FOR IMPROVING THE QUALITY OF MATERNAL AND NEWBORN HEALTH SERVICES [Venue: Hall - B]

Chairpersons: Sapumal Dhanapala and Khalid Yunis

**OP-12: Training Module for Primary Health Care Worker for Improved Recognition and Follow Up for Persons with a Congenital Disorder** Colleen Aldous University of KwaZulu Natal, South Africa

OP-13: Implementation of Expanded Operational Guidelines on Use of Antenatal Corticosteroids: Perspective of Program Implementers and Health Care Professionals in Northern India

Mona Duggal, Post Graduate Institute of Medical Education and Research, Chandigarh, India

OP-14: The Impact of Different Intervention Packages on Facility Intervention Package on Health Facility Deliveries Utilization and Other Maternal and Newborn Care Practices in Uganda

Nathan Isabirye, Makerere University School of Public Health, Uganda

**OP-15: Best Wishes Program: Use of Mobile Phone Technology (mHealth) for Care Coordination Directed towards Birth Defects Prevention** Ram Thapa, Karuna Foundation, Nepal

**OP-16: The Baby Buddy App: A Digital Health Intervention to Bridge Neonatal Health Inequalities in The United Kingdom.** Nilushka Perera, Best Beginnings, UK

#### SYMPOSIUM 6 - QUALITY OF LIFE STRATEGIES TO REDUCE MORTALITY AND IMPROVE PHYSICAL AND DEVELOPMENTAL OUTCOMES OF CHILDREN WITH BIRTH DEFECTS [Venue: Hall - C]

Chairpersons: Saraji Wijesekara and Nathalie Roos

**OP-17: Outcomes of Infants with Structural Birth Defects in Galle, Sri Lanka** Janithra De Silva, University of Ruhuna, Sri Lanka

**OP-18: Quality of Life of Primary Caregivers having Children with Congenital Heart Diseases Awaiting Cardiac Surgery Attending the Cardiology Clinic at the Lady Ridgeway Hospital for Children, Colombo, Sri Lanka** Pavithra Warnakulasooriya, University of Colombo, Sri Lanka

**OP-19: Thalidomide and Zika Virus: Lessons for Health and Welfare Systems of Low and Middle Income Countries** Anita Kar, Birth Defects and Childhood Disability Research Centre, Pune, India

**OP-20: Birth Defects in South-East Asian Regional Countries; Challenges and Progress in Research, Policies and Programmes: A Scoping Review** Tonima Islam Trisa, icddr,b, Bangladesh
# 25 FEBRUARY 2020: CONFERENCE DAY 2

#### 08:30 AM – 10:00 AM: PLENARY 5 - UNADDRESSED CHALLENGES OF BIRTH DEFECTS [Venue: Hall - A]

Chairpersons: Sanath P Lamabadusuriya and Peggy Honein

**Eliminating Stigma and Discrimination Against Persons with Birth Defects and Disabilities** Leivan Bauwens, International Federation for Spina Bifida and Hydrocephalus, Belgium

**Advocating for National Newborn Screening Programs** Carmencita Padilla, University of the Philippines Manila, Philippines

**Unaddressed Challenges of Birth Defects Prevention & Control in Sri Lanka** Chithramlee de Silva, Ministry of Health, Sri Lanka

**Mobilizing Resources for Care of Children with Congenital Heath Disease in Sri Lanka** Duminda Samarasingha, Lady Ridgeway Hospital for Children, Sri Lanka

#### 10.00 AM – 11.00 AM: PLENARY 6 - ADDRESSING BIRTH DEFECTS FROM THE LENSE OF HUMAN RIGHTS & INCLUSION [Venue: Hall - A]

Organised by United Nations Fund for Population Activities (UNFPA)

**Welcome Address** Ritsu Nacken, Representative, UNFPA Sri Lanka

#### Introduction to the Panelists

Janitha Rukmal, Co-founder, ENABLE Lanka foundation

# Family Planning in the Context of Preventing Birth Defects, Maternal Newborn and Child Health

Athula Kaluarachchi, President, Sri Lanka College of Obstetricians and Gynecologists

#### **SRHR of People Living with Disability – The Sociological and the Rights Aspect** Subhanghi Herath, Department of Sociology, University of Colombo, Sri Lanka

**Bringing Disability to the Fore Front in the Era of SDGS** Shyamani Hettiarachchi, Department of Disability Studies, University of Kelaniya, Sri Lanka

#### Conclusions

Madusha Dissanayake, Assistant Representative, UNFPA Sri Lanka

#### 11.00 AM - 11.30 AM: POSTER SESSION 5 & 6, NETWORKING, TEA

#### 11:30 AM - 01:00 PM: SIMULTANEOUS SYMPOSIA [7-9]

#### SYMPOSIUM 7 - STRENGTHENING BIRTH DEFECTS SURVEILLANCE FOR SAVING AND IMPROVING LIVES. [Venue: Hall - A]

Organised by the World Health Organization, Geneva

Chairpersons: Anuruddha Padeniya and Pablo Duran

**Strengthening Services for Managing Birth Defects** Ornella Lincetto, World Health Organization, Switzerland

**Current Status of Global Birth Defects Surveillance: Results from 2018-2019 WHO Birth Defects Surveillance Systems Mapping Project** Vijaya Kancherla, Center for Spina Bifida Prevention Emory University, USA

**Birth Defects Surveillance - Experiences from the Recent Zika Outbreak** Cara Mai, Centers for Disease Control and Prevention, USA

**Introduction to the Updated WHO Birth Defects Atlas – What is New?** Nathalie Roos, World Health Organization, Switzerland

Country Perspective from Uganda: Parents Voices: Children Living with a Birth Defect

Sylivia Kyomuhendo and Okello Harrison, Infants' Health Foundation, Uganda

**Moderated Discussion with Questions and Answers** Rajesh Mehta, South East Asia Regional Office, World Health Organization, India

#### SYMPOSIUM 8 - DIAGNOSTIC STRATEGIES: GENETIC TESTING, NEWBORN SCREENING, PATHOLOGICAL EVALUATIONS [Venue: Hall - B]

Chairpersons: Nirmala Sirisena and Carmencita Padilla

**OP-21: Molecular Signature of Preterm Premature Rupture of Fetal Membrane** Jing Pan, SMU-NYSIBR Joint Translational Research Center for Developmental Disabilities, Guangzhou, China

**OP-22: The Saudi Human Genome Program (SHGP) and Precision Medicine in Saudi Arabia** 

Aida I Al Aqeel, Prince Sultan Military Medical City, Saudi Arabia

#### **OP-23: Genetic Service for Birth Defects at the Dutch Caribbean Islands** Eline Verberne, University of Amsterdam, Netherlands

**OP-24: Minimally Invasive Autopsy in the Evaluation of Fetal Malformations** Nitika Langeh, All India Institute of Medical Sciences, New Delhi, India

#### SYMPOSIUM 9 - NETWORKING AND PARTNERSHIPS FOR BIRTH DEFECTS RELATED RESEARCH, PREVENTION AND CARE [Venue: Hall - C]

Chairpersons: Ruwan Ferdinando and Neelam Aggarwal

# **OP-25: Creating and Strengthening Global Partnerships to End Disability from Clubfoot**

Alaric Aroojis, Miracle Feet, Chapel Hill, NC, USA

**OP-26: Combining the Strength of Government and NGOs, Providing Comprehensive Care for the Infants: An Encouraging Experience from China** Xinliang Zhao, Peking University, Beijing, China

**OP-27: Thailand National Plan for Prevention and Care of Birth Defects & Disabilities** 

Pornswan Wasant, Mahidol University, Bangkok, Thailand

**OP-28: The Neonate Fund: Delivering Hope through Facilitating Quality NICU Care** Pascale Nakad, American University of Beirut, Beirut, Lebanon

OP-29: Knowledge on Advanced Maternal Age, Offspring Outcomes and Attitudes Towards Advanced Maternal Age among Pregnant Mothers in the Colombo Municipal Area.

Sashiprabha Nawaratne, Ministry of Health, Sri Lanka

#### 01:00 PM - 02:00 PM: LUNCH

02:00 PM – 03:00 PM: PLENARY 7 - PREMATURE BIRTH [Venue: Hall - A]

Chairpersons: Kapila Jayaratne and Ornella Lincetto

Burden of Preterm Birth in LMICs

Razia Pendse, World Health Organization, Sri Lanka

Scientific Advances in Prevention of Preterm Births Kelle Moley, March of Dimes, USA

Advances in Care of a Baby Born Too Soon or Too Sick Ornella Lincetto, World Health Organization, Switzerland

#### 03:00 PM - 04.00 PM: PLENARY 8 - DIGITAL HEALTH STANDARDS AND INTERVENTIONS FOR IMPROVING BIRTH OUTCOMES [Venue: Hall - A]

Chairpersons: Vajira H. W. Dissanayake and Rahul Gupta

**Birth Defects and Rare Disorders Terminology** Matthew Darlison, WHO Collaborating Centre for Community Genetics, University College London, UK

**A Semantic Information Standard for Global Child Health** Liesbeth Siderius, Paediatrician, GGD IJsselland, Netherlands Development of the Global Birth Defects App for Birth Defect Description and Coding. International Committee for Congenital Anomaly Surveillance Tools EUROCAT, ECLAMC, ICBD, CDC, WHO-SEARO, WHO-TDR, MUHJU Aminkeng Leke, Ulster University, UK

04.00 PM - 04.30 PM: POSTER SESSION 7 & 8, NETWORKING, TEA

#### 04:30 PM - 06:00 PM: SIMULTANEOUS SYMPOSIA [10-12]

#### SYMPOSIUM 10 - COUNTING BIRTH DEFECTS: HOSPITAL AND POPULATION BASED **SURVEILLANCE** [Venue: Hall - A]

Chairpersons: Kaushalya Kasthuriarachchi and Boris Groisman

#### OP-30: Study of Birth Defects in Government Referral Hospital, Maldives 2016-2018

Ahmed Faisal, Indira Gandhi Memorial Hospital, Male, Maldives

**OP-31: A Descriptive Study on Feto-Infant Structural Anomalies in Maternal** Diabetes Mellitus: An Autopsy Study at a Tertiary Care Setting. S. P. R. V. Lakmini, University of Colombo, Sri Lanka

#### OP-32: Analysis of Stillbirth in a Tertiary Care Hospital of Delhi: A Contribution to **NNBD** Project

Abha Singh, Lady Hardinge Medical College, New Delhi, India

#### **OP-33:** Incidence, disease spectrum, and genetic defects in congenital heart disease in China: Prenatal ultrasound screening identified 18,171 affected fetuses from 2,452,249 pregnancies

Nanbert Zhong, New York State Institute for Basic Research in Developmental Disabilities, New York, USA

## **OP-34: Birth defects in Congenital Rubella Syndrome - A Hospital Based Surveillance at Tertiary Care Center**

Kuldeep Singh, All India Institute of Medical Science, Jodhpur, India

#### **OP-35: Neural Tube Defects in China, 2007-2017: Data from the National Population-based Birth Defects Surveillance** Changfei Deng, Sichuan University, Chengdu, China

#### SYMPOSIUM 11 - EPIDEMIOLOGIC APPROACHES IN THE STUDY OF PRETERM BIRTH AND **BIRTH DEFECTS** [Venue: Hall - B]

Chairpersons: Rohini Seneviratne and Madhulika Kabra

#### **OP-36: Global, Regional, and National Levels and Trends in Congenital Birth** Defects, 1990-2019 Lauren B. Wilner, University of Washington, Seattle, USA

#### **OP-37: Bronchopulmonary Dysplasia, Other Comorbidities, and Mortality Rates** Associated with Extremely Premature Births in a Large Medical Records Database in the United States

Csaba Siffel, Shire- a Takeda Company, Lexington, MA, USA

**OP-38: Epidemiological Profiling from Scientific First Principles - Progress and Prospects of the Modell Global Database of Congenital Disorders** Matthew W Darlison, University College London, London, UK

#### OP-39: Prevalence and Mortality among Infants with Congenital Diaphragmatic Hernia: A Multi-Registry Analysis

Wendy N. Nembhard, University of Arkansas for Medical Sciences, Little Rock, Arkansas, USA

## OP-40: Population Medical Genetics approach in clusters of Mucopolysaccharidoses identified in Latin America

Francyne Kubaski, Hospital del Niño de Panamá, Panamá, Panamá

# **OP-41: Spina Bifida Phenotypes and Sex-Specific Prevalence among Offspring of Folic Acid Users**

Cynthia Moore, US Centers for Disease Control and Prevention, Atlanta, GA, USA

# SYMPOSIUM 12 - FOOD FORTIFICATION FOR PREVENTION OF BIRTH DEFECTS [Venue: Hall - C]

Chairpersons: Renuka Jayatissa and Homero Martinez

#### **OP-42: High Potential for Reducing Folic Acid-Preventable Spina Bifida and Anencephaly, and Related Stillbirth and Child Mortality, in Ethiopia** Vijaya Kancherla, Emory University Rollins School of Public Health, Atlanta, Georgia, USA

#### OP-43: Folate and Vitamin B12 status in Women of Reproductive Age in Rural Areas in Haryana, Northern India

Reena Das, Postgraduate Institute of Medical Education and Research, Chandigarh, India

#### OP-44: Awareness and Factors Associated with Reported Intake of Folic Acid-Fortified Flour Among Women of Reproductive Age in Ifakara, Morogoro Region, Tanzania: A Cross-Sectional Study

Ipyana Frank Mwandelile, Muhimbili University of Health and Allied Sciences, Dar essalaam, Tanzania

#### **OP-45: Potential Level of Global Prevention of Neural Tube Defects with Folic Acid-Fortified Salt**

Vijaya Kancherla, Emory University Rollins School of Public Health, Atlanta, Georgia, USA

#### 7.30 PM Onwards - CONFERENCE DINNER & BEACH PARTY [Mount Lavenia Hotel]

## 26 FEBRUARY 2020: CONFERENCE DAY 3

#### 08.30 AM - 10.00AM: SIMULTANEOUS SYMPOSIA [13-15]

SYMPOSIUM 13 - SHARING UNIQUE SRI LANKAN EXPERIENCES [Venue: Hall - A]

Chairpersons: Chithramalee de Silva and Indika Karunathilake

## Caring for a Newborn with Birth Defects: Approach to Evaluation & Management in Sri Lanka

Surantha Perera, Castle Street Hospital for Women, Sri Lanka

**Caring for Children in Conflict Situations - Sri Lanka's Experience** Sujewa Amarasena, University of Ruhuna, Sri Lanka

The Electronic Reproductive Health Information Management System (eRHMIS) -The Country Wide Digital System for Monitoring Maternal and Child Health Services in Sri Lanka Kaushalya Kasturiarachchi, Ministry of Health, Sri Lanka

**Genetic Testing for Birth Defects - the Sri Lankan Experience** Vajira H. W. Dissanayake, University of Colombo, Sri Lanka

#### SYMPOSIUM 14 - PRECONCEPTION CARE: ENGAGING CIVIL SOCIETY [Venue: Hall - B]

Organised by Preparing for Life

Chairpersons: Saveen Gamage and Mona Elnaka

**The Role of Civil Society in Preconception Education** Symone Detmar, Preparing for life, Netherlands

**The Role of Zonta in Engaging Civil Society** Dineshani Hettiarachchi, Zonta, Sri Lanka

**Youth Engagement in Preconception Care** Carmencita Padilla, University of Philippines Manila, Philippines

**A School Based Program in the Netherlands** Symone Detmar, Preparing for life, Netherlands

**Lessons from Voluntarism** Rufus Adducul, Philippines

#### SYMPOSIUM 15 - REDUCING RISK FACTORS FOR ADVERSE BIRTH OUTCOMES [Venue: Hall - C]

Chairpersons: Ananda Lamahewage and Helen Malhrebe

**OP-46: Changing Spectrum and Risk Factor Association in Newborn Babies with Birth Defects in a Tertiary Care Hospital in North India** Shobhna Gupta, V.M.M.C. and Safdarjung Hospital, New Delhi, India

**OP-47: Pattern and Risk Factor Associated with Congenital Anomalies in Newborn Babies Born in Dhulikhel Hospital** Srijana Dongol Singh, Dhulikhel Hospital, Dhulikhel, Nepal

## **OP-48: Specific Birth Defects in Pregnancies of Women with Diabetes - National Birth Defects Prevention Study, 1997-2011** Cindy Moore, US Centers for Disease Control and Prevention, Atlanta, GA, USA

#### OP-49: Prevalence, Pattern and the Association of Major Structural Congenital Anomalies with Maternal and Fetal Factors at the Neonatal Care Unit (NICU/SCBU) of a Tertiary Hospital in Colombo, Sri Lanka Hasani Hewavitharana, Castle Street Hospital for Women, Colombo, Sri Lanka

Hasam Hewavitharana, Castle Street Hospital for Women, Colombo, Sri Lanka

**OP-50: Pattern of Congenital Malformations and Associated Risk Factors among Neonates Admitted at Kenyatta National Hospital, Kenya** Everlyn Machogu, University of Nairobi, Kenya

#### 10.00 AM - 10.30 AM: POSTER SESSION 9 & 10, NETWORKING, TEA

# 10.30 AM – 12.30 PM: PLENARY 8 - THE VOICE OF THE COMMUNITY & PARENT ORGANIZATIONS [Venue: Hall - A]

Chairpersons: Kapila Jayaratne, Saliman Walani, Vajira H. W. Dissanayake

**Parent Perceptions, Fear, Support and Legal Protection on Children with Disability** Lasanthi Daskon, International Foundation for Electoral Systems (IFES), Sri Lanka

**Stories of Stigma and Isolation Experienced by Persons with Birth Defects in India** Anita Kar, Birth Defects and Childhood Disability Research Centre, Pune, India

**Giving Patients and Parents of Premature and Sick Babies in Low Resource Settings a Voice** Selina Bentoom, African Foundation for Premature Babies and Neonatal Care (AFPNC), Ghana

# Giving Women and Children a Voice for Improving their Birth Outcomes Through "Our Dream Initiative" In Egypt

Mona Elnaka, Egyptian Members Association of Royal College of Pediatrics and Child Health - 'EMARCPCH', Egypt

#### **Food and Birth Defects**

Anuruddha Padeniya, Lady Ridgeway Hospital for Children, Colombo, Sri Lanka

**Multidisciplinary Team Involvement in Children with Disabilities in Sri Lanka** Samanmali Sumanasena, University of Kelaniya, Sri Lanka

#### 12:30 PM - 1.00 PM

**CLOSING CEREMONY** 

12:30 PM - 02:00 PM

# LUNCH AND DEPARTURE FOR THE VISIT TO 'AYATI' - THE NATIONAL CENTER FOR CHILDREN WITH DISSABILITIES, UNIVERSITY OF KELANIYA

# **INVITED SPEAKER PROFILES**



**Rufus Adducul** obtained his degree in B.S. Biology in Genetics from the University of the Philippine in Los Banos in 2013. His interest in human genetics started when he became an active member of the Volunteer Youth Leaders for Health – Philippines (VYLH-Philippines) and attended several activities that involve interaction with patients diagnosed with genetic disorders. He was recently elected as the National President of VYLH-Philippines, a network of youth leaders in the forefront of the campaign directed to increase the public's awareness on the significance of folic acid supplementation in the prevention of birth defects, and on the importance of newborn screening in saving babies from death and mental retardation. The network also advocates for the passage of the Rare Disease Act, an act addressing the needs of patients with rare and orphan disorders.



**Sujeewa Amarasena** MBBS, MD, DCH (Colombo), DCH (Sydney) is the Vice Chancellor and Senior Professor in Paediatrics at the University of Ruhuna in Sri Lanka. He has special interest in newborn screening, birth defects surveillance, and child protection and has many publications in those fields for which he has won national awards. Prior to assuming duties as Vice Chancellor, he was Dean of the Faculty of Medicine of the General Sir John Kotelawala Defense University and Dean of the Faculty of Allied Health Sciences of the University of Ruhuna.



**Lieven Bauwens** is president of the International Federation for Spina Bifida and Hydrocephalus (IF), and the chair of Child-Help Belgium and serves as director of Child-Help International. Child-Help is a group of charities dedicated to helping children with Spina Bifida and Hydrocephalus in developing countries. He is a global advocate for the rights of persons with the disabilities while engaging actively in primary prevention of these impairments. He is a board member of the Food Fortification Initiative, the International Disability Alliance, Rare Diseases Europe (EURORDIS) and a founding partner of the global alliance for Spina Bifida and Hydrocephalus, PUSH!. A background in Architecture and Business Administration, Lieven got involved in IF as brother of a young man with Spina Bifida and Hydrocephalus.



**Selina Bentoom** is a mother of a surviving preterm child born in Africa. She is an international speaker, advocate for Maternal and Newborn Health, and the Founder/Executive Director of the African Foundation for Premature Babies and Neonatal Care (AFPNC). She is a member of the Advocacy & Communication Sub-Committee for National Newborn and Child Health Advocacy & Communication Strategy. She was featured and contributed to a 2018 joint report by World Health Organization and UNICEF titled 'Survive & Thrive: Transforming care for every small and sick baby'. In Africa, she has pioneered several intervention initiatives, policy, research projects in her capacity as a Parent Representative and AFPNC president. Selina is a Parent Advisory Board member of the EFCNI and a member of the founding committee of the Global Alliance for Newborn Care (GLANCE).In January 2019, she became the first African and youngest woman to receive a prestigious award from EFCNI for her outstanding work.



**Arnold Christianson** graduated MBChB (Birmingham) in Zimbabwe in 1974, & trained in Paediatrics at the University of Cape Town. He obtained his MRCP (UK) in 1981 and was elected FRCP Edin in 1998. Initially a consultant neonatologist in Cape Town he entered private Paediatrics practice in Durban for 6 years. Returning to academia to do neuro-developmental Paediatrics in 1990 in Johannesburg, he switched to medical genetics in 1992. His research interests were dysmorphology, the epidemiology of birth defects & developing care & prevention services for people with birth defects in developing nations. A professor of medical genetics from 1997 he became Professor & Head of Medical Genetics, University of the Witwatersrand, in 2008, retiring

in 2015. He published 59 peer reviewed articles, 11 book chapters and was an expert advisor at 10 WHO meetings, of which one he chaired and another co-chaired. In 2015 he obtained an MA (Wits) in Applied Ethics.



**Matthew Darlison** BA, MA, PhD is the informatics lead and co-director (with Professor Bernadette Modell) of the WHO Collaborating Centre (CC) for Community Genetics at UCL. Originally trained as a linguist, Matthew joined the CC at UCL - then for the \_Community Control of Hereditary Disorders\_ - in 1997, to take forward new work developing approaches to online dissemination of materials to support genetic counselling at scale. This work led into Matthew's PhD in health informatics (2009) and to engagement with many facets of genetic science and service development and delivery at many levels of health services. The CC for Community Genetics at UCL is designated through the Regional Office for the Eastern Mediterranean, and current work

includes the development of (a) methods for estimating the epidemiology of congenital disorders on the basis of demographic and other data sources (to benchmark and address gaps in empirical data) and (b) tools and approaches to support primary care health professionals in delivering the key information interventions that form the gateway to more sophisticated genetic services. Matthew is based in the UCL Institute of Health Informatics, where his role combines teaching on, and providing learning support to, the UCL Graduate Programmes in Health Informatics, Health Data Science and Health Data Analytics



**Lasanthi Daskon** is the Deputy Country Director for the International Foundation for Electoral Systems (IFES) in Sri Lanka. She is an Attorneyat-Law with a background in human rights and has over fifteen years of experience working to promote the empowerment of marginalized groups, with a particular focus on rights of persons with disabilities. She holds a holds a master's degree on Human Rights from the Faculty of Graduate Studies, University of Colombo. Through her work with IFES Sri Lanka, Daskon supports the Election Commission of Sri Lanka as well as civil society partners in their efforts to further promote inclusive elections. Daskon is also a visiting lecturer at the Department of Disability Studies at the University of Kelaniya where she teaches a core module on Disability Theory and Concepts. She has contributed to

numerous research publications focusing on rights of children with disabilities, inclusive education, political inclusion, and access to justice among other areas.



**Chithramlee de Silva** MBBS, MSc MD is a Specialist in Public Health. She is a Medical Graduate and obtained post graduate qualifications MSc in 1991 and MD in Community Medicine in 1996 from the Postgraduate Institute of Medicine, Colombo and Diploma in Geriatrics and Gerontology from the University of Malta in 1997. She is currently the Director of Maternal and Child Health at the Family Health Bureau of the Ministry of Health, Sri Lanka. In this position she is responsible for national level implementation of reproductive, maternal, newborn, child, youth and women's health programmes. She was the former Director Mental Health (2015-2018), where she was involved in mental health programme management including prevention of suicide and

substance abuse. She was the President of the Perinatal Society of Sri Lanka in 2011 and the President of the Sri Lanka College of Community Physicians in 2014/2015. Her significant contributions to national development include the services she rendered to the fields of Maternal and Child Health, Monitoring and Evaluation, Human Resource Development, Non-communicable Diseases and Mental Health. Dr. de Silva has represented Sri Lanka at many seminars and conferences nationally and internationally. She has numerous published scientific papers, internationally and nationally and operational manuals and reports to her credit.



**Symone Detmar** is the Head of Program on Youth, Growing Up Healthy and Safely, TNO Child Health, Netherlands. Symone is a nurse and medical anthropologist. She has a PhD in quality of life research. She has more than 30 years' experience in the field of health research and practice, specifically in the area of maternal and child health, with scientific (more than 50 peer reviewed articles) and management experience. She was coordinator of several (inter)national studies in the field of perinatal screening, quality of life of children and the societal aspects of genomics research. She is member of several committees in the field of promoting health and safety for children from – 10 months. Currently she is program manager of the business line "Youth, Growing

Up Healthy and Safely" at TNO, where she is responsible for the strategic course involving 50 professional researchers. Within this business line specific attention is towards strengthening the perinatal period. In addition she is co-chair of the Preparing for Life Foundation.



*Vidya Jyothi* Vajira H. W. Dissanayake is the Chair, Senior Professor and Head of Department of Anatomy; Director of the Human Genetics Unit; and the Chairperson of the Specialty Board in Biomedical Informatics at the University of Colombo, Sri Lanka. He has been spearheading the development of clinical and diagnostics services, education, training and research in the field of Genetics and Genomics in Sri Lanka since 2004. In addition to developing the services in Sri Lanka he had contributed to establishment of services in Nepal for which he was honoured with an Honourary Visiting Professorship by National Academy of Medical Sciences, Kathmandu Nepal in 2018. Prof. Dissanayake has coauthored over 9 book chapters, 130 journal papers and 260 conference papers. He has received numerous national and

international awards for research including the Young Investigator Award of the Asia Oceania Federation of Obstetrics and Gynecology for his pioneering work in genetics of pre-eclampsia. He was elected a fellow of the National Academy of Sciences of Sri Lanka in 2013 and received the national titular honor of *Viday Jayothi* for his contribution to science and medicine in Sri Lanka.

He is an Executive Board Member of the Global Genomic Medicine Collaborative (G2MC); Board Member of the Forum for Ethical Review Committees in Asia and the Western Pacific (FERCAP); President of the Asia Pacific Association for Medical Informatics (APAMI); Vice President (Asia Pacific) of the International Medical Informatics Association (IMIA); Chairman of the Commonwealth Center for Digital Health (CWCDH); and Immediate Past President of the Commonwealth Medical Association (CMA).



**Madusha Dissanayake** is a Social Anthropologist with more than fifteen years of post-qualifying experience and expertise in the areas of public policy, social and behaviour change communication strategies, public health, gender, human rights and international relations. She has developed innovative programmes, established several communitybased networks and developed capacities of organizations, communities and individual women, youth and marginalized communities in several countries. Twenty-four years ago, Madu started her career as a Health & Science teacher then worked in the Government, Non-Government & Academia in the UK and Sri Lanka. Before she joined UNFPA as the Assistant Representative, she worked as

the Director for Public Affairs, Policy and Advocacy at the Family Planning Association of Sri Lanka.



**Pablo Duran** is a Medical Doctor and PhD from University of Buenos Aires; MPH from University of El Salvador (Buenos Aires, Argentina), and specialized in Pediatrics at the Children's Hospital Pedro de Elizalde, Fellow in International Nutrition and Nutritional Epidemiology (Cornell University, Ithaca, NY, USA), with more than 20 years of experience. Associate Researcher. Council of Research in Health at the Ministry of Health, Buenos Aires, Argentina. Regional Advisor on Perinatal Health at the Latin American Center for Perinatology (CLAP/SMR), Pan American Health Organization/World Health Organization, performing activities initially in the perinatal area and newborns, in coordination with other specific areas of Family and Community Health, developing technical

cooperation activities in related topics to the health of the newborn and its influence on the course of life. Dr. Duran is Associate Professor of Public Health at the School of Public Health, University of Buenos Aires.



**Mona Elnaka** MD, MBBCH, IBCLC, HRD is Executive Manager of Egyptian Members Association of Royal College of Pediatrics and Child Health -'EMARCPCH' - an Egyptian NGO established in 2007. Dr. Elnaka joined EMARCPCH in 2019, after having worked in a variety of positions in the Egyptian Ministry of Health and Population, including; First Undersecretary head of Primary Health Care Sector, General Director of Children with Special needs Directorate, General Director of the Egyptian National Neonatal Screening Program. She obtained her MBBCh degree from Medical School in Ain Shams University, Cairo, 1981, and then got her Masters degree in Pediatrics from the same

medical school, 1987. She also got a human resource management diploma from the American University in Cairo. She received training of Leadership in Strategic Health Communication, organized by Bloomberg School of Public Health, Johns Hopkins University and also completed the Executive Program for Emerging Leaders of the Ministry of Health conducted by Harvard School of Public Health. She is also certified as international Lactation Consultant since year 2002.

She actively participated in a study for Screening for birth defects among newborns performed in East Alexandria, Egypt, on 19530 neonates born over one-year period.



**Priyadarshani Galappatthy** Professor Priyadarshani Galappatthy, MBBS(Col), MD(SL), MRCP(UK), DipMedTox(Cardiff), FCCP, FRCP(Lond) is a Specialist in General Medicine and a Senior Professor in Pharmacology at the Faculty of Medicine, University of Colombo. She is a member of the consultative group of the Global Patient Safety Challenge on 'Medication Without Harm', of the World Health Organisation and has taken the lead in developing and implementing a national action plan on medication safety, with the Ministry of Health.



**Boris Groisman** is a medical geneticist and epidemiologist from Argentina. He's a member of the coordination of the National Network of Congenital Anomalies of Argentina (RENAC), a birth defects surveillance program covering around 300,000 births per year in the country. His areas of expertise are epidemiological analysis, information and communications technology, public health surveillance, e-learning and medical genetics.

Boris Groisman is a member of the Executive Committee of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR). He regularly participates as faculty in international training programs on birth defects surveillance.



**Rahul Gupta** MD, MPH, MBA, FACP, is the Senior Vice President and Chief Medical and Health Officer at March of Dimes, the nation's leader in mom and baby health. In this role, Dr. Gupta provides strategic oversight for March of Dimes medical and public health efforts to improve the health of all moms and babies. Before joining the March of Dimes, Dr. Gupta served as West Virginia's Health Commissioner. As the Chief Health Officer, he led the state's opioid crisis response efforts and launched a number of pioneering public health initiatives. Dr. Gupta, a specialist in internal medicine and preventive medicine, served as an academic faculty in Tennessee and Alabama before moving to West Virginia originally in 2009 to lead the Kanawha-Charleston Health Department. He is also adjunct professor in the Department of Health

Policy, Management and Leadership in the School of Public Health at West Virginia University and visiting faculty at TH Chan Harvard School of Public Health. Dr. Gupta was born in India and grew up in Maryland and the suburbs of Washington, DC. He completed medical school at University of Delhi, and subsequently did his internship and residency at St. Joseph Hospital, Northwestern University in Chicago, IL. He earned a master's degree in public health from the University of Alabama-Birmingham and a global master's of business administration degree from the London School of Business and Finance.



**Okello Harrison** is the Co-Founder and CEO of the Infants' Health Foundation, Uganda. He holds a bachelor's degree in Community Based Rehabilitation from Kyambogo University, Uganda. He grew up from Namayingo district - Uganda where there was no health facilities and one had to always travel many miles at a great cost to access the nearby health facility, as a result his family and community muchly relied on traditional herbalists and birth attendants, and witch doctors for health services and information. His newborn twin sisters both died at the traditional birth attendants' home during delivery. He was motivated to co-found Infants' Health Foundation. He is always at the forefront of leading the organization in bringing the basic lifesaving health care to

the underserved people in their unreached communities.



**Subhanghi Herath** PhD is a Professor in Sociology at the Department of Sociology University of Colombo and has served at the University of Colombo for 35 years. She obtained her PhD in Sociology in 1997 from the University of Waterloo, Canada on a Canadian Commonwealth Scholarship specializing in gender studies. She also received a British Commonwealth Postdoctoral Fellowship in 2004. She was the key person in developing social work studies in the Department of Sociology in 2008 engaging in a number of related field including ageing, disability, environment, violence, women and children's issues. She has served as a consultant in a large number of governmental and non-governmental institutions in the country and has taught in several universities abroad.



**Dineshani Hettiarachchi** MBBS, MSc, MCGP is a Lecturer in the Department of Anatomy and the Human Genetic Unit at the Faculty of Medicine, University of Colombo. Her qualifications include MBBS (Manipal), MSc in Regenerative Medicine (Colombo), Diploma in Advances in Stem Cell Biology (Doctoral School Module, Institute Pasteur, France) MCGP (Member of College of General Practitioners of Sri Lanka) and Certification for Teaching Higher Education, CTHE / SEDA UK. She is currently reading for her PhD on Rare Undiagnosed Disorders. She is also an invited steering committee member of the

Global Genomic Medicine Collaborative (G2MC) initiative as a young investigator. She writes a weekly health column in the Sunday Morning paper. Additionally, she is an alumni member of the Global Shapers Colombo Hub an initiative of the World Economic Forum and an active member of Zonta international. Dineshani was felicitated at the alumni recognition segment at the District 25 Zonta centennial conference for being the charter President of the Z club and winning the international award for young women in public affairs. She is also head of Human Resources and Administration team at the Enable Lanka Foundation, Sri Lanka.



**Shyamani Hettiarachchi** is a Speech & Language Therapist and Dramatherapist, and Senior Lecturer in the Department of Disability Studies at the Faculty of Medicine, University of Kelaniya. She graduated from the University of Kelaniya and completed her postgraduate studies in the UK. Her special research interests include child speech disorders, paediatric dysphagia, access to education and the intersectionality between poverty, gender and disability.



**Margaret (Peggy) Honein** PhD, MPH is an epidemiologist and Director of CDC's Division of Birth Defects and Infant Disorders. Dr. Honein received her BS degree in Biology from the University of California, Riverside in 1986, her MPH from the University of California, Los Angeles (UCLA) in 1992, and her PhD in Epidemiology from UCLA in 1995, and joined CDC as an EIS officer in 1997. She completed her doctoral dissertation research on HIV among tuberculosis patients in the East African country Djibouti. Dr. Honein served as the co-lead for the Pregnancy and Birth Defects Task Force throughout over 20 months of CDC's Emergency Zika Response; in this capacity, she developed and

directed work to advance understanding of and mitigate the impact of Zika virus infection during pregnancy. Dr. Honein also served as the Epidemiology Lead for the Maternal Health team during the 2009/10 Influenza Pandemic Response, a critical component of CDC's efforts to protect the health of pregnant women and infants. Dr. Honein has published over 150 scientific papers in the field of birth defects and maternal and child health. Her research interests include understanding the role of substance exposure in pregnancy on birth defects, assessing the safety or risk of medication use and vaccine use during pregnancy, identifying congenital infections with adverse neonatal and infant outcomes, understanding longer term outcomes and costs associated with congenital heart defects and other birth defects, and addressing the needs of vulnerable populations (including pregnant women and infants) in emergency responses. She has extensively mentored junior scientists at CDC over the past 22 years.



**Kapila Jayaratne** MBBS, MSc, DCH, MD is a Consultant Community Physician and the National Programme Manager for Maternal & Child Morbidity & Mortality Surveillance at the Family Health Bureau of the Ministry of Health, Sri Lanka. Dr. Jayaratne graduated from the Faculty of Medicine, University of Colombo and obtained masters and doctorate in Community Medicine from the Postgraduate Institute of Medicine of University of Colombo. He had his post-doctoral training at the University of Melbourne, Australia. He is the national focal point for surveillance of maternal deaths, maternal suicides, maternal near

misses, infant deaths, birth defects and injury-related child deaths. Dr. Jayaratne has carried out numerous studies on maternal and child health. He has contributed to reshaping maternal and child health service delivery by translating lessons learnt out of maternal and feto-infant deaths in Sri Lanka into action. He was the President of the Perinatal Society of Sri Lanka in 2016 and the Secretary General of the Sri Lanka Medical Association in 2019.



**Renuka Jayatissa** MBBS, MSc, MD is the Head of the Department of Nutrition at the Medical Research Institute, Ministry of Health, Sri Lanka. She obtained her MBBS, MSc and MD in Community Medicine from University of Colombo and received public health and clinical nutrition training in the UK. She has worked as a Nutrition Specialist for the UNICEF in Colombo for 5 years. She is an advisor to the multi sectoral nutrition action plan at Presidential Secretariat. She is the current President of Sri Lanka Medical Nutrition Association and the Past President of Nutrition Society of Sri Lanka.



