International Clearinghouse for Birth Defects
Surveillance and Research

51st Annual Meeting of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR)

November 2 – 5, 2025
Mount Grace Hotel & Spa, Magaliesburg
South Africa

Organized by the ICBDSR, the sub-Saharan Congenital Anomalies Network (sSCAN) and the Centre for Human Metabolomics, North West University (NWU)

Annual Meeting Planning Committee
(in alphabetical order)

Lorenzo D. Botto
Colleen Fifield
Emma Kalk
Helen Malherbe
Cecilia Mellado
Anke Rißmann
Mary Ann Thomas

ICBDSR website: www.icbdsr.org

The ICBDSR Executive Committee and ICBD Staff thank the **sub-Saharan Congenital Anomalies Network (sSCAN)** and the **Centre for Human Metabolomics, North West University (CHM/NWU), South Africa** for their cooperation and support and **all ICBDSR members** who work in birth defects surveillance and research, to achieve our common goal of improving primary prevention and care of birth defects.

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sSCAN
Sub-Saharan African Congenital Anomalies Network

NWU® Centre for Human Metabolomics

The ICBDSR is a Promoting Organization of

www.worldbirthdefectsday.org

Programme

Pre-Conference Workshop:
Quality counts, visualization matters: from accurate diagnoses to impactful data visualization

Sunday 2nd November 2025, Rosewood Chambers	
0930 – 1000	Registration, pretest, introductions
1000 – 1300	Clinical-epidemiologist case studies (small groups with discussion)
1300 – 1400	Lunch
1400 – 1515	Data quality and quality improvement: case study (small groups with discussion)
1515 – 1530	Break
1530 – 1700	Data visualization and story telling: goals, target audience, formats, practice
1630 – 1830	ICBDSR Meeting Registration
1900 – 2100	Welcome Reception (Terrace) & Buffet Dinner (Twist Restaurant)

ICBDSR Annual Meeting
Rosewood Chambers

Monday 3rd November 2025	
0800 – 0830	ICBDSR Meeting Registration
	Session 1: Opening **Moderators: Emma Kalk & Helen Malherbe**
0830 – 0840	**Welcome** *Emma Kalk, University of Cape Town & sSCAN* *Helen Malherbe, North-West University (NWU) & sSCAN*
0840 – 0900	**Keynote and Official Opening** *Binu Luke, Lead: Desmond Tutu School of Medicine, NWU*
0900 – 0930	**ICBDSR Executive Committee Update & Annual Report** *Anke Rissmann, ICBDSR Chair & Lorenzo Botto, ICBD Director*
0930 – 1000	***J. David Erickson Lecture*** **Birth Defects in Africa: A hidden Public Health Crisis** *Philippa Musoke* Department of Paediatrics and Child Health, Makerere University, and a Co-Principal Investigator (PI) for the Johns Hopkins University-Uganda, Kampala Uganda
1000 – 1030	***Keynote Lecture*** **Spina bifida: tip of an iceberg** *Graham Fieggan* Helen and Morris Mauerberger Professor and Head; Director, Neuroscience Institute, University of Cape Town; Head of Clinical Department: Neurosurgery, Groote Schuur Hospital and Red Cross War Memorial Children's Hospital, Cape Town, South Africa
1030 – 11:00	Break & Posters
1100 – 1120 (including Q&A)	**Keynote Lecture:** **"So what"? Finding meaning and purpose in data** *Lorenzo Botto, ICBD Director*
1120 – 1130	**Lisi-Hurtado Award & Prizegiving**
1130 – 1150 (including Q&A)	**Lisi-Hurtado Award Winner** Outcomes of Neural Tube Defects Among Children Under 15 Years in Eastern Ethiopia: A Descriptive Report. *Samrawit Tegene*
1150 – 1210 (including Q&A)	**Lisi-Hurtado Award Winner** Does previous antenatal care improve folic acid uptake in subsequent pregnancies? A systematic review and meta-analysis. *Utibe Ebong*
1210 – 1215	Questions & discussion
	Session 2: Birth defect programmes: global, regional and national **Moderator: Cecilia Mellado**
1215 – 1230	Burden of Birth Defects: Review of methods and results (WHO). *Hannah Blencowe*
1230 – 1245	The Sub-Saharan Congenital Anomalies Network (sSCAN). *Emma Kalk*
1245 – 1300	Ubomi Buhle Project & Global Birth Defects App. *Diane Lavies*
1300 – 1400	Lunch
	Session 3: Descriptive epidemiology and research of birth defects and rare diseases **Moderator: Florence Rouget**
1400 – 1415	Unmasking the burden of Neural Tube Defects in eastern Ethiopia, the SALT project. *Lola Madrid*