**Athula Kaluarachchi** MBBS, MS, MRCPI, FSLCOG, FRCOG is Professor in Obstetrics and Gynecology at the Faculty of Medicine, University of Colombo. Professor Athula Kaluarachchi is a Senior Lecturer in Obstetrics & Gynecology at Faculty of Medicine, University of Colombo. He is also an Honorary Consultant Obstetrician and Gynecologist at the National Hospital of Sri Lanka and the De Soysa Hospital for Women. Having obtained his MBBS in 1985, Professor Kaluarachchi received his Master of Surgery at the Post Graduate Institute of Medicine, University of Colombo in 1990. He has authored over 30 peer reviewed publications in his field. In his 35 year-long career, Professor

Kaluarachchi has focused on developing the skills of others, working in teaching hospitals of Sri Lanka as well as the United Kingdom. His research interests include Infertility, Advanced Fertility Treatments & Medical Disorders in Pregnancy. Professor Kaularachchi is the immediate past President of the Sri Lanka College of Obstetricians and Gynecologists.



**Vijaya Kancherla** PhD is the Research Assistant Professor in the Department of Epidemiology at Emory University Rollins School of Public Health. She also serves as an Epidemiologist at the Center for Spina Bifida Prevention at Emory University. Dr. Kancherla has received her doctoral degree in Epidemiology from the University of Iowa, and completed a post-doctoral training as Pierre Decouflé Fellow with the National Center on Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention. Her current work includes birth defects surveillance, risk factors and health outcomes research,

prevention, and policy. Additionally, she is an affiliate member of the International Clearinghouse on Birth Defects Surveillance and Research. She is engaged in the Global WHO Birth Defects Technical Working Group, assisting in WHO efforts to increase awareness and research for birth defects globally.



**Anita Kar** PhD is the Founder-Director of the Birth Defects and Childhood Disability Research Centre, Pune, and Visiting Faculty (formerly Professor and Director) at the School of Health Sciences of the University of Pune, India. Dr Kar has a background in genetics, public health and epidemiology. Her studies place birth defects in a public health framework, researching the magnitude of birth defects and childhood disability in India, analysis of policies and services, describing the impact on parents and children in terms of prevalence and consequences of disability, co-morbidities, access to care, quality of life and economic impact. The overarching goal of the research is to identify the components and delivery mechanisms for a child-, parent and family-centric birth defects service. She has over 50 publications in

international and national journals and has an edited volume *Birth Defects in India: Epidemiology and Public Health* (forthcoming, Springer Nature). She is currently developing online content entitled *Birth defects in India and low middle income countries*, where individual modules explore the challenges and approaches to developing a birth defects service in the background of global health agendas, and the public health context of low and low-middle income countries.



**Kaushalya Kasturiarachchi** MBBS, MSc, MD is a Consultant Community Physician at the Family Health Bureau of the Ministry of Health, Sri Lanka. She is the Consultant In charge of the Monitoring and Evaluation Unit. She graduated MBBS from University of Peradeniya, Sri Lanka in 1998, and had her MSc in Community Medicine in 2001 and MD in Community Medicine in 2010 from the Postgraduate Institute of Medicine, University of Colombo. She has been involved in training of undergraduates in both Community Medicine and Health Informatics fields and has supervised MSc and MD students. She has also involved in development of national guidelines and policies. She has published number of research articles in international and local journals including BMC infectious Diseases. Dr. Kasturiaratchi has held many positions and memberships in academic organizations including Perinatal Society of

Sri Lanka and College of Community Physicians of Sri Lanka. Currently, she is the President of the Perinatal Society of Sri Lanka and a Council Member of the College of Community Physicians of Sri Lanka.



**Sylivia Kyomuhend** is a bachelor's degree holder in Community Based Rehabilitation from Kyambogo University, Uganda and the Founder & Director of Infants' Health Foundation, a grassroots level organization that aims at increasing access to & utilization of maternal child healthcare services to low income mothers and children living far from health facilities in remote hardest to reach villages of Uganda. She is a part time lecturer at Kyambogo University in the Department of Community & Disability Studies. Sylivia has won various awards and fellowships including Top Female Entrepreneur of the Year and 1st Runner-up in the recent Total Startupper Competitions She is a member

of the CIVICUS Alliance, Consortium of Grassroots based Development Partners - Uganda, NCD Alliance, and World Birth Defects Day Movement among others.



**Dhavendra Kumar** MBBS, MD, DCH (RCPSI), MMedSci, FRCPI, FRCP, FRCPCH, FACMG, DSc (HC) is one of the globally acknowledged clinical geneticist and a leader in genomic and molecular medicine. His special interests in clinical genetics include genetic diseases of children, inherited conditions of heart and blood vessels, genomic medicine and genomic applications in global healthcare. He has authored/edited many textbooks including 'Genetic Disorders of the Indian Subcontinent', 'Principles and Practice of Genomic Medicine', 'Cardiovascular Genetics & Genomics- Principles and Clinical Practice', 'Genomics and Health in Developing World' and 'Clinical Molecular

Medicine-Principles & Practice'. His book 'Medical & Health Genomics' is acknowledged by the British Medical Association Annual Medical Book Awards. He founded and led new genetic/genomic journals- Genomic Medicine/Human Genomics & Applied and Translational Genomics. Currently he is the Chief Serial Editor for 'Advances in Genetics' and serves on the editorial board of many biomedical journals. He is the Founding Editor in Chief of the new biomedical journal 'Genomic and Molecular Cardiology'. He is acknowledged with the benchmark practice of 'multi-disciplinary care for inherited/familial heart diseases. He founded and leads the 'Global Familial Heart Challenge' project for promotion; early detection, management and prevention of familial/ inherited heart diseases globally. Professor Kumar established and led as the Course Director of the wholly distance learning Post Graduate Certificate and Diploma course in Genomic Medicine and Healthcare of the University of South Wales. He founded and leads the

Indo-UK Genetic Education Forum and the UK India Genomic Medicine Alliance (UKIGMA). His new project, the Global Consortium for Genomic Education (GC4GE) aims to enhance and empower healthcare providers in the developing world through genetic/genomic education and training. He founded and leads the Genomic Medicine Foundation (UK), a 'not for profit' charitable organization that aims to support genomic applications in medicine, healthcare and socio-economic welfare through scholarship and fellowship.



**Aminkeng Leke** holds a PhD in Pharmacoepidemiology from Ulster University-UK. From 2007-2018, he served in various administrative and academic roles at Biaka University Institute of Buea-Cameroon; including his last position as DVC for Research, Cooperation, Quality. He has experience in projects focused on maternal medication use and safety in pregnancy in Cameroon and risk congenital anomalies related to antibiotics using the EUROCAT database. Currently, working at Ulster University to develop an App for the accurate description and coding of congenital anomalies, including the WHO Pregnancy Registry project in Africa. He is a FAIMER fellow (2011) – USA.



**Ornella Lincetto** is an Italian neonatologist, paediatrician and public health expert working with the World Health Organization, Maternal Newborn Child and Adolescent Health and Ageing Department, Geneva, Switzerland, as senior medical officer Newborn Health. She currently leads the Department's work on newborn health policies, practices and programmes and coordinates ENAP activities on behalf of WHO. She joined WHO in 2000, first in Geneva as newborn health expert and later as team leader Maternal Newborn Child and Adolescent Health and Nutrition at WHO country offices of Lao PDR, Vietnam and Papua New Guinea, and as WHO Representative in Bhutan. Before joining WHO, Dr Lincetto worked for various NGOs as project coordinator in Angola, public health expert in the Philippines, and as neonatologist and

researcher in Mozambique and Italy.



**Homero Martinez** is a clinical pediatrician with a PhD in International Nutrition from Cornell University. He is currently based at Nutrition International in Canada, where hey heads the secretariat for the Folate Task Team, a multi-disciplinary group to implement a global strategy for the control of folate deficiency and neural tube defects in low- and middle-income countries. Homero collaborates on a number of institutional 's initiatives related to technical assistance on nutrition and health, building partnerships, and institutional strengthening. In the past Dr. Martinez has held various teaching positions, and has published over 140 peer-reviewed articles, 54 book chapters, and edited 13 books,

with over 3,000 citations to his work.



**Rajesh Mehta** works with World Health Organization – South-East Asia Regional Office, New Delhi as the Regional Adviser-Newborn, Child and Adolescent Health since 2010. He supports 11 countries in South-East Asia in designing and implementation of these programmes. Before this he was at India Country Office of WHO as National Professional Officer and Coordinator for Family and Community Health. He managed WHO programmes to support the national plans in India related for maternal & reproductive health, newborn & child health, adolescent health, nutrition, gender, and nursing-midwifery. He has been managing Regional Birth Defects initiative and Regional initiative for improving quality of care for RMNCAH, Point of Care Continuous Quality

Improvement (POCQI) and collaborative learning platform. He is a Pediatrician by training, has been a teacher and has published many scientific papers, has contributed chapters in several text books and national and international training packages. He is a Salzburg Global Fellow on Early Child Development and also a recipient of Fellow of Indian Academy of Pediatrics (FIAP).



**Cara Mai** DrPH, MPH is a health scientist with the National Center on Birth Defects and Developmental Disabilities, U.S. Centers for Disease Control and Prevention (CDC) in Atlanta, Georgia. Dr. Mai has worked at CDC for the past two decades and provides technical assistance to develop and enhance birth defects surveillance programs. She was detailed to the CDC Zika emergency response to lead state-based surveillance of birth defects related to Zika virus. Her areas of interest include surveillance methodology, data utilization, dissemination, and evaluation.



**Kelle Moley** is the Chief Scientific Officer and Senior Vice President of the March of Dimes. She was the Vice Chair for Research and the Chief of the Division of Basic and Translational Science Research in the Department of Obstetrics and Gynecology, at Washington University in St. Louis, USA where she spent her entire 30-year academic career. She was also the Director of the Center for Reproductive Health Sciences also at Washington University. As an independent physician-scientist for the last 25 years, her work has impacted our understanding of reproductive performance and glucose metabolism in diabetic and obese animal models and this is directly applicable to the pathophysiology of diabetes related disorders in their offspring. In addition to her research endeavors, she is the former Co-Director of the

Washington University Institute of Clinical and Translational Sciences and was the Fellowship Director of Reproductive Endocrinology and Infertility for 12 years. She was the President of the Society of Reproductive Investigation in 2013-14 and was the Chair of the NIH Standing study section "Pregnancy and Neonatology" from 2011-2014. She also PI of the national NIH K12 Reproductive Scientist Development Program, which resides at Washington University in St. Louis. She was elected to the National Academy of Medicine in 2014.



**Cynthia Moore** MD, PhD is the Chief Medical Officer for the Division of Birth Defects and Infant Disorders in the National Center on Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention. She is a pediatrician and clinical geneticist. Her career has spanned over two and a half decades and has primarily focused on finding the causes of birth defects. Her recent contributions include characterizing the clinical syndrome associated with congenital Zika infection. Dr. Moore has authored or co-authored more than 150 scientific publications and has lectured extensively in the US and internationally.



**Gita Mishra** is an NHMRC Principal Research Fellow and Professor of Life Course Epidemiology at the School of Public Heath, University of Queensland. She is Director of the Australian Longitudinal Study on Women' Health, a major national study running since 1996 that has informed development of policy related to women's health. Prof Mishra also leads the InterLACE project, a major international consortium on reproductive health and chronic disease. She is internationally recognised for her contribution to research on life course epidemiology and women's health. Her specific focus is on the factors that affect reproductive health from menarche to menopause, and the influence of reproductive health across the life course. In 2017, she was presented

an honorary membership of Sigma International, a global nursing organization, for her contribution to women's health and was elected as a Fellow of the Australian Academy of Health and Medical Sciences (FAHMS).



**Ritsu Nacken** is the Country Representative for the UNFPA in Sri Lanka since 2017. As a passionate international civil servant of UNFPA, she has led a number of policy advocacy initiatives related to population policies, gender-based violence and youth development. During her over 20 years of service with the United Nations. Through these assignments, she contributed to a number of joint UN initiatives, including policy advocacy, strategy development and capacity building. Ritsu has a Master's degree in non-profit management from the Milano Graduate School of International Affairs, Management and Urban Policy in New York, and a Bachelor of Arts degree from the International Christian

University in Tokyo. She is originally from Japan.



**Anuruddha Padeniya** MBBS, DCH, MD is a Consultant Paediatric Neurologist at the Lady Ridgeway Hospital for Children, Colombo and the Academic Head of the Department of Paediatrics, Faculty of Medicine and Allied Sciences, University of Rajarata. In addition to his clinical obligations, as a professional and an activist, mainly in policy development, he has extended his leadership beyond his discipline to the field of healthcare in general, human resource structuring, human resource development. He has taken initiative of many public health problems at national level. He was honored with the Prestigious Eisenhower Fellowship in 2012 in recognition of his outstanding leadership shown in the field of health and other sectors. He is also the President of Professionals' National Forum and playing a major role in formulating the National Trade Policy.



**Carmencita Padilla** MD, MAHPS Dr Padilla is Professor of Pediatrics and Chancellor of University of the Philippines Manila. Dr Padilla is a pioneer in genetics in the Philippines and the Asia Pacific region. In the Philippines, she is responsible for setting up the genetic services at the Philippine General Hospital in 1990 and the various genetic laboratories now housed at the National Institutes of Health-Philippines. She is also responsible for setting up of newborn screening services, currently available in 7000+ health facilities in the country. In the Asia Pacific region, Dr. Padilla is part of the pioneering group that established the Asia Pacific Society for Human Genetics and served as President in 2008-2010. Dr. Padilla is Council member of Human Genome Organization

(HUGO), an international organization of scientists from 69 countries. In 2010, she was appointed country representative to the inter academy Partnership (Health), a global network of more than 60 academies in the world. Dr Padilla has more than 120 publications. In the area of policy making, she is responsible for the Newborn Screening Act of 2004 (Republic Act 9288) and Rare Disease Act of 2016 (Republic Act 10747). In 2008, she was elected to the National Academy of Science and Technology.



**Razia Pendse** took over as WHO Representative to Sri Lanka in May 2017. Dr Pendse, who is a national of India, has been with WHO since 2004. She has held various positions at WHO Regional Office for South-East Asia and WHO headquarters in Geneva. She has over 20 years of experience working in the development sector, is passionate about promoting health and wellbeing for all and aspires for an equitable, peaceful and sustainable world where no one is left behind. Dr Pendse received her Doctor of Medicine (MD) in Pediatrics from the University of Rajasthan, India; and Masters in Public Health (MPH) from the University of London, UK.



**Surantha Perera,** Consultant Paediatrician at Castle Street Teaching Hospital for Women, graduated from Faculty of Medicine Colombo 1997. He obtained his MD in Paediatrics in 2009. He was awarded fellowship in Royal College of Physicians in Edinburgh in 2014. He became a Fellow in Royal College of Paediatrics and Child Health in 2015. He is the President of Perinatal Society of Sri Lanka (PSSL) and the President of the Perinatal Association of Private Hospitals (PAPHSL). He is also the Past Secretary and Council Member of Sri Lanka College of Paediatricians (SLCP) and Council Member in Sri Lanka Medical

Association. He holds posts in many other forums in Sri Lanka. He is a member of several National Technical Advisory Committees of Ministry of Health in Sri Lanka. He is a co-investigator in several national level Innovative Projects under Ministry of Health. He is a Course Instructor in APLS, NLS and ENBC and Post Graduate trainer in MD Paediatrics. There are several book publications and research under his name.



**Neena Raina** is a public health specialist, working as Senior Advisor, Reproductive, Maternal, Newborn, Child & Adolescent health and Ageing programme in WHO, Regional Office for South-East Asia. She joined WHO in 1998 and has worked a coordinator- Health through Life course and Regional Advisor, Child and Adolescent Health and development. She has been working in WHO for the past 21 years. Dr Raina worked in government and non-government sector at senior level before joining WHO. She has worked in the areas of reproductive, maternal, newborn,

child & adolescent health, immunization, gender-based violence for the past 40 years. She has published about 200 scientific articles in journals/chapters in books.



**Nathalie Roos** is currently affiliated to Karolinska Institutet in Stockholm Sweden and the Division of Clinical Epidemiology working with nationwide health registers. She has a medical degree from the University of Uppsala, Sweden and her postgraduate training as an Obstetrician and Gynecologist at the Royal Karolinska University Hospital in Stockholm, Sweden. She did her PhD studies at the department of Women's and Children's health at Karolinska Institutet, Stockholm, Sweden focusing on perinatal epidemiology. Dr Nathalie Roos was formerly technical officer at the World Health Organization (WHO) team at the Department of Maternal, newborn, Child and

Adolescent Health (MCA Department) in the Epidemiology, Monitoring and Evaluation team (EME), in Geneva, Switzerland since 2015 and prior to that, at WHO country office in Burkina Faso working broadly on Maternal, Newborn and Child Health programs supporting technically the Ministry of Health.



**Janitha Rukmal** is the Co-founder, ENABLE Lanka Foundation, Sri Lanka which is Committed to enable the citizens of Sri Lanka with skills and entitlements through innovative solutions and approaches. Janith has a Bachelor of Arts Degree from the University of Sri Jayewardenepura in Sri Lanka & holds a Masters in Development Studies and Public Policy from the Open University of Sri Lanka. He has served as an Interpreter and Translator at the Presidential Secretariat and as a Consultant and Researcher at the Centre for Monitoring Election Violence. He is a senior member of the technical working group for disability inclusive elections and the youth committee of elections affiliated to the Election Commission of Sri Lanka. He is also a Youth

advisor on Disability and inclusion for the International Youth Alliance for Peace. Janith has published a Policy brief on political rights and representation of persons with disabilities in Sri Lanka under the patronage of Centre for Monitoring Election Violence. He has been an International representative of Leonard Cheshire: Represented Young Voices from Sri Lanka at the Global Young Voices Conference held in Nairobi in October 2013, and he also Served as a coordinating advocate for combating HIV in mobile populations at the United Youth Consortium under UNAIDS Sri Lanka 2015-16. He is a member of the founding directors' team of "Without Borders" that won the Global Trailblazer's Award from the Harvard Social Innovation Collaborative and was recognized by Her Majesty Queen's Young Leaders' Programme. Janith also received the Global Trailblazer's Award from the Harvard Social Innovation Collaborative



**Duminda Samarasinghe** MBBS, DCH, MD is the first board certified Paediatric Cardiologist in Sri Lanka. He is the Consultant Paediatric Cardiologist at the Lady Ridgeway Hospital for Children the premier Paediatric hospital in Sri Lanka. He was the President of the President Sri Lanka Heart Association 2018-2019 and Chairman of the Children's Heart Project of Sri Lanka since 2012. He was instrumental in designing, creating and establishing a fully functional database system at the Cardiology Unit at Lady Ridgeway Hospital which contains data of over 100,000 patients recorded over last 10 years. His team won the Cardiology Team of the Year award at the BMJ Awards South Asia awards in 2015.



**Liesbeth Siderius**, pediatrician aspires to optimize care for children with rare and disabling conditions. Trained in pediatrics and genetics she has experience of working in hospital (secondary) pediatrics, clinical (academic) genetics and preventive (primary) child health care in the Netherlands. She has been a volunteer for patient organizations working with and for them. Acknowledging the mere existence of patient organization and their expertise needs permanent advocacy. With the establishment of a rare disease working group of the European Academy of Pediatrics the importance of rare conditions in pediatrics was endorsed. Thanks to collaborations, networks, presentations as congresses, membership of European Expert Commission on rare

disease, a broad view on child health care in Europe and beyond has been developed.



**Samanmali Sumanasena** MBBS, DCH, MSc, MD, MRCPCH in the Professor in Paediatric Disabilities at the Department of Disability Studies, Faculty of Medicine, University of Kelaniya. She is the Past Chair of the International Committee of the American Academy of Cerebral Palsy and Developmental Medicine and Past President of the Sri Lanka Association for Child Development. Her recent research involved development of a hospital based surveillance system for Sri Lanka on cerebral palsy in collaboration with CP alliance Australia, capacity building of community health service providers on the MhGAP tool for autism and other developmental disorders with funding from the World Health Organization: and a randomize control trail evaluating the

impact of a multi-sensory tool for children with reading difficulties in the Gampaha district of Sri Lanka with funding from the National Research Council.



**Sonia Trikha** MD (Ob/Gyn), MPH is Director Health Services and Executive Director of the State Health Systems Resource Centre in Haryana, India. Currently, she heads the Quality Improvement initiative for over 350 public health facilities with a focus on improvement of processes related to clinical care and patient safety. She heads the capacity building initiative for the government for in-service doctors, nurses, paramedical personnel and first responders for RMNCH+A, Quality Assurance, M and E, and health policy and planning. She is also leading the food fortification initiatives in Haryana. Dr. Trikha has over 20 years of experience in managing large Reproductive, Maternal, Newborn Health, Family Planning & HIV AIDS programs in international organizations, public and not-for-profit sectors. She has worked as a

Maternal & Neonatal Health and HIV Specialist with UNICEF India. She was the focal point for 'Making Pregnancy Safer' and Reproductive Health and Research with WHO India. She has also led the HIV Prevention and Control Program for Chandigarh administration.



**Salimah R. Walani** PhD, MPH, RN is Vice President of Global Programs at March of Dimes. She leads the organization's global initiatives by working with international public and private partners. Her work focusses on advancing the health of women and girls in low and middleincome countries and improving prevention and care in the areas of premature birth and birth defects. Dr. Walani has served on numerous technical advisory committees to inform policy and programs for the health of women and children. She is a known leader in birth defects advocacy and has played a key role the World Birth Defects Day Movement. She joined March of Dimes in 2012 after having worked in a variety of healthcare and academic settings, including the New York City

Department of Health as an epidemiologist and as director of a New York University operated mobile health clinic that provided healthcare to immigrant children in Brooklyn, NY. Originally from Pakistan, Salimah Walani acquired her nursing and midwifery education from Aga Khan University in Karachi. She now holds a PhD from New York University, a Masters of Public Health from Harvard University and a Masters of Nursing Science from Simmons College in Boston. She has held academic positions at Aga Khan University, Felician University, Pace University and University of New York.

# **ABSTRACTS OF ORAL PRESENTATIONS**

# SYMPOSIUM 2 - LIFE COURSE APPROACHES AND PROGRAMS FOR IMPROVING THE HEALTH OF WOMEN AND GIRLS BEFORE, DURING AND AFTER PREGNANCY

### **OP-01**

Workplace Wellness across the Lifespan: A Pilot Program for Health Promotion in a Middle-Income Country

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**Introduction and objectives:** A health education program focusing on LINC factors: Lifestyle, Infection, Nutrition and Family Planning to be implemented in Lebanon and the Philippines. To assess the impact of the program, participants completed a Knowledge and Behavior Assessment Questionnaire (KBAQ) before and after implementation. KBAQ assessed health knowledge and behavior. To assess retention and long-term impact KBAQ was conducted at 6 months post-test. A multidisciplinary team of specialists delivered the sessions at a local Bank in Beirut, Lebanese project site. A booklet containing key health messages in Arabic and English was designed, and provided to participants.

**Results:** A total of 91 participants were enrolled with 50 females. Of them, 95% of participants were above 25 years of age, 98% had a university degree and 66% were married. Participants were divided into two groups and total of 22 lectures were provided from April to August 2018. The total knowledge score improved significantly from 64.2 to 74.3 and maintained significant at 6-month post 71.7 (P-value <0.001). Additionally, emotional score (P-value=0.005) improved while smoking decreased (P-value=0.021) at post-test and were maintained at 6 month. Positive feedback was received from the participants on the material, lectures and booklet. The average session evaluation score was 4.6/5 (4.4 - 4.9).

**Conclusions:** This wellness program showed significant improvement in employees' health knowledge and behavior. The analysis shows a high impact of the program on knowledge through an effect-size of 0.576.

Keywords: Workplace Wellness Programs, health literacy, Lifestyle changes

### OP-02 Workplace Wellness Modules Across the Lifespan: Utilizing e-Learning Access to Knowledge Padilla C.<sup>1\*</sup>, Sur A.L.<sup>1</sup>, Bandalaria M.<sup>2</sup>, Adducul R.T.<sup>3</sup>, Walani S.<sup>4</sup>

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**Introduction and Objectives:** The Institute of Human Genetics, National Institutes of Health, University of the Philippines Manila in collaboration with March of Dimes and American University of Beirut of Lebanon, successfully piloted a gender and culturally appropriate health education curricula consisting of 14 modules focused on preconception health utilizing the LINC (Lifestyle, Infection, Nutrition, Contraception) framework. Modules highlighted strategies to prevent prematurity and birth defects. This was implemented in a textile company and a university utilizing face-to-face approach for instruction. The results of the initial implementation showed enhanced knowledge based on pre-and post- test scores. To maximize the utilization and to widen its reach, the modules will utilize e-learning modalities. The objective of this paper is to present the process of conversion of the original modules into bite-size learning parts for online/offline access either through an app or through a dedicated website.

**Methods:** Two teams were created. The first team reviewed and finalized the content of the modules for bite-size learning parts. The outputs were reviewed by content experts before turning over to the second team. The second team developed the structure and design of the interactive online and offline e-learning program. The modules divided into bite-size learning parts were pre-tested by the first team before deployment to project sites. Knowledge change for the modules was measured through pre-tests and post-tests.

**Results:** With bite-size learning parts, utilization was increased. Data analysis using pre-tests and post- tests scores will be presented.

**Conclusions:** The project showed that e-learning modalities can increase utilization leading to increased acquisition of health knowledge at their most convenient time. E-learning which highlights remote access to knowledge is an effective alternative to traditional learning. As a result, there was enhancement of knowledge of workplace wellness.

Keywords: workplace wellness, LINC, e-learning

## OP-03 Readiness for Implementation of Preconception Care in Uganda Nathan Isabirye<sup>1\*</sup>, Agnes Kayego<sup>1</sup>, Agnes Nyabigambo<sup>1</sup>, Peter Waiswa<sup>1</sup>

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**Introduction and objectives:** In many resource limited setting, preconception care is not prioritized. Consequently, a gap is created in the continuum of care and may lead to adverse birth outcomes for mothers and newborns. Preconception care is imperative to reduce health risks and may empower women so as to have positive experiences of pregnancy. Little is known about the current health system and program context under which preconception care could be integrated. We assessed the policy and district context readiness for implementation of preconception care in Uganda.

**Methods:** Cross sectional study was conducted with a desk review, key informant interviews with local experts and a field assessment of a cohort of 300 pregnant and recently delivered women in Uganda. Phase1 will inform Phase 2 in designing and piloting of a preconception health care package. Desk review involved search of data bases limited to published and unpublished reports in English, and low-income countries. Search terms were categorized, combined using Boolean operators. Data was extracted and analyzed thematically.

**Results:** The study is still ongoing. Preliminary findings show that preconception care is neglected in Uganda's health system and literature is scanty. Uganda has no policy on preconception health and care. In existence are highlights of guidelines on preconception care embedded in other national guidelines. Components of antenatal and postnatal care exist in the continuum, with a missing link between postnatal and the next conception. Coverage of key maternal and newborn indicators is still poor. Communities and health workers have limited knowledge on preconception care. Risk factors for neonatal deaths in Uganda include; unplanned/short birth intervals pregnancies, teenage pregnancies, medical conditions, malnutrition, parity of +5, malaria in pregnancy, antepartum and intrapartum anaemia, preeclampsia, non-facility deliveries and poor birth monitoring, Infectious diseases and environmental smoke exposure.

**Conclusion:** This study is among the very few studies on preconception health and care in Uganda and in Sub Saharan Africa. There is an urgent need to integrate preconception care in maternal newborn and child health policies and programs in Uganda and other low income countries.

**Keywords:** preconception care, Uganda, readiness, neonatal death

### OP-04 Preconception Health Care Services Delivered in Selected Medical Officer Areas of Kandy District Sri Lanka Wijekoon W.M.C.R.<sup>1</sup>, Dharmarathne S.<sup>2</sup>, Ubeysekara V.G.S.C<sup>\*</sup>.<sup>3</sup>, Wickramasinghe I.P.<sup>4</sup>

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**Introduction and Objectives:** Rate of reduction of maternal and neonatal mortality had slowed down recently highlighting recent evidence that further reduction of these rates requires effective pre-conceptional interventions. Although the concept of Pre-conception care had been established recently, a detailed description has not been done on its service delivery in the Sri Lankan context. The study was conducted to describe the Pre-conception care delivered to primiparous women of Yatinuwara, Warallagama and Udunuwara Medical Officer of Health Areas of Kandy district before their first live pregnancy.

**Methods:** A descriptive cross-sectional study was conducted in community antenatal clinics. Primiparous women below 32 weeks of gestation living in selected MOH areas for more than 6 months were included. 218 participants were recruited according to population size. Data on prepregnancy health care services were collected using a consensually validated pre-tested interviewer-administered structured questionnaire. Univariate analysis for components of preconception care and bivariate analysis for identified variables of participants were performed. **Results:** Two hundred and seventeen participants responded. The proportion of 0.95(95%CI. 0.92-0.98) was above adolescent age and 0.12(95%CI, 0.08-0.16) had pre-conceptual medical consultations. The proportion of 0.3(95%CI,0.23-0.36) underwent pre-conceptual measurement of BMI and 0.52(95%CI,0.45-0.58) had pre-conceptual folic acid supplementation for at least three months. The proportion of 0.96(95%CI, 0.93-0.99) had rubella vaccination and 0.23(95% CI,0.17-0.29) had participated to at least one pre-pregnancy health education session either in government sector or private sector. The proportion of 0.03(95%CI, 0.007-0.05) married to close relative but none had undergone genetic counseling. The proportion of 0.91(95% CI,0.87-0.95) had planned pregnancies and 0.16(95%CI,0.11-0.21) had pre-conceptional screening for chronic diseases. The private sector had delivered 74.2% of total pre-pregnancy consultations. Statistically, no significant difference found between socio-demographic factors of receivers and none receivers of pre-conception care at the probability of 0.05 level.

**Conclusions:** Special attention should be given to improve preconception medical consultations, screening for chronic diseases and preconceptional health education services as a low proportion of participants received those services. Efficient utilization of National Preconception Health care package of the public sector and proper advocacy of private sector may improve the service delivery.

**Keywords:** Preconception care, Pre-pregnancy care, Pre-Pregnancy preparedness

## OP-05 Impact of Antenatal Care on Stillbirth at a Tertiary Care Referral Hospital of India Gaikwad H.<sup>1</sup>, Choudhary N.<sup>1\*</sup>, Mittal P.<sup>1</sup>, Jayathilaka C.A.<sup>2</sup>, Raina N.<sup>2</sup>

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**Introduction and objectives:** Stillbirth is a major cause of perinatal death in India and there has been wide variation in stillbirth rate across India. Deciphering the cause of still birth will help in formulating prevention strategies to bring down the stillbirth rate. Our objective is to explore the antepartum and intrapartum causes and risk factors of stillbirths.

**Methods:** This was a prospective observational study done under WHO-SEARO project. This study included all stillbirths occurring in Safdarjung hospital from August 2015 to December 2018. Verbal autopsies were done, and thorough antenatal records were documented and analysed.

**Results:** Out of 1,09,578 deliveries, 2689 were the stillbirths (2.4%). Among these, 1583 (58.9%) were unsupervised and unbooked pregnancies, whereas 1106 (41.1%) pregnancies were supervised and booked. The mean maternal age was  $25.9\pm4.5$  years, whereas mean body mass index (BMI) was 24.2 kg/m<sup>2</sup>. There was no significant difference between maternal age and BMI in booked and unbooked pregnancies. Significantly lower percentage of patients in unbooked pregnancies had iron-folic acid intake; significantly higher percentage of mothers had anaemia, hypertension, diabetes and antepartum haemorrhage compared to booked pregnancies.

Among antepartum causes, hypertension (23.4%), abruption (18.5%) and infection (3.4%) were the leading causes for still births. Maternal factors had contributed significantly higher percentage of still birth in unbooked pregnancies as compared to booked pregnancies. Intrauterine growth retardation (24.1%), birth defects (5.6%), and extreme prematurity (3.4%) were the most common fetal causes of still birth. Intrapartum causes constituted 38% of total stillbirths; higher percentage was in the unbooked pregnancies. Cord complications (16.6%), fetal distress (8.4%) and chorioamnionitis (1.2%) were leading intrapartum causes of still birth. In 21% of stillbirth, no cause was attributed. Approximately, 89% patients with stillbirth had delayed seeking of health care facility.

**Conclusions:** The most common cause of stillbirth was abruptio placentae in this cohort. Various maternal related factors are significantly higher in unbooked pregnancies. Reducing these factors might reduce the prevalence of stillbirth.

**Keywords:** Still birth, Antenatal care, Unbooked pregnancy

## SYMPOSIUM 3 - COUNTING BIRTH DEFECTS: INNOVATIONS IN SURVEILLANCE

### **OP-06**

Global Birth Defect Inventory of Resources for Surveillance and Research International Committee for Congenital Anomaly Surveillance Tools<sup>1</sup>, Dolk H.<sup>2\*</sup>, Aminkeng L.<sup>2</sup>

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**Introduction and Objectives**: The Zika outbreak has created more awareness about the public health significance of birth defects and intensified the need for better birth defect surveillance and research around the world, especially in low resource settings. Apart from the financial and human resources required to accommodate this shift, it <del>this</del> requires effective sharing of documentary, software, online and training resources. As part of the Zika PLAN project, the International Committee for Congenital Anomaly Surveillance Tools (comprising members from Europe, Latin America, Africa, Asia and the USA) has put together an inventory of existing birth defect surveillance resources.

**Methods**: Information was sourced from expert organizations around the world, and by internet search, to provide an inventory of resources that can facilitate birth defect surveillance and associated activities. Each resource is briefly described, with an internet link to where it can be found.

**Results**: The Global Birth Defects website (https://globalbirthdefects.tghn.org/) has been created in December 2018 to host the continuously updated inventory. The inventory is divided into 8 sections. 1) details of all major international networks and organizations for birth defects surveillance and teratology information 2) details of manuals and online atlases for congenital anomaly diagnosis and (ICD) coding, in multiple languages. 3) video material for training in neonatal surface examination and head circumference measurement 4) guidelines, common dataset specifications and coding systems for registration and surveillance of congenital anomalies, and associated freely available software 5) specialist resources for diagnosis of genetic syndromes 6) training materials and courses 7) public health tools to strengthen national policies on birth defect prevention and care, including estimations of burden, attributable fraction, types of policy action and tools for their implementation, including in relation to folic acid and Zika 8) a selection of information resources written specifically for parents.

**Conclusion**: The inventory provides a central place where a wide variety of resources can be readily identified to help establish/improve birth defect surveillance, and ultimately prevention and care. The sitting of the inventory within the Global Health Network serves to "mainstream" birth defect prevention as part of wider public health activity.

Keywords: Birth Defect Surveillance, Networks, Registries, Online resources, Training, Global.

## OP-07 A Reflection of Birth Defect Surveillance in Bangladesh: Challenges and Opportunities Shahidullah M.<sup>1\*</sup>, Mannan M.A.<sup>1\*</sup>, Dey S.K.<sup>1</sup>, Moni S.C.<sup>1</sup>, Shabuj K.H.<sup>1</sup>, Jahan I.<sup>1</sup>

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**Introduction and objectives:** Birth defects are an upcoming problem for developing countries where infections related deaths have been reduced substantially. Newborn birth defects surveillance program is now known as "Strengthening and Expansion of Newborn Birth Defects (NBBD) Surveillance in Bangladesh" funded by WHO-SEARO to ascertain the magnitude of birth defects. Objective of the study was to determine the magnitude and types of birth defects in the nodal center, Bangabandhu Sheikh Mujib Medical University (BSMMU).

**Methods:** BSMMU has been participating as a nodal center since the inception of the NBBD in the country with technical support from WHO-Bangladesh and SEARO. Newborns were evaluated at delivery room, post-natal ward and in NICU at BSMMU and data were entered into NBBD database. Data of all newborns including birth defects were analyzed from NBBD database from January 2018 to December 2018.

**Results:** In BSMMU, 1,878 babies were born during the study period, of which 91 had birth defects with the prevalence of 4.8%. Among them, 51% were preterm and 50% were low birth weight. Twelve percent had consanguinity. Thirty percent had maternal age >30 years. Birth defects were significantly more frequent among male newborns than females. Anomalous infants were significantly more premature and having low birth weight than that of term and normal birth weight. According to the ICD code 10, birth defect involving musculoskeletal system was 35%, CNS malformation 24%, defects of face/neck/eye/ear 9.7%, defects of circulatory system 8.4%, facial cleft 6.3% and others 16%.

**Conclusion:** Prevalence of the birth defects during the period of study in BSMMU was 4.8%. Birth defects involving musculoskeletal system were the commonest birth defects.

Keywords: Newborn, Birth Defect, Surveillance, Bangladesh

### **OP-08**

Integrating Birth Defect Surveillance Midwives in to Existing Labor Ward Structures: A Comparison between Government and Private Not-For-Profit Hospitals in Kampala, Uganda

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**Introduction and Objectives:** Hospital based birth defects (BD) surveillance systems requires substantial resources and staffing to implement and maintain. In Uganda, a BD surveillance system was implemented in one government hospital and three private-not-for-profit hospitals that deliver approximately 50,000 births annually. We describe the different staffing approaches that were taken, problems encountered with each approach and actions taken to resolve these problems.

**Methods:** The government hospital had the highest number of annual deliveries (30,000), hence midwives at this facility could not be burdened with additional surveillance activities. Therefore, additional midwives were recruited and specifically trained to conduct surveillance activities. These surveillance midwives worked alongside the government hospital midwives, conducted surveillance activities, assisted with routine deliveries, and were paid a monthly salary. At the private-not-for-profit hospitals, midwives already working on the maternity wards were trained to work as surveillance midwives. These midwives conducted surveillance activities in addition to their routine hospital responsibilities and were compensated for surveillance activities.

**Results:** In the government hospital, integrating new surveillance midwives into the existing labour ward services was problematic due to inadequate space to accommodate the new staff and their equipment, high delivery workload of the existing staff, and poor interpersonal relationships between the existing and new midwives. In the private-not-for-profit hospitals, challenges included adding additional activities to the workload of the existing midwives, surveillance midwives being transferred to other wards, fitting surveillance activities into existing hospital systems, and coordinating BD surveillance activities with different multiple stakeholders. Challenges at both types of hospitals were overcome by negotiating with the hospital administrations to acquire adequate space, developing unique ways of coordinating BD surveillance activities, and midwives' shift schedules to better accommodate BD surveillance activities, and training on good interpersonal skills to improve interpersonal relationships. In the private-not-for-profit hospitals, additional activities included continuous training and retraining of staff.

**Conclusions:** Implementation of a successful BD surveillance system into an existing hospital structure is a daunting task. The delivery volume at the participating hospitals may dictate which approach is feasible. Regardless of which approach is taken, intensive follow up will be needed and should be planned for.

**Keywords:** Birth Defect, Surveillance, Midwives, Government hospital, Private-non-for-profit hospital

**OP-09** 

# Pattern of Congenital Anomalies among Women Opting for Elective Termination of Pregnancy for Fetal Anomalies (ETOPFA) Sharma B.<sup>1\*</sup>, Aggarwal N.<sup>1</sup>, Suri V.<sup>1</sup>, Kakkar N.<sup>1</sup>, Chaudhary N.<sup>1</sup>, Raina N.<sup>2</sup>, Mehta R.<sup>2</sup>

Sharina B.<sup>1</sup> , Aggarwai N.<sup>1</sup>, Suri V.<sup>1</sup>, Kakkar N.<sup>1</sup>, Chauunary N.<sup>1</sup>, Raina N.<sup>2</sup>, Menta R.<sup>2</sup>

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**Introduction and objectives:** India has the largest number of infants born with genetic disorders or birth defects in the world with a prevalence of 6-7%, which is around 1.7 million annually. With the availability of prenatal screening and diagnostic techniques, it is possible to diagnose a wide variety of structural and genetic abnormalities before the period of viability i.e. before 20 week. In order to have a complete spectrum of birth defects it is important to include previable birth defects opting for pregnancy termination. This study was done to know the prevalence of previable birth defects and pattern of occurrence.

**Methods:** All the women opted for ETOPFA were included after informed consent. The case ascertainment of birth defects or any other chromosomal anomaly was done on the basis of prenatal ultrasound, fetal Echocardiography or genetic testing (chorionic villous sampling or amniocentesis). The demographic and clinical information was gathered from clinical record and open history. All parents were given the option of autopsy and results were compared with the prenatal diagnosis.

**Results:** A total of 1184 cases of previable birth defects were reported in four years and congenital malformation of central nervous system was the commonest (54%). Out of 1184 cases 746 were having isolated anomalies whereas in rest multiple systems were involved. A total of 1622 types of birth defects were noted in these fetuses. 34.7% women were primigravida and in the age group of 20 -30 years. The autopsy rate among pre viable fetuses was almost 90%. During this study period 5012 cases of birth defects among viable fetuses ( both live and stillborns) were reported from 3,42,770 births from same hospitals, so by capturing the previable birth defects data the birth prevalence have increased from 152.2 to 188.0 per 10,000 live births.

**Conclusions:** The prevalence of neural tube defects a preventable birth defect is alarmingly high in women opting for ETOPFA and to capture the real magnitude of congenital anomalies, previable birth defects needs to be identified and reported along with other birth defects. This would also help to assess the coverage of universal prenatal screening and quality of preconception & antenatal care.

Keywords: Birth defects, Previable, ETOPFA, Preconception, Screening
#### OP-10 Comparison of Modelled and Observed Data for Isolated Oral Facial Clefts in South Africa Malherbe H.<sup>1\*</sup>, Darlison M.<sup>2</sup>, Modell B.<sup>2</sup> and Aldous C.<sup>1</sup>

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**Introduction and Objectives:** Orofacial clefts (OFCs) are the most common craniofacial malformation but consistent empirical data is lacking for SA. Modelling offers a reliable means of estimating numbers until surveillance systems are adequately developed. This study compares modelled OFC data for 2012 with observed OFC data collected for 2013/14 as a means to assess the modelling methodology applied and the resulting estimates.

**Methods:** Modell Global Database of Congenital Disorders (MGDb) methods were adapted and used to quantify the burden of isolated OFCs in SA for 2012 for children under-5. The MGDb uses prevalence data from well-established surveillance systems and local demographic data to generate country/provincial estimates for OFCs 1) in the absence of care and 2) factoring in available care to evaluate the effect of interventions on birth outcomes/survival. Access to services was quantified using the infant mortality rate (IMR) as a proxy. These modelled estimates were compared with observed (secondary) data for OFC for 2013/14 collected from 11 specialized academic centres across SA as part of a separate PhD study.

**Results:** Factoring in available care of 30% for 2012, MGDb estimated 250 affected live births with a birth prevalence of 0.22 per 1 000 live births. Under-5 mortality was estimated at 50%, with 2% dying from other causes and 48% surviving with disability. Observed data collected for 2013 and 2014 indicated a birth prevalence of 0.36 and 0.24 per 1 000 live births respectively. This study quantifies the scale of the burden of isolated OFCs in SA for 2012 using the MGDb method.

**Conclusions**: Results from comparison with observed data supports this method as a tool to generate estimates for policy makers to use in service planning until improved surveillance yields accurate data.

**Keywords:** Oral facial clefts; South Africa; congenital disorders; modelling; Modell Global Database; estimates

# Overcoming the Defect: A Perinatal Review of Congenital Anomalies in 6984 Consecutive Deliveries in a Tertiary Center in Western Nepal Aryal S.<sup>1\*</sup>, Shrestha D.<sup>1</sup>, Aryal S.<sup>2</sup>

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**Introduction and objectives:** Neonatal congenital anomalies are the 17th cause of global disease burden and are a traumatizing experience to a mother and an overwhelming encumber to the family. Congenital anomalies in low income countries are high due to prevalence of nutritional deficiencies, intrauterine infections, exposure to teratogen and unsupervised self medication. This study was done with the aim to find the association of various antenatal risk factors and birth defects.

**Methods:** A prospective study was done for a duration of thirty months, (June 2016-December 2018) at Lumbini Medical College Teaching Hospital, Nepal. All neonates with congenital anomalies were examined and antenatal history reviewed. Consanguinity, intrauterine infections, presence of anemia, history of drug intake was noted. In cases of stillborn with anomalies, mode of delivery, labor complications and the three delays in maternal morbidities namely, delay in seeking care, reaching care and receiving care were also noted. Logistic regression analysis was used to determine the association of various antenatal factors and congenital anomalies.

**Results:** There were 65 congenital anomalies out of 6984 deliveries during the study period, out of which 43 were live births and 22 were still births. The incidence of congenital anomalies was 9.3 per 1000 births. Commonest anomaly 11 (25.5%) amongst live births was of musculoskeletal system (Q65-Q79) and commonest anomaly 10 (45.4%) amongst still births was of nervous system (Q00-Q07). Regression analysis showed that consanguineous marriage and lack of folic acid supplements were independent factors predicting congenital anomalies. In cases of still births with anomalies, most deliveries were vaginal (97%) without any maternal complications. Among the three delays, delay in deciding to seek care was significantly associated (p=0.023) with stillbirths with birth defects.

**Conclusions:** Birth defects are common in this part of the country. Factors like consanguinity and intake of folic acid are associated modifiable factors which can be useful in preventing congenital anomalies. Encouraging early antenatal visits might help in early detection of anomalies. This will provide an opportunity for timely counseling so that it is less traumatic to the mother and the family.

Keywords: Birth defect, Folic acid, Risk factors, Still birth

# SYMPOSIUM 5 REORIENTING HEALTHCARE SYSTEM FOR IMPROVING THE QUALITY OF MATERNAL AND NEWBORN HEALTH SERVICES

#### **OP-12**

Training Module for Primary Health Care Worker for Improved Recognition and Follow Up for Persons with a Congenital Disorder Aldous C<sup>1</sup>, Malherbe H<sup>\*1</sup>

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**Introduction:** Optimal care for a person with a congenital disorder requires early diagnosis and prompt referral to specialist services. In most low and middle income countries patients present to primary health care facilities and the first health care worker they will have contact with would be most likely be a nurse. It warrants that nurses working in primary health care facilities should be trained to identify of congenital disorders and to follow up procedures including initial treatment and appropriate referral. This aim to explore the effectiveness of a nurse training programme in genetics: the Medical Genetics Education Programme (MGEP).

**Methods:** MGEP was developed in 2003 to equip registered nursing staff with basic primary healthcare medical genetic education MGEP. This was implemented in two parts; MGEP 1 and MGEP 2. MGEP 1 focused on theory, over a period of four months with one contact day per month. An examination was taken at the end of the course. Successful MGEP 1 participants could undertake MGEP 2, a 2-week contact course focusing on basic clinical diagnosis and genetic counselling.

**Results:** MGEP 1 and 2 resulted in an improvement in skills and knowledge of nurses. A pre- and post-course evaluation of MGEP 1 and 2 showed an improvement in knowledge from 48% to 75%, and skills marks improving from 4.5% to 86%. Challenge was that, MGEP 2 required nurses to spend 10 consecutive working days away from their duties. A request from the National Department of Health was made to merge the two MGEP components and offer the new course on a one day a month basis for six months.

**Conclusions:** MGEP was revised to meet NDoH requirements, merging knowledge acquisition with skills development. The new version of MGEP will be field tested in a site in South Africa. **Keywords:** Genetics, Nurse training, MGEP, Primary health care

#### Implementation of Expanded Operational Guidelines on Use of Antenatal Corticosteroids: Perspective of Program Implementers and Health Care Professionals in Northern India Duggal M.<sup>1\*</sup>, Kankaria A.<sup>1</sup>, Sarkar D.<sup>1\*</sup>, Kumar M.<sup>1</sup>, Kumar P.<sup>1</sup>, Litch J.A.<sup>2</sup>

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**Introduction and Objectives:** The existing Government of India Operational Guideline on use of Antenatal Corticosteroids (ACS) for Preterm Labour was expanded in 2018 to include safe and effective use of ACS for threatened preterm birth by all levels of health care provider and level of facility. In order to understand the implementation of the expanded guideline this study collected the perspectives of program implementers and health care professions regarding useful aspects of expanded guideline and opportunities of improvement.

**Methods:** This cross-sectional study was conducted in Hisar and Ambala, Haryana, India between January - February 2019. Data were collected using semi-structured questionnaire with openended questions among facility leaders, program implementers and health care providers. A total of 124 interviews were carried out. ATLAS.ti 8 software was used to code the data and thematic analysis was undertaken in an iterative process.

**Results:** The findings demonstrated that the guideline was easy to understand as it consists adequate content, represented knowledge about indication and contraindication, increased clarity on facility readiness criteria and authorization of facility for ACS use. Healthcare professionals reported that they became aware of critical window period of 24 - 34 weeks and importance of accurate gestational age estimation for ACS administration. Suggested areas of improvement was availability of the guideline in the local language of health care providers and the need for continuous capacity building among health care providers. Further, the study highlighted that regular discussions on ACS related parameters and cases studies should be held during monthly meetings with facility leadership for improving adherence of expanded guideline. **Conclusion:** Pilot testing of the implementation of the expanded guideline before finalization and national scale-up was an innovative approach that resulted in further revision of a guideline at regular intervals based on the experience gained during implementation that considers the local service delivery context and emerging evidence.

**Keywords:** Threatened preterm birth, Antenatal Corticosteroids, implementation and qualitative research, operational guideline.

# The Impact of Different Intervention Packages on Facility Intervention on Health Facility Deliveries Utilization and other Maternal and Newborn Care Practices in Uganda Waiswa P.<sup>1\*</sup>, Isabirye N<sup>1</sup>., Muhumuza R.K.<sup>1</sup>, Ekirapa E.<sup>1</sup>

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**Introduction and objectives**: Understanding how to improve utilisation and quality of maternal and neonatal care services is urgently required in reducing the stagnated neonatal and mortality rates. We assessed the impact of implementing an integrated community-facility intervention package on health facility utilization and other maternal and newborn care practices.

**Methods**: This was a pre and post-test two year quasi experimental study with three trial arms: 1) Full intervention component consisting of Community Health Worker (CHW)s, transport vouchers for pregnant women, and bonus payments for health facilities based on performance, and health systems strengthening; 2) CHWs and health systems strengthening; and 3) CHWs only. A total of 2009 and 2098 participants were interviewed at baseline and end line. Descriptive statistics were generated for variables disaggregated by study areas and data collection period. Generalised linear model with logit link function and binomial family was used to assess outcome determinants.

**Results**: The interventions led to improvement in knowledge of pregnancy danger signs (43.07%, 37.64%), VHT home visits while pregnant (19.07%, 25.61%), Health facility delivery (2.11%, 4.01%) and at least 4 ANC attendance (3.32%, 6.96%). VHT home visits after delivery (-10.02%, -16.84%), Knowing at least 3 newborn danger signs (-14.72%, -1.1%), birth preparedness (-4.27%, -4.45%) and access to voucher (-5.53%, 0.58%) improved in the control than intervention areas. Being a resident is the intervention area was positively associated to health facility delivery and at least 4 ANC attendances, but negatively associated to ANC attendance in the first trimester. Factors positively associated to health service utilisation included; level of education, ANC attendance, access to voucher, wealth index, and adequate preparation for birth,

**Conclusions**: The study led to improvements in health service utilization, knowledge of pregnancy danger signs and VHT visits while pregnant. However, the study did not improve other components like; birth preparedness, VHT visits immediately after birth and knowledge on newborn danger signs. The findings underscore the strong influence of health care systems in improving health service utilization. There is need to understand further the dynamics of scaling up interventions using an integrated approach aimed at increasing demand and access to health care services.

Keywords: quality maternal and neonatal care services, health service utilization , Uganda

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#### Best Wishes Program: Use of Mobile Phone Technology (mHealth) for Care Co-ordination Directed Towards Birth Defects Prevention Sapkota D.R.<sup>1</sup>, Thapa R.K.<sup>1\*</sup>, Shrestha M.<sup>1</sup>

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**Introduction and Objectives:** Karuna Foundation Nepal, in direct partnership with local governments, has been implementing a community based inclusive development model called Inspire2Care. Best Wishes Program is a pivotal component of the model that employs mobile phone technology to establish safe motherhood care coordination directed towards birth defects prevention.

**Methods:** The program uses a mobile technology to register pregnant women and track them for Antenatal Care Checkup, institutional delivery and post-natal checkup including the newborn checkup. The trained Female Community Health Volunteer (FCHV) sends a mobile message to the Gateway Phone to register the identified pregnant woman. The system then generates a unique registration number. For ANC visits, the system automatically sends a reminder to the volunteer so that FCHVs could follow pregnant woman for checkups. Upon the reminder, the volunteer reaches the pregnant lady and makes sure that she visits the health facility. This continues until the pregnant woman delivers at health institution and undertakes postnatal services and newborn checkup.

**Results:** The program trained 1,032 FCHVs from 48 health facilities from ten (rural) municipalities in Eastern Nepal. These FCHVs have been continuously tracking and following up the pregnant women for the services. Amidst the poor mobile connectivity in many parts of the municipalities, the results have been increasingly promising both terms of service coverage. 1<sup>st</sup> ANC visit coverage has increased from 80% (2015) to 85% (2018) and 4<sup>th</sup> ANC visit coverage has been improved from 30% to 44% in the same period. Proportion of institutional deliveries rose from 63.5% to 81% from 2015 to 2018

**Conclusions:** The use of digital technologies offers new opportunities to improve people's health. If digital technologies are to be sustained and integrated into health systems, they must be able to demonstrate long term improvements over the traditional ways of delivering health services. **Keywords:** Mobile Phone, Technology, Birth Defects, Prevention

# OP-16 The Baby Buddy App: A Digital Health Intervention to Bridge Neonatal Health Inequalities in the United Kingdom. Baum A.<sup>1</sup>, Perera N.<sup>2\*</sup>, and Papain S.<sup>1</sup>

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**Introduction and objectives:** A healthy start to life is the key to reducing intergenerational disadvantage. Despite the availability of universal health care, evidence shows the existence of health inequity in maternal and child health. The public health charity Best Beginnings continues to use a multiphase approach based on co creation during the development and implementation of the Baby Buddy app. A separate series of resources, Small Wonders, which focuses on premature babies and their wellbeing, that are now embedded within the app.

**Methods:** This study reviewed the impact and reach of the Small Wonders videos embedded within the Baby Buddy app using a mixed methods approach. The development and implementation of the app into local care pathways and its reach and impact was examined using feedback from surveys, focus groups and health professionals training to use to the app and the in-app analytics.

**Results:** With over 220,000 registrations in the UK, Baby Buddy is an evidence-based app with personalised daily information and over 300 video clips and 500 FAQs. The app contains almost 40 films that are designed for parents and health professionals to support premature and sick babies with over 22,000 views up to April 2019. The process of co-creation was valued within health professionals and parents alike in improving outcomes related to breastfeeding self-efficacy, bonding and attunement and strengthening multidisciplinary working. Uptake trends of the app indicate it reaching seldom heard communities which includes younger mothers, minority ethnic groups and mothers whose first language is not English. The in-app analytics of the Baby Buddy app shows that 98% (n=3961) of the users feel confident in taking care of their baby after using the app.

**Conclusions:** Baby Buddy is a strong example to show the effectiveness of using co-creation to design and implement a complex digital health intervention. The co-creation approach of the Baby Buddy app has underpinned the app's success in supporting public health priorities such as neonatal health and well-being and the initiatives of the charity have a strong potential to bridge inequalities in child health in the United Kingdom.

Keywords: Digital health, premature babies, apps, neonatal health, co-creation

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# OP-17 Outcomes of Infants with Structural Birth Defects in Galle, Sri Lanka De Silva J.<sup>1\*</sup>, Amarasena S.<sup>2</sup>, Jayaratne K.<sup>3</sup> and Perera B.<sup>1</sup>

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**Introduction and objectives:** Birth defects is a significant health problem among children. The outcomes of structural birth defects (SBD) vary owing to the severity of the defect and availability of the management options. Outcomes of children with SBD are vital to identify the severity of the problem, to make policies and to allocate services to manage SBD.

**Methods:** A prospective longitudinal follow-up study was carried out to follow-up 620 SBD in a sample of 315 infants during their infancy. They were not diagnosed with any genetic syndrome. At the end of the first year of life the outcomes of the infants and the outcomes of the SBD were evaluated. The infant outcomes were measured as alive or dead. The outcomes of the SBD were measured as completely corrected, partially corrected (residual defect or residual impairment present) and uncorrected. Data were analyzed using SPSS version 20.

**Results:** There were 179(56.8%) male infants. Fifty-six (17.8%) were preterm. Three (1%) infants and 6(1%) SBD were lost to follow-up. The commonest defects were the cardiovascular (CVS) defects (n=398, 64.2%) followed by the central nervous system defects (n=52, 8.4%). The commonest CVS defects were atrial septal defect (ASD) (n=219), patent ductus arteriosus (PDA) (n=79) and ventricular septal defect (VSD) (n=48). Forty-four (14%) infants died during infancy. They had a total of 141 defects and 79 out of them were lethal. The most common lethal defects were the CVS defects (n=44, 55.7%) followed by CNS defects (n=13, 16.4%). Among the 473 SBD found among live infants 134(28.3%), 71(15.0% and 268(56.6%) were completely corrected, partially corrected and uncorrected respectively. Among the completely corrected defects 77 were resolved spontaneously (including 43 ASD, 6 VSD, 24 PDA) and the rest were managed surgically (n=55) and medically (n=2). Among the infants diagnosed with CVS defects needing surgical correction (n=35), 23 underwent surgery and 5 of them died. Three infants among the rest of the 12 infants, died while awaiting surgery.

**Conclusions:** The rate of correction of SBD among infants in Sri Lanka is suboptimal. There is a need to increase the health care services available to correct SBD in infants.

Keywords: Structural birth defects, outcomes, Galle, Sri Lanka

#### OP-18 Quality of Life of Primary Caregivers having Children with Congenital Heart Diseases Awaiting Cardiac Surgery Attending the Cardiology Clinic at the Lady Ridgeway Hospital for Children, Colombo, Sri Lanka Warnakulasooriya P.<sup>1\*</sup>, Kasturiaratchi K.<sup>2</sup>

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**Introduction and objectives:** Congenital heart disease (CHD) is the commonest type of birth defect, of which the estimated prevalence is around 8-12/1000 worldwide. Nearly 4% of all neonatal deaths are due to CHD. Caregivers of children with CHD are easy victims of low quality of life (QOL) and instability of household cost. Lady Ridgeway Hospital (LRH) is the largest government hospital for children in Sri Lanka and the only tertiary care referral center for children with CHD in the country. According to hospital data, over 40% of these children do not have appropriate access to early intervention and are compelled to wait until they get their turn. This study was conducted to determine the quality of life and household economic cost of primary caregivers having children with CHD awaiting cardiac surgery attending the cardiology clinics in LRH.

**Methods:** This was a descriptive cross-sectional study conducted using consecutive convenient sampling method over three months at cardiology clinics in LRH (July 2017 to September 2017). The sample consisted of 422 caregivers having children with CHD awaiting cardiac surgeries. An interviewer-administered questionnaire was utilized to obtain data. Pre-tested validated WHOQOL-BREFF was used as a tool for assessing QOL. Costing related questions assessed the cost borne by the caregiver attending clinic and they were prepared following extensive review of literature and by consulting local experts. Univariate analysis and logistic regression were performed to detect associations.

**Results:** Mean score of the QOL was comparatively low in all domains except in social - relationship domain. Quality of life was associated with age of the caregivers, educational level, marital status, number of children, income, type of heart disease and postponement of the surgery. Median direct expenditure per clinic visit was LKR 1800 (range of LKR 1175 to 3000). Median indirect cost was Rs. 1000. Out of all care givers, 28.7% of the care givers were falling in to catastrophic expenditure during that particular month of clinic visit.

**Conclusions:** Quality of life of primary caregivers was comparatively low in all domains except in social-relationship domain. Nearly a third are having catastrophic expenditure on clinic visit alone. Physician and health care managers should make a proper planning to improve the QOL of affected caregivers.

**Keywords:** quality of life, primary care givers, household economic cost, children, congenital heart disease, cardiac surgery

### OP-19 Thalidomide and Zika virus: Lessons for Health and Welfare Systems of Low and Middle-Income Countries Kar A.<sup>1\*</sup>

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**Introduction and objectives:** The thalidomide episode of the 1960's and the Zika virus outbreak five decades later illustrate the catastrophic potential of teratogens. This paper reviews the health and welfare system responses that were put in place in affected countries to address the epidemic of children with disabilities. The study aims to identify the health and welfare system responses, policies, services and regulations that were put in place, and to describe the key essential components of a birth defects service in low and middle-income countries.

**Methods:** A narrative review was carried out by using selected Keywords that examined the magnitude of the outbreaks, needs of affected children, caregivers' needs and the health and welfare system activities. The outbreak responses were analyzed to identify the key health and social welfare systems that are essential for a service for birth defects and developmental disabilities.

**Results:** The analysis showed the key components of a service to address birth defects and developmental disabilities. The need for a birth defects surveillance system that can detect the teratogenic exposure as early as possible was revealed. Birth defects service preparedness identified the need for not only medical services, but also rehabilitation services for care for children with disabilities. Another essential function was the need to include psychosocial support for caregivers. Review of the thalidomide literature identified the need for social welfare measures including financial support, caregiver support such as respite care, support for transportation, linkages to special education and vocational training.

**Conclusions:** The review informed the key essential components of a service for birth defects and developmental disabilities, underlining the need for an integrated health and social welfare service, that can provide a holistic set of services to address the consequences of these conditions. **Keywords:** Thalidomide, Zika virus, health system, welfare system, birth defect and childhood disability service

# Birth defect in South-East Asian Regional Countries; Challenges and Progress in Research, Policies and Programmes: A Scoping Review Trisa T.I.<sup>1\*</sup>, Hoque D.M.E.<sup>1</sup>, Shabnab S.<sup>1</sup>, El Arifeen S.<sup>1</sup>

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**Introduction and Objectives:** Every year 8.1 million are born with a serious birth defect globally. Of these, 2.3 million are from South East Asia region (SEAR). To achieve Sustainable Developmental Goals, birth defect should be addressed. This review was conducted to identify gaps, challenges and progresses in the existing research, policies and programmes undertaken for the prevention and management of birth defects in SEAR countries and to suggest possible solutions and scopes for future research.

**Method:** A scoping review was conducted following PRISMA-P guideline. An extensive search of published and unpublished grey literatures and reports was done using PubMed, Cinahl, Web of science and Google Scholar. Cochrane extraction form was adopted for data extraction and SIGN checklists were used for assessing study quality. Narrative synthesis was conducted following Poppy J guidelines.

**Results:** 14308 abstracts were selected primarily based on the search strategy. After reviewing the abstracts, 22 articles were selected for narrative synthesis based on inclusion and exclusion criteria. Following adoption of the regional strategic framework (2012–2016) at country level, wide variation in progress and challenges was identified. Among all SEAR countries, Sri Lanka is advancing in reporting birth defect since nearly 100% babies are born in facilities. The coverage of births registered varied from 34% in Nepal to almost 100% in Bhutan. The burden of birth defect prevalence in 2006 was highest in India (64.3%) and lowest in DPR Korea (54.1%). Findings are based on hospital survey and it does not reflect the actual situation except for Sri Lanka. In terms of prevention & management gap exists in surveillance, genetic services, screening programmes during pre-pregnancy and pregnancy. Since National Neonatal Perinatal Database has been launched by WHO-SEARO in 6 SEAR countries in 2007-08, India has made impressive advancement in terms of reporting, generating evidence and publications on birth defect.

**Conclusion:** There are plenty of scope for research on the current status of birth defects. To address birth defect, WHO regional office should increase technical and financial support and advocacy to bring attention of the policy makers on preventing and treating birth defect at country level.

Keywords: Birth defect, Congenital Anomaly, SEARO, South East Asia

#### SYMPOSIUM 8 DIAGNOSTIC STRATEGIES: GENETIC TESTING, NEWBORN SCREENING, PATHOLOGICAL EVALUATIONS

#### OP-21 Molecular Signature of Preterm Premature Rupture of Fetal Membrane Pan J.<sup>1\*</sup>, Zhong N.<sup>1,2</sup>

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**Introduction and Objectives:** Preterm premature rupture of fetal membrane (pPROM) presents the dominant feature of spontaneous preterm birth (sPTB), which accounts for 5-10% live birth annually. The long-term morbidity of surviving preterm infants is significantly higher than that of term neonates. The causes of sPTB are complex and not fully understood. Placenta, the maternal and fetal interface, is the only environmental core of fetal intrauterine life, mediates fetal nutrient uptake, waste elimination and oxygen exchange via umbilical cord. Therefore, the function of placenta is strongly associated with preterm birth. In this study, the molecular signature of preterm birth placenta was assessed and compared to full-term placenta by proteomic profiling. **Methods:** Four groups of fetal membrane were included in the study: A: spontaneous preterm labor (sPTL, N=5); B: preterm premature rupture of membrane (pPROM, N=5); C: Full-term birth (FTB, N=5); D: Full-term premature rupture of membrane (fPROM, N=5). Placenta membrane and placenta tissues were dissected, placenta membranes were used for lab free quantitative proteome study. Maxquant and Perseus were used for protein quantitation and statistical analysis. Both placenta membrane and placenta tissue samples were used to validate proteomic discovery. Results: Proteomics analysis identified 2,800 proteins cross four groups. Twenty-nine of them show statistical difference between preterm and full-term control groups. Among these differentially expressed proteins are 1) proteins involved inflammation (HPGD), T cell activation (PTPRC), macrophage activation (CAPG, CD14 and CD163), cell adhesion (ICAM, ITGAM), 2) proteolysis (CTSG, ELANE and MMP9), 3) antioxidant (MPO), 4) extracellular matrix (ECM) proteins (APMAP, COL4A1, LAMA2, LMNB1, LMNB2, FBLN2 and CSRP1) and 5) glycolysis (PKM, ADPGK), fatty acid synthesis (ACOX1, ACSL3) and energy metabolism (ATP6AP1, CYBB).

**Conclusions:** Molecular signature study of preterm placenta membranes revealed inflammation as major event which is consistent with previous findings. Proteolysis may play an important role for placenta membrane rupture. ECMs have been altered in preterm placenta membrane due to proteolysis. Metabolism was also altered in preterm placenta membrane. The molecular changes in placenta membrane were more significant than in placenta.

Keywords: Preterm premature rupture of felt fetal membrane, pPROM, Molecular signature study

# OP-22 The Saudi Human Genome Program (SHGP) and Precision Medicine in Saudi Arabia Aida I Al-Aqeel <sup>1\*</sup>

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**Introduction and objectives**: Saudi Arabia has a high burden of rare diseases, mostly due to the high rate of consanguineous marriages (around 60% of the marriages), resulting in severe inherited disorders manifesting early in life, affecting ~8% of births in the Kingdom. Common diseases with a significant genetic contribution, including diabetes, cardiovascular disease, asthma and others that manifest later in life affect over 20-30% of the population. The Saudi Human Genome Program (SHGP) was launched in 2014 with the goal of identifying genetic variation in the population and improving personal care in Saudi Arabia.

**Methods**: The SHGP targets the sequencing of 100,000 samples over five years. It has sequenced over 25,000 samples from patients and family members with inherited diseases. This has resulted in a large knowledge database of genetic variants, which identifies pathological and normal variants present in the Saudi population.

**Results:** In our experience over 25 years genomic variants has helped in implementing several families based prevention programs including pre-implantation genetic diagnosis, prenatal diagnosis, and population screening programs including newborn screening and pre-marital screening. It has also helped us in establishing personalized therapeutic strategies in several genetic metabolic disorders. With the pre-marital screening program approximately 60% of couples found to carry variants likely to result in affected children choose not to proceed with marriage. This has saved healthcare cost of more than SAR 3 billion on an annual basis with reduction in social burden to the community and health care professionals.

**Conclusions**: Establishing the SHGP and personalized clinical care was necessary to provide the necessary infrastructure to solve cases and understand disease in the Saudi population and to implement strategies for prevention and therapy within an Islamic ethical framework to reduce the burden of these diseases in the community and establish acknowledge based economy.

Keywords: Saudi Human Genome Program; Consanguineous marriage, Screening

# OP-23 Genetic Service for Birth Defects at the Dutch Caribbean Islands Verberne E.A.<sup>1\*</sup>, Caribbean genetic pediatricians<sup>2</sup>, Mannens M.M.A.M.<sup>1</sup>, van Haelst M.M.<sup>1</sup>

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**Introduction and objectives**: Small Island Developing States (SIDS) are islands of the Caribbean Sea and the Atlantic, Indian and Pacific Oceans. They face challenges due to small size and relative remoteness, including limited access to (specialized) health care like medical genetics. We established a genetic service for pediatric patients in Curacao, Aruba, Bonaire and St. Maarten (Dutch Caribbean Islands). The aim of this study is to evaluate the diagnostic and clinical outcome of 7-year genetic care service for this pediatric population.

**Methods**: Twice per year the clinical geneticist evaluates patients with birth defects at the pediatric clinic. If indicated, blood samples are sent to the Netherlands for genetic testing. For this study, we retrospectively included all children (age <18 years) that had a genetic consultation between November 2011 and November 2018. We excluded children who are already received a definitive diagnosis previously and healthy children that were referred for carrier testing. Because of the relatively isolated population we hypothesized that specific conditions (caused by autosomal recessive or founder mutations) could be more frequently identified amongst these children.

**Results**: A total of 479 children were included in this study. We were able to establish a definitive clinical and/or molecular diagnosis among 24%. Molecular diagnoses included autosomal dominant conditions (40%), autosomal recessive conditions (10%), microdeletion/-duplication syndromes (32%), X-linked conditions (7%), aneuploidy (6%) and other (5%).

**Conclusions**: Since genetic results are still pending for 51% of the included patients, a final diagnosis could be much higher than 24% of the children. Although the population structure of small islands could suggest a higher level of recessive conditions, we were unable to show the same in this sample in Dutch Caribbean islands. A genetic diagnosis helps in ending the diagnostic odyssey for parents and enables them to make informed reproductive choices. We suggest that this strategy with a visiting clinical geneticist can be used to provide genetic testing for birth defects in other small island developing states (SIDS).

**Keywords**: genetic analysis, diagnostic strategy, birth defects, small island developing states (SIDS)

# OP-24 Minimally Invasive Autopsy in the Evaluation of Fetal Malformations Langeh N<sup>1</sup>\*, Gupta N.<sup>1</sup>, Sharma A.<sup>1</sup>, Mridha A.<sup>1</sup>, Jana M.<sup>1</sup>, Barwad A.<sup>1</sup>, Dadhwal V.<sup>1</sup>, Kabra M.<sup>1</sup>

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**Introduction and objectives:** Minimally invasive autopsy (MIA) using post-mortem magnetic resonance imaging (PMMR) with ancillary investigations is reported to be as accurate as conventional autopsy in fetuses with structural abnormalities. This study was planned to assess the feasibility of implementing PMMR and to assess its accuracy as compared to conventional autopsy.

**Methods:** PMMR and/or conventional autopsy were performed on fetuses (14-20weeks gestation) and IUD/stillbirths (>20weeks gestation) with malformation over 2y 3m period. Feasibility of capturing these fetuses, median time to MRI, autopsy, and reporting were assessed. Concordance between MIA with autopsy was assessed in terms of agreement [Kappa coefficient (k)], sensitivity, specificity, positive predictive value and negative predictive value

**Results:** We enrolled 171 cases [fetal malformations (100) and IUD/stillbirths with malformation (71)] opting MIA followed by autopsy (81%, n=139), only MIA (10%, n=17), only autopsy (6%, n=10) and neither (3%, n=5). The median time between delivery and PMMR was 23h 33min and between delivery and autopsy was 27h 30min. Concordance between MIA and autopsy was assessed in 133 cases where both MIA and conventional autopsy results were available. The diagnostic accuracy of MIA for abnormalities of brain and spinal cord, cardiovascular system, respiratory system, abdomen and genitourinary system when compared with conventional autopsy as the gold standard was 90.7%, 90.6%, 96.9%, 99.2% and 97.6%, respectively. The sensitivity, specificity, positive predictive value and negative predictive value were respectively: 88.4%, 100%, 100%, 68.4% for brain and spinal cord; 92.1%, 76.9%, 97.2% and 52.6% for cardiovascular system; 99.2%, 25%, 97.6 and 50% for respiratory system; 99.1%, 100%, 100%, and 94.7% for abdomen and 99%, 92.5%, 98% and 96.1% for genitourinary system. The findings of MIA were in agreement with conventional autopsy in 90.7% [k-0.7] brain and spinal cord, 90.6% [k-0.5] cardiovascular, 96.9% [k-0.3] respiratory, 99.2% [k-0.9] abdomen and 97.6% [k-0.9] genitourinary anomalies

**Conclusion:** Although conventional autopsy is the gold standard, PMMR is feasible in a tertiary care setting and can be considered as a potential alternative for post-mortem assessment, especially in settings with limited facility of perinatal autopsy and parental refusal.

**Keywords:** Minimally invasive autopsy, post-mortem MRI, conventional autopsy, virtual autopsy, fetal malformations

#### SYMPOSIUM 9 NETWORKING AND PARTNERSHIPS FOR BIRTH DEFECTS RELATED RESEARCH, PREVENTION AND CARE

#### **OP-25**

**Creating and Strengthening Global Partnerships to End Disability from Clubfoot Pettitt-Schieber C**<sup>1</sup>**, Everhart J.**<sup>1</sup>**, Owen R., Mosende M.C.**<sup>2</sup>**, Aroojis A.**<sup>1\*</sup> <sup>1</sup>Miracle Feet, Chapel Hill, NC, USA, <sup>2</sup>Global Club Foot Initiative \*Correspondence: Christie.Pettitt-Schieber@miraclefeet.org

**Introduction and objectives:** Annually, 174,000 babies are born with clubfoot, primarily in Low and Middle Income Countries (LMIC) where fewer than 15% can access effective treatment. Several countries have successfully implemented national plans aimed at awareness, detection, referral and treatment of babies born with clubfoot deformity. The success of these programs is due to a validated, low cost and effective treatment called the Ponseti method. A group of NGOs working through collaborations with ministries of health have undertaken a cohesive effort through a global consortium to support improved quality and reach of the Ponseti method in LMIC.

**Methods:** In 2009 the Global Clubfoot Initiative (GCI) was created with eight founding members who were collaboratively developing clubfoot programs in LMIC. GCI has grown to 30 members working across Africa, South Asia, and Latin America. In 2017, GCI launched an ambitious strategy, Run Free 2030, to ensure that >70% of children in LMIC have access to treatment by 2030. This strategic model strengthened the consortium's work by providing agreed-upon goals at the highest level.

**Results:** With a team of global experts, GCI developed a standardized Ponseti method curriculum in English, Spanish and French, enabling programs to scale high quality training for efficient upskilling of healthcare providers. Using a train-the-trainer model, seven trainings have created a group of over 100 local trainers in 30+ countries. A blended-learning approach utilizing inperson and online training allows GCI members to train in basic clinic skills and provide ongoing mentoring. A mobile-based patient record management system helps ensure quality treatment through monitoring of globally-accepted indicators. GCI webinars on advanced treatment topics provide continuing education. GCI advisory boards representative of multiple countries, professions and organizations offer guidelines for development of best practices, including parent education to increase retention and advocacy strategies for sustainability. GCI publishes a biennial report on clubfoot treatment in LMIC, measuring progress toward its global strategy.

**Conclusions:** Shared expertise has enabled GCI to provide valuable programmatic guidance for strengthening service delivery of clubfoot treatment. This consortium provides a collaborative model that could be replicated for treatment of other birth defects and prevention of related disabilities in LMIC.