1415 – 1430	Burden and Risk Profile of Neural Tube Defects in Colombia: Evidence from ECLAMC Surveillance, 1982–2022. *Karen Sarmiento*
1430 – 1445	The Prevalence of Neural Tube Defects in Eastern Ethiopia, a Population-Based Surveillance. *Helina Heluf*
1445 – 1500	Enhancing Rare Birth Defect Surveillance: Using Unaffected Birth Intervals to Detect Shifts in Microcephaly Occurrence. *Robert Serunjogi*
1500 – 1515	Epidemiology of Rare Diseases in Brazil: the Brazilian Rare Diseases Network perspective. *Claudia Fernandes Lorea*
1515 – 1530	Breaking Down the Burden of Rare Diseases in South Africa: A National Point Prevalence Study Using Orphanet Methodology. *Helen Malherbe*
1530 – 1545	Correlation of population health and economic indicators with presence of congenital anomaly registries – a global secondary data analysis. *Martina Schatz*
1545 – 1600	Break & Posters
	Session 4: Birth defect surveillance and research in low- and middle-income countries **Moderator: Sarah Stevens**
1600 – 1615	Emerging Oropouche Virus Infection in Pregnancy: Vertical Transmission and Perinatal Outcomes in Brazil, 2023–2025. *Lavinia Schuler-Faccini*
1615 – 1630	Prevalence and types of congenital anomalies in eight villages in Pune district, India. *Anita Kar*
1630 – 1645	Cleft Lip and Palate in Colombia: Trends, Birth Outcomes, and Perinatal Determinants from a Four-Decade Case-Control Study. *Karen Sarmiento*
1645 – 1700	Evaluating the data quality of the birth defects surveillance system in Tshwane district, Gauteng, South Africa 2023. *Vuyiswa Lebese*
1700 – 1715	A Decade of Clinic-Integrated Clubfoot Surveillance in South Africa: A Locally Adapted Model for Data Collection and Service Strengthening in Resource-Constrained Public Health Systems. *Karen Moss*
1715 – 1730	Developing and Validating a Neonatal Screening Tool for Congenital Anomalies to be used in Low- and Middle-Income Country Settings. *Hana Gebreselassie*
1730 – 1745	Integration and Sustainability of Birth Defect Surveillance in Participating Hospitals: Summary of Consultative Meetings Held with Frontline Health Workers in Kampala, Uganda. *Joyce Namale-Matovu*
1745 – 1800	Racial and Ethnic Differences in the Prevalence of Congenital Heart Defects: A Scoping Review. *Wendy Nembhard*
1900 – 2130	Marimba Band & Traditional Braai (Terrace)

Tuesday 4th November 2025	
	Session 5: Risk factors, prenatal screening, early pregnancy, birth, newborn screening and postnatal outcomes **Moderator: Karen Sarmiento**
0830 – 0845	Estimating the burden of risk factors and the potential for prevention: the PEACE tool. *Lorenzo Botto*
0845 – 0900	Using congenital anomaly registration to monitor the evaluative roll out of non-invasive prenatal testing (NIPT) for trisomy 21, 18 and 13 in England as part of a national screening programme. *Sarah Stevens*
0900 – 0915	NIPT for sexual chromosome abnormalities; case series. *Lucian Pop*
0915 – 0930	Neonatal Jaundice-Associated Mortality as a Sentinel Indicator of G6PD Deficiency Burden in Brazil. *Lavinia Schuler-Faccini*
0930 – 0945	Prevalence and risk factors associated with major congenital disorders identified through the UBOMI BUHLE pregnancy exposure registry in Soweto, South Africa. *Dorothy Sefara*
0945 – 1000	Enhancing screening of congenital anomalies in low resource settings: Insights from the MiMBa Pregnancy Exposure Registry in Kenya. *Kennedy Weseka*
1000 – 1015	Newborn Screening in South Africa: Current Status, Challenges, and Opportunities for Expansion. *Chris Vorster*
1015 – 1030	Merging clinic-based research with population-based surveillance: the Ponseti clubfoot network. *Paul Romitti*
1030 – 1100	Break & Posters
	Session 6: Innovative Solutions for Common Goals **Moderators: Mary Ann Thomas & Anke Rissmann**
1100 – 1215	**Round Table Session** Panel Discussion and Action Plan of Topics Pre-selected by Birth Defect Surveillance Networks (EUROCAT/NBDPN/ICBDSR/sSCAN/Ubomi Buhle/ReLAMC)
1215 – 1300	ICBDSR New Report Format *Lorenzo Botto*
1300 – 1400	Lunch
14:00 – 18:00	Optional Tour: Cradle of Humankind