Keywords: LMIC, Ponseti method, Club foot, train-the-trainer model

## Combining the Strength of Government and NGOS, Providing Comprehensive Care for the Infants: An Encouraging Experience from China Zhao X.<sup>1\*</sup>, Wang P.<sup>1</sup>, Tao X.<sup>1</sup>, Zhong N.<sup>1</sup>

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China was well known for its population size. The government has been fighting against the birth defect and unbalanced distribution of quality perinatal health care resources for decades. The public health department of the government overlooks the birth defect related matter, establishes rules and regulations, and supports the basic medical expenditure, and runs health insurance. The involvement of non-governmental organizations greatly boosted such progress, improved the condition birth defect treatment, and strengthened the birth defect prevention.

The NGOs can provide medical and financial aids, which could be an ideal supplement of public health insurance. In the year 2018, two major NGOs has reimbursed over 1.5 million infants of newborn screening, 489 cases were confirmed supported. The Chinese Foundation of Birth Defect Prevention and Aid service covers 29/34 regions of China, and has issued 100 million Yuan to 9900 metabolic disorder or malformation children for their treatment. Unlike the stodgy and tedious governmental guidance, huge varieties of eye-catching educational material were produced by NGOs, and could be easily distributed to the end users. Modern communication media like We chat, QQ, Weibo, Tik Tok, were used by NGOs. The NGOs has spectrum of funding source; thus, they have more capability to set up a reasonable and responsible project in a time-efficient manner. In 2018, eight NGOs has trained 3700 healthcare professionals, donated 23 mass spectrometry labs and 4 genetic diagnostic labs.

In conclusion, the public health department of government has achieved a remarkable accomplishment that brings national birth defect rate from 8% to less than 5%. However the government approach to birth defect control need improvements. The NGOs can apply their advantage, work with the government, and compliment the country's efforts to control the birth defects in China.

Keywords : Birth defect, Government, NGO, aid, prevention

# Thailand National Plan for Prevention and Care of Birth Defects & Disabilities Wasant P<sup>1\*</sup>

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Thailand Birth Defects Task Force' developed to establish a system of prevention and care of birth defects according to Sixty-Third World Health Assembly Resolutions in 2010. Thailand National Network for Prevention and Care of Birth Defects & Disabilities began in May 2011 supported by ThaiHealth. It is a national network consisting of 8 medical institutes, National Health Security Office (NHSO) and Birth Defects Association (Thailand). First Phase (2011-2014) (i)development of strategic map, master plan and care map (ii)completion of manuals and care map for 5 chosen common birth defects (BD) for provincial and community hospitals (iii) memorandum of understanding between 4 Ministries (Health, Education, Social Welfare and Human Security and Interior), NHSO & ThaiHealth including Birth Defects Association (Thailand) (iv) pilot program on BDR Online in 22/77 provinces in collaboration with NHSO (v) initiation of eleven health districts model in 11 provinces focusing on holistic approach on prevention and care of 5 BD (vi)"Thailand Country Action Plan" initiated by Department of Medical Services, Ministry of Public Health(MOPH) in February 2012 (vii)systematic approach including working with policymakers and stakeholders at 3 levels (country, provincial and community). Second Phase (2015-2017) (i) 'Thailand development of provincial & district health system' expanded into 26/77 provinces, 26 model district hospitals including Bangkok Metropolitan focusing on holistic care of 5 chosen common disorders (DS, NTD, CL/CP, LA, DMD) (ii) expansion of 'BDR Online' to Bangkok Metropolitan (iii) NHSO agreed to add folate supplementation to Universal healthcare benefit (iv) prevention of birth defects with folate supplementation in women of child-bearing age initiated at sub-district health centers (vi) folate fortification discussed among healthcare professionals, policymakers and stakeholders for possible implementation (vii) 'Thailand Country Action Plan' declared by Minister, MOPH as a National health policy on October 9, 2017. (ix) importance of Birth Defects & Disabilities emphasized in Precongress Workshop on Birth Defects 12th Asia-Pacific Conference on Human Genetics (APCHG 2017), November 8-10, 2017, Bangkok, Thailand. It is concluded that 'Thailand National Plan for Prevention and Care of Birth Defects and Disabilities' successfully moves toward national agenda via Thailand National Health Assembly in 2019.

Keywords: birth defects & disabilities, prevention and care, national plan

#### OP-28 The Neonate Fund: Delivering Hope through Facilitating Quality NICU Care Pascale Nakad P.<sup>1,2\*</sup>, Sawwaf R.<sup>2</sup>, and Yunis K.A.<sup>1,2</sup>

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**Introduction and Objectives:** Access to quality Neonatal Intensive Care is significantly limited in middle and low-income countries due to high cost and absence or minimal health care and third-party coverage. This has direct impact on neonatal survival as well as short and long-term morbidities. In Lebanon, many families are unable to have third-party coverage; besides insurance companies have limited coverage, insufficient to settle huge bills. The Neonate Fund (NF) was established in 2012 at the American University of Beirut Medical Center (AUBMC) to provide financial support to needy families of premature and sick newborns admitted to NICU. The NICU is a level 4-referral unit providing state of the art care with highly specialized equipment and experienced multidisciplinary team of healthcare professionals.

**Methods:** Patients seeking financial assistance are referred by the NF coordinating office for an assessment of eligibility by the social services department at AUBMC. Patients are classified into three eligibility categories: highly eligible, eligible and partially eligible, based on four criteria: financial status, newborn's medical condition, estimated length of stay in NICU and availability of other coverage. Based on the assessment, and the bill, the NF assists with a percentage of the bill at a hospital-discounted rate.

**Results**: Between January 2012 and April 2019, 423 newborns were financially assisted by the Fund, of which 27% were transfers. In total, 64% were premature, specifically, 40% extreme preterm (<32 weeks), 17% very preterm (32-33 Weeks) and 43% late preterm (34-36 weeks). 75% of all cases were assessed as highly eligible, 8% eligible and 16% partially eligible. The average amount of support per patient decreased by eligibility: \$13,350 (up to:\$80,000), \$5,350 (up to:\$25,000), \$2,592 (up to:\$17,000) for highly eligible, eligible and partially eligible respectively.

**Conclusions:** Financial support by NF provides access for needy premature and sick newborns admitted to the NICU at AUBMC to receive the quality critical care they need. This results in better health outcomes, and survival among extremely premature newborns, as low as 24 weeks of gestation, who would have died had they been admitted to a lower level NICU. The goal however remains to provide and spread universal health care across the country and at the highest standards.

Keywords: Neonate Fund, Financial support, Quality Care, Neonatal Intensive care unit

#### Knowledge on Advanced Maternal Age, Offspring Outcomes and Attitudes Towards Advanced Maternal Age among Pregnant Mothers in the Colombo Municipal Area. Nawaratne S. D<sup>1\*</sup>, Vidanapathirana H. M J. P<sup>2</sup>

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**Introduction and Objectives:** Advanced maternal age (AMA), which is defined as 35 years and older, is a strong risk factor for birth defects. According to 2016 DHS, 11.8% of total births were by women in AMA in Sri Lanka. Reasons for child bearing in the later part of reproductive age can be diverse. The aims of this study were to assess the knowledge on the risk, describe attitudes and assess correlates of AMA among pregnant mothers.

**Methods:** Descriptive cross-sectional study was carried out among 423 pregnant women attending antenatal clinics in Colombo Municipal area, Sri Lanka. Participants selected from 14 clinics by two stage systematic sampling. Pretested interviewer administered questionnaire was used. Knowledge score calculated on a 0-10 scale. Marks for each question were added and converted into a percentage. Responses to statements on attitudes were on 5-point Likert scale.

**Results:** The proportion of pregnant women in AMA was 14.4% in the sample. Only 57.4% mothers in AMA and 48.9% of all participants knew that Chromosomal abnormalities of offspring are increased with age. Sixty four percent mothers in AMA and 65% of all participants did not know that the likelihood of having a child with Down's syndrome increases with age. Thirty six percent of total participants and 29.5% of mothers in AMA knew risk of still births increases in AMA. Only 20.3% participants had 'good' level of overall knowledge (>75 marks) on risk of AMA, while 49.0% had 'very poor' to 'poor knowledge' (<50 marks). Positive attitude observed among 67.1% of the respondents. Participants' perceived older age improves readiness to be a parent by having established relationships, financial stability, established career and being intellectual more mature. 22.5% had negative attitudes towards AMA, while 10.4% were neutral in their views towards AMA.

**Conclusions:** Majority of participants are unaware of the risk of AMA; but, carried a positive attitude towards AMA. Health care providers must evaluate knowledge and attitudes on AMA among pregnant women to implement effective risk communication strategies among them. **Keywords:** Advanced Maternal Age, Knowledge, Attitudes

## SYMPOSIUM 10 COUNTING BIRTH DEFECTS: HOSPITAL AND POPULATION BASED SURVEILLANCE

#### OP-30 Study of Birth Defects in Government Referral Hospital, Maldives 2016-2018 Faisal A.<sup>1\*</sup>, Ibrahim N.<sup>1</sup>, Rasheed H.A.<sup>1</sup>, Karna P.<sup>2</sup>, Raina N.<sup>2</sup>

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**Introduction and objectives:** Maldives has made tremendous progress in reducing child mortality in the MDG era, but now with low mortality rates, there is an epidemiological shift in causes of child mortality. Birth Defects account for up to 30% of neonatal mortality (WHO 2015). Indira Gandhi memorial hospital (IGMH) is the tertiary apex government referral hospital in the Maldives, where almost half of all deliveries in the country occur, and almost all birth defects are ultimately referred.

The study was carried out to describe the epidemiology of birth defects identified in live births born at the time of birth and those that were referred to IGMH.

**Methods:** Data from 3 sources were used. They include World Health Organization Regional Office for South-East Asia (WHO-SEARO) Newborn and Birth Defects Database (NBBD) (IGMH inputs data to this since November 2015), NICU admissions and discharge registry and referral abroad registry. Data of all intramural and extramural live births were included form January 2016 to December 2018.

**Results:** A total of 7287 live births were documented IGMH in 3 years. The number of live births with a birth defect was 462 (6.3%). Total anomalies detected were 689, out of which 551 were found, inborn babies. There were 65 lethal, 519 significant but not lethal and 105 mild anomalies. Most defects were from the cardiovascular system (28%), followed by the musculoskeletal system (14%) of which club foot (33 cases) was the most common anomaly. There was a similar incidence in dermatological, genital, abdominal, head and neck anomalies. All of them contributed 10% of birth defects as an individual category. There were 21 newborns that required cardiac surgery out of which 19 babies had a good outcome. There were 67 neonatal deaths during the 3 years period. The main cause of neonatal mortality was birth defects (31% of deaths), 3 of them were neural tube defects. There were 21 cases of Down syndrome as well.

**Conclusions:** Birth Defects are common and a significant cause of neonatal mortality and morbidity in the Maldives. Treatment measures have saved most of the babies with lethal anomalies. There is significant need to improve diagnostic and treatment modalities in managing birth defects.

Keywords: Birth defects, Maldives

#### OP-31 A Descriptive Study on Feto-Infant Structural Anomalies in Maternal Diabetes Mellitus: An Autopsy Study at a Tertiary Care Setting. Lakmini S.P.R.V.<sup>1\*</sup>, Priyani A.A.H.<sup>1</sup>, de Silva M. V. C.<sup>1</sup>

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**Introduction and Objectives:** Maternal diabetes mellitus is known to cause number of adverse pregnancy outcomes such as miscarriages, preterm births, stillbirths and major congenital malformations. Objective of this study was to describe the pattern of structural anomalies in feto-infantal autopsies in the presence of maternal diabetes mellitus and to find the association between maternal diabetes mellitus and presence of congenital malformations.

**Methods:** Data were gathered from 522 autopsies of feto-infantal deaths performed at Department of Pathology, University of Colombo, Sri Lanka from January 2007 to December 2018. Details of the maternal medical conditions were available in 214 cases which were extracted and analyzed retrospectively in this study. Autopsies were performed according to the standard protocols and none of the autopsies had extensive autolytic changes obscuring the gross anomalies. Data analysis was done using Statistical Package for Social Sciences and chi square test was performed to explore the associations.

**Results:** Study included 214 autopsies (49 second trimester miscarriages(T2M), 121 stillbirths, 44 neonatal deaths (ND)). Maternal age ranged from 18 to 45 year with a mean age of 30.03 (SD5.57) years. 31.8% (n=68) mothers had diabetes mellitus (pre-gestational diabetes mellitus: 43/68, gestational diabetes mellitus: 25/68). Out of the cases 32.4% (n=22/68) had structural anomalies (T2M:3/22=13.6%, stillbirths: 8/22=36.4% and ND: 11/22=50%), in which 50% were males 45.5% were females 4.5% had undetermined sex. Isolated system defects were present in 59.1% (n=13/22) and multisystem defects were seen in 40.9% (n=9/22). Lung hypoplasia was the commonest isolated defect (n=5) followed by atrial septal defect (n=3), hypoplastic left heart (n=2), holoprocencephaly (n=1), absence of fingers (n=1), abdominal wall defect (n=1). 26.7% (n=39/146) of autopsies without a history of maternal diabetes mellitus were positive for congenital anomalies (isolated system defects: 16/39=41%, multiple system defects: 23/39=59%). Hence, there was no statistically significant association between maternal diabetes mellitus and the presence of structural anomalies (p=0.412).

**Conclusions:** Isolated system defects were commoner than multisystem defects in feto-infant deaths with maternal diabetes mellitus. Lung hypoplasia was the commonest isolated system defect. Though there is no statistically significant correlation between maternal diabetes mellitus and presence of structural anomalies further studies are needed.

Keywords: structural anomalies, pre-gestational diabetes mellitus, gestational diabetes mellitus

### Analysis of Stillbirth in a Tertiary Care Hospital of Delhi: A Contribution To NNBD Project Singh A.<sup>1\*</sup>, Kumar M.<sup>1</sup>, Raina N.<sup>2</sup>, Mehta R.<sup>2</sup>

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**Introduction and Objectives:** Developing countries account for 98 % of total stillbirths worldwide but still have received very little attention while making programs or policy. This study intended to collect data on epidemiological profile of cases experiencing stillbirths, to assess associated antenatal high-risk factors and to find out the probable cause of stillbirth.

**Methods:** This cross sectional, observational study, done as a part of NNBD study with WHO SEARO support after ethical clearance from the institution. The study included all stillbirths which occurred in the hospital during August 2015 to November 2018. Antenatal records were reviewed, maternal investigations were done. Baby was examined after delivery. Pre-structured proforma was filled and the relevant condition found was classified under CODAC system of stillbirth classification.

**Results:** In a period of 3 years there were 46,816 deliveries of which, 1239 (26/1000) were stillborn. Most of the women (44.7%), were between 24-28 years of age and majority (42.2%) were primigravida. The most common associated factor was pre-eclampsia (20.7%) and fetal growth restriction was observed in 20.2% cases. More than half of them delivered (54.7%) after 32 weeks. At admission fetal heart was absent in 78.8% cases. Majority had fresh stillbirth (56%). Nearly 80% weighed less than or equal to 2500g. An intrapartum cause was present in 152/1239 (12.3%) cases. Maternal cause was noted in 300/1239 (24.2%) cases. Fetal cause in 359/1239 (28.9%) cases, placental and cord origins were suspected in 263/1239 (21.2%) and fetal death was unexplained in 165/1239 (13.3%) cases. Among the maternal causes the most common was preeclampsia in 183/1239 (14.8%) and FGR followed by infection including fever 71/1239 (5.7%), the most common infection was hepatitis. Among the fetal causes birth defect was the most common 219/1239(17.7%) followed by extreme prematurity in 87/1239 (7.0%).

**Conclusions:** Recording and analyses of stillbirth in every institution is the first step towards reducing the burden of preventable stillbirth in developing countries. Preeclampsia and fetal growth restriction were most common maternal factors while birth defects were the most important fetal cause of stillbirth.

Keywords: Still Birth, India, Birth defects, NNBD

Incidence, Disease Spectrum, and Genetic Defects in Congenital Heart Disease in China: Prenatal Ultrasound Screening Identified 18,171 Affected Fetuses from 2,452,249 Pregnancies

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**Introduction and Objectives**: Congenital heart disease (CHD), the most common birth defect in China, may result in an economic loss of 13 billion RMB yuan annually in China. However, few studies of fetal CHD, which is quite different from neonatal CHD, have reported the incidence disease spectrum, and genetic deficits of CHD in China.

**Methods:** Cross-sectional prenatal ultrasound screening for fetal CHD was performed in 2,452,249 pregnancies with a standardized protocol (based on ISUOG guidelines), to assess CHD. Screening was performed by 182 prenatal ultrasound specialists, who were enrolled from 92 hospitals across all provinces/autonomous regions of China, except Tibet, where no data were available. These prenatal ultrasound specialists were trained in the National Prenatal Diagnosis Center of Chinese Health Ministry.

**Results:** A total of 18,171 pregnancies with fetal CHD were detected, giving an incidence rate of 7.4/1,000, similar to the global figure. Among these cases, 5.64% had a high family risk, 20.30% received serological screening, 9.1% received chromosome or gene testing and 32.8% were combined with extra-cardiac malformations. The most common congenital heart abnormalities, in descending order, were VSD, TOF, AVSD, DORV, TGA, SV, HLHS, and COA/IAA. Among these, TOF, AVSD, and DORV were prone to be associated with extra-cardiac abnormalities (ECAs). A total of 32.8% (5,961/18,171) of pregnancies showed fetal CHD accompanied by ECAs. The most common ECAs were single umbilical artery, cleft palate, neck lymphoma/fetal edema, thoracic/abdominal effusion, Dandy-walker malformation, choroid plexus cysts, cisterna magna, club foot, ventriculomegaly, and omphalocele. Of prenatally identified fetal CHD cases, 68.8% (12,976/18,171) were followed up, and 1,965 cases were followed up until birth, of which 88% (1,730) were consistent with prenatal diagnosis. Karyotyping was applied to 1,659 (9.1%) pregnancies in this cohort, and in 374, chromosomal abnormalities were identified, including trisomy 21 and trisomy 18 as the most common. Deletion/duplication analysis with microarray identified microduplication in VSD, ASD, TA; large duplication in SV, TOF, VSD, HLHS, CVR; microdeletion in VSD, TOF, DORV, AS, TGA, PTA, IAA, and HRHS; and uniparental disomy in VSD, IAA, and persistent left superior vena.

**Conclusions:** This study reports the findings of the largest cohort of pregnancies screened for fetal CHD by prenatal ultrasound in China.

Keywords: Incidence, disease spectrum, congenital heart disease, prenatal ultrasound screening

# OP-34 Birth Defects in Congenital Rubella Syndrome - A Hospital Based Surveillance at Tertiary Care Center Singh P.<sup>1</sup>, Singh K.<sup>2\*</sup>, Vyas V.<sup>3</sup>, Yadav G.<sup>4</sup>, Nag V.L.<sup>5</sup>

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**Introduction and objectives:** Birth defects pose heavy burden on the family, society and nation due to huge cost in management. Some birth defects are preventable like Congenital Rubella Syndrome (CRS). However prevention is possible if burden and spectrum of disease is well known. Prevalence of CRS is not known in many developing countries like India. The study was carried out to determine the spectrum and burden of CRS in a tertiary care center in Western Rajasthan India.

**Methods:** Study site is a tertiary care center in Western India and also a sentinel site for CRS surveillance. Inclusion Criteria were Structural cardiac defects confirmed by ECHO, Hearing impairment confirmed by BERA or ASSR, Cataract / micro-ophthalmos / micro-cornea/ congenital glaucoma /pigmentary retinopathy. Maternal history of suspected or confirmed rubella in pregnancy. And a strong clinical suspicion of microcephaly/ developmental delay/ hepatosplenomegaly/ jaundice or purpura. All suspected cases were subjected to clinical examination, [ENT, Eye, cardiac etc] and 1ml blood was collected for Rubella antibody. Oropharyngeal swab for PCR was collected from 2017 onwards. Known/suspected genetic syndromes, anomalies not due to rubella eg- neural tube defects- anencephaly, meningo-myelocele, etc, were excluded from the study data was collected from 2016 till Feb 2019 and analyzed for incidence/prevalence, and type of anomaly.

**Results:** A total of 180 suspected CRS cases were screened during the study period. One hundred and sixteen were less than 3 months old. Thirty-three were lab confirmed by PCR. Congenital heart defects accounted for 87.2%, eye signs for 19.4%, hearing impairment for 14.4%. PDA was the most common anomaly noted in the study.

**Conclusions:** The prevalence/incidence of CRS in this study was 18.3%. The commonest birth defect was structural heart defect followed by eye and hearing defects. Rubella is a preventable viral illness by vaccination.

Keywords: Congenital Rubella Syndrome, CRS, heart defects, congenital cataract

# OP-35 Neural Tube Defects in China, 2007-2017: Data from the National Population-based Birth Defects Surveillance Deng C.<sup>1,2\*</sup>, Dai L.<sup>1,2</sup>, Li X.<sup>1,2</sup>, Zhu J.<sup>1,2</sup>

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**Introduction and objectives:** Neural Tube Defect (NTD) is one of the most common birth defects and raised concern worldwide. With the widely implement of maternal peri conceptional supplement of folic acid and prenatal screening and ultrasound diagnosis in China, little is known about its current epidemiology.

**Methods:** Data were collected from the National Population-based Birth Defects Registry from 2007 to 2017(October 2006 to September 2007 refers to 2007, and so on). All newborns (live or stillbirths aged 28 weeks of gestation or more) born to women lived in 64 counties across China for at least one year were recruited and followed until 42 days after birth. NTDs refers to anencephaly, spina bifida (with or without hydrocephaly) and encephalocele. All the cases were coded by International Classification of Diseases-10 (ICD-10).

**Results:** A total of 889 infants with NTDs were identified from 3,744,949 consecutive births during 2007-2017. The prevalence of anencephaly, spina bifida, encephalocele and overall NTDs was 0.54, 1.55, 0.29 and 2.37 per 10,000 respectively, with a decreasing trend. The reducing curve of the prevalence of spina bifida was sharper than anencephaly and encephalocele from 2007 to 2017. 69.30%, 39.65%, 55.14% and 48.25% of infants with anencephaly, spina bifida, encephalocele and overall NTDs were premature. The low-birth-weight proportion of the infants with anencephaly, spina bifida, encephalocele and overall NTDs was 72.77%, 35.52%, 52.33% and 46.01% respectively. The infants affected by NTDs experienced a relatively high mortality (72.33%), particularly for encephalocele cases (95.05%).

**Conclusions:** The downward tendency of NTDs in China indicates that prenatal prevention made great progress and should be further strengthened due to the severe birth outcomes.

Keywords: Neural tube defects, birth defects surveillance

### SIMULTANEOUS SYMPOSIUM 11 EPIDEMIOLOGIC APPROACHES IN THE STUDY OF PRETERM BIRTH AND BIRTH DEFECTS

#### **OP-36**

# Global, Regional, and National Levels and Trends in Congenital Birth Defects, 1990-2019 Wilner L.<sup>1\*</sup>, Kassebaum N.<sup>1</sup>

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**Introduction and objectives:** The Global Burden of Diseases, Injuries and Risks (GBD) 2017 study is a comprehensive effort to evaluate comparative health loss that quantifies mortality and morbidity from congenital birth defects globally for ages 0-69 years, by country, age, sex, and year.

**Methods:** Cause-of-death ensemble modeling (CODEm) tested all covariate combinations to maximize out-of-sample predictive validity of cause fraction and rate models informed by vital registration, surveillance, verbal autopsy, and CHAMPS/MITS data, all of which was corrected for completeness and misclassification. Nonfatal disease burden was modeled in DisMod-MR 2.1, a Bayesian meta-regression tool, for 29 different defect-specific models informed by registry data, ICD-coded administrative data, and published studies that were extracted and standardized to the reference case definition, including correction for compositional bias in some sources that only report a subset of defects.

**Results:** There were 5.296 million (95% CI: 4.953 – 5.703 million) babies born with congenital birth defects in 1990, and that was similar in 2017 with 5.445 million (95% CI: 5.088 – 5.844 million) babies born with congenital birth defects. This translates to a rate of approximately 3,866 per 100,000 live births (95% CI: 3,615 – 4,163 per 100,000) in 2017 and 3,922 per 100,000 live births (95% CI:3,615 – 4,163 per 100,000) in 2019. However, deaths due to congenital birth defects across all ages significantly decreased from 826,877 (95% CI: 743,550 – 996,616) in 1990 to 584,861 (95% CI: 556,252 – 618,250) in 2017. Accordingly, prevalence of congenital birth defects for all ages has increased from 74.80 million people (95% CI: 70.99 – 79.12 million) in 1990 to 100.4 million people (95% CI: 95.15 – 106.5 million) in 2017. Among the sub-causes of congenital birth defects, congenital musculoskeletal defects have been the most prevalent, and congenital heart defects have been responsible for the most deaths.

**Conclusions:** While incidence of congenital birth defects has remained relatively constant, there has been a dramatic increase in the number of people living with congenital birth defects. This necessitates increased support services for these individual living with birth defects around the world.

Keywords: congenital birth defects, global burden of disease, health metrics, disability

# Bronchopulmonary Dysplasia, other Comorbidities, and Mortality Rates Associated with Extremely Premature Births in a Large Medical Records Database in the United States Siffel C.<sup>1\*</sup>, Sarda S.P.<sup>1</sup>, Chen H.<sup>2</sup>, Ferber J.<sup>2</sup>, Kuzniewicz M.W.<sup>2</sup>, Li D-K.<sup>2</sup>

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**Introduction and objectives:** Extremely premature (EP) births (<28 weeks gestational age [GA]) account for ~5% of preterm (<37 weeks) births globally. Its complications are recognized as a leading cause of deaths in children under 5 years of age. Incidence estimates for bronchopulmonary dysplasia (BPD) among EP infants ranges 10–89% (Europe 10–73%; North America 18–89%; Asia 18–82%). This study aims to assess prevalence and mortality rates of EP births in the US, and rates of comorbidities associated with EP births namely BPD, intraventricular hemorrhage [IVH], and retinopathy of prematurity [ROP].

**Methods:** This was a population-based cohort study of liveborn EP infants in the electronic medical record database of Kaiser Permanente Northern California between January 1997 and December 2016. Newborns with major congenital malformations were excluded. Endpoints included prevalence of EP births, comorbidities identified during birth hospitalization and/or up to 2 years of age, and mortality/survival rates at 2 years of age.

**Results:** From 1997–2016, 679,159 total births were recorded, and 59,217 infants (8.7%) were born preterm. Of those, 5588 (9.4%) had major birth defects and were excluded. A total of 2154 (0.32%; 95% CI: 0.30, 0.33) infants were EP. BPD was present in 34.3%, IVH in 22.7%, ROP in 37.1%; and 26.1% had one comorbidity, 30.9% had  $\geq$ 2 comorbidities, while 43.0% survived with no comorbidities during birth hospitalization. Survival rates increased with increasing GA at birth. Survival rate of infants born at 23 weeks GA was 7.6% (95% CI, 4.80–11.17), which increased to 91.7% (88.90–93.83) for those born at 27 weeks GA. The overall cumulative mortality rate increased from 29.5% at  $\leq$ 1 day of age (all events with or without active resuscitation) to 42.5% at 2 years of age in EP infants. The cumulative mortality rate among EP infants with a BPD or an IVH diagnosis (with/without other comorbidity) was 9.1% and 29.9%, respectively.

**Conclusion:** This retrospective analysis demonstrated the burden of comorbidities that occur in EP infants and the high mortality associated with EP births. Funded by Takeda.

**Disclosures:** Csaba Siffel and Sujata P Sarda are employees of and hold shares in Takeda. H. Chen, J. Ferber, M Kuzniewicz, D-K Li are employees of Kaiser Permanente Northern California, which was contracted through Shire, a Takeda company, to perform this study.

#### OP-38 Epidemiological Profiling from Scientific First Principles - Progress and Prospects of the Modell Global Database of Congenital Disorders Darlison M.W.<sup>1\*</sup>, Modell B.<sup>1</sup>

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**Introduction and objectives:** The Modell Global Database (MGDb) grew out of work with WHO /Geneva in the early 1980s and has been supported more recently by the WHO Eastern Mediterranean Regional Office as a tool to support policy advocacy and benchmarking of existing services. It was the source of WHO epidemiological estimates for many years, and the basis of the 2006 March of Dimes report and of many of the estimates contained within the Born healthy health needs assessment toolkit for congenital disorders. This presentation will provide an update on progress since the last ICBD in Bogota, Colombia.

**Methods:** The MGDb uses its published methods, many of which are novel, to integrate data from many sources to generate country-level estimates of adverse birth outcomes, birth prevalence, early mortality and disability associated with early-onset congenital disorders that predispose to death or significant disability in the absence of care. Informatics methods are now under development (a) to improve access to the raw data in tabular and visual forms and (b) to support adaptation and application of the approach at national and sub-national levels.

**Results:** Having published the underlying methods, the project is now moving on to explore uses of the estimates, particularly in relation to (a) overcoming inertia in the policymaking process and (b) benchmarking existing surveillance systems, particularly in settings where complete ascertainment may be difficult or impossible.

**Conclusions:** The availability of global-, regional- and country-level data supplemented by country profile data is a first step towards mobilising the outputs of the MGDb for practical use. **Keywords:** Epidemiology, congenital disorders, birth defects, policy, services.

Prevalence and Mortality among Infants with Congenital Diaphragmatic Hernia: A Multi Registry Analysis

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**Introduction and Objectives:** Congenital diaphragmatic hernia (CDH) is a severe birth defect characterized by an incompletely formed diaphragm with a high mortality rate. The aim of this study is to examine the global prevalence and mortality trends associated with CDH.

**Methods:** Twenty-five hospital- and population-based surveillance programs in 19 countries from members of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) provided data for a birth defects surveillance study between the years 1974 and 2015. Participants were cases with CDH that resulted in live births, stillbirths, or elective termination of pregnancy for fetal anomalies. Prevalence and Kaplan-Meier mortality rates were calculated for all births by type of registry. Mortality was examined by isolated and non-isolated case status (multiple congenital anomalies and syndromic cases combined) for the programs where data was available.

**Results:** The total prevalence of CDH was estimated to be 2.7 per 10,000 total births (95% Confidence Interval: 2.6, 2.8) between years 1974 and 2015. The overall cumulative percent mortality of CDH was 37.2%, with hospital-based registries having a higher number of live births with CDH result in death than population-based registries (45.1% compared to 33.6%). The majority of deaths due to CDH occurred among infants aged 2 to 6 days for both registry types (36.3% for hospital-based and 12.1% for population-based). Multiple and syndromic cases of CDH had higher one-week mortality rates (44.9%) than isolated cases (28.4%) for both hospital- and population-based registries.

**Conclusions:** The global prevalence of infants with CDH remains high, with an elevated mortality rate, especially during the first week of life. Further research is needed to examine the differences between population- and hospital-based registries and the 'hidden mortality' that might be present.

Keywords: Congenital diaphragmatic hernia, prevalence, mortality, international, registry

Population Medical Genetics Approach in Clusters of Muco-Polysaccharidoses Identified in Latin America

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**Introduction and objectives**: The Mucopolysaccharidoses (MPS) are 11 rare genetic disorders caused by deficiency of specific lysosomal enzymes resulting in the accumulation of undegraded glycosaminoglycans (GAGs), leading to several clinical consequences. The combined incidence for all MPS subtypes is approximately 1:25,000 live births. Clusters of these diseases have been identified in areas with high consanguinity rates and/or founder effect associated to endogamy. Methods: The MPS Brazil Network, associated to the Brazilian Institute of Population Medical Genetics (INAGEMP), identified several MPS clusters in Latin America and investigated them by biochemical and molecular analyses. Enzyme assays and DNA extraction for molecular analyses were performed mainly in dried blood spot samples. **Results**: Three clusters were confirmed in Brazil, of MPS IIIC (state of Paraíba), MPS IVA (state of Paraíba) and MPS VI (state of Bahia). Two clusters were identified in Ecuador: MPS IIIB (state of Manabi) and MPS IVA (state of Pastaza). A cluster of MPS VI was also identified in the Dominican Republic. Other clusters are being investigated in Haiti (MPS VI), Panamá (MPS IVA), and Brazil (MPS IIIB, Minas Gerais state). Haplotype analyses are underway to better characterize mutation profiles, and results already available for the clusters of MPS IVA and MPS VI in Brazil indicate founder effects with common ancestors. As one example of the benefits of cluster identification, a newborn screening program for MPS VI was implemented in the specific Brazilian region in order to provide early identification and treatment of affected newborns. Measures to increase awareness of the community, provide training to health care personnel, as well as genetic counseling and prenatal diagnosis, could also be offered.

**Conclusions**: Examples of MPS clusters were identified in Latin America and it is likely that several others are still unreported. The identification and characterization of MPS clusters provides a better understanding of how they were originated and enable to provide preventive and management measures to the affected communities.

**Keywords**: Population medical genetics; mucopolysaccharidoses; clusters; lysosomal storage disorders; inborn errors of metabolism

# Spina Bifida Phenotypes and Sex-Specific Prevalence among Offspring of Folic Acid Users Moore C.A.<sup>1\*</sup>, Patimeteeporn C.<sup>2</sup>, Yeung L.F.<sup>1</sup>, Crider K.<sup>1</sup>, and BerryR.J.<sup>1</sup>

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**Introduction and Objectives:** Periconceptional consumption of folic acid (FA) can reduce the risk of a neural tube defect (NTD)-affected pregnancy; however, information about the effect of FA on sex-specific or phenotype-specific spina bifida (SB) prevalence is still emerging. Data from the China-U.S. NTD Prevention Project were used to evaluate these effects in a community intervention trial conducted in northern China, a high prevalence area.

**Methods:** From 1993 through 1995, women were asked to take a daily pill containing 400µg FA alone. Birth defects were ascertained by a population-based surveillance system. SB lesion level was estimated using anatomic landmarks on photographs. Periconceptional use of FA was based on monthly records of supplement use. Birth prevalence estimates for SB were calculated and compared across FA supplement users and non-users and by sex in addition to prevalence of high (cervicothoracic) vs. low (lumbosacral) level of SB. The association between level of the lesion and FA exposure among the entire cohort was examined in an adjusted regression model.

**Results:** Among 21,536 women who took FA at any time before and during early pregnancy and 56,541 women who did not take FA, the prevalence of SB was 7.9 per 10,000 pregnancies and 19.3 per 10,000, respectively (risk reduction ~59%, p<0.01); SB prevalence among female offspring was 8.8 and 24.2 per 10,000 pregnancies, respectively (risk reduction ~64%, p<0.01), and the prevalence among male offspring was 8.0 and 17.0 per 10,000, respectively (risk reduction ~53%, p<0.01). The prevalence of high SB lesions among offspring was 3.3 and 11.0 per 10,000 pregnancies, respectively (risk reduction ~70%, p<0.01), and prevalence of low SB lesions among offspring was 4.6 and 8.3 per 10,000 pregnancies, respectively (risk reduction ~45%, p=0.1).

**Conclusions:** These findings suggest that FA effect on neural tube closure is not limited to any one SB phenotype; however, it appears that the effect may differ by level of lesion and offspring sex. Examination of these variations in FA effects may be helpful in identifying factors important in the pathogenesis of SB and other neural tube defects.

Keywords: Folic acid, Neural tube defects, Spina bifida

### SIMULTANEOUS SYMPOSIUM 12 FOOD FORTIFICATION FOR PREVENTION OF BIRTH DEFECTS

#### **OP-42**

# High Potential for Reducing Folic Acid Preventable Spina Bifida and Anencephaly, and Related Stillbirth and Child Mortality in Ethiopia Kancherla V.<sup>1\*</sup>, Dixon M.<sup>1</sup>, Magana T.<sup>2</sup>, Mulugeta A.<sup>3</sup>, Oakley, Jr G.P..<sup>1</sup>

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**Introduction and Objectives:** Spina bifida and anencephaly are two severe and often fatal birth defects of spine and brain and associated with high perinatal and postnatal mortality, especially in resource-poor settings. There is evidence that a majority of cases of spina bifida and anencephaly can be prevented by ensuring adequate maternal folic acid intake before and early after conception. Recent studies in Ethiopia show a profoundly large epidemic of spina bifida and anencephaly. Reductions can be achieved in stillbirth and child mortality rates in Ethiopia by preventing folic acid-preventable spina bifida and anencephaly through effective interventions.

**Methods:** We estimated percent reductions in stillbirth, neonatal, infant, and under-five child mortality rates that would have occurred in Ethiopia in the year 2016 had all folic acid-preventable spina bifida and anencephaly been prevented. We also estimated the contributions of these reductions toward Ethiopia's Year 2030 Every Newborn Action Plan (ENAP)goal on stillbirth, and Sustainable Development Goal (SDG) on child mortality rates. Based on recent data from Ethiopia, we considered the current prevalence of spina bifida and anencephaly in Ethiopia to be 13 per 1,000 total births, and the preventable rate to be 12.5 per 1,000 total births.

**Results:** Folic acid interventions would have prevented about 41,610 cases of folic acidpreventable spina bifida and anencephaly-affected pregnancies and a related 31,830 stillbirths and 7,335 under-five child deaths. These reductions contribute to the reduction in stillbirths needed to achieve Ethiopia's ENAP goal addressing stillbirth and reductions in neonatal and under-five mortality, needed to achieve the SDG addressing preventable child mortality.

**Conclusions:** Primary prevention of spina bifida and anencephaly can contribute to timely reductions in current stillbirth, neonatal and under-five mortality rates in Ethiopia. Effective folic acid interventions, including fortification of wheat and maize flour and salt, along with folic acid pill supplementation program, have a potential for controlling the epidemic and achieving reductions in stillbirth and child mortality associated with birth defects in Ethiopia, and contribute towards reaching SDG on preventable child mortality.

**Keywords:** Anencephaly; Mortality; Neural Tube Defects; Spina Bifida; Sustainable Development Goals

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Folate and Vitamin B12 Status in Women of Reproductive Age in Rural Areas in Haryana, Northern India

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**Introduction and Objectives:** Maternal folate insufficiency and vitamin B12 deficiency are known risk factors for neural tube defects. We conducted a biomarker survey to assess the baseline prevalence of folate deficiency and insufficiency and vitamin B12 deficiency in women of reproductive age prior to the start of a fortification program in Haryana, India.

**Methods:** A multi-stage cluster probability biomarker survey was conducted. Participants were non-pregnant women of reproductive age (18-49 years) who resided in rural areas of two subdistricts in Ambala District in Haryana. Venous blood samples were collected among 866 women. Plasma, serum, and red blood cells (RBC) were separated by centrifugation, processed, and stored at <-80°C until analysis. RBC and serum folate concentrations were measured using microbiologic assay and serum vitamin B12 was measured via chemiluminescence. Serum folate deficiency was defined as serum folate <7 nmol/L; RBC folate deficiency and insufficiency were defined as RBC folate <305 nmol/L and <748 nmol/L, respectively. Vitamin B12 deficiency was defined as vitamin B12 <200 pg/mL and vitamin B12 marginal deficiency was defined as vitamin B12  $\geq$ 200 and <300 pg/mL. We calculated geometric mean concentrations for serum and RBC folate and serum vitamin B12, and prevalence for serum folate deficiency, RBC folate deficiency, and vitamin B12 deficiency.

**Results:** Geometric mean concentrations for serum folate, RBC folate, and serum vitamin B12 were 12.3 (95% CI: 11.8, 12.9) nmol/L, 544 (95% CI: 516, 573) nmol/L, and 190 (95% CI: 176, 206) pg/mL, respectively. Prevalence of folate deficiency was 11.3% (95% CI: 9.2, 13.9) for serum folate and 9.7% (95% CI: 7.8, 12.0) for RBC folate, and prevalence of RBC folate insufficiency was 78.6% (95% CI: 74.8, 82.5). A total of 58.3% (95% CI: 54.2, 62.5) of women were vitamin B12 deficient (<200 pg/mL) and 22.9% (95% CI: 19.7, 26.1) were marginally deficient for vitamin B12.

**Conclusions:** The magnitude of folate insufficiency and vitamin B12 deficiency in this Northern Indian population is a substantial public health concern. The findings from the survey help establish the baseline for a planned fortification program aimed at reducing these micronutrient deficiencies.

Keywords: Folate, Folic acid, Vitamin B12, micronutrient deficiencies, fortification

### Awareness and Factors Associated with Reported Intake of Folic Acid-Fortified Flour among Women of Reproductive Age in Ifakara, Morogoro Region, Tanzania: A Cross-Sectional Study

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Introduction and objectives: Folic acid fortification of staple foods has been in place in many countries for over two decades. Studies have shown that folic acid fortification can significantly reduce the incidence of neural tube defects. Tanzania adopted a mandatory fortification policy for commercially-produced wheat and maize flour in 2011. We determined factors influencing the intake of folic acid-fortified flour among women of reproductive age (WRA).

Methods: We conducted a cross-sectional study among WRA during March-April 2017 in Ifakara Town Council, Morogoro Region. Multistage cluster sampling was used to select study participants. We used a questionnaire to capture information on demographics, awareness of folic acid, awareness of existence of folic acid fortified flour in community and intake of folic acid fortified flour. Intake was defined as reported consumption of folic acid fortified flour products at least once within seven days before interview. Univariate, bivariate, and multivariable logistic analyses were done to evaluate factors associated with the intake of folic acid fortified flour.

**Results:** The median age of the 698 participating WRA was 30 years (range: 18-49). Awareness of folic acid and folic acid fortified flour was 6.9% (95% CI: 5.2%-9.0%) and 7.5% (95% CI: 5.7%-9.6%). respectively. Consumption of folic acid fortified flour was 63.3% (95% CI: 59.7%-66.8%). Independent factors associated with intake included being employed (adjusted odds ratio (aOR)=1.91; 95% CI: 1.19-3.06), having no children (nulliparity) (aOR=2.59; 95% CI: 1.36-4.95) or having 1-4 children (aOR=1.98; 95% CI: 1.17-3.33) and folic acid awareness (aOR=2.53; 95% CI: 1.30-4.92).

**Conclusions:** Folic acid-fortified flour was used by most respondents in our study despite low awareness of the existence of folic acid fortified flour in the community. Being employed, having fewer than five children and folic acid awareness were independent factors associated with intake. We recommend doing further studies on blood folate level among women of reproductive age in Ifakara to assess fortification program effectiveness.

Keywords: Intake, folic acid, fortification, flour, women of reproductive age

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#### OP-45 Potential Level of Global Prevention of Neural Tube Defects with Folic Acid-Fortified Salt Kancherla V.<sup>1\*</sup>, Tsang B.<sup>2,3</sup>, Wagh K.<sup>4</sup>, Dixon M.<sup>1</sup>, Oakley, Jr. G.P.<sup>1</sup>

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**Introduction and Objectives:** Mandatory fortification of staples like wheat and maize with folic acid is effective in preventing folic acid-preventable spina bifida and anencephaly (FAP SBA) by providing women of reproductive age with adequate folic acid before and early after conception. Our objective was to estimate the proportion of FAP SBA that could be prevented worldwide provided the mandatory fortification of salt with folic acid is implemented in countries where there is a high coverage of iodine-fortified salt.

**Methods:** Using existing data sources for the year 2017, we identified countries with information on the percentage of households consuming iodized salt and on average salt intake. We assumed mandatory folic acid fortification of salt at 20 ppm and that at least 150 mcg of folic acid per day fully protects against FAP SBA, reducing the prevalence of spina bifida and anencephaly to 0.5 per 1,000 live births from their current high prevalence. We modeled the proportion of FAP SBA that could be prevented annually through mandatory fortification of salt with folic acid.

**Results:** A total of 100 countries had data for our analysis, of which 37 were implementing mandatory folic acid fortification of wheat or maize. The remaining 63 countries had a high potential for salt fortification with folic acid, where the majority of households consumed iodized salt. We estimated that approximately 180,000 cases of FAP SBA can be prevented through mandatory fortification of salt with folic acid. Globally, a high proportion of FAP SBA (64%) can be prevented annually by fortifying salt with folic acid. Countries in Africa and Asia can utilize their existing salt fortification-infrastructure for FAP SBA prevention.

**Conclusions:** Primary prevention of FAP SBA through iodine and folic acid fortified salt can address current gaps in prevention in countries where wheat or maize flour are not consumed as staples or infrastructure for centrally processed cereal grain products does not exist. Salt fortification with folic acid can contribute to timely reductions in current stillbirth, neonatal and under-five mortality rates in countries associated with FAP SBA, and contribute towards reaching Sustainable Development Goals on preventable child mortality and disability.

Keywords: Folic Acid, Fortification, Neural Tube Defects, Salt, Spina Bifida

#### SIMULTANEOUS SYMPOSIUM 15 REDUCING RISK FACTORS FOR ADVERSE BIRTH OUTCOMES

#### **OP-46**

Changing Spectrum and Risk Factor Association in Newborn Babies with Birth Defects in A Tertiary Care Hospital in North India Gupta S.<sup>1\*</sup>, Chellani H.K. <sup>1</sup>, Gupte N.<sup>1</sup>, Raina N.<sup>2</sup>, Mehta R.<sup>2</sup>

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**Introduction and Objectives:** To determine the spectrum of birth defects and association of maternal and neonatal risk factors in live babies born with birth defects

**Methods:** Hospital based, case control study at the NICU, VMMC & SJH, New Delhi All babies born with structural birth defect in the hospital over a 12 month period were included. The study group (n=297) consisted of babies born with a structural birth defect/s while control group (n=594) of two live healthy babies born immediately after the study case. All cases were followed till discharge/death in the hospital. Outcome was measured for the presence of risk factors associated with occurrence of birth defects from the maternal history as per the predesigned proforma and neonatal clinical examination. Quantitative data was analysed by using SPSS version 14.0 and qualitative by Chi square test/Fisher exact test. P value less than 0.05 was considered significant.

**Results:** Out of 28,504 births, 27,652 were live births and 852 were still births. Prevalence of birth defects among live births was 1.07%. Out of the total defects, congenital heart disease and related birth defects were highest (18.4%) as compared to 11.8% in previous year, followed by defects of the nervous system (15%) and digestive system (8.58%).

Among the risk factors studied, maternal hypothyroidism (p=0.002, OR=3.33), obesity (p=0.027, OR=3.06), maternal fever (p=0.002, OR=5.14), smoking exposure during pregnancy (p=0.006, OR=2.1), poor antenatal care (p=0.0001) were significantly associated antenatal factors. Significant association was also seen with male gender (p=0.0001), preterm baby <34 weeks (p= 0.0001, OR= 11.84), small for gestational age baby (p=0.00001, OR=2.18 and with instrumental delivery (p=0.0001, OR=22.02). However, no association was seen with respect to multiple pregnancy or with IVF conception.

**Conclusions:** The study shows the change in the spectrum of predominant birth defects from neutral tube defects to congenital heart disease. The higher incidence of Congenital heart disease was noted because of the improved availability of diagnostic facilities in our hospital in the last one year. This study identifies some of the modifiable risk factors associated with birth defects and indicates the importance of good antenatal care to address such risk factors thereby preventing some of the birth defects.

Keywords: birth defects, risk factors, newborn

#### OP-47 Pattern and Risk Factor Associated with Congenital Anomalies in Newborn Babies Born in Dhulikhel Hospital Singh S.<sup>1\*</sup>

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Introduction and objectives: Birth defects are defined as structural and functional defects that develop during the organogenesis periods and present at birth or detected later in life. They are an important cause of morbidity and mortality in infants. The known causes of birth defects are mostly genetic effect modified by environmental factors which may be prevented. The main objective of the study was to examine patterns and the association of certain risk factors with birth defects occurring in newborns and infants seeking care in Dhulikhel Hospital, Kavre, Nepal. **Methods:** This is a hospital based, prospective, cross sectional study involving 219 newborns admitted in the Dhulikhel Hospital, Kathmandu University Hospital. We enrolled infants admitted at Neonatal intensive care unit, Neonatal ward, and neonates staying with mother in post natal wards, from 1stJanuary 2015 to 31stDecember 2017. Data were entered and analyzed using SPSS, version 23. The association between congenital anomalies and socio-demographics were assessed applying binomial logistic regression analysis for the study and reference group and chi square test was done between the studv and reference group. **Results:** The most frequent birth defect was cardiovascular systems 29(26.8%) followed by musculoskeletal system 17(15.7%) and face 15(13.8%). Less than 4 in number of Antenatal visit (p=0.017), not using folic acid during early pregnancy (p<0.001) and low socioeconomic conditions (p<0.001) were significantly associated with congenital malformations. Among all congenital anomalies complex congenital heart disease had poor outcome.

**Conclusion:** The most common congenital defects involved cardiovascular and musculoskeletal system. Poor antenatal visit, lack of folic acid during peri conceptional age and low socioeconomic conditions were the most common risk factors associated with congenital anomalies of this study sample.

Keywords: Congenital anomalies, Risk factors, Newborn

**OP-48** 

Specific Birth Defects in Pregnancies of Women with Diabetes - National Birth Defects Prevention Study, 1997-2011

Tinker S.C.<sup>1</sup>, Gilboa S.M.<sup>1</sup>, Lakmini S.P.R.V.<sup>1\*</sup>, Priyani A.A.H.<sup>1</sup>, de Silva M. V. C.<sup>1</sup>, Moore C.A.<sup>1\*</sup>, Waller D.K.<sup>2</sup>, Simeone R.M.<sup>1</sup>, Kim S.Y.<sup>1</sup>, Jamieson D.J.<sup>3</sup>, Botto L.D.<sup>4</sup>, Reefhuis J.<sup>1</sup>, and the National Birth Defects Prevention Study

<sup>1</sup>National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, GA, USA; <sup>2</sup>UTHealth, School of Public Health, The University of Texas Health Science Center at Houston, Houston, TX, USA; <sup>3</sup>Department of Gynecology and Obstetrics, Emory University School of Medicine. Atlanta, GA, USA; <sup>4</sup>Division of Medical Genetics, Department of Pediatrics, University of Utah School of Medicine, Salt Lake City, UT, USA \*Correspondence: cam0@cdc.gov

**Introduction and Objectives:** Diabetes is associated with an increased risk for many birth defects and is likely to have an increasing impact on birth defect prevalence due to the global rise in diabetes in recent decades. Strong associations for pregestational diabetes with several birth defects were previously reported from the initial six years of the National Birth Defects Prevention Study (NBDPS), but few exposures among some of the less common birth defects led to unstable estimates with wide confidence intervals. Our objective was to use data augmented from additional years of NBDPS to provide updated and more precise estimates of the association between diabetes and birth defects, including some defects not previously assessed.

**Methods:** We analyzed data on deliveries from October 1997 through December 2011. Mothers of case and control infants were interviewed about their health conditions and exposures during pregnancy, including diagnosis of pregestational (type 1 or type 2) diabetes before the index pregnancy or gestational diabetes during the index pregnancy. Using logistic regression, we separately assessed the association between pregestational and gestational diabetes with specific categories of structural birth defects for which there were at least three exposed case infants – with at least five exposed case infants, we calculated adjusted odds ratios (ORs); with three or four exposed cases, crude ORs.

**Results:** Pre-gestational diabetes was reported by 0.6 percent of mothers of control infants (71 / 11,447) and 2.5 percent of mothers of case infants (775 / 31,007). Gestational diabetes during the index pregnancy was reported by 4.7 percent of mothers of control infants (536 / 11,447) and 5.3 percent of mothers of case infants (1,653 / 31,007). Pre-gestational diabetes was associated with strong, statistically significant ORs (range: 2.5 to 80.2) for 46 of 50 birth defects considered; for gestational diabetes, statistically significant ORs were fewer (12 of 56) and of smaller magnitude (range: 1.3 to 2.1).

**Conclusions:** Pre-gestational diabetes is associated with markedly increased risk for many specific birth defects. Because glycemic control before pregnancy is associated with a reduced risk for birth defects, preconception care is an important prevention opportunity.

Keywords: Birth defects, Diabetes, Pregnancy

#### OP-49 Prevalence, Pattern and the Association of Major Structural Congenital Anomalies with Maternal and Fetal Factors at the Neonatal Care Unit (NICU/SCBU) of a Tertiary Hospital in Colombo, Sri Lanka Hewavitharana H.<sup>1\*</sup>, Perera S.<sup>1</sup>

<sup>1</sup>Neonatology Unit, Castle Street Hospital for Women, Colombo, Sri Lanka \*Correspondence: hewavitharanahasani@gmail.com

**Introduction and objectives:** Congenital malformations are structural or functional anomalies that are present at the time of birth and represent defects in morphogenesis during early fetal life. In Sri Lanka, congenital anomalies are the second leading cause of deaths among babies. This study was done to assess the prevalence, pattern and association of maternal and fetal factors on clinically detectable major structural congenital malformations, among live babies delivered at a Tertiary Care Hospital in Sri Lanka.

**Methods:** All live babies born with major structural congenital anomalies, over a period of one year, at Neonatal Unit, Castle Street Hospital for Women, Sri Lanka, were included in this retrospective observational study. Diagnosis of congenital anomalies was based on clinical evaluation and complementary studies. A data collection tool was used to collect sociodemographic data, risk factors associated with congenital malformations, from maternal and neonatal records and analysed with Statistical Software.

**Results:** There were 178 babies with structural malformations among 11653 deliveries and 11656 live births, with a prevalence of 1.53%. Majority (71%) of the babies were born to mothers between 20 to 35 years of age. Highest frequency of congenital anomalies occurred in the cardiovascular system (42%) followed by musculoskeletal system (15%). Involvement of multiple systems was seen in 46 babies. Of the mothers, 1% had a previous baby with malformation and 15% had a previous abortion. Statistical significance was seen in plurality, birth weight and maturity. No statistical significance seen in parity, sex of the baby and the presence of medical conditions such as Diabetes and Hypertension.

**Conclusions:** The prevalence of major structural anomalies in the study sample was within values reported in literature. Heart defects was the most prevalent anomaly detected and there was a significant association with birth weight, maturity and plurality. Emphasis should be made on the prevention, awareness and evaluation of high-risk populations by early prenatal diagnosis of congenital anomalies. Public health actions should also be strengthened to improve the wellbeing of affected babies.

**Keywords:** Structural malformations, birth defects, congenital anomalies, fetal factors, maternal factors

#### **OP-50**

#### Pattern of Congenital Malformations and Associated Risk Factors among Neonates Admitted at Kenyatta National Hospital, Kenya Machogu E.M.<sup>1\*</sup>, Musoke R.<sup>1</sup>, and Aluvaala J.M.<sup>1</sup>

<sup>1</sup>Department of Pediatrics and Child Health, University of Nairobi, Kenya \*Correspondence: evemachogu@gmail.com

Introduction and Objectives : Congenital malformations are structural or functional abnormalities that may be detected prenatally, at birth or later in life. They are a major cause of neonatal and child mortality, disability and chronic illness. There are few local studies on the subject and Kenya, a developing country, also lacks a surveillance system for birth defects. This study aimed to describe the structural defects and associated risk factors among neonates admitted at Kenvatta National Hospital(KNH), a public tertiary referral hospital that offers specialist neonatal care in Kenya. The objectives of this paper were to describe the types and frequency of structural congenital malformations among neonates admitted to the newborn unit and pediatric medical wards at Kenyatta National Hospital and to evaluate associated risk factors. Methods: A cross sectional descriptive survey of all newborns with and without congenital malformations was carried out. Consecutive sampling was done till the sample size was achieved. 300 newborns were recruited at admission upon consent from their mothers. The details of physical examination of the babies and diagnostic imaging done were recorded in a data abstract tool. Mothers were interviewed to obtain information on parental sociodemographic/biophysical characteristics and risk factors for birth defects.

**Results:** Of the 300 newborns studied, 68 were found to have various congenital malformations. The most common malformations were congenital heart defects (27%) followed by gastrointestinal defects (21%) and neural tube defects (17%). Congenital malformations were a major reason for referral and admission to KNH. Maternal HIV status (OR=3.20; 95%CI, 1.16-8.83, p=0.025) and late introduction of multivitamin use in pregnancy (COR=13.71, 95%CI, 1.53-122.69, p=0.019) were the most significant associated factors.

**Conclusions:** Birth defects are a major contributor to neonatal morbidity. About 22.6% of the babies examined had various malformations. The data gained from this study can be used to start a birth defects register in the hospital and information on risk factors could also be used in prevention of these defects. A birth registry would help in surveillance of congenital anomalies and follow up of affected children.

Keywords: Congenital malformations, associated risk

### **POSTER SESSIONS**

#### SESSION 1 – POSTER STATION 1 24 FEBRUARY 2019: 11.00 AM – 11.30 AM

Chairperson: Lahiru Prabodha and Helen Dolk

PP-01: A Case Series of Three Sri Lankan Families with Rare Autosomal Recessive Congenital Neuromuscular Disorders Identified Through Exome Sequencing

PP-02: Structural Anomalies of Feto-Infant Deaths: An Autopsy Study at a Tertiary Care Center

PP-03: Effectiveness of Early Intervention for Neonates at Risk of Future Motor Deficits, in a Multi-Disciplinary Local Paediatric Clinic in a Tertiary Care Hospital, Sri Lanka

PP-04: Analysis of Polymorphisms in Osteopontin Gene and Risk of Urolithiasis: A Case-Control Study and Meta-Analysis

PP-05: Analysis of the GJB2 Gene and its Mutated Protein in Syndromic Hearing Loss Patients of Gilgit-Baltistan

PP-06: Association Between Genetic Variants of MTR Gene and Congenital Heart Disease: Evidence from a Family-Based Case-Control Study in Chinese Population

#### SESSION 2 – POSTER STATION 2 24 FEBRUARY 2019: 11.00 AM – 11.30 AM

Chairperson: Dilhani Samarasekara and Joyce Namale Matovu

PP-07: Barriers Facing Youths in Accessing Youth-Friendly Sexual and Reproductive Health Services in Tanzania: A Case Study from Kilimanjaro Christian Medical University College

PP-08: Birth Defects and Small for Gestational Age (SGA) Status is There a Link?

PP-09: Birth Defects Surveillance in Lebanon: Who are the Most Affected Neonates?

PP-10: Candidates for Prenatal Prediction of Adverse Outcome of Pregnancy: Cytokines and Chemokines Shared among Spontaneous Miscarriages, Stillbirths, and Preterm Births with Transcriptomic Approach

PP-11: Center–Based Statistics of Cleft Lip, Alveolus and Palate Patients in Aden, Yemen

PP-12: Characteristics of Neonates who are Undergoing Early Interventions for Future Neurodevelopmental Deficits at Colombo-South Teaching Hospital, Sri Lanka

#### SESSION 3 – POSTER STATION 3 24 FEBRUARY 2019: 4.00 PM – 4.30 PM

Chairperson: Sajith Edirisinghe and Matthew Darlison

PP-13: Clinical Description of Maternal Infections During Pregnancy and Congenital Defects in Their Offspring in Bogotá and Cali, Colombia 2001-2018

PP-14: Clinical Spectrum of Congenital Anomalies and its Contributing Factors among Infants Presenting to a Tertiary Care Hospital

PP-15: Comparison of Implementation Status of Revised Antenatal Care Guidelines in Matara District During 2015 and 2019.

PP-16: Congenital Surgical Malformation in Gabriel Touré Teaching Hospital in Bamako, Mali: Epidemiological and Clinical Aspects

PP-17: Congenital Unilateral Hypoplasia of Depressor Angularis Oris - A Rare Case

PP-18: Association Between Children Diagnosed with Autism Spectrum Disorder with Sensory Difficulties and Determinants oif Mother's Reduced Mobility During Pregnancy- A Pilot Study at a Tertiary Care Institution, Sri Lanka

#### SESSION 4 – POSTER STATION 4 24 FEBRUARY 2019: 4.00 PM – 4.30 PM

Chairperson: Piyara Rathnayake and Tahmina Banu

PP-19: Developmental Neurotoxicity Potential Through Chronic Exposure to Dietary Lead in CKD Hotspots in Sri Lanka

PP-20: Effectiveness of Health Education Handbook Designed to Increase Knowledge on Birth Defects among Public Health Midwives in Southern Province of Sri Lanka

PP-21: Epidemics of Spina Bifida and Anencephaly in Tigray, Northern Ethiopia: Hospital-Based Studies.

PP-22: (Epi)Genetic Variants of the Sarcomere-Desmosome are Associated with Premature Utero-Contraction in Spontaneous Preterm Birth

PP-23: Exploring Perinatal Mental Health in Sri Lanka: A Brief Needs Assessment

PP-24: Factors Related to Growth of the Infants with Structural Birth Defects in Galle, Sri Lanka

#### SESSION 5 - POSTER STATION 1 25 FEBRUARY 2019: 11.00 AM - 11.30 AM

Chairperson: Navoda Atapattu and Symone Detmar

PP-25: First Trimester Traditional Medication Use in Pregnancy in Cameroon: A Multi-Hospital Survey

PP-26: Frequency of Cardiac & Neural Congenital Anomalies: Findings from a Cross Sectional Study at Children Hospital, Lahore

PP-27: Gastrointestinal and Genitourinary Birth Defects in Chittagong, Bangladesh: Results From an Urban and a Rural Center

PP-28: Genetic Analysis of Children with Congenital Ocular Anomalies in Three Ecological Regions of Nepal.

PP-29: Health Barriers Detected in Patients with Congenital Anomalies That Affect Hearing or Vision

PP-30: Hospital Based Surveillance of Congenital Malformations at a Tertiary Care Centre in North India: An Initial Experience

#### SESSION 6 – POSTER STATION 2 25 FEBRUARY 2019: 11.00 AM – 11.30 AM

Chairperson: Nishani Lucus and Scott Montgomery

PP-31: Hospital Based Surveillance on Birth Defects in Nyamira County and Referral Hospital

PP-32: Incidence of Low Weight at Birth and Stunting at Childhood: A Case Study of Malda District, India

PP-33: Integrating Rare Diseases in the Public Health Delivery System Through Legislation

PP-34: International Classifications as a Tool for Interoperability in Child Health

PP-35: Major Congenital Anomalies Associations Through Graph Theory

PP-36: Major Congenital Anomalies: A Retrospective Regional Study in Three Hospitals in Cali – Colombia

#### SESSION 7 – POSTER STATION 3 25 FEBRUARY 2019: 4.00 PM – 4.30 PM

Chairperson: Asiri Hewamalage and Paula Hurtado

PP-37: Management of Craniofacial Clefts

PP-38: Nutrition Status of Children with Cerebral Palsy: Usefulness of New United States Cerebral Palsy Growth Chart

PP-39: Oral Health Care Package for High Caries Risk Children with Birth Defects

PP-40: Pioneering a Youth-Led Community-Based. Preconception Health Awareness Campaign Utilizing the LINC Framework: The Volunteer Youth Leaders for Health (VYLH): Philippines Experience

PP-41: Prevalence and Spectrum of Congenital Anomalies in a Tertiary Care Centre of North India: 20 Years' Experience

#### SESSION 8 – POSTER STATION 4 25 FEBRUARY 2019: 4.00 PM – 4.30 PM

Chairperson: Santhushya Fernando and Ignacio Zarante

PP-42: Prevalence of Renal Cystic Disease in 467,575 Births in the Surveillance Program of Congenital Defects Since 2003 to 2018 in Bogotá- Cali

PP-43: Prevalence of Talipes Equinovarus, Congenital Dislocation of the Hip, Cleft Lip/Cleft Palate, Down Syndrome and Neural Tube Defects among Live Newborns in Anuradhapura, Sri Lanka.

PP-44: Prevalence of Various Birth Defects in Northern India- A Multicentric New-Born Birth Defect Surveillance (NBBD)

PP-45: Prevalence, Risk Factors and Outcome of Congenital Birth Defects among Live Neonates in a Tertiary Care Hospital in North India

PP-46: Profile of Nutritional Status among Children with Cerebral Palsy in Tertiary Care Rehabilitation Center, Colombo, Sri Lanka.

PP-47: Quality of Life and Household Economic Burden of Primary Caregivers Having Children with Congenital Heart Disease Awaiting Cardiac Surgery Attending the Cardiology Clinic at the Lady Ridgeway Hospital for Children, Sri Lanka

PP-48: Referral Mechanism for Periconception Folic Acid Supplementation to Newly Married Couples to Prevent Neural Tube Defects

#### SESSION 9 - POSTER STATION 1 26 FEBRUARY 2019: 10.00 AM - 10.30 AM

Chairperson: Palitha Kumarapperuma and Nirmala Sirisena

PP-49: Relative Prevalence and Outcome of Antenatally Detected Congenital Malformations Using ICD -10 Codes in a Tertiary Care Hospital of India

PP-50: Role of Maternal Blood Oxidative Stress Markers and Inflammatory Markers in Unexplained Stillbirth: A Pilot Case-Control Study

PP-51: An Intervention to Improve the Health Status of Low-Income Women Living in Remote Hardest to Reach Areas of Uganda Before, During and after Pregnancy

PP-52: Surgical Care for Myelomeningocele and Congenital Hydrocephalus in a Large Urban Teaching Hospital in Ethiopia

PP-53: Synaptic Vesicle Associated Transcripts (SVATS) in Circulating Exosomes of Autistic Children and Physio-Pathological Pregnancies

PP-54: The Need for Domestic Violence Policies among Lebanese Mothers and Displaced Syrian Mothers.

#### SESSION 10 - POSTER STATION 2 26 FEBRUARY 2019: 10.00 AM - 10.30 AM

Chairperson: Sumithra Tissera and Nanbert Zhong

PP-55: Two Siblings with Autosomal Recessive Primary Microcephaly due to Novel Compound Heterozygous Null Variants in the ASPM Gene

PP-56: Use of Antenatal Corticosteroids for Threatened Preterm Birth: From Implementation Research to Policy Revision

PP-57: Validation of Two Parent-Reported Autism Spectrum Disorders Screening Tools M-CHAT-R and SCQ in Bamako, Mali.

PP-58: What Happens to Children with Birth Defects and Developmental Disabilities after They are Referred from the Community to Tertiary Care Centers by the Rashtriya Bal Swasthya Karyakram in India? A Follow Up Study

PP-59: Whole Exome Sequence Analysis of the Family Affected with Marfan Syndrome

**ABSTRACTS OF POSTER PRESENTATIONS** 

A Case Series of Three Sri Lankan Families with Rare Autosomal Recessive Congenital Neuromuscular Disorders Identified Through Exome Sequencing

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**Introduction and objectives:** Whole exome sequencing (WES) for the rapid and accurate diagnosis of rare undiagnosed neuromuscular disorders is now well established in the clinical setting. Herein, we report three Sri Lankan families with rare congenital neuromuscular disorders who were diagnosed using WES.

**Methods:** Trio WES was performed at the National Institutes of Health Intramural Sequencing Center using the Illumina TruSeq Exome Enrichment Kit and Illumina HiSeq 2000 sequencing instruments. Variants were filtered for 3 different segregation scenarios [dominant (de novo), recessive (homozygous and compound heterozygous)] using a customized Structured Query Language script with the following parameters: minimum allele frequency below 0.5% in the Exome Aggregate Consortium, Exome Variant Server databases and the laboratory's aggregate exome variant database with 587 exomes; Combined Annotation Dependent Depletion score above 20; and coverage above 10 reads.

**Results:** Family 1: novel likelv pathogenic homozygous missense variant [NM 001848:c.1667G>T;NP\_001839.2:p.Gly556Val] in COL6A1 gene, associated with Ullrich congenital muscular dystrophy-1[MIM#254090] in 2 girls from a consanguineous family, aged 7years and 10-years with progressive proximal myopathy and joint hyperlaxity. Family 2: pathogenic homozygous missense variant [NM\_003560.3:c.1799G>A;NP\_003551.2:p.Arg600Gln] in PLA2G6 gene, associated with infantile neuroaxonal dystrophy-1[MIM#256600] in a 3-yearold girl with global developmental delay and mixed upper and lower motor neuron lesions. 3: Family novel likely pathogenic homozygous missense variant [ENST00000368399:c.221A>G;p.Tyr74Cys] in DPM3 gene, associated with congenital disorder of glycosylation, type-I0[MIM#612937] in a 4-year-old boy with recurrent generalized seizures and myopathic features. The parents were heterozygote carriers for the respective variants in all three families.

**Conclusions:** Trio WES ended the diagnostic odysseys of three families with rare undiagnosed neuromuscular disorders. Establishing a precise diagnosis facilitated appropriate clinical management and accurate genetic counseling of the affected individuals.

Keywords: Myopathy, Neuromuscular, Undiagnosed, Variants

## Structural Anomalies of Feto-Infant Deaths: An Autopsy Study at a Tertiary Care Center Lakmini S.P.R.V.<sup>1\*</sup>, Priyani A.A.H.<sup>1</sup>, de Silva M. V. C.<sup>1</sup>

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**Introduction and Objectives:** Feto-infantal deaths are still a major public health concern despite the slower reduction in neonatal mortality and stillbirth rate. This study describes the pattern of structural anomalies in feto-infant deaths detected at autopsy and its correlation with selected fetal and maternal clinical parameters.

**Methods:** 522 autopsy reports, at the Department of Pathology, University of Colombo from 2007 to 2018 were retrospectively analyzed. Data were categorized under second trimester miscarriages(T2M), stillbirths, neonatal deaths (ND) and analyzed using SPSS and chi square test for associations.

**Results:** 156 T2M, 254 stillbirths and 112 ND were included. None of the stillbirths had extensive autolysis obscuring gross anomalies. Structural anomalies were seen in 166 (31.8%): T2M-22.3%(n=37), 34.3%(n=57) were stillbirths and 43.4% (n=72) were ND. Cardiovascular anomalies were the commonest (T2M:13/37=35.1%, stillbirths: 27/57=47.4%, ND: 36/72=50%) followed by respiratory, musculoskeletal, genitourinary, nervous system and gastro-intestinal anomalies. Same pattern was seen in all three categories of deaths. Atrial septal defect was the most common individual defect (34/76=44.7%) followed by hypoplastic left heart (11/76=14.5%), hypoplastic right heart (6/76=7.9%), hypoplastic aortic arch (5/76=6.6%), coarctation of aorta (2/76=2.6%), tetralogy of Fallot (1/76=1.3%), and transposition of great arteries (1/76=1.3%). In respiratory system 96.4% (n=53/55) lung hypoplasia, 1.8% (n=1/55) choanal atresia, and in musculoskeletal system 58.3% (n=28/48) foot deformities, 20.8% (10/48) abdominal wall defects were present. Cystic diseases of the kidney was the commonest genitourinary anomaly (15/45=33.3%) followed by renal agenesis (12/45=26.7%), horseshoe kidney (5/45=11.1%), hypoplastic kidney (3/45=6.7%). Anencephaly and spinabifida (6/26=18.8%), imperforated anus (12/21=57.1%) were the commonest anomaly in their respective systems.

54.7% were males, 42.9% were females and 2.4% had ambiguous genitalia. Maternal age ranged from 17-45years with a mean age of 29.4 (SD 5.58) years. 81.4% of mothers were <35years of age. Prevalence of congenital anomalies in Para(P)-1, P2, P3, P4, P5, and P6 were 30.1%, 37.2%, 30.1%, 33.3%, 28.6%, and 20% respectively. There is no statistically significant correlation between the presence of anomalies and fetal gender (p=0.070), increasing maternal age (p=0.8) or increasing parity (p=0.795).

**Conclusions:** Cardiovascular defects were the commonest anomalies among feto-infant deaths. Statistically significant associations were not observed between the presence of anomalies and the fetal sex, increasing maternal age or parity.

Keywords: Structural anomalies, fetal, infant, autopsy

#### Effectiveness of Early Intervention for Neonates at Risk of Future Motor Deficits, in a Multi-Disciplinary Local Paediatric Clinic in a Tertiary Care Hospital, Sri Lanka Wijesekara D.S.<sup>1</sup>, Palliyaguru T.D.N.<sup>2\*</sup>, Peiris P.L.S.<sup>2\*</sup>, Fernando D.S.<sup>2</sup>, Fonseka W.A.D.N.<sup>2</sup>, Yapa Y.M.S.P.<sup>2</sup>, Perera D.L.D.D.<sup>2</sup>

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**Introduction and Objectives:** University Paediatric Multi-Disciplinary Team (MDT) clinic at Colombo-south teaching hospital (CSTH) recruit neonates at birth who are at risk for future neurodevelopmental deficit (e.g.: prematurity, birth asphyxia, neonatal meningitis, sepsis) to initiate early interventions which include physiotherapy, occupational therapy and other treatment modalities. Alberta Infant Motor Scale (AIMS) being the practiced tool at the clinic, is a norm referenced, validated, observational scale that is used to gauge motor development. Objective of the study was to assess the effectiveness of early intervention for motor development using AIMS score and the corresponding centile improvement.

**Methods:** Serial AIMS scores and centiles were obtained from the clinic records of 46 patients who were recruited at the paediatric MDT clinic, CSTH in the past two years for early interventions. Mean values of the AIMS scores and centiles at first three MDT visits and descriptive statistics were calculated and comparison of means was performed with paired sample t test by Statistical Package for Social Sciences version 16.0.

**Results:** Of the study sample, 50% had an AIMS of <10th centile at the first visit at a mean age of 4 months compared to the 22.8% at the 2nd visit at mean age of 7 months. Comparison of mean values in AIMS centiles at first two visits revealed a statistical significance (p < 0.001). Comparison of means of AIM scores at 1st and 2nd visits as well as 1st and 3rd visit revealed a strong positive correlation with a statistical significance (p < 0.001). Of the infants, 27.2% had improved motor development with early interventions between 1st and 2nd clinic visits over a period of three months.

**Conclusions:** Early intervention significantly improves the motor development of infants who are at risk for future neuro-developmental deficits.

**Keywords:** AIMS, Early Interventions, MDT, Motor deficits

#### Analysis of Polymorphisms in Osteopontin Gene and Risk of Urolithiasis: A Case-Control Study and Meta-Analysis

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**Introduction and objectives:** Urolithiasis is a common urological problem (worldwide prevalence of 4-20%) causing high patient morbidity and associated healthcare burden. The reported etiology of urolithiasis is multifactorial involving both, environmental and genetic risk factors, with heritability of 50%. Osteopontin (OPN) is a urinary macromolecule potentially involved in regulation of renal stone formation. Polymorphisms of OPN gene have been investigated for their association with urolithiasis but with inconsistent results. We conducted the present study to determine any association between OPN gene polymorphisms and urolithiasis using case-control study of Pakistani subjects and meta-analysis of previous literature.

**Methods:** We recruited 235 patients presenting with urolithiasis from five tertiary care hospitals in Punjab, Pakistan. Healthy controls (n=248) without antecedents of urolithiasis were enrolled as the control group. All subjects were genotyped for six OPN polymorphisms to investigate association with urolithiasis. A systematic review of literature and a meta-analysis was performed. Pooled odds ratios (ORs) were calculated after heterogeneity analysis using fixed or random effect models.

**Results:** Results of indigenous case-control study indicated a significant association of 3 OPN polymorphisms (OR = 3.14; p = 0.006 for rs2853744 in a dominant model, OR = 1.78; p = 0.006 and OR = 1.60; p = 0.012 for rs11730582 and rs11439060, respectively, in a recessive model) with urolithiasis. We also observed a 1.68-fold positive association of a tri-allelic haplotype of these OPN polymorphisms (G-C-dG) with risk of urolithiasis (OR = 1.68; p = 0.0079). However, no association was evident when data were stratified according to gender, age at first presentation, stone recurrence, family history of urolithiasis, parental consanguinity and stone multiplicity. The overall results from meta-analysis, which included 4 studies, suggested no significant association of OPN polymorphisms with susceptibility of urolithiasis except for rs2853744 (OR = 1.37; p = 0.004).

**Conclusion:** The present study reports significant association of 3 OPN polymorphisms with urolithiasis for the first time from South Asia. However, increased risk of urolithiasis phenotype is suggested only for rs2853744 after meta-analysis of pooled studies.

Keywords: Candidate gene association study, Secreted Phosphoprotein 1, Renal Calculi, Pakistan

#### PP-05 Analysis of the GJB2 Gene and its Mutated Protein in Syndromic Hearing Loss Patients of Gilgit-Baltistan Ali E.<sup>1</sup>, Hussain N.<sup>1\*</sup>

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**Introduction and objectives:** Pakistani population is a unique population for the study of recessive genetic diseases due to a higher rate of consanguinity. Immunogenetic and other environmental factors are the causes of hearing loss that is further categorized into SHL (30%) and NSHL (70%). GJB2 mutation is one of the main cause of hearing loss in different populations including Pakistan. The GJB2 gene encodes a gap junction protein that is involved in the homeostasis of the inner ear through recycling of potassium ion. The aim of this research was to find out mutations in GJB2 gene.

**Methods:** Both control and patient samples were collected from Gilgit - Baltistan for DNA isolation and PCR was done by using specific primer while sequencing was done by Sanger sequencing. Mutations were detected by a software Mutation Surveyor and BLAST. Protein structures of both control and mutated samples were constructed by PHYRE2 and visualized by a software PyMOL.

**Results:** The detected mutations were 380G>A (R127H), 457G>A (V153I), 36T>C (G12G), 496C>T (L166L), 650 delA and 79G>A (V27I). This study can help deaf patients through drug designing by using various oxidants.

**Conclusions:** The rate of GJB2 mutations in Pakistani population is different in different areas but this study shows a high rate (11.66%) as compared to Tibet population of China.

Keywords: Sensory, Gap junction protein, Mutations, Oxidants, Drug designing

#### Association Between Genetic Variants of MTR Gene and Congenital Heart Disease: Evidence from a Family-Based Case-Control Study in Chinese Population Deng C.<sup>1,2\*</sup>, Deng Y.<sup>1,2</sup>, Xie L.<sup>2</sup>, Liu H.<sup>2</sup>, Dai L.<sup>1,2</sup>

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**Introduction and Objectives:** MTR, a key enzyme in folate metabolism, plays important role in removing homocysteine (Hcy) to metionina by catalysing the transfer of a methyl group from 5-methyltetrahydrofolate to Hcy. Past studies reported genetic polymorphisms of MTR gene are associated with congenital heart diseases (CHDs), however, the results remain inconsistent. The aim of our study is to explore the association between MTR gene and CHDs, including the relative risks and parent-of-origin effect.

**Methods:** A family-based case-control study including160 CHD case-parent triads and 208 control-parent triads were conducted. Genomic DNA was extracted from peripheral blood according to the manu (The QIAamp DNA Blood Mini Kit) and the SNPs were genotyped using a commercial custom-by-design 2x48-Plex SNPscanTM Kit. All cases were classified into three subgroups: (1)septal defects(SPD);(2) conotruncal(CTD), including conotruncal, right-sided obstructive, left-sided obstructive;(3) other cardiac abnormalities. The parent-of-origin effect was estimated by R package Haplin.

**Results:** The child's C allele of rs1770449 and A allele of rs1050993 of the MTR gene were associated with an increased risk of total CHDs, SPD and CTD respectively. The relative risks associated with inheriting one copy of C allele of rs1770449 or A allele of rs1050993 were both 2.17 (95%CI:1.37,3.46) and these were both 5.31 (95%CI:2.29,12.8) when inheriting two copies in total CHDs. The relative risk among offspring who inherited two copies of haplotype CAA in MTR (rs1770449-rs1805087-rs1050993) was 9.6 (95%CI:1.8,54.6) in CTD group. No parent-of-origin effect was observed.

**Conclusions:** Our study indicates that two MTR SNPs (rs1770449 and rs1050993) may contribute to CHDs. These findings provide evidence about CHD etiology. They need to be verified functionally in the further large-scale study.

**Keywords:** Congenital heart diseases, Genetic polymorphism, Parent-of-origin effect, Relative risk

#### Barriers Facing Youths in Accessing Youth-Friendly Sexual and Reproductive Health Services in Tanzania: A Case Study from Kilimanjaro Christian Medical University College Saria V.F.<sup>1\*</sup>, Alloyce J.P.<sup>1</sup>

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**Introduction and objectives:** Youth-friendly services are those that effectively attract young people, address their needs appropriately and responsively, and succeed in their accessibility of sexual and reproductive health services. Many youths are at risk of reproductive health consequences including unwanted pregnancy which is related to maternal and fetal complications. This study aimed to identify the barriers facing youths in accessing Youth-Friendly Sexual and Reproductive Health Services (YFSRHS) among youths in Tanzania.

**Methods:** A cross-sectional study was conducted among college students at Kilimanjaro Christian Medical University College selected by random sampling method.

**Results:** A total of 138 participants were studied. Their mean age (SD) was 23 (1.2) years and 56.5% were males. About 40 (29.0%) participants knew the meaning of YFSRHS and 37 (26.8%) participants had ever visited health facilities concerning YFSRHS. Among them, 54.5% participants had ever utilized the services like voluntary counselling and testing. Sources of information of YFSRHS include social media, health providers, friends and relatives. Barriers facing youths in accessing YFSRHS were limited knowledge among youths and lack of information, poor accessibility of YFSRHS centres, cultural affiliations and financial barriers.

**Conclusions:** Many youths face challenges in accessing YFSRHS. The major barriers comprised low knowledge, cultural barriers, inadequate information and lack of YFSRHS centres. There is a need to set-up interventions that will overcome barriers facing youths in accessing sexual and reproductive health services, such as incorporating various institutes where majority of youths gather, including teaching institutes, church and organising seminars and mass education through radio, television and social media. There is a need to refine the health policy to facilitate and prioritize YFSRHS in Tanzania.

Keywords: Youth-Friendly, Sexual and Reproductive Health Services, Barriers

#### PP-08 Birth Defects and Small for Gestational Age (SGA) Status is There a Link? Jain A.<sup>1</sup>\*, Sharma A.<sup>1</sup>, Chopra A.<sup>1</sup>, Pal S.<sup>1</sup>, Jain N.<sup>1</sup>

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**Introduction and Objectives:** Birth defects have been counted and described in various surveillance programs in different health facilities and National Health Programs. Various factors like environmental exposure, nutrition, teratogens have been linked to the causation of these defects. However, the birth defects and Small for Gestational Age (SGA) status has not been investigated systematically. This study reports the incidence of SGA among babies with birth defects reported annually in a tertiary referral government hospital.

**Methods:** This is a descriptive study from the surveillance records of the babies born with birth defects in a tertiary referral hospital in India. The weight of the babies was measured within 24 hours of birth with the same weighting machine (SECA 2000) and the average of 3 recordings was obtained. The gestational age of the babies was estimated by the first trimester ultrasound scan wherever available. When this was not available the same was assessed by the Expanded New Ballard score of gestational assessment. The Intergrowth -21 Newborn Cross-Sectional Growth charts were used to classify the babies as AGA (Appropriate for gestational age) or SGA (Small for gestational Age).

**Results:** There were 279 babies with birth defects born in 2017. This is 3.4% of the total annual births. 27.4% of these babies were SGA. The mean gestational age, head circumference, father's age, mother's age of AGA babies was 37.1±3, 32.7±2.6, 29.9±4.3, 26.1±4.1 respectively and was comparable to the SGA babies Viz 37.5±1.9, 31.8±2.7, 29.5±5.7, 26.1±4.8 respectively. Consanguinity was more common in SGA babies (9.21%) compared to AGA babies (3.94%). The deaths in AGA and SGA babies with birth defects was comparable (14.29% vs 14.47%).

**Conclusion:** A larger proportion of the babies born with birth defect are Small for gestational age (SGA). Most of the baseline characteristics of these babies are similar to AGA babies. The association of this intrauterine malnutrition and fetal growth failure on the occurrence of birth defect needs to be further studied.

**Keywords:** Small for gestational age, appropriate for gestational age, birth defects, consanguinity, India

#### PP-09 Birth Defects Surveillance in Lebanon: Who are the Most Affected Neonates? Yunis K.A.<sup>1,2\*</sup>, El Hayek G.Y.<sup>1,2</sup>, Al Bizri A.<sup>1,2</sup>, Nakad P.<sup>1,2</sup>, El Rafei R.<sup>2</sup>, and Ammar W.<sup>3</sup>

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**Introduction and objectives:** Birth defects (BD) still pose a serious burden in low and middleincome countries, where hospital and community based surveillance systems are rare and crucial. In Lebanon, a National Birth Defects Surveillance System (NBDSS) was implemented in 2012 by the Ministry of Public Health in collaboration with the National Collaborative Perinatal Neonatal Network. The ongoing Syrian crisis, generating an influx of refugees burdened the Lebanese healthcare system with an increase in the number of deliveries. The objective of this study is to compare Lebanese and Syrian reported BD cases.

**Methods:** Data analysis was performed on NBDSS data collected between January 2013 and December 2017. Nationality was defined according to the father's origin. The rate of birth defects was calculated, and bivariate analysis was conducted to compare socio-demographic characteristics and birth defects by nationalities.

Results: From 2013-2017 there were 3,288 cases of reported BD, of which 67% are Lebanese and 33% are Syrians. The overall BD rate for the five years was 6 per 1000 live birth. The most common BD was musculoskeletal (24.6%) and the least common was identified syndromes (1.4%). The bivariate analysis showed that Syrians (35.4%) have more Central Nervous System BD compared to Lebanese (18.5%, p-value≤0.001). On the other hand, Lebanese have more cardiovascular BD (18.4% versus 10.5%, p-value $\leq 0.001$ ); genitourinary BD (21.0% versus 16.2%, p-value=0.002), abnormalities (10.2%) chromosomal versus 6.5%, p-value≤0.001), gastrointestinal BD (9.3% versus 7.1%; p-value=0.043), and respiratory BD (3.2% versus 1.7%, p-value=0.020). Syrian mothers and fathers were younger (Mean±SD: 26.40±6.67 and 32.0±13.02 years old respectively) than Lebanese (Mean±SD: 29.22±6.44 and 35.03±7.10 respectively). About half (51.3%) of Syrian mothers did not take folic acid, compared to 28.3% of Lebanese mothers (p-value≤0.001). Also, 51.3% of Syrian babies versus 26.9% of Lebanese resulted from consanguineous marriages (p-value≤0.001).

**Conclusions:** Our findings show that Syrian newborns have less BD than Lebanese, despite their dire socio-demographic and medical conditions. However, different issues arise with the reporting system, which is only hospital-based, probably missing displaced Syrians giving birth in homes, camps clinics, and primary health care centers. Additionally, with the increasing number of deliveries in hospitals, some centers might have been underreporting the cases of BD.

Keywords: Birth Defects, surveillance, prevalence, refugee, Arab

#### PP-10 Candidates for Prenatal Prediction of Adverse Outcome of Pregnancy: Cytokines and Chemokines Shared among Spontaneous Miscarriages, Stillbirths, and Preterm Births with Transcriptomic Approach

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**Introduction and Objectives:** Infection and inflammation have been determined as the etiological factors for the adverse outcome of pregnancies in spontaneous miscarriage, stillbirth, and preterm. We hypothesize that the adverse outcome of pregnancies may share pathogenic pathway(s) at the condition of infection/inflammation.

**Methods:** Transcriptomics with bioinformatic analysis have been applied to investigate the molecular mechanism associated with infection and inflammation via assessing the gene expression profiles in miscarriages (MC), stillbirths (SB), or spontaneous preterm birth (sPTB). The bioinformatic core-analysis was run by Ingenuity Pathway Analysis (IPA) software. Online STRING (https://string-db.org) was applied to study gene associations. Statistical significance was determined by cut off with change of fold (CF) >=2 and p value =< 0.01.

**Results:** The differential expression profiles of mRNAs were generated from eight groups. Variant comparisons were performed with [sPB vs X], where the X represents different outcome of pregnancies. The top-30 significantly expressed genes were used for comparison assessment. 274 pathways were identified if the pathway was present at least in two groups of comparison. Similarly, if a gene appeared at least in two groups within a pathway, it would be considered as the common genes being shared (CGBS). Totally, 20 CGBS have been identified. Among which, 13 genes are associated with the biological process of cellular response to organic substance or chemical stimulus. Analysis of reactomic pathways showed that 11 proteins were associated with immune system. Nine genes, which are CXCL10, EIF2AK2, FOS, FOXO1, HLA-DRB1, IL33, IL6ST, IRF9, and MEF2C, were characterized to be involved in cytokine signaling pathway in immune system. Further clustering of these genes using the Markov Cluster Algorithm (MCA) determined that FOS, MEF2C, FCER1G and NFAT5 displayed high enrichment with inflammation and five genes (CXCL10, HLA-DRB1, IRF9, EIF2AK2, and IL33) were involved in influenza A pathway.

**Conclusion:** Novel cytokines and chemokines that associated with infection and inflammation pathways have been identified to be shared among spontaneous miscarriages, stillbirths, and preterm births, which may present a subset of biomarker candidates for prenatal detection at early stage pf pregnancies and facilitate prevention and intervention of adverse outcome of pregnancies.

Keywords: adverse outcome of pregnancy, prenatal prediction, cytokines, chemokines

#### PP-11 Center–Based Statistics of Cleft Lip, Alveolus and Palate Patients in Aden, Yemen Esmail A.<sup>1\*</sup>, Abdo M.<sup>1</sup>, Gundlach K.<sup>2</sup>

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**Introduction and Objectives:** Cleft lip and/or palate (CL/P) is a common human congenital defect, promptly recognized at birth. Until recently, services to repair clefts were hardly available in Yemen, and many parents are still unaware that a child with a cleft can be effectively treated. In Yemen, the prevalence of CL/P is unknown due to a lack of a birth-defect register and lack of national surveys. Therefore, the purpose of this study was to report the types and patterns of cleft lip alveolus, and palate as seen in a Cleft Lip and Palate Centre, Aden, Yemen.

**Methods:** Retrospective, centre-based study was conducted at the Cleft Lip and Palate Centre, Aden University, Yemen. Statistical evaluation of the data from all cleft patients who were registered at or referred to this centre during the years 2005-2011

**Results:** A total of 1110 cleft patients were seen during the period studied (2005-2011). Of these, there were 183 (16.48%) with a cleft lip, 144 (12.98) with a cleft of lip and

alveolus, 288 (20.54%) had a cleft palate, and 555 (50%) had a combination of cleft lip and palate. Among the patients with an isolated cleft palate there were 102 (9.2%) cases with a cleft of the soft palate only; these were found most often in girls. The ages of the patients were between one day and 40 years. The clefts were found more often in males

(57.3% males and 42.7% females). This difference was statistically significant (p=0.005). There were statistically significant sex differences noted in evaluating various cleft types. About 201 (18%) had a positive family history of clefts. Among the risk factors considered, consanguineous marriages were the most frequent (48%).

**Conclusions:** The prevalence rate of orofacial cleft types among this Yemeni sample was similar to previously reported prevalence rates in white Caucasians. The present study did not find many cases with medication before or during pregnancy, and the incidence of positive family histories was also similar to other published studies on cleft lip / palate.

Keywords: Cleft lip, Cleft palate, Yemen, Consanguineous marriages

#### Characteristics of Neonates who are Undergoing Early Interventions for Future Neurodevelopmental Deficits at Colombo-South Teaching Hospital, Sri Lanka Wijesekara D.S.<sup>1</sup>, Peiris P.L.S.<sup>1\*</sup>, Fernando D.S.<sup>1\*</sup>, Palliyaguru T.D.N.<sup>1</sup>, Fonseka W.A.D.N.<sup>1</sup>, Yapa Y.M.S.P.<sup>2</sup>, Perera D.L.D.D.<sup>2</sup>

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**Introduction and Objectives:** There is growing evidence that interventions which are commenced in the early neonatal period minimize the probability of future neurodevelopmental deficits. Paediatric multi-disciplinary team (MDT) clinic at Colombo-South teaching hospital (CSTH) recruits neonates with risk factors for future neurodevelopmental defects to initiate therapies including physiotherapy, occupational therapy, speech therapy and other relevant treatments from birth and review them regularly. The objective of this study was to identify the frequencies of neonatal risk factors among infants undergoing early interventions.

**Methods:** A descriptive study was conducted taking into consideration of all the patients recruited for early intervention within a period of 2 years from 01/2017 to 01/2019 to the paediatric MDT clinic at the University paediatric unit of Colombo-South teaching hospital. Data of 108 patients was collected from the records maintained by the clinic team. Characteristics of the sample were analyzed using SPSS version 21.0.

**Results:** Among the 108 subjects 65.7% were preterm, compared to 34.2% term babies. Out of the preterm babies 12.6% were extreme preterm babies (less than 28 weeks). Forty five percent babies were born within 28-32weeeks of gestation. More than one third (39.8%) of the 108 subjects were admitted to SCBU for further management at birth. Among the 43 subjects who were admitted to SCBU, 37.9% required an artificial ventilation method. Nearly half (45.3%) of the babies were born with low birth weight (LBW). Commonest risk factors for early interventions other than prematurity and LBW, were in order respiratory distress, neonatal jaundice, birth asphyxia, neonatal meningitis which were 46.2%, 39.8%, 17.5% and 15.7% respectively. 31% of babies had five or more risk factors whereas 20% had two risk factors.

**Conclusions:** Majority of the babies who were recruited to the MDT were preterm and were born with low birth weight (LBW). Commonest risk factors for early interventions other than prematurity and LBW were respiratory distress, neonatal jaundice, birth asphyxia and neonatal meningitis.

Keywords: Early intervention, Neurodevelopmental defects, Neonatal risk factors

#### Clinical Description of Maternal Infections During Pregnancy and Congenital Defects in Their Offspring in Bogotá and Cali, Colombia 2001-2018

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**Introduction and Objectives:** The teratogenic role of infections has been a matter of discussion to produce congenital abnormalities (CA). We aimed to describe a cohort of patients with CA and their prenatal exposure to acute maternal infections during pregnancy from the Surveillance Program of Congenital Defects (SPCD) in two Colombian cities.

**Methods:** Data were collected from the Bogota and Cali SPCD from 2001 to 2018, using the Latin American Collaborative Study for Congenital Malformations (ECLAMC) protocol. We reviewed the database and extracted data related to patients with CA. The variables considered for the analysis were sex, weight, prenatal diagnosis, other associated malformations and live or death born. Maternal infections were classified based on compromised systems (Respiratory tract, gastrointestinal tract, urinary tract, genital organs, skin, and soft tissues). Data were analyzed using frequency distribution.

**Results:** We identified 90,460 CA from 474,697 births and the rate was 381.1 per 10,000 live births (95% CI: 225.7 – 602.8). We found 3,200 mothers (17.6%) with acute maternal infections in all trimesters. Males were the most affected by CA (55.2%). The most common maternal infection among our cases was urinary tract infections (50.0%) followed by genital organ infections (43.5%) and respiratory tract infections (2.5%). CA with higher prevalence were ear malformation causing hearing loss (5.5%), talipes equinovarus (3.6%) and polydactyly (2.8%).

**Conclusions:** Timely screening and treatment are the best available measures in order to reduce the rate of infection and the associated adverse health outcomes. In sexually active women, urinary tract infections and sexually transmitted infections are frequent and increasing in prevalence. We recommend providing adequate antipyretic therapy as a practical preventive for the CA.

**Keywords:** Maternal infectious diseases, fever, birth defects, case-control study.

#### Clinical Spectrum of Congenital Anomalies and its Contributing Factors among Infants Presenting to a Tertiary Care Hospital

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**Introduction and Objectives**: Congenital anomalies affect 1 in 33 infants globally resulting in 3.2 million disabilities and deaths in newborns every year. Various genetic and environmental factors contribute to anomalies distribution in a certain population. Currently there is no local data available to compare risk factors of congenital anomalies and their comparison in normal infants. Aim of this study was to determine the frequency of various types of congenital anomalies in infants and their contributing factors.

**Methods**: This was a cross sectional study conducted at pediatrics department of a tertiary care hospital that included sample of 994 patients taken through non probability consecutive sampling. All infants of both gender presenting to pediatric unit for routine checkup or vaccination and having congenital anomalies were enrolled. Statistical analysis was done using SPSS 21 with descriptive analysis and Chi-square tests.

**Results:** Out of 994 patients, 392(39.4%) were females and 602 (60.4%) were males. Results also showed that 362 patients (36.4%) had genito-urinary system abnormalities, 232 patients (23.3%) had congenital anomalies in central nervous system, 222 patients (22.3%) had congenital anomalies in musculo-skeletal system, 94 children (9.5%) had gastro-intestinal system anomalies, 84 (8.5%) had cardio vascular system anomalies. Among five important contributing factors for congenital anomalies (extreme maternal age i.e. <20 years and >40 years at time of conception, history of consanguity, preterm births, maternal smoking and family history of birth defects), no statistically significant results were found which depicts they are equally distributed among patients with different anomalies.

**Conclusion**: It is concluded that congenital anomalies of genito-urinary system are highest followed by those of central nervous system and musculo skeletal system in this study sample. **Keywords:** congenital anomalies, genito-urinary system, central nervous system

## Comparison of Implementation Status of Revised Antenatal Care Guidelines in Matara District During 2015 and 2019.

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**Introduction and Objectives:** Antenatal care should be provided according to a standard guideline. Revised Sri Lankan antenatal care guidelines provides optimum standardized care for better outcome of the pregnancy. Present study was aimed to compare implementation of revised antenatal care guidelines in Matara district in 2015 and 2019.

**Methods:** A descriptive cross-sectional study was carried out among pregnant mothers who received antenatal care from registration to 36 weeks of gestation from health care workers of Matara district and admitted to District General Hospital, Matara in 2015 and 2019. Data was compared to see the difference. A sample of 403 pregnant mothers was selected in 2015 and a sample of 100 pregnant mothers in 2019. All the pregnant mothers registered in the admission book of the obstetric wards from the date of data collection were recruited until the allocated sample size was achieved. Women who delivered within a short time after the admission were taken for the study at the postnatal ward before discharging from the hospital. A pretested interviewer administered questionnaire was used to collect data and checklist was used to assess the documentation of the services provided.

**Results:** Majority of pregnant mothers had sought care from the MOH system and it showed some improvement over the years (81.6% in 2015 and 88.6% in 2019). Six percent of pregnant mothers were not visited by PHM during the pregnancy in 2015 and this was four percent in 2019. In 2015, 10.5% of pregnant mothers and 48.6% of their husbands never attended to antenatal sessions conducted by public health staff and these figures were 9.1% and 45.4% in 2019. Most of the documentation improved with time except plan for the delivery and maintaining of symphisio-fundal height graph.

**Conclusions:** Implementation status of revised antenatal care guidelines were improved with time.

Keywords: Antenatal guidelines, antenatal care, public health midwife, Sri Lanka

Congenital Surgical Malformation in Gabriel Touré Teaching Hospital in Bamako, Mali: Epidemiological and Clinical Aspects

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**Introduction and Objectives:** Congenital surgical malformations are common causes of neonatal morbidity and mortality. The aim of the study was to access epidemiological, clinical and prognostic characteristics of newborns admitted with those pathologies to the Pediatric Department of Gabriel Touré Teaching Hospital in Bamako, the capital city of Mali.

**Methods:** We conducted a one-year prospective study from January 1st, 2016 to December 31st, 2016 in the neonatology service in the Pediatric Department of Gabriel Touré Teaching Hospital in Bamako. All neonates from 0 to 28 days hospitalized for a surgical emergency were included.

**Results:** One hundred and fourteen cases of congenital surgical malformations were recorded (3% of hospitalizations). The average maternal age was 24 years old. Sex ratio was 1.1. The average consultation time was 7.5 days. In 55% of cases, newborns were premature. The antenatal diagnosis was made in seven patients. Digestive diseases predominated with 72.9% with mainly omphalocele (17.5%), anorectal malformations (13.2%), and laparochisis (12.3%). Hirshprung's disease was found in two newborns (1.8%). Surgery was performed in 59 neonates (51.8%). The pre and postoperative lethality was 55.26%. The overall case fatality rate was 49.1%. The most lethal pathology was laparoshisis (100%). Factors associated with mortality were maternal insctruction level (p = 0.00), vaginal delivery (p = 0.01), and laparoschisis (p = 0.000).

**Conclusions:** Evidence based policies by the health authorities to reduce mortality related to neonatal congenital surgical pathologies is needed.

Keywords: Newborn, Congenital malformation, Mortality

#### PP-17 Congenital Unilateral Hypoplasia of Depressor Angularis Oris -A Rare Case Dharmasoma U.W.N.K.<sup>1\*</sup>, Dissanayaka D.M.B.P.

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Asymmetric face is estimated to occur in 0.2%–0.6% of infants. Congenital unilateral hypoplasia of depressor angularis oris has been implicated in the pathogenesis of asymmetric crying face since1931. Diagnosis can be established by the clinical picture and/or an electromyography study. It is associated with a wide variety of anomalies involving cardiovascular, gastrointestinal, genitourinary, skeletal, and central nervous systems. Accurate diagnosis of this subtle condition ensures proper management and screening for associated anomalies.

A male neonate was born to a 31yrs old primi mother with gestational diabetes mellitus at POG 37+5 via elective cesarean section with term Intra Uterine Growth Retardation. He was found to have lower lip asymmetry during crying along with drooling from one side of mouth, forehead wrinkling and nasolabial fold depth. The eye closure remains intact and equal on both sides. Also, he had R/pre auricular skin tag and asymptomatic hypoglycaemia. Clinical examination revealed it is a defect in depressor angularis oris muscle but not a facial nerve palsy causing asymmetrical crying face. Brain and kidneys were ultrasonically normal, no eye defects were identified and systemic examination were normal. Correction surgery and sphincteroplasty was done at the age of 2 years and facial symmetry achieved.

It is a rare case but combination of high clinical suspicion leading to early diagnosis and thorough search for abnormalities in other systems ensure proper management and prevention of complications in children with asymmetric crying face.

Keywords: Congenital unilateral hypoplasia, depressor angularis oris, case study

#### Association Between Children Diagnosed with Autism Spectrum Disorder with Sensory Difficulties and Determinants oif Mother's Reduced Mobility During Pregnancy- A Pilot Study at a Tertiary Care Institution, Sri Lanka Adhikary B.T.N.<sup>1</sup>, Wijesekara D.S.<sup>1,2\*</sup>

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**Introduction and Objectives:** Autism is a condition with a prevalence of 1/93 in year 2000 in Sri Lanka. According to DSM-5 (Diagnostic and Statistical Manual) autism diagnosis involves deficits in the areas of social interactions, communications and restricted, repetitive patterns of behaviour and interests. Behaviours such as rocking, spinning, and hand-flapping are believed to occur as a result of neurological dysfunction in the central nervous system due to poor integration of neural connections. Evidence shows that development of sensory system starts during pregnancy and it matures during childhood. While the basic physical structure of the sensory receptors (i.e. eyes, ears etc.) develops early in gestation and most of the neurosensory development occurs in the second and third trimester. It is important to know if autism with sensory difficulties could be influenced by maternal factors of immobility or reduced mobility during pregnancy.

**Methods:** This is a pilot study carried out in autism clinic at the Teaching Hospital Colombo South. Seventeen mothers of children diagnosed with autism spectrum disorder participated in the study. Demographic data and determinants of mobility of mothers during pregnancy such as previous miscarriages, illness during pregnancy, medications during pregnancy (other than vitamin supplements, calcium and iron) occupation, rest, exercise and self-assessment of mobility were assessed using an interviewer administered questionnaire. Two tailed Pearson correlation coefficient was used to calculate the data.

**Results:** Levels of significance between autism spectrum disorder and previous miscarriages is 0.42, illness during pregnancy 0.304, medications during pregnancy 0.478.occupation in pregnancy 0.55, bed rest 0.272, exercise 0.812 and self-assessment of level of mobility 0.552.(correlation is significant at the 0.05 level (2 tailed)

**Conclusions:** According to the pilot study the null hypothesis is accepted in all determinants assessed. Hence there is no significant association of Autism spectrum disorder vs. miscarriages, illness and medications during pregnancy bed rest, occupation in pregnancy, exercise and self-assessment of mobility.

Keywords: Autism spectrum disorder, sensory difficulties, immobilization, pregnancy

#### PP-19 Developmental Neurotoxicity Potential Through Chronic Exposure to Dietary Lead in CKD Hotspots in Sri Lanka Ananda Jayalal T.B.<sup>1\*</sup>

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**Introduction and Objectives:** Lead has been found in considerable quantities in the blood of people living in chronic kidney disease (CKD) hotspots, and their food is contaminated with lead. The European Food safety Agency (EFSA) Panel on Contaminants in the Food Chain (CONTAM) considers developmental neurotoxicity ( $1.2\mu g/dL$ ), chronic kidney disease ( $1.5\mu g/dL$ ) and systolic blood pressure ( $3.6\mu g/dL$ ) as the most potent adverse effects of lead. The objective of this study is to assess the impact of dietary exposure to the developmental neurotoxicity which reflects by Intelligent Quotient (IQ).

**Methods:** Study was carried out in Madawchchiya and Padaviya, two areas with the highest CKD prevalence in Sri Lanka. Lead levels were determined in food samples (n = 277) obtained from CKD households. Blood lead was tested in 184 members of those households. Rice samples (n = 196) were collected from paddy fields of the Padaviya with GPS coordinates for lead determination.

**Results:** The mean lead content of 277 food samples out of which 65 were rice, from households were 216  $\pm$  223 ug/kg. Minimum, maximum and mean lead in rice were 50, 790, 118 µg/kg respectively in the 196 samples from fields. Blood lead ranged from 2.33 (LOD: 3.3) to 8.8 µg/dL with a mean of 3.35 + 1.47 µg/dL. Estimated mean exposure to lead per week in a standard 60kg male consuming a typical diet for these communities was 928µg.

**Conclusions:** Dietary exposure is sufficient to cause conditions such as neurotoxicity, chronic kidney disease, systolic blood pressure, and possibly other effects such as Low Birth Weight and malnutrition. The blood levels are sufficient to expose the fetus to adverse effects of lead. Retarded neurodevelopment leading to lower IQ appears to be a serious concern. Immediate public health interventions are warranted to lower the described exposure to reduce serious health and social consequences.

Keywords: Lead, chronic kidney disease (CKD) hotspots, food contamination with lead

# Effectiveness of Health Education Handbook Designed to Increase Knowledge on Birth Defects among Public Health Midwives in Southern Province of Sri Lanka De Silva J.<sup>1\*</sup>, Amarasena S.<sup>2</sup>, Jayaratne K.<sup>3</sup> and Perera B.<sup>1</sup>

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**Introduction and objectives:** Birth defects is a significant health problem among children. Prevention of birth defects is cost effective than spending resources to correct BD in newborns. Increasing knowledge and relevant skills in primary health care workers on birth defects is urgently needed to prevent birth defects in Sri Lanka.

**Methods:** An interventional study was carried out to evaluate the effectiveness of a health education handbook prepared to increase knowledge on birth defects among the Public Health Midwives (PHMs) of Southern province of Sri Lanka. A self-administered questionnaire was used to evaluate the knowledge of PHMs of Bope-Poddala (intervention group, n=16) and Matara municipal council health unit area, (control group, n=17). Post-evaluation was carried-out for both the intervention and control groups using the same questionnaire. The range of scores on the knowledge on birth defects, on associated factors and on prevention and management of birth defects were 0-10, 0-21 and 0-10 respectively. Data were analyzed using SPSS version 20. Means of categorical knowledge on birth defects were calculated and t test was used to identify the differences between means. Significance level was considered as p<0.05.

**Results:** The mean scores obtained on the knowledge on birth defects, on associated factors of birth defects and on prevention and management of birth defects were 6.69, 14.69 and 8.19 respectively by the intervention group during pre-evaluation. The respective mean scores were 8.88, 18.63 and 9.19 (p<0.001) after the intervention. Among the control sample the mean scores of knowledge on birth defects, on associated factors and on prevention and management were 8.00, 14.53 and 7.65 in pre-evaluation and they were 7.07, 14.73 and 8.20 in the post-evaluation (p>0.05). The PHMs highlighted the importance of handbook as the first concise health education material they received on birth defects and they suggested distribution of the handbook among the other PHMs of Sri Lanka.

**Conclusions:** The developed health education handbook is effective in increasing the knowledge on birth defects, on associated factors of birth defects and on prevention and management of birth defects among PHMs of Southern Province of Sri Lanka.

Keywords: Birth defects, knowledge, health education, Public Health Midwives, Sri Lanka

#### PP-21 Epidemics of Spina Bifida and Anencephaly in Tigray, Northern Ethiopia: Hospital-Based Studies. Berihu B.<sup>1\*</sup>

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**Introduction and Objectives:** Neural tube defects (NTDs) are the major causes of fetal loss and considerable disabilities in infants. Prior to this study there was no significant research on the incidence of NTDs in the Tigray region of Ethiopia. The objective of this paper is **t**o determine the incidence and clinical pattern of the NTDs.

**Methods:** A hospital-based cross-sectional study was conducted from October 2016 to June 2017. All pregnancy outcomes were examined for any externally visible birth defects and neurological integrity by trained midwives under the supervision of senior obstetrics and gynecology and a neurosurgeon. Data were collected using a survey tool to collect maternal and newborn demographic data and a checklist developed to capture newborns with Neural Tube Defects. Data were analyzed using SPSS version 20. The prevalence of NTDs was calculated per 10,000 births.

**Results:** Out of the 14,903 births during the study period, a total of 195 infants were born afflicted with NTDs. The burden of infants with anencephaly and spina bifida was 66.4 and 64.4 per 10, 000 births, respectively. The overall incidence rate of NTDs in this study was 131 per 10, 000 births of which 23% were liveborn and 77% were stillborn. The highest burden of Neural Tube Defects was observed in Adigrat Hospital from Eastern Zone of Tigray (174 per 10,000 births) and Lemlem Karl Hospital from Southern Zone of Tigray (304 per 10,000 births) compared to Kahsay Abera Hospital from Western Zone (72.8 per 10,000 births) and Sihul Hospital from North Western Zone of Tigray (69.8 per 10,000 births).

**Conclusion:** Assuming that the non-folic acid preventable rate should be 5 per 10,000 births in developed countries such as United States, China and Canada. Prevalence rate reported by the present study is 131 per 10,000 births, a rate that is 26 times higher than rates proposed in developed countries. This just emphasizes the urgency to implement effective programs to get all women of reproductive age to have adequate folic acid to prevent all of folic acid-preventable spina bifida and anencephaly, which would prevent 96% (125/130) of spina bifida and anencephaly in the Tigray Provence.

Keywords: Anencephaly, Folate, Incidence, Neural Tube Defects, Spina bifida, Supplementation

#### PP-22 (Epi)Genetic Variants of the Sarcomere-Desmosome are Associated with Premature Utero-Contraction in Spontaneous Preterm Birth Wang J.<sup>1</sup>, Zhong N.<sup>2\*</sup>

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**Introduction and Objectives**: Spontaneous preterm birth (sPTB), the leading cause of neonatal mortality and morbidity, is a syndrome with clinical and genetic heterogeneity. Spontaneous preterm labor (sPTL) and pre-labor premature rupture of membranes (pPROM) are the most common forms of sPTB. Our previous epigenomic studies identified various pathogenic pathways involved in sPTB, particularly in the fetus-originated rupture of chorioamniotic membranes. However, few studies have focused on genetic and epigenetic defects and pathogenic mechanisms associated with premature uterine contraction (PUC).

**Methods:** Cases of sPTBs, along with controls, were selected from an existing biobank from a Chinese pregnancy cohort. An integrated omics approach of systems biology was employed. Exome sequencing was conducted to uncover gene structural variations, including single nucleotide variants (SNVs) and pathogenic variants. Microarrays were used to analyze the GMPs and DEPs of placentas. DEPs of proteins in fetal membranes were assessed using proteomic technology. Data mining with interpretation was employed to focus on genetic loci/genes related to uterine muscle contraction, and specifically on genes associated with sarcomeres and desmosomes. The possible molecular mechanisms associated with PUC and to characterize the epigenetic regulation of the genome-wide methylation profile (GMP), and the differential expression profile (DEP), of long noncoding RNA (lncRNA) in PUC associated with sPTB have been investigated.

**Results:** Thirteen SNVs and pathogenic variants were identified in the sarcomere gene, TTN, from 146 women with sPTL. DEPs of five lncRNAs were identified from loci that overlap with four sarcomeric genes. Longitudinally, the lncRNA TPM3 was found to significantly regulate the messenger RNA (mRNA) of TPM3 in the placenta, compared to maternal blood. The majority of GMPs related to PUC were also identified in the CpG promoters of sarcomeric genes/loci. DEP of PCU mRNAs showed 22 genes associated with the sarcomere and three with the desmosome.

**Conclusions:** The results demonstrated that PUC of sPTB was associated mainly with pathogenic variants of the TTN gene and that transcription of sarcomeric PUC genes is likely regulated by epigenetic factors, including methylation and lncRNA.

**Keywords**: (epi)Genetic variants, sarcomere-desmosome, premature utero-contraction, spontaneous preterm birth

#### PP-23 Exploring Perinatal Mental Health in Sri Lanka: A Brief Needs Assessment N. Perera<sup>1\*</sup>, and K. Booso<sup>2</sup>

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**Introduction and objectives:** Perinatal mental health has been recognized as a public health priority by the World Health Organization as an estimated 15.6% of women from low and middle-income countries experience mental ill-health during pregnancy, whilst 19.8% of women experience mental ill-health following childbirth. Perinatal mental ill-health is found to increase the risk of maternal suicide, poor birth outcomes and influence a child's overall development. The maternal mental health burden in Sri Lanka is highly under reported and warrants further investigation.

**Methods:** The paper presents a needs analysis for perinatal mental health support in Sri Lanka, based on a workshop conducted in Colombo, Sri Lanka. The paper includes the analysis of a feedback form and discussion-based findings from mothers. Insights from the facilitators were also considered in the analysis.

**Results:** The workshop survey was completed by 14 mothers from the Colombo district, who attended a forum on maternal health and wellbeing. The sample (N=14) comprised of women from upper-middle-class families, between the ages of 21 and 52. Majority of the sample consisted of first-time mothers. A key insight from the analyses was that only a small minority (2/14) of mothers had received information on mental health and wellbeing from their healthcare professionals during their pregnancy. The main source of information were healthcare professionals while some also relied on the internet, apps, and loved ones for information on pregnancy. Marital satisfaction, family dynamics and cultural expectations are key issues that were discussed. Gaps in information were in the areas of breastfeeding and mental health.

**Conclusions:** These preliminary findings of the study poses a significant concern to the lack of awareness of maternal mental health in Sri Lanka. It provides useful insights towards building a health campaign to develop awareness and bridge support gaps for perinatal mental health in Sri Lanka. It also highlights the need for a shift in doctor-patient communication during pregnancy and the effective channels of communication which can be used to strengthen impact. Further qualitative exploration is encouraged to distil the varying factors in maternal mental health burden in Sri Lanka

Keywords: Perinatal Mental Health, Sri Lanka, Needs Survey
# Factors Related to Growth of the Infants with Structural Birth Defects in Galle, Sri Lanka De Silva J.<sup>1\*</sup>, Amarasena S.<sup>2</sup>, Jayaratne K.<sup>3</sup> and Perera B.<sup>1</sup>

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**Introduction and objectives:** Birth defects in children is a significant public health problem. Birth defects are known to adversely affect the growth of the infants. This study was carried out to assess the growth and factors related to impaired growth of infants with structural birth defects.

**Methods:** Three hundred and fifteen (n=315) infants with structural birth defects (without genetic syndromes) from Galle district were prospectively followed up for a period of one year Their growth at the age of 12 months was assessed. The growth charts in the Child Health Development Record of Sri Lankan children were used as the references. Data analysis was done using SPSS version 20. Descriptive statistics and univariate and multivariate analysis were performed to identify the factors associated with impaired growth. Significant level was set as p<0.05.

**Results:** Two hundred and sixty-eight infants (85%) were alive at the age of one year. One hundred and fifty-nine (n=159, 59.3%) were males, 44(16.4%) were preterm and 77(28.7%) had low birth weight. The majority (58.6%) had cardiovascular defects followed by defects involving multiple systems (21.6%). The prevalence of normal growth, underweight, stunting, wasting and both stunting and wasting by the age of one year were 72.4%, 3.0%, 8.2%, 9.0% and 7.5% respectively. Low birth weight (OR=3.6, 95% CI= 1.8-7.4), presence of developmental delay (OR=3.1, 95% CI= 1.4-6.8) presence of three or more birth defects (OR=3.4, 95%CI= 1.2-10.0) and presence of infection requiring in hospital management (OR=3.9, 95%CI= 1.6-9.2) were the independent associates for growth impairment among infants with structural birth defects. **Conclusions:** A significant proportion of infants with structural birth defects have growth impairment. Special attention should be paid by the caregivers and the healthcare workers on the nutrition and growth of the infants with structural birth defects from the date of diagnosis. **Keywords:** Structural birth defects, Growth impairment, Underweight, Stunting, Wasting

# First Trimester Traditional Medication Use in Pregnancy in Cameroon: A Multi-Hospital Survey

## Leke Z.A.<sup>1\*</sup>, Dolk H.<sup>1</sup>, Loane M.<sup>1</sup>, Casson K.<sup>1</sup>, Maboh M.N.<sup>3</sup>, Maeya S.E.<sup>3</sup>, Dibo L.<sup>2</sup>, Nyenti B.P.<sup>3</sup>, Obale A.<sup>2</sup>, Etiendem D.<sup>3</sup>

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**Introduction and Objectives:** There is a paucity of epidemiological data on medication use in pregnancy in Cameroon to inform medication safety measures.

**Methods**: Between February and August 2015, 795 pregnant women attending 20 randomly selected urban or rural hospitals in South West Cameroon for antenatal (ANC) or other care were interviewed on first trimester orthodox (OTM) and traditional (TM) medication use using structured questionnaires.

**Results:** A total of 293 (36.9%) women reported use of TM during the first trimester, 77.8% of whom used it in combination with OTM. Herbs were the main component of TM, often taken collectively or sometimes mixed with other substances of animal origin and allowed to ferment before consumption. The most common indications for taking TM were for prevention/treatment of anaemia (26.3%), treatment of maternal diseases/ailments/fever (22.8%), general cleansing/enema (13.4%) and to ease labour/delivery process (10.1%). TM were taken for long periods (months). The most common (48.5%) factor influencing women's decision to take TM was a perception that TM was more potent compared to OTM. TM were mainly self-prescribed (33.3%), but relatives other than their mother (27.8%) and mothers of the women (18.8%) also played a major role in prescribing. The majority of women (67.3%) believed that TM was always safe to take in pregnancy and 34.8% had not received or could not remember (a further 35.0%) receiving any safety advice during pregnancy. In a logistic regression analysis adjusting for confounders, health unit (p<0.001), rural setting type (p=0.024), and a belief in the safety of TM (p<0.001) were statistically significantly associated with use of TM.

**Conclusions:** We identified a very high prevalence of TM use compared to studies in other African countries, raising many safety concerns. Unlike in western countries with modernised packaged forms, TM are still mainly available in African countries in the crude form. The assumed safety in pregnancy and strong belief in the efficacy of these TM by pregnant women in our study is very concerning and needs urgent attention. Discussions around pharmacovigilance systems in pregnancy in Africa and other similar contexts need to factor in the TM use and safety concerns identified in our study.

**Keywords:** Herbal medication, Traditional medication, First trimester, Pregnancy, Cameroon.

### PP-26 Frequency of Cardiac & Neural Congenital Anomalies: Findings from a Cross Sectional Study at Children Hospital, Lahore Jamil U.<sup>1</sup>, Kiran K.<sup>2\*</sup>, Kazmi T.H.<sup>1</sup>, Malik A.<sup>3</sup>, Bhatti Z.<sup>4</sup>

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**Introduction and Objectives:** Congenital anomalies (CAs) represent one of the main causes of fetal death, infant mortality and long-term disability. According to the WHO, each year approximately 3.2 million of children worldwide are born with CA and approximately 300,000 newborns with a diagnosis of CA die. The objectives of this paper are to describe main CAs and commonly seen factors with CAs among the paediatric patients at Children's hospital and Institute of Child Health, Lahore.

**Methods:** Cross sectional study was conducted in the Children's Hospital and Institute of Child Health Lahore, Pakistan. Hospital receives patients from all over Punjab. Hospital also receives referred cases from KPK & Gilgit Baltistan provinces of Pakistan. Draining areas in Punjab consist of different degrees of urbanization and socioeconomic status. The study sample of 100 children suffering from CAs were registered in Children's Hospital Lahore during the period of 11 March 2019 - 27 April 2019. Patients were given questionnaire that was formulated to ascertain underlying risk factor.

**Results:** Leading cardiac anomalies detected were ventricular Septal Defect (42%), Atrial Septal Defect (12%), Patent Ductus Arteriosus (13%) & Tetralogy of Fallot (4%). Leading neural anomalies detected were Hydrocephalus (2%) & Down Syndrome (1%).CA was more common in female children with a male to female ratio of 1:3. At the time of the birth, 29% of children with CA were underweight. In 56% of children with CA there was a history of use of ground water for drinking as compare to 2% who used boiled water. Diabetes Mellitus (2%) & Hypertension (13%) were the commonest diseases found in mothers of children with CA. History of Hypertension before pregnancy was seen in 9% of cases. 56% females gave history of using different medication during pregnancy. (60%) of couples gave history of marriage among relatives. The same among grandparents was 59%. Grandparents of father of 40% of children were cousins. Grandparents of mother of 39% children were cousins.

**Conclusion:** Among children with CA, half of their mothers used different medication and ground water. Forty percent of parents were related to each other before marriage. **Keywords:** Ground water, Grandparents, ventricular septal defect

## PP-27 Gastrointestinal and Genitourinary Birth Defects in Chittagong, Bangladesh: Results From an Urban and a Rural Center Banu T.<sup>1</sup>, Aziz T.T.<sup>1\*</sup>, Pulock O.S.<sup>1</sup>, Hayee S.<sup>1</sup>

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**Introduction and Objectives:** Birth defects are one of the key contributors to global mortality, morbidity and childhood and adulthood disability. In lower- and middle-income countries like Bangladesh the burden related to health problems caused by birth defects are higher than the high-income countries. The aim of this study was to compare Gastrointestinal (GIT) and Genitourinary (GU) birth defects in an urban and a rural setting of Chittagong, Bangladesh.

**Method:** a prospective observational study was done in Chittagong Research Institute for Children Surgery (CRICS) and Dr. Matin AMRAH Community Hospital, Chandanaish, Bangladesh over a period of 17 months. The data collection was done by face to face interviews and through medical records.

**Results:** 1123 Birth Defects out of 1634 patients were collected. 77.5% of the birth defects were related to GIT and GU systems. 281 (62.4%) GIT birth defects were found in urban and 169 (37.6%) from rural area. GU birth defects were 298 (68.3%) in urban and 138 (31.7%) in rural area. In urban area, Male female ratio was 4.3:1. Also in urban area, Neonates were 5.15%, Infant 21.85%, Toddler 24.87%, Children 39.08%, Adolescent 7.99% and Adult 1.07%. In rural area, Male female ratio was 6:1. And neonates were 3.3%, infant 16.3%, toddler 26.4%, children 40%, adolescent 10.4% and adult 3.6%. In case of GIT, Inguinal Hernia, Ano-rectal Malformation and Umbilical hernia were commonly seen in both urban and rural areas. In urban patients, Pelviureteric junction obstruction and Posterior urethral valve were common. And in rural patients, Hypospadias, Undescended testes and Phimosis were mostly seen in GU Birth Defects. GIT and GU Birth defects were more commonly seen in rural areas than in urban areas (OR= 2.19, P < 0.0001).

**Conclusion:** GU birth defects relating to upper urinary tract were commonly seen in urban area and lower urinary tract and external genitalia anomalies were commonly seen in rural area. In case of GIT Birth Defects, Inguinal Hernia, Anorectal Malformation and Umbilical hernia were equally seen in both urban and rural settings.

Keywords: Birth Defects, Bangladesh, Genitourinary, Gastrointestinal, Congenital Anomaly

## PP-28 Genetic Analysis of Children with Congenital Ocular Anomalies in Three Ecological Regions of Nepal. Thakur N1\*

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**Introduction and Objectives:** Vision disorders are one of the most common disabilities to affect children. It has been estimated that there are 1.4 million blind children worldwide, two third of whom live in developing countries like Nepal. WHO considers childhood blindness to be a priority, as it is listed in the WHO's Vision 2020 program. Congenital ocular anomalies are one of the important causes of childhood ocular morbidity and blindness. Most of these anomalies are difficult to treat or treatment still not available. Genetic testing can make a very positive impact on individuals and families affected with inherited eye disease in a number of ways. When properly performed, interpreted, and acted on, genetic tests can improve the accuracy of diagnoses and prognosis, can improve the accuracy of genetic counseling, can reduce the risk of disease occurrence or recurrence in families at risk.

**Methods:** 25 clinically diagnosed children with congenital ocular anomalies were taken for genetic analysis after taking written informed consent with the parents. Five types of congenital anomalies were studied i.e. congenital ptosis, microphthalmus, congenital cataract, coloboma and Crouzen syndrome. Polymerase chain reaction and cycle sequencing was performed on selected mutations.

**Results:** Among 30 Children, 11 (36.7%) were female while 19 (63.3%) were male. Among the total children, 14 (46.7%) of the children had congenital ptosis, 6 (20.0%) of the children had congenital cataract, 4 (13.3%) of the children had coloboma, 3 (10.0%) of the children had microphthalmous, and each child had nevus of Ota, proptosis ( Crouzen syndrome) and retinal dystrophy separately. Ten Single Nucleotide Polymorphism among seven genes for five congenital eye disorders listed above were tested. Among them one missense alteration G12411T of ZFHX4 gene in patient with congenital ptosis was found. Another missense variation T>C P.Y374C of STRA6 gene in patient with microphthalmus were also identified whereas others were found to be normal.

**Conclusion:** This study was able to find two mutations among two genes in the cohort of Nepalese population. Though there was various limitation of this study, it is a milestone for further analysis of genetic factors to understand the causes of various congenital anomalies.

Keywords: Congenital ocular anomalies, Genetic analysis, children

Health Barriers Detected in Patients with Congenital Anomalies That Affect Hearing Or Vision

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**Introduction and objectives**: The opportune medical attention allows to reduce mortality and disability in patients with congenital anomalies, however, there is an increase of barriers in the health services for this vulnerable population. We set out to identify health barriers in patients with visual or auditory congenital anomalies (CA).

**Methods**: Patients born between August 2016 and June 2018 with CA that affected hearing or vision were selected. The parents were contacted by phone, the informed consent was signed and the patients were clinically assessed. Information was obtained from the Comprehensive Care Program for Families with Orphan or Visual Hearing Diseases. The information was recorded in Redcap and analyzed in Microsoft Excel® 2016.

**Results**: Seventy children with CA with visual or auditory involvement were included. Of them, 82.9% presented at least one barrier in health care and according to the Tanahashi model. We detected barriers with availability as the most frequent (45.1%), the appointment schedule for the first time with a specialist doctor being the most referred barrier (70%) followed by scheduling the appointment for the examinations (65.7%).

**Conclusions**: The role of the regulatory institutions of the service providers is important to ensure adequate and timely care, to reduce mortality and disability.

**Keywords**: Public health, epidemiology, congenital anomalies; Barriers to health care; view; hearing

## Hospital Based Surveillance of Congenital Malformations at a Tertiary Care Centre in North India: An Initial Experience

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**Introduction and Objectives:** Birth defects are common, costly and critical. Birth defect surveillance is used for epidemiological, planning, educational and healthcare purposes. We share our initial experience of hospital-based surveillance for the malformations in fetuses, stillbirths and livebirths and evaluate the spectrum of different types of birth defects at a tertiary care center in North India.

**Methods:** The study was conducted from June 2017 to December 2018. Number of all the fetuses undergoing medical termination of pregnancy, stillbirths and live births were noted. Elective terminations of pregnancy for fetal anomalies (ETOPFA) (<20 weeks), stillbirths with malformations (>20 weeks) and newborns with birth defects were included. Detailed examination of all the fetuses and neonates for congenital malformations was performed soon after birth. Autopsy was performed wherever applicable after an informed consent by parents. Relevant maternal details, risk factors, fetal weight, sex, gestational age, examination findings, dysmorphism and autopsy findings were also documented.

**Results:** A total of 250 fetuses and babies with congenital anomalies (ETOPFA-79/122, stillbirths with malformations-54/93, Live births with malformations- 117/4075) were included. The malformations were classified as isolated and multiple (>1 malformations). The incidence of isolated defects was more as compared to multiple malformations/syndromic (75.2% vs 24.8%) in live births as compared to the fetuses and still births (47.3% vs 52.6%). In the former, CNS was the most commonly involved system (20/63;31.7%), with neural tube defects (11/63;17.4%) being more common in fetuses and stillbirths, while in live births, cardiac defects were commoner (31/88;35.2%) followed by musculoskeletal deformities (18/88;20.4%) genitourinary system abnormalities (9/88;10.2%) and central nervous system (8/88;9.1%). Follow up for live birth outcomes showed a survival rate of about 72%.

**Conclusions:** The study highlights the spectrum of malformations across various life course stages. Neural tube defects are the most prevalent and preventable birth defect. Although the prevalence of various types of birth defect varies, the proportion remains almost the same during and after pregnancy, demanding better antenatal surveillance and proper genetic counseling to prevent recurrences.

Keywords: Hospital based surveillance, Birth defects; congenital anomalies; malformations

## PP-31 Hospital Based Surveillance on Birth Defects in Nyamira County and Referral Hospital Ondieki J.<sup>1\*</sup>, Ateka C<sup>1</sup>, Basweti A.<sup>1</sup>

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**Introduction and Objectives:** Birth defect is any anomaly found on a baby at birth which occurs during intra-uterine life. Birth defects are the fourth leading cause of neonatal deaths in the world and it is estimated that 7.9 million children are born with major birth defects every year globally. The predisposing factors of birth defects include; environmental agents, infectious agents, lifestyle factors, medications/drugs, occupational exposures, genetic factors and nutritional factors. The objective of this study is to ascertain the number of children born with birth defects in Nyamira County Referral Hospital (NCRH) between April 2017 and April 2019

**Methods:** Sensitization on birth defects surveillance was done in NCRH on April 2017, whereby 50 health care workers from; theatre, Newborn Units (NBU), maternity, Maternal and Child Health clinic (MCH) were sensitized. Then hospital-based surveillance on birth defects was carried out on 4676 children born between 15th April 2017 and 30 April 2019 in the hospital. Active case finding approach was employed and so all babies born in the hospital were examined to exclude any anomalies. Thereafter mothers of affected children were interviewed to determine the probable cause of the defects. The findings were analyzed quantitatively and qualitatively and presented in figures and percentages

**Results:** 25 out of 4676 children born in the hospital over the period had birth defects. The defects included; hydrocephalus- 5cases (20%), tallipes- 6 cases (24%), spina- bifida - 2 cases (8%), cleft palate- 4 cases (16%), cranioschidis omphalocele- 2 cases (8%), Turner's syndrome-1 case (4%), left ear congenital atresia-1 case (4%), hypospadia-2 cases (8%), amniotic band syndrome-1 case (4%) and encephaly-1 case (4%). 0% of mothers took folic acid supplements before conception, 32% never took the folic both before and during pregnancy, 68% took the folic acid during pregnancy. 1 mother (4%) had family history of birth defects, 24 mothers (96%) had no family history of birth defects. 70% of the mothers started antenatal clinic at 2nd trimester while the remaining 30% at 3rd trimester.

**Conclusions:** Mothers attending NCRH did not get pre-conception care and started antenatal care late hence risk of birth defects.

Keywords: Hospital based surveillance, birth defects, Pre-conception care

## PP-32 Incidence of Low Weight at Birth and Stunting at Childhood: A Case Study of Malda District, India Rayhan S.K.<sup>1\*</sup>, Banerjee A.<sup>1</sup>

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**Introduction and Objectives:** Despite higher economic growth and several policy interventions, malnutrition in children below five years of age remains a major concern in India. Although, several studies have examined the association between nutritional status and socioeconomic and demographic factors especially at macro level, the effect of low birth weight (LBW) as a risk factor is little known at macro and micro level in India. Also, the effect of the Integrated Child Development Services (ICDS) Scheme which was launched in 1975 to improve the nutritional status of children is hardly known. Therefore, a case study was designed to examine the risk factors of stunting among pre-school children at the micro level.

**Methods:** A cross-sectional survey was conducted on a sample of 731 from 42 communities nested within nine villages/wards in Malda district, one of the backward districts of India. A semistructured questionnaire was used to collect data on maternal and child socio-demographic characteristics, and child's anthropometry. Anthropometric indices of Height-for-age Z-scores (HAZ) was used to classify a child as stunting. Bivariate and multilevel logistic analyses were performed to investigate the risk factors of stunting.

**Results:** About, 40% pre-school children (36-59 months) are stunted in the study region. This percentage varies widely across the communities, social groups, religious groups and mother's working status. Results of multilevel analysis revealed that children born with LBW (<2500 kg) are 2.2 times more likely to become stunted at childhood compared to their counterpart. LBW was the strongest predictor of a child's nutritional status (OR=2.22; p<0.001) after adjusting for other factors. ICDS does not have any significant effect on nutritional status. Children of bidi workers are also at higher risk of being stunted (OR=1.92;p<0.010). Other important risk factors of stunting are preceding birth interval, duration of breastfeeding, and mother's level of education. On the other hand, there is a significant variation (14%, SE 0.14) in stunting observed due to community/neighbourhood effect.

**Conclusion:** For improving the child's nutritional status, special attention needs to be paid for children born with LBW and intervention should be taken for mothers who are bidi workers. **Keywords:** Low birth weight, stunting, India

## PP-33 Integrating Rare Diseases in the Public Health Delivery System Through Legislation Padilla C.<sup>1\*</sup>, Lynn Sur A.<sup>1</sup>, Magdaraog C.<sup>2</sup>

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**Introduction and Objectives:** In the Philippines, a rare disease affects 1: 20,000 of the population. The Newborn Screening Law of 2004 (RA 9288) covers rare metabolic disorders. However, other rare genetic disorders are not covered by this law. Due to rarity, patients have been misdiagnosed or diagnosed late due to lack of clinical skills among the physicians. If with diagnosis, treatment was not accessible due to high cost of medicine and its unavailability locally. Drawing inspiration from the Newborn Screening Law, lobby efforts for a legislation for rare disease began in 2008. The law was passed after an 8-year journey. The objective of this talk is to present the policy development of the Philippine Rare Disease Act of 2016 (RA 10747).

**Methods:** The process for policy development for rare disease involved: a) consultation with various stakeholders; b) review of Rare Disease laws in other countries; c) preparation of draft bills; d) identification of champions within the legislative body; e) lobby efforts for the passage of the bill in both houses; f) enactment of the law; g) formulation of the Implementing Rules and Regulations (IRR) and; h) translation of the policy at all levels.

**Results:** The Rare Disease Act of 2016 (RA 10747) mandates that persons with rare disease have the right to survival and full and healthy development through access to timely health information and adequate medical care that is comprehensive, integrated and sustainable. The Department of Health (DOH) is the lead agency and the law defines the roles of other national service agencies in its implementation.

**Conclusions:** A national policy for Rare Disease is a powerful tool to mandate all local and national service agencies, major stakeholders, health professionals, societies and civil societies to address the needs of patients with rare disease. The Rare Disease Act of 2016 (RA 10747) served as basis in establishing a comprehensive program that will address the special needs of patients with rare disease and their families.

Keywords: Rare disease, Rare disease law, Policy

## PP-34 International Classifications as a Tool for Interoperability in Child Health Siderius L.<sup>1\*</sup>, Poulissen P.<sup>1</sup>, de Graauw M.<sup>2</sup>

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**Introduction and Objectives:** Some 93 million children under 15 years of age live with a moderate or severe disability (WHO, 2015). Children with autism, developmental delay, cerebral palsy, hearing deficits and visual impairment may have a rare condition with specific (preventable) health risks. The WHO Maternal and Child Health (MCH) handbook contains essential information, kept by the family, to promote and maintain health of mothers and children. Technically much is possible. Still most individual data are locked in IT systems. On the other hand in lower and middle income countries the use of electronic data in medical care is still on the verge of development. This study aims to explore, how can electronic patient recording systems improve health outcomes of children.

**Methods:** Patient representatives of six rare conditions describe how to gather lifelong information in Unified Modeling Language in https://rarecare.world. Recognizable features and diagnostic test are listed to find symptoms of a rare condition in primary care. Characteristic symptoms are linked with a set of international terminologies applicable in primary care practice: the International Classification of Primary Care, Human Phenotype Ontology and Standard for identifying health measurements, observations, and documents. Each symptom is linked with a disease using the International Classification of Diseases. Diseases are or are part of one of more rare condition. For each condition an application programming interface (API) is provided.

**Results:** The oculo-auricular-vertebral spectrum (OAVS) presents with several features recognizable by parents and primary care professionals. An electronic MCH record shared with primary child health services linked with the API allows integrated and continued care from birth into a adulthood for children with birth defects. Vaccination can be added with the Anatomical Therapeutic Chemical Classification System and disabilities with the International Classification of Functioning, Disability and Health. Pooled anonymized data can fill international registries such as European Surveillance of Congenital Anomalies to improve the quality of care.

**Conclusion:** Regardless of the chronic condition, the development of a generic set of tools will allow service providers to effectively manage chronic illness and improve health outcomes of affected children. To support personal health management the MCH needs a tight integration with international terminologies.

Keywords: E-health, patient-centered care, primary care, rare diseases, MCH

## PP-35 Major Congenital Anomalies Associations Through Graph Theory Elias D.<sup>1,2\*</sup>, Campaña H.<sup>1,2,3</sup>, Rittler M.<sup>1</sup>, Cosentino V.<sup>1,2</sup>, Poletta F.<sup>1,2</sup>, Santos R.<sup>1,4</sup>, Pawluk M.<sup>1,2</sup>, Gimenez L.<sup>1,2</sup>, Ratowiecki J.<sup>1,2</sup>, Gili J.<sup>1,2,5</sup>, Camelo J.L.<sup>1,2</sup>

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**Introduction and Objectives:** The identification of new major congenital anomalies complexes and associations is an important activity in birth defects surveillance. To date, two approaches have been used to analyze associations. The first focuses on a specific anomaly and determines the association degree with other anomalies. The second approach was based on the cases clustering considering each anomaly as a binary variable. The graph theory, offers an integrating approach, allows to analyze the complete set of anomalies and each of them. A graph is constituted by nodes that represent entities and edges that represent the relationship between entities. In recent years this approach has enriched many study areas, for example, proteins interactions and diseases associations. The objective of this work is to analyze the feasibility of this approach to study major anomalies complexes and associations.

**Methods:** This study was based on the Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC) registers. We used 118244 cases with at least one major anomaly recorded between 1967 and 2017. We used the ECLAMC anomaly coding system, which has 207 codes. To determine the association strength between anomalies we used the volume-adjusted Chi-Squared independence test. The graph was partitioned using the Infomap method.

**Results:** The major congenital anomalies graph had 74 nodes and 175 edges. Its degree distribution presented a greater adjustment to an Exponential distribution than to a Poisson and Power distributions, therefore the graph obtained differ from random graphs. The graph partition generated 9 anomalies groups, the partition modularity was 0,53. The anomalies complexes identified through graph theory would correspond to those found in the literature, such as Patau syndrome. Atresia and stenosis of rectum and anus, Anophthalmia/Microphthalmia and Ambiguous genitalia were the more associated anomalies (> 15).

**Conclusions:** These results imply that it is feasible to use graph theory for the study of major congenital anomalies associations.

Keywords: Graph Theory, Congenital Anomalies Associations, ECLAMC

## PP-36 Major Congenital Anomalies: A Retrospective Regional Study in Three Hospitals in Cali – Colombia Orozco-Chamorro C.<sup>1</sup>, Madariaga I.<sup>2</sup>, Holguín J.<sup>3</sup>, Zarante I.<sup>2</sup>, Hurtado-Villa P.<sup>1\*</sup>

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**Introduction and objectives:** Congenital malformations or birth defects correspond to antenatal genesis' situations existing at birth, potentially impacting infant's health, growth and/or survival. It's a wide and heterogeneous group of diseases. Malformations can be classified as major and minor. Generally, major malformations have medical consequences, significant effects on function and/or social implications. These often require surgical repair or could be life-threatening. The objective of this study was to evaluate the postnatal prevalence of major congenital anomalies and malformation patterns in the population from three hospitals since 2011 to 2018 from the Surveillance Program of Congenital Defects (SPCD) in a Colombian city.

**Methods:** Information was taken from Cali Birth Defects Surveillance Program database using Latin-American Collaborative Study of Congenital Malformations (ECLAMC) criteria. Data from 69,977 births across three hospitals in Cali was reviewed using the following inclusion criteria: live births, with a probable (phenotypical characteristics) or confirmed diagnosis. We reviewed data of births from January 2011 to December 2018 in Cali and analyzed variables as sex, prenatal diagnosis, type of diagnosis and live or death born. Major congenital anomalies were classified according to organ system, gender, maternal age and prenatal diagnosis.

**Results:** There were 1171cases of major birth defects among 69,977 live births (LB), giving a prevalence of 1.67/100 LB. Major congenital anomalies were more frequent in males 58,75% (n=688). Anomalies of the upper limbs and lower limbs were the most common defect (29.4%), followed by genital's anomalies (13,6%), urinary system's anomalies and chromosomal anomalies (11.6%). Only, 11% (n=129) cases had prenatal diagnosis

**Conclusions:** The prevalence, type and frequency of anomalies reported during this period coincide with the other published work, including male predominance. The commonest congenital anomalies in Cali in the last eight years are the anomalies of limbs.

Keywords: Case-control studies; Congenital malformation; Birth Defects; surveillance

## PP-37 Management of Craniofacial Clefts Balaji S.M\*

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Orofacial clefts (OFCs) are common and treatable birth defects. The role of cleft surgeons in treating individuals with OFC and craniofacial anomalies is to provide comprehensive therapeutic health care and aim to restore the missing areas.

The spectrum of Indian OFC varies from simple cleft lip to complex OFC involving multiple Tessier's clefting. Recreating Asian noses is tougher in Tessier type 0 clefts - managing nasal musculature, mengiocele, hypertelorism and absence of midline structures are difficult to treat. As no two OFC cases are the same, treatment of each Tessier's 0 cleft needs to be customized and determined as per functional and esthetical needs of the patient. Involvement of orbital compartment as orbital clefts, isolated or as oro-orbital or oro-orbito-nasal clefts requires customized treatment. Resultant orbital dystopias, medial and lateral canthus correction, correction of mongloid slants are some of the secondary corrections that might be required. Sometimes, regeneration of missing bone and soft tissues might be necessitated.

Indian cleft surgeons often get to meet patients at an older age than is the Western norm, which automatically complicates intervention as growth of tissues would be well advanced with greater maturation of soft and skeletal tissues, establishment of voice and feeding habits along with the effects of repeated infections besides various other factors. Management of typical OFC cases are being discussed with representative cases. For a few cases, traditional, basic, gold standard treatments are provided while for others, latest technological advances including guided bone regeneration techniques are used. The presentation intends, by utilizing 25 years of OFC treating experience, to highlight the type of OFC care in this part of the world with appropriate examples. Management of Indian patients from birth to adulthood will be presented and discussed with the main goal to provide the best care and improve the quality of their lives.

Keywords: OFC care, Asian, role of cleft surgeon

## Nutrition Status of Children with Cerebral Palsy: Usefulness of New United States Cerebral Palsy Growth Chart Mallawaarachchi M.S.S.<sup>1</sup>, Jayatissa R.<sup>2</sup>, Jagoda J.S.<sup>3</sup>, Tennakoon G.M.J.<sup>4\*</sup>, Mallawaarachchi W.P.P.K.<sup>5</sup>

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**Introduction and Objectives:** Nutritional status is a key factor affecting the quality of life in a child with Cerebral Palsy (CP). Early identification and management of malnutrition in CP children is important for long term care. The objective of the study was to assess the nutritional status of children with CP with new CP growth chart in United States (US) compared with WHO growth chart to identify the most useful tool.

**Methods:** A cross sectional study was conducted in 102 children with cerebral palsy attending a rehabilitation clinic in a tertiary care hospital in Colombo, Sri Lanka. Interviewer administered questionnaires was administered to the mother or to the immediate care giver to collect relevant information. A 24-hour diet recall was also done. The level of Gross Motor Function Classification System (GMFCS) was used and the level of GMFCS was categorized into five, walks independently without limitations, walks independently with limitations, walks independently using a handheld mobility device, self-mobility with limitations, may use powered mobility and no head control with transported in a manual wheelchair. Weight and height/length was measured and plotted in both weight-for-height for children under 5 years and BMI-for-Age for children above 5 years using WHO growth charts and US CP charts according to the level of GMFCS.

**Results:** Malnutrition by GMFCS of the study population was 1.0%, 5.8%, 8.7%, 4.8%, and 22.3% in level 1, 2, 3, 4 and 5 respectively. Overall malnutrition in the study population was 53% using WHO charts and 43% using US CP growth charts (kappa 0.76). 24-hour dietary recall revealed 33.7% and 63.5% of them had low protein and calorie intake respectively.

**Conclusions:** High level of malnutrition was observed in CP children after applying both WHO and US growth charts both of which had good agreement. It is useful to detect malnutrition with the level of GMFCS for targeted interventions.

Keywords: US cerebral palsy Growth chart, WHO Growth chart

## PP-39 Oral Health Care Package for High Caries Risk Children with Birth Defects Perera I.R.<sup>1\*</sup>, Wickramaratne P.W.N.<sup>2</sup>, Gajanayake C.<sup>3</sup>, Herath E.M.U.C.K.<sup>4</sup>, Perera M.L.<sup>5</sup>

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**Introduction and Objectives:** Advancements in medical technologies and services have enhanced life spans of better quality for children with birth defects. Sri Lanka as a lower middleincome country, aspired to ensure universal health coverage for children including those with birth defects. They carry a high oral disease burden which could negatively impact on their existing medical conditions. Nevertheless, oral health status among children with birth defects is often being overlooked compounded by their existing prioritized health needs. Against this backdrop, this analysis is aimed to describe characteristics related to oral health among a sample of children with birth defects presented to a multi-specialty tertiary care public dental hospital and on a special oral health care package for high caries risk children in the above sample.

**Methods:** Sample contained 79 children with birth defects who visited the Preventive Oral Health Unit of the National Dental Hospital Sri Lanka from 1st January 2017 to 29<sup>th</sup> April 2019

Data were extracted from unit's data base and analysed using SPSS-21 package. The level of cooperation of a child for oral health care was assessed by child's compliance with mouth examination and provision of simple dental treatment in a child-friendly dental setting.

**Results:** Children with birth defects carried a high burden of untreated dental caries. The mean number of non-cavitated decayed teeth was  $2.65 \pm 3.62$ , while cavitated decayed teeth was  $2.97 \pm 2.97$  and pulp exposed teeth was  $3.59 \pm 3.60$  respectively. All children practiced unhealthy dietary habits with daily consumption of biscuits, buns, toffees and chocolates. Of children, 50.6% deemed cooperative for receiving simple dental care. All parents and other care givers were counselled on healthy dietary pattern and optimal oral hygiene practices. Cooperative children with non-cavitated and cavitated dental caries were provided with professional fluoride application, fissure sealant application for permanent molars and restorations. Uncooperative children with symptomatic pulp exposed teeth received dental care under general anesthesia for pulp therapy or extractions in extreme cases.

**Conclusions:** The innovative oral health care package was comprised of oral health promotion combined with simple and advanced oral health care provision. Since children with birth defects are more prone to dental caries, such proactive innovative approaches could immensely benefit those children.

**Keywords:** Birth defects, children, high dental caries burden, oral health promotion, oral health care package

Pioneering a Youth-Led Community-Based. Preconception Health Awareness Campaign Utilizing the LINC Framework: The Volunteer Youth Leaders for Health (VYLH): Philippines Experience

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**Introduction and Objectives:** The Volunteer Youth Leaders for Health (VYLH)-Philippines is a network of youth that fosters volunteerism and public service to improve birth outcomes through advocacy. In the absence of a national program on preconception healthcare in the country, the first youth-led community-based preconception health awareness campaign was piloted to promote healthy lifestyle, infection prevention, good nutrition, and responsible parenthood or the LINC framework among the local health workers, youth volunteers, and reproductive age women residing in the municipality of Los Baños, Laguna, Philippines.

**Methods:** The project focused on four major components: 1) Partnerships with the local government unit, municipal health office, and youth organizations; 2) Assessment of baseline knowledge on preconception health in the community; 3) Development of culture and gender-sensitive modules and information materials on preconception care; and 4) Conduct of health education activities on preconception care. Knowledge increase among participants of the health education series was analyzed through percent change from pre- and post-session test results. Social media were also utilized to increase reach of awareness campaign.

**Results:** Survey revealed low level of baseline knowledge on preconception health among women of reproductive age (20-35 years) in the community, nonetheless 97% agree that there is a need for health education services for the reproductive age. At the end of project implementation, the estimated Filipino reached by the awareness campaign is 10,421 (9,249 Filipino youth). There was a significant knowledge increase among the 24 local health workers (84.5%) and 32 youth volunteers (46%) trained as program facilitators and peer educators. For the target women residents of reproductive age (20-35 years) in the community, a knowledge increment of 45.5% was observed after the five-day health education series.

**Conclusions:** The youth plays a significant role in initiating awareness on preconception health to improve birth outcomes and promote wellness in the county given that better birth outcome is relative to the knowledge on preconception health especially of those in the reproductive age. It is highly envisioned that with the success of this project, this initiative can be further replicated and used as a novel model for youth involvement in preconception health promotion.

Keywords: Preconception Health, LINC framework, VYLH, Youth-Led, Awareness Campaign

## Prevalence and Spectrum of Congenital Anomalies in a Tertiary Care Centre of North India: 20 Years' Experience Kumar P.<sup>1</sup>, Kumar J.<sup>1\*</sup>, Saini S.S.<sup>1</sup>, Sundaram V.<sup>1</sup>, Mukhopadhyay K.<sup>1</sup>, Dutta S.<sup>1</sup>, Kakkar N.<sup>1</sup>

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**Introduction and Objectives:** The frequency and type of Congenital Anomalies (CAs) vary in different populations due to variations in ethnicity, socioeconomic status, nutrition, environmental factors, maternal age and lifestyle. It is not clear if there has been a change in the prevalence or pattern of CAs in the last couple of decades along with the changes in care patterns. For the implementation of preventive strategies and treatment services, a systematic assessment of CAs and the exact estimate of region-wise prevalence are needed. We aimed to determine the prevalence, trend, spectrum and outcomes of CAs at a tertiary care center over two decades.

**Methods:** Prospectively collected data of all live births from January 1998 till December 2017 was screened for infants with major CAs. International Statistical Classification of Diseases and Related Health Problems (ICD-10) was used for uniformity. Further sub-categorization was done as per World Health Organization birth defects surveillance manual.

**Results:** Of 86,850 live births, 1578 [1.82%, (95% CI 1.73-1.91%)] neonates had a major CA. The overall prevalence of CAs was 182 (95% CI 171 -193) per 10,000 live births. There was no significant change in proportion of CAs over two decades. CA related death rate was 6.78 per 1,000 live births and no significant change was observed in the contribution of CAs to overall mortality over two decades. Anomalies of circulatory system were the commonest (28%) followed by musculoskeletal system (18.6%), urinary system (14.3%), respiratory system (9.0%), nervous system (8.5%) and digestive system (6.5%). The prevalence of circulatory system (Q20-Q28) anomalies was 71.5 (95% CI 65-77) per 10,000 live births and most of them are amenable to corrective surgery. Majority of anomalies were malformation (90.7%) followed by deformation, dysplasia and disruption. According to clinical presentation, most (69.6%) presented as an isolated anomaly.

**Conclusions:** CAs are responsible for significant mortality and morbidity, supporting the need for national surveillance program and birth defect services. A significant proportion of CA is either preventable or amenable to corrective surgeries.

Keywords: Birth defect, Congenital anomaly, Malformation, Mortality, Neonate

### PP-42 Prevalence of Renal Cystic Disease in 467,575 Births in the Surveillance Program of Congenital Defects Since 2003 to 2018 in Bogotá- Cali Madariaga I.<sup>1</sup>, Orozco C.<sup>2</sup>, Hurtado P.<sup>2</sup>, Holguín J.<sup>3</sup>, Rodríguez C.<sup>4</sup>, Zarante I.<sup>1\*</sup>

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**Introduction and objectives:** Renal cystic disease (RCD) refers to a heterogenic kidney pathology involving cystic lesions in kidney parenchyma, differing in location, heritage and clinical overcomes among them. Ultrasound represents the least invasive way to find out these diagnoses during prenatal screening. The purpose of this study was evaluating the postnatal prevalence of RCD and its relation to sociodemographic factors in a cohort since 2003 to 2018 from the Surveillance Program of Congenital Defects (SPCD) in two Colombian cities.

**Methods:** Data were collected from the Bogota and Cali SPCD since 2003 to 2018, using the Latin American Collaborative Study for Congenital Malformations (ECLAMC) protocol. We reviewed the database and extracted data related to patients with "kidney polycystic disease" (KPD) and "multicystic dysplasic kidney" (MDK), obtaining 55 cases. We proceeded to analyze variables as sex, weight, prenatal diagnosis, other malformation associated and live or death born.

**Results:** 467,575 births were reviewed, 54 patients with renal cystic disease, with a prevalence of 1.15 per 10,000 (95% CI; 0.87-1.51) being more frequent in males 53, 7% (n=29). From this, 50% were isolated (n=27) and 50% (n=27) had another malformation associated. Of them, 96.2 % (n= 52) were live births and 3,7% (n=2) corresponds to stillborn and 37% (n=18) of the cases had prenatal diagnosis.

**Conclusions:** Prevalence of RCD is lower than the rates already reported in literature (1 per 1000 to 1 per 10000 in dominant KPD vs recessive KPD respectively and 1 per 4300 births in MDK). These cases were classified as IIb\* (SPCD database classification) corresponding to a high risk for mortality and disability. Diagnosing this pathology can be done with ultrasound prenatal screening (prenatal ultrasound sensitivity 88% in urological anomalies) being possible facilitating an early treatment and prevent fatal outcomes.

Keywords: Renal cystic disease, ECLAMC, Surveillance Program of Congenital Defects

## Prevalence of Talipes Equinovarus, Congenital Dislocation of the Hip, Cleft Lip/Cleft Palate, Down Syndrome and Neural Tube Defects among Live Newborns in Anuradhapura, Sri Lanka.

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**Introduction and Objectives**: The study was conducted 17 years ago in 2002-2003. It was presented at the Anuradhapura Clinical Society meeting in 2003. The findings were published in the Sri Lanka Journal of Child Health in 2007. Only a handful of papers on this subject have been published since then. A birth defect registry was suggested in the discussion. This is a landmark study of historical importance. The objective of the original study was to determine the prevalence of talipes equinovarus, congenital dislocation of the hip, cleft lip/cleft palate, Down syndrome and neural tube defects among live newborns at Anuradhapura General Hospital, Sri Lanka.

**Methods:** Babies born from February 2002 to January 2003 were examined for the above five congenital anomalies. Anomalies were recorded in a register and live birth prevalence rate (LBPR) for each anomaly were calculated.

**Results:** 9105 newborns were examined for the 5 selected congenital anomalies. LBPR per 10,000 live births of the 5 anomalies were as follows: talipes equinovarus 31.8, congenital dislocation of hip 26.3, cleft lip/cleft palate 21.9, Down syndrome 20.8 and neural tube defects 13.2.

**Conclusions:** Live birth prevalence rates for talipes equinovarus, congenital dislocation of hip, cleft lip/cleft palate, Down syndrome and neural tube defects were 31.8, 26.3, 21.9, 20.8 and 13.2 per 10000 live births respectively.

**Keywords:** Prevalence, Talipes equinovarus, Congenital dislocation of hip, Cleft lip/palate, Down syndrome, Neural tube defects, Anuradhapura, Sri Lanka

## Prevalence of Various Birth Defects in Northern India- A Multicentric New-Born Birth Defect Surveillance (NBBD)

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**Introduction and objectives:** Congenital anomalies were not considered a significant contributor to mortality in Low and middle income countries as the proportion of other causes like perinatal complication, infections and malnutrition was very high. Over years improvement in health care services resulted in appropriate management of intrapartum complications, prematurity and low birth weight which ultimately increases the contribution of congenital anomalies. This epidemiological transition argues for the need for the data on magnitude, type of congenital anomalies and distribution in particular geographical area. In 2014 WHO SEARO created an online database for the surveillance of birth defects at birth for both live and stillbirths. This paper is an attempt to look for the pattern of different congenital anomalies and birth prevalence in 10 identified hospitals of Northern India over a period of 4 years (2015-2018)

**Methods:** This is a prospective Newborn birth defects surveillance being done in 220 hospitals of nine countries and in this paper the data of northern India which include 10 identified hospitals with high delivery load is analyzed. The ascertainment of birth defects cases were done by prenatal ultrasound, clinical examination, neonatal ultrasound and other investigations. The standard structured birth defect surveillance tool is used for data variable and uploaded online. **Results:** A total of 5012 cases of birth defects were reported from 3, 42.770 births from 10

identified hospitals over a period of four years. The calculated birth prevalence is 152.1 per 10,000 live births. Out of 5012 cases of birth defects 641 (12.7%) were reported among stillbirths. The common were of central nervous system (27.3%), musculoskeletal (22.6%) and cardiovascular system (13.8%).

**Conclusions:** This data identifies a very high prevalence of birth defects in northern India and maximum number is of neural tube defects which are preventable. The birth defect services like preconception folic acid, universal prenatal screening and preventive strategies like folic acid fortification would lead our way for preventing this huge burden.

Keyword: Birth defects, Prevalence, Birth prevalence, Neural tube defect, Folic acid

# Prevalence, Risk Factors and Outcome of Congenital Birth Defects among Live Neonates in a Tertiary Care Hospital in North India

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**Introduction and Objectives:** India accounts for the largest share of congenital birth defects (CBD) in the world. CBD accounts for 13-16% of neonatal deaths here. Being a medical centre with a large birth cohort, we conducted a study to find the prevalence, characteristics, risk factors and outcome of neonates with CBD.

**Methods:** A cross sectional study was conducted among consecutively born live neonates in of King George's Medical University, India from Aug 2018 to April 2019. The neonates were examined for congenital anomalies and mothers were interviewed for demographic variables. Of the 5455 babies delivered in study period of 8 months, 69 had CBD.

**Results:** The most common CBD was congenital talipes equinovarus (CTEV) - 35(53.8%) followed by meningomyelocele- (MMC) 17(26.2%) and congenital heart disease- 9 (13.8%). Most mothers (63.1%) were in 20-30 years age group, housewives (78.5%), resident of urban area (63.1%), multifarious (60%), literate (75.4%), received antenatal care (89.2%) and without consanguinity (95.4%). CBD neonates were mostly term gestation (61.5%), appropriate for gestational age (89.7%), male female ratio of 1.3. MMC cases were associated with illiterate mothers (p=0.002), rural residence (p=0.029), without antenatal care (p=0.004), and without folic acid supplementation (p=0.003). Only 34 of 65 (52.1%) were discharged, 16/65 (24.6%) died and 15/65 (23.1%) left hospital against medical advice (LAMA) despite counselling. MMC constituted 11 of 15 of LAMA (73.3%) (p <0.001).

**Conclusions:** Prevalence of CBD was 1.2 % in our study, with musculoskeletal system (CTEV), predominant system being involved followed by nervous system (MMC). The outcome of patients was dismal especially the MMC patients, which points towards the better preventive strategies like folate supplementation of mother and better palliation of these cases in our set up. **Keywords:** Congenital birth defect, Live neonate, Outcome, Prevalence, Risk factors.

## Profile of Nutritional Status among Children with Cerebral Palsy in Tertiary Care Rehabilitation Center, Colombo, Sri Lanka

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**Introduction and Objectives:** Nutritional status is a key factor affecting the quality of life in a child with Cerebral Palsy. The objective of the study was to evaluate the nutritional status of children with cerebral palsy with relevance to current defined growth parameters.

**Methods:** Descriptive cross-sectional study was among 104 children with cerebral palsy, attending a rehabilitation clinic in a tertiary care hospital in Colombo, Sri Lanka. Interviewer administered questionnaires to collect primary data and growth parameters were measured.

**Results:** Mean age was 6 years with male preponderance (68%). Mother (87%) was the main care giver and overall care was satisfactory (81%). Majority of the children (54%) were suffering from other medical conditions and seizure disorders (51%) were the commonest amongst them. Children with GMFCS (Gross Motor Function Classification System) level 4 and 5 with severely limited independent physical limitations were common (61%).

In consideration of nutritional status according to weight for height or body mass index (BMI), malnutrition (53.9%) was the commonest with 20%, 25% and 55% with severe acute malnutrition, moderate acute malnutrition and chronic malnutrition, respectively. While 38.7% had normal nutritional status, 7.7% were overweight or obese. Although majority (66%) had adequate protein intake (p=0.03), energy intake was low in 63% (p<0.001). Normal solid diet (77%) was administered orally (99%) and food supplementation was given for 51% (p=0.04) and 67% had no vitamin supplementation.

**Conclusion:** Malnutrition is common among children with Cerebral palsy presented tertiary care rehabilitation center in Colombo.

Keywords: Malnutrition, Cerebral Palsy, Sri Lanka

## Quality of Life and Household Economic Burden of Primary Caregivers Having Children with Congenital Heart Disease Awaiting Cardiac Surgery Attending the Cardiology Clinic at the Lady Ridgeway Hospital for Children, Sri Lanka Warnakulasooriya P.<sup>1\*</sup>, Kasturiaratchi K.<sup>2</sup>

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**Introduction and objectives:** Congenital heart disease (CHD) is the commonest type of birth defect, of which the estimated prevalence is around 8-12/1000 worldwide. Nearly 4% of all neonatal deaths are due to CHD. Caregivers of children with CHD are easy victims of low quality of life (QOL) and instability of household cost. Lady Ridgeway Hospital (LRH) is the largest government hospital for children in Sri Lanka and the only tertiary care referral center for children with CHD in the country. According to hospital data, over 40% of these children do not have appropriate access to early intervention and are compelled to wait until they get their turn. This study was conducted to determine the quality of life and household economic cost of primary caregivers having children with CHD awaiting cardiac surgery attending the cardiology clinics in LRH.

**Methods:** This was a descriptive cross-sectional study conducted using consecutive convenient sampling method over three months at cardiology clinics in LRH (July 2017 to September 2017). The sample consisted of 422 caregivers having children with CHD awaiting cardiac surgeries. An interviewer-administered questionnaire was utilized to obtain data. Pre-tested validated WHOQOL-BREFF was used as a tool for assessing QOL. Costing related questions assessed the cost borne by the caregiver attending clinic and they were prepared following extensive review of literature and by consulting local experts. Univariate analysis and logistic regression were performed to detect associations.

**Results:** Mean score of the QOL was comparatively low in all domains except in social - relationship domain. Quality of life was associated with age of the caregivers, educational level, marital status, number of children, income, type of heart disease and postponement of the surgery. Median direct expenditure per clinic visit was LKR 1800 (range of LKR 1175 to 3000). Median indirect cost was Rs. 1000. Out of all care givers, 28.7% of the care givers were falling in to catastrophic expenditure during that particular month of clinic visit.

**Conclusions:** Quality of life of primary caregivers was comparatively low in all domains except in social-relationship domain. Nearly a third are having catastrophic expenditure on clinic visit alone. Physician and health care managers should make a proper planning to improve the QOL of affected caregivers.

**Keywords:** quality of life, primary care givers, household economic cost, children, congenital heart disease, cardiac surgery

## Referral Mechanism for Periconception Folic Acid Supplementation to Newly Married Couples to Prevent Neural Tube Defects Giri Y.<sup>1</sup>, Regmi K.<sup>1,2\*</sup>, Rajthala S.<sup>1</sup>, Koirala S.<sup>1</sup>

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**Introduction and Objectives:** Birth defects specifically congenital anomalies represent fourth largest cause of neonatal death in Nepal. Currently, food fortification is the only approach implemented to tackle growing burden of neural tube defects at population level. However, periconception folic acid supplementation is a well-established intervention and can significantly reduces incidence of neural tube defects and other birth defects. Identification and tracking of couple intending to plan pregnancy for preconception care is extremely challenging as more than fifty percent of the pregnancies go unplanned. Therefore, we aimed to develop the referral mechanism to encourage newly married women plan their pregnancy and bring them to health facility for periconception care counselling and folic acid supplementation.

**Methods:** A rural municipality setting in eastern Nepal, consisting two primary health care centers, three birthing centers, two community health unit and 11 local government wards was selected to develop and pilot the referral mechanism for folic acid supplementation. Facility and local government readiness assessment and mapping were done using modified version of health facility readiness assessment tool. Stakeholders such as members of local government, skilled birth attendant, health workers, female community health volunteers, vital registration authorities were interviewed, oriented and re-oriented to establish the linkage between local government wards and health facility.

**Results:** Vital registration unit of local government, antenatal care clinic, and female community health volunteers were identified as the institutional point of contact. A referral tool (best wished card) was developed with high internal consistency. The ward marriage registration office was identified to issue the referral tool and antenatal clinic nurse as reacting to referral tool. Skill birth attendant provided folic acid with the brief preconception care counselling. Piloting of the tool revealed 20%(n=97) of the total pregnancy (N=493) newly married couple from all local government wards were referred to the health facility for periconception folic acid supplementation.

**Conclusions:** Referral linkage and locally adopted referral tool were found useful to bring newly married women for utilization of preconception folic acid supplementation. Tracking the utilization of folic acid and its impact on selected pregnancy outcomes including neonatal death, still birth, neural tube defects could be future research priorities.

**Keywords:** Preconception care, Folic Acid supplementation, Neural tube defects, Birth defects, Prevention.

# Relative Prevalence and Outcome of Antenatally Detected Congenital Malformations Using ICD -10 Codes in a Tertiary Care Hospital of India Kumar M.<sup>1\*</sup>, Singh A.<sup>1</sup>, Mehta R.<sup>2</sup>, Raina N.<sup>2</sup>

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**Introduction & Objectives:** The developing countries together constitute more than 80% of the world population and have far greater burden of mortality arising from congenital anomaly. The use of ICD-10CM for disease classification is important for uniformity in the surveillance systems. To find out the relative prevalence of antenatally diagnosed congenital malformations using ICD -10 CM and to determine their postnatal outcome in a tertiary care hospital of India.

**Methods:** This prospective study was conducted as a part of WHO SEARO project from April 2014- March 2018 and included all women with prenatally detected major congenital malformations. At delivery, the cases with birth defect were classified according to the ICD -10 classification system, the outcome was assessed in terms of survival at birth.

**Results:** In the study period of five years, there were total 1012 cases with antenatally diagnosed birth defect, 95 of them were lost to follow up, 917 were followed till delivery. The mean gestational age of presentation was 27.3 weeks, only 40.1% babies were delivered live, 31.5% were stillborn. About 260 women (28.4%) came before 20week gestation and underwent pregnancy termination. The CNS anomalies (ICD 00-07) were most common (47%), and contributed to the stillbirths (17.3%). The survival was more than 50% in cases with renal (ICD 60-64), digestive (ICD 30-45) and respiratory system malformations (ICD 30-34), it was least in hydrops (ICD 87) and chromosomal abnormality (ICD 90-99).

**Conclusion:** The ICD-10-CM code sets have updated medical terminology and classification of diseases and allows comparison between units or countries. In India, the majority of congenital anomalies present late in gestation. The CNS anomalies were most common and contributed most to the stillbirths.

Keywords: Birth defect, ICD -10 CM coding, India, Stillbirth, postnatal outcome

# Role of Maternal Blood Oxidative Stress Markers and Inflammatory Markers in Unexplained Stillbirth: A Pilot Case-Control Study Aggarwal R.<sup>1\*</sup>, Kumari A.<sup>1</sup>, and Mehndiratta M.<sup>1</sup>

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**Introduction and objectives:** Stillbirth is defined as "fetal death at 22 weeks of gestation or birth weight of 500 grams. Despite extensive work-up, around 25-60% of stillbirths remain unexplained. Hence it becomes difficult to counsel the parents regarding the cause of stillbirth and the future risk of stillbirth in subsequent pregnancy. The risk of unexplained stillbirth increases late in pregnancy, suggesting that placental aging is an etiological factor which is associated with oxidative damage. Other studies have documented inflammatory lesions in ~37 to 40% of placentas from stillbirth. We hypothesize that oxidative stress and inflammation has a role to play in unexplained stillbirth. This study aims to identify oxidative stress markers i.e., telomere length and relative mtDNA content and markers of inflammation i.e., hsCRP and Ferritin in maternal blood samples in cases of unexplained stillbirth and to compare their levels with healthy controls.

**Methods:** A case control study was conducted at the Department of Obstetrics &Gynecology, Biochemistry and Pathology, University college of Medical Sciences, Delhi. Pregnant women with stillbirth with gestational age of 22 weeks with negative work-up were considered 'unexplained stillbirth' and included as cases. Controls were gestational age matched low risk pregnant women. Telomere length was compared by unpaired t-test. Since relative mitochondrial DNA content, hsCRP and ferritin had skewed distribution, they were compared using Mann-Whitney U test.

**Results:** Both telomere length (220.04 $\pm$ 77.02 versus 281.17 $\pm$ 82.15kb; p=0.004) and relative mtDNA content(169.88 $\pm$ 82.75 versus 257.68 $\pm$ 162.69; p=0.011) were significantly reduced in stillbirth compared to livebirths suggesting increased oxidative stress. Serum ferritin levels (143.73 $\pm$ 78.05 versus 80.39 $\pm$ 42.03ng/ml; p=<0.001) and hsCRP levels (19.88 $\pm$ 6.32 versus 5.95 $\pm$ 5.39mg/L; p=<0.001) were significantly increased suggesting role of inflammation in stillbirth.

**Conclusions:** The association of telomere length, relative mtDNA content, serum ferritin and hsCRP with unexplained stillbirth proving the role of oxidative damage and inflammation, calls for early treatment measures to ameliorate this stress at the beginning of pregnancy. Methods for detecting placental aging through these markers may have clinical utility in predicting those at high risk of stillbirth who may benefit from prophylactic caesarean section.

**Keywords:** Unexplained stillbirth, telomere length, relative mitochondrial DNA content, Ferritin, hsCRP

# An Intervention to Improve the Health Status of Low-Income Women Living in Remote Hardest to Reach Areas of Uganda Before, During and after Pregnancy Sylivia K.<sup>1\*</sup> and Harrison O.<sup>1</sup>

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**Introduction and Objectives:** Research in Uganda shows that long distances to health facilities and poor roads affect women living in remote, hardest to reach areas to effectively access and utilize health care services throughout pregnancy stages, but little has been done to address these challenges. We addressed this through our motorcycle project, of which the aim was to increase access to and utilization of quality and timely maternal child health care services among low income women in remote hardest to reach villages by providing motorcycles to nurses to provide home-based care.

**Methods:** In 2017, we carried out a qualitative study in remote Namayingo district for better understanding of the problem women faced in accessing health care. In-depth interviews were conducted with 45 women. Two focus group discussions were carried out among purposively selected community leaders and health workers to obtain comparative data. Analysis of responses revealed that the challenges were high transport costs to government hospitals and that nearby private clinics were expensive. With the findings of the qualitative study, an intervention was designed to use motorcycles to transport nurses to remote villages to provide home based maternal-child health care services including education on a healthy lifestyle, vaccination, family planning, counselling, medical checkup and treatment of illnesses and appropriate referrals.

**Results:** Preconception health information was given to 480 mothers and 169 pregnant women have received antenatal care. 126 pregnant women transported to health facilities for delivery and are currently receiving postnatal checkups. Sexually transmitted infection cases were diagnosed among 102 women and treated and transported to hospitals in cases of referrals.

**Conclusions:** Our motorcycle project has helped to bring healthcare to mothers and children living in remote hardest to reach areas.

Keywords: Accessibility, Universal Health coverage, Hard to reach areas, Maternal care

## PP-52 Surgical Care for Myelomeningocele and Congenital Hydrocephalus in a Large Urban Teaching Hospital in Ethiopia Kancherla V.<sup>1\*</sup>, Koning M.<sup>2</sup>, Koning J.<sup>2</sup>, Woldemr M.<sup>3</sup>, Biluts H.<sup>3</sup>

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**Introduction and Objectives:** Myelomeningocele (open spina bifida) and congenital hydrocephalus are two serious and common birth defects. They are associated with high morbidity and mortality, especially in resource-poor settings. Early surgical intervention, within 48 hours after birth, in a tertiary care hospital, is a recommended guideline to prevent health complications and death among those affected.

**Methods:** In our descriptive cross-sectional study design, we examined selected clinical characteristics among children who were treated in a large urban teaching tertiary care hospital in Addis Ababa, Ethiopia. Data were collected from surgical care clinical records from January to June, 2017. We examined sex, age at surgery, diagnosis documented for surgery, and the type of surgical repair. Data were summarized using basic statistics, including univariate analysis for age at surgery, and frequencies and percentages for selected categorical variables that were available for analysis.

**Results:** There were 250 surgeries recorded during the study period, ranging from 7-14 surgeries per week, conducted on consecutive Saturdays. The sex ratio was 1:1. Overall, 78% of the diagnoses were related to myelomeningocele, and the remaining were for hydrocephaly (16%), post-myelomeningocele hydrocephaly (4%), and encephalocele (2%). Majority of surgical procedures were conducted for myelomeningocele repair (75%). Shunt repairs constituted about 15% of surgeries. The median age at surgery was 10.5 days, and overall age range at surgery was between 1 day and 8 years. Specific age distribution for the time of surgical repair was 8% at age 1-2 days, 68% at ages 3 days to 1 month; 13% at ages over 1 month to 6 months; 8% at ages 6 months to 1 year; 1.5% at ages over 1 year to 5 years, and 1.5% at ages 5 years to 10 years. Less than 10% of all surgeries for myelomeningocele occurred during the recommended period (i.e., within 48 hours after birth).

**Conclusions:** There is a need to integrate prenatal diagnosis for myelomeningocele and hydrocephalus with timely neurosurgery immediately after birth. High risk pregnancies with myelomeningocele should be identified early, and referred to deliver in tertiary care hospitals with a trained medical personnel specialized in pediatric neurosurgery.

**Keywords:** Clinical Care; Hydrocephalus; Neural Tube Defects; Spina Bifida; Surgery

## PP-53 Synaptic Vesicle Associated Transcripts (SVATS) in Circulating Exosomes of Autistic Children and Physio-Pathological Pregnancies Pan J.<sup>1</sup>, Zhong N.<sup>1,2\*</sup>

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**Introduction and Objectives:** Impaired synaptic vesicle recycling represents a critical node for disease pathologies underlying the development of autism. The developing brain in utero and during the early years of childhood is highly vulnerable to environmental influences. Our earlier study identified a subset of significant alterations of long noncoding RNAs (lncRNAs) and messenger RNAs (mRNAs) of synaptic vesicle associated transcripts (SVATs) in autism. We further assessed SVATs in exosomes (EXs) for a purpose of applying SVATs as the biomarkers for the early prediction of autism.

**Methods:** Genotyping of SVATs were performed in the peripheral EXs that were isolated from circulating plasma of autism children and women with physio-pathological pregnancies. SVATs wrapped in EXs were identified by RNA-seq quantitated with qRT-PCR, among normal full-term birth (FTB), full-term premature rupture of fetal membrane (PROM), pre-term premature rupture of fetal membrane (pPROM), spontaneous preterm labor (sPTL), preeclampsia (PE), and gestational diabetes mellitus (GDM).

**Results:** In addition to that circulating EXs were determined from placental trophoblasts, Western-blot of neuronal biomarkers Nestin and L1CAM demonstrated that a portion of EXs was derived from neurons. Significant gene expression profiles (GEPs) of six-pairs of SVATs of mRNA-lncRNA in plasma EXs of autistic children further confirmed their associations with autism. Longitudinally quantitative measurement from 11 gestational week (GW) of the 2nd trimester to the term of birth, demonstrated there were two expression peaks in physiological pregnancies. GEP of SVATs in sPTL was significantly different from that in PE and DGM.

**Conclusions:** Our results showed that the ASD-associated SVATs can be measured in plasma EXs for a minimal invasive real-time evaluation of intrauterine, in addition to postnatal, and fetal development. Our studies demonstrate that assessment of SVATs in maternal EXs opened a new avenue for early prediction of neurodevelopment during physio-pathological pregnancies. **Keywords**: Synaptic vesicle associated transcripts, SVATs, exosomes, autistic children

#### The Need for Domestic Violence Policies among Lebanese Mothers and Displaced Syrian Mothers Tamim H 1 El Havek C V <sup>2,3\*</sup> Nakad P <sup>2,3</sup> Alameh M <sup>3,4</sup> Al Hamod D <sup>3,5</sup> Adra A <sup>3,6</sup> Theis F <sup>3,7</sup>

# Tamim H.<sup>1</sup>, El Hayek G.Y.<sup>2,3\*</sup>, Nakad P.<sup>2,3</sup>, Alameh M.<sup>3,4</sup>, Al Hamod D.<sup>3,5</sup>, Adra A.<sup>3,6</sup>, Tleis F.<sup>3,7</sup>, and Yunis K.A.<sup>2,3</sup>

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**Introduction and Objectives:** Recently, violence against women is under the spotlight throughout the Middle-East. In Lebanon, a number of civil society initiatives have attempted to break the silence, surrounding domestic violence, resulting in a weak new law. This study looks at factors associated with the importance of domestic violence policies for families' wellbeing.

**Methods:** A cross sectional study (October 2017-June 2018) was conducted in four hospitals part of the National Collaborative Perinatal Neonatal Network in Lebanon, on 1,792 mothers (57.9% Lebanese Mothers (Lbm) and 42.1% displaced Syrian Mothers (dSm)). The outcome variable was importance of domestic violence policies for family wellbeing, ranked from 0 (lowest) to 10 (highest). A stepwise multivariable linear regression was performed to examine factors associated with the importance of domestic violence policies. The independent variables in the models were related to socio-demographic characteristics of both parents, and the family composition.

**Results:** The collected sample was distributed as follows: 42.1% dSm giving birth in a hospital in the southern part of Mount Lebanon, 22.3%, 19.8%, and 15.7% Lbm giving birth in central Beirut, east Beirut, and the North of Lebanon respectively. Hospital, education of the father and having an external family living at home were the only variables that were retained in the stepwise linear regression model. Compared to mothers who delivered in central Beirut, on average the importance of domestic violence policies for family's wellbeing were lower for the hospital in Mount Lebanon (beta:-6.77, 95%CI:-7.04,-6.49) and east Beirut (beta:-0.92, 95%CI:-1.24,-0.59) but higher for mothers who delivered in the North (beta:0.61, 95%CI:0.30,0.90). Additionally, a decrease in the domestic violence importance for family's wellbeing was observed for fathers with an intermediate or high school education compared to elementary school education (beta:-0.26, 95%CI:-0.50,-0.03). Also, having external family members living at home showed a reduction in the importance of domestic violence policies for family swellbeing were lower for family's wellbeing (beta:-3.6, 95%CI:-0.62,-0.70).

**Conclusions:** Unlike LBM, DSM do not rate domestic violence policies for families' wellbeing high. Also, the presence of external family members and having an intermediate or high school education affect rating of policies for families' wellbeing.

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Keywords: Domestic violence, Arab, displaced mothers, displaced population

## Two Siblings with Autosomal Recessive Primary Microcephaly due to Novel Compound Heterozygous Null Variants in the ASPM Gene

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**Introduction and objectives:** Pathogenic variants in the ASPM (abnormal spindle-like, microcephaly-associated) [MIM:605481] gene located on chromosome 1q31.3 are known to be associated with autosomal recessive primary microcephaly-5 (MCPH5) [MIM:608716]. Herein, we report a Sri Lankan family with 2 siblings with primary microcephaly and severe intellectual disability in whom novel compound heterozygous null variants in the ASPM gene were identified. **Methods:** A 9-year-old girl and her 5-year-old brother with primary microcephaly and severe intellectual disability were referred for genetic evaluation. Whole exome sequencing (WES) was performed on both siblings and their parents. Data were analyzed utilizing the computational resources at the National Institutes of Health high-performance computing Biowulf cluster.

**Results:** The 2 children were born to a non-consanguineous couple. Their elder sibling and both parents were apparently healthy with no significant family history of any congenital anomalies. The 2 siblings presented with primary microcephaly (occipitofrontal circumference less than 3 standard deviations of the age-related mean), severe intellectual disability, speech delay and absence seizures. Computed tomography scan of the brain showed fused lambdoid sutures and narrowing of the sagittal and coronal sutures in the female proband and premature fusion of the metopic suture in the male proband. The electroencephalograms in both children were normal. WES identified novel compound heterozygous null variants in the ASPM gene [NM\_018136.4]: a paternally inherited pathogenic nonsense variant c.7212delC:p.(Gln2438Argfs\*20).

**Conclusions:** We report novel compound heterozygous null variants in the ASPM gene in 2 siblings with autosomal recessive primary microcephaly-5. Establishing the precise genetic diagnosis is beneficial for clinical management and provision of accurate genetic counseling for families affected with primary microcephaly.

**Keywords:** Autosomal recessive, Compound heterozygous, Primary microcephaly, Rare disorders, Whole exome sequencing

### PP-56 Use of Antenatal Corticosteroids for Threatened Preterm Birth: From Implementation Research to Policy Revision Kankaria A.<sup>1\*</sup>, Duggal M.<sup>1</sup>, Sarkar D.<sup>1</sup>, Chauhan A.<sup>2</sup>, Litch J.A.<sup>3</sup>, Kumar P.<sup>1</sup>

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**Introduction and Objectives:** The WHO recommends that antenatal corticosteroids (ACS) should only be administered where the specific conditions are met at facility level: accurate assessment of gestational age, presence of imminent preterm birth, absence of maternal infection, availability of adequate childbirth care and preterm newborn care. Considering this, the objective of the study was to assess of facility readiness regard to current ACS utilization practices and to expand the existing Government of India Operational Guideline on use of ACS for Preterm Labour. **Methods:** This study used mixed method approach to assess current use of ACS in Hisar and Ambala, Haryana, India. The facility leadership, programme implementers, facility assessment, provider interview, clinical verification, maternal recall interview, HMIS indicator tools were used in the study. At 37 facilities, a total of 20 leaders, 86 health care providers, 19 mothers with preterm delivery who received ACS and 7 program officers/implementers were interviewed were studied. Based on the findings, critical areas were identified that urged for expansion of existing operational guideline of ACS. Consequently, an Advisory was realized in order to adhere prerequisite conditions for use of ACS.

**Results:** The findings shown that healthcare professional were not aware of the appropriate weeks of gestational age (GA) for administration of ACS. Job aids were lacking at all levels of the health facilities, and use of ACS did not adhere to standard guidelines. The knowledge regarding clinical indications and contraindications were deficient among different health care providers. The quality of care for ACS use was lacking across facility levels. The assessment results further emphasized the need for better supervision, clarification of the role of providers, authorization of level of facility for providing ACS, and development of ACS specific indicators used in monitoring performance.

**Conclusions:** As findings suggested a critical need of standard protocols, job–aids and adequately trained healthcare providers, the Government of India Operational Guideline was expanded emphasizing accurate GA for use of ACS, development of algorithm and decision tool for guideline adherence, and broaden the scope from spontaneous preterm birth to all conditions at high risk of preterm birth.

Keywords: Threatened preterm birth, ACS, facility readiness, national guideline, health policy

## Validation of Two Parent-Reported Autism Spectrum Disorders Screening Tools M-CHAT-R And SCQ in Bamako, Mali.

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**Introduction and Objectives**: Early screening is` crucial for early autism spectrum disorders (ASD) diagnosis and intervention. ASD screening tools have mostly been constructed based on the Western cultural context. This study aims to validate the modified checklist for autism in toddlers-Revised (M-CHAT-R) and the social communication questionnaire (SCQ) in the Malian sociocultural context for ASD screening.

**Methods:** We administered M-CHAT-R and SCQ in 947 toddlers aged 16-30 months old at the district and community health centers in Bamako and 120 patients (60 autistic and 60 age and sex matched controls) aged  $\geq$ 4 years old at the psychiatry department in Bamako. Toddlers at moderate to high risk of ASD underwent M-CHAT-R/F and clinical evaluation by an ASD multidisciplinary team. M-CHAT-R and SCQ were evaluated for cultural appropriateness by Malian anthropologists. The sensitivity, specificity, Positive Predictive Value (PPV), Negative Predictive Value (NPV)were determined for both tools. Health professionals have been trained on how to use M-CHAT-R and SCQ for ASD screening in Bamako.

**Results**: We found for the M-CHAT-R, a sensitivity of 50%, a specificity of 100%, a PPV of 100% and a NPV of 87%. The SCQ had a sensitivity of 71%, a specificity of 72%, a PPV of 73% and a NPV of 70%. We have found four out of twenty items on the M-CHAT-R that were culturally inappropriate in the Malian context. M-CHAT-R and SCQ can be used for early autism screening in Mali. We plan to train Malian physicians in chief and pediatricians at the district hospitals across the country to integrate the early ASD screening into the national health system.

**Conclusion:** M-CHAT-R has a perfect specificity and SCQ a fair diagnostic accuracy for ASD in Mali.

**Keywords**: AMALDEME, Malian association for mental deficiencies; ASD, Autism spectrum disorders

## PP-58 What Happens to Children with Birth Defects and Developmental Disabilities after They are Referred From the Community to Tertiary Care Centers by the Rashtriya Bal Swasthya Karyakram in India? A Follow Up Study Radhakrishnan B.<sup>1\*</sup>, Girase T.<sup>1</sup>, Ujagare D.<sup>1</sup>, Anita Kar A.<sup>2</sup>

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**Introduction and Objectives:** The Rashtriya Bal Swasthya Karyakram (RBSK) is a communitybased screening and early intervention programme for specific types of birth defects and developmental disabilities in India. Mobile health teams screen children in community settings and refer suspected cases to the District Early Intervention Centre and other tertiary care centres. While the initial contact at the referral centre is mentored by RBSK personnel, subsequent adherence to medical or habilitation advice is unknown. The objective of this study was to determine the uptake of services by caregivers of children with birth defects and developmental disabilities, current treatment status, and the factors influencing uptake and adherence to referral advice. Methods: A follow up study was conducted in Pune district, India. Caregivers of a random sample of 115 children were interviewed using a semi structured questionnaire.

**Results:** The highest uptake of services was for congenital heart defects. For developmental delays, caregivers followed referral advice, but discontinuation of treatment was evident for conditions requiring rehabilitation services. Loss of wages due to long distance for travel, long waiting time for therapies, difficulty in transporting the child, not being informed about the referral and already using private medical services at the time of diagnosis were reasons influencing uptake of referral advice or adherence to treatment.

**Conclusions:** While screening and referral component of the RBSK service are in place, the key challenge appears to be continuation of treatment. When the reasons behind discontinuation of services are considered, community-based rehabilitation strategies might lead to better outcomes. At the community level, medical and rehabilitation services need to be integrated with social welfare services including education and social welfare opportunities for the affected child. **Keywords:** Birth defects, Developmental disabilities, India, National programme

## PP-59 Whole Exome Sequence Analysis of the Family Affected with Marfan Syndrome Babar M.E.<sup>1</sup>, Ali A.<sup>1\*</sup>, Ashraf M.U.<sup>2</sup>

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**Introduction and Objectives:** Marfan syndrome is a disorder that affects the connective tissue in many parts of the body. Connective tissue provides strength and flexibility to structures such as bones, ligaments, muscles, blood vessels, and heart valves. The signs and symptoms of Marfan syndrome vary widely in severity, timing of onset, and rate of progression. We aimed to identify the causative mutation in the family.

**Methods:** For this study, we identified and registered the consanguineous families having more than three affected with symptoms after obtaining written informed consent. Blood samples were collected from parents, patients and other family members. DNA was extracted using organic methods. Illumina platform was used for whole exome sequencing with 100x coverage and more than 6GB data. The sequence data was analyzed using Genious tool. After cleaning the data, variant calling was performed.

**Results:** We identified mutation in the fibrillin-1 (FBN1) gene potentially the main cause of the disease in the sequenced family. The mutation found in the gene c.4408T>C mutation (p.Cys1470Arg). This identified mutation is already reported in the literature.

**Conclusion:** Pre-marital screening of the FBN1 gene could be helpful to reduce the disease risk in Pakistani families.

Keywords: Marfan Syndrome, connective tissue, fibrillin-1, exome sequencing
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