Wednesday 5th November 2025		
0830 – 1030	**Session 7: Birth defect surveillance and research in low- and middle-income countries II** **Moderator: Wendy Nembhard**	
0830 – 0845	A systematic review of the long-term outcomes of neural tube defects in the Eastern African region. *Hope Mwangudzah*	
0845 – 0900	Descriptive Analysis of Congenital Anomalies Among Admitted Newborns across hospitals implementing with the NEST360 Alliance. *Keisuke Ogasawara*	
0900 – 0915	Privacy-Enhancing Technologies for International Registry Collaboration: An England-US Pilot *Sarah Stevens*	
0915 – 0930	Biobanks as Catalysts for Birth Defect and Rare Disease Programs: Linking Diagnostics, Treatment, Research, and Awareness in LMIC. *Engela Helena Conradie*	
0930 – 0945	Missed Opportunities: Addressing Diagnostic Delays in Structural Birth Defects Among Ethiopian Children Through Screening and System Strengthening. *Hiwote Assefa*	
0945 – 1000	Preliminary declaration for publication on ICBDSR annual meeting *Helen Malherbe & Emma Kalk*	
1015 – 1030	Wrap up and close of meeting *Outgoing & Incoming ICBDSR Chairs*	
1030 – 1100	Break & Posters	
1100 – 1300	ICBDSR Business Meeting/Election & Future Annual Report Discussion *ICBDSR members only*	sSCAN Business Meeting *sSCAN members only*
1300 – 1400	Lunch & Departure	

Sunday November 2nd 2025

Pre-Annual Meeting Workshop

Rosewood Chambers

Quality counts, visualization matters: from accurate diagnoses to impactful data visualization

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Monday, 3rd November

Session I – Oral Presentations

Moderators:
Emma Kalk and ***Helen Malherbe***

J. David Erickson Lecture
Birth Defects in Africa: A Hidden Public Health Crisis
Philippa Musoke
Department of Paediatrics and Child Health, Makerere University, and a Co-Principal Investigator (PI) for the Johns Hopkins University-Uganda, Kampala Uganda

Keynote Lecture
Spina bifida: tip of an iceberg
Graham Fieggan
Helen and Morris Mauerberger Professor and Head;
Director, Neuroscience Institute, University of Cape Town;
Head of Clinical Department: Neurosurgery, Groote Schuur Hospital and Red Cross War Memorial Children's Hospital, Cape Town, South Africa

Keynote Lecture:
"So what"? Finding meaning and purpose in data.
Lorenzo Botto, ICBD Director

Lisi-Hurtado Award Winner
Outcomes of Neural Tube Defects Among Children Under 15 Years in Eastern Ethiopia: A Descriptive Report.
Samrawit Tegene

Lisi-Hurtado Award Winner
Does previous antenatal care improve folic acid uptake in subsequent pregnancies? A systematic review and meta-analysis.
Utibe Ebong

J. David Erickson Lecture
Birth Defects in Africa: A Hidden Public Health Crisis
Philippa Musoke
Department of Paediatrics and Child Health, Makerere University, and a Co-Principal Investigator (PI) for the Johns Hopkins University-Uganda, Kampala Uganda

Email of corresponding Author: pmusoke@mujhu.org

Globally, an estimated 8 million infants are born with a birth defect (BD), and approximately 3.3 million children die by 5 years of age each year. Birth defects contribute to significant morbidity and mortality in infants and children in low and middle-income countries. Prevalence rates of birth defects vary between regions, ranging from 2.8 - 69/1000 live births in north America and south east Asia respectively. In Africa, a systematic review and meta-analysis of BD reported a pooled prevalence of 23.5 (95% CI 20-25.9) per 1000 newborns. Sub-analyses demonstrated a considerable difference in prevalence per country, with 9.6% in Kenya and 1.7% in South Africa. Not taking folic acid, maternal illness and drug use had increased odds of BD. The sub-Saharan African Congenital Anomalies network conducted a scoping review of birth defects in Africa. The included articles originated from 22 of the 49 sub-Saharan African countries, with 88% investigating a single BD, 56.9% focused on burden, but only 3.5% covered BD

surveillance. Commonly reported external birth defects include neural tube defects (1-3/1000), talipes equinovarus (clubfoot), and cleft lip and palate. National Birth Defect Registries are limited, BD surveillance coverage inadequate, with hospital-based programs rather than the more comprehensive community-based surveillance. Paucity of BD prevalence data in Africa, associated stigma, and inadequate health services lead to underreporting of BD, high mortality, and chronic disability. To address these gaps, developing triple BD surveillance programs will include prevention, early diagnosis, and appropriate treatment and care for infants born with BD. Ultimately, the successful implementation of national BD surveillance programs and support for the sub-Saharan African Congenital Anomalies Network should improve the survival and quality of life of children born with BD in Africa.

Keynote Lecture

Spina bifida: tip of an iceberg

Graham Fieggan
Helen and Morris Mauerberger Professor and Head;
Director, Neuroscience Institute, University of Cape Town;
Head of Clinical Department: Neurosurgery, Groote Schuur Hospital and Red Cross War Memorial Children's Hospital, Cape Town, South Africa

Email of corresponding Author: graham.fieggen@uct.ac.za

Spina bifida is almost unique among central nervous system disorders in that the lesion is visible externally. In many respects however, this is the tip of an iceberg, and this presentation will address the modern challenges posed by this ancient disease.

The most common form of spina bifida is myelomeningocele (mmc) or open spinal dysraphism which results from defective primary neurulation, or incomplete closure of the neural tube. Although less common, closed spinal dysraphism presents a bewildering array of variations, further confused by complex and sometimes archaic terminology.

Perhaps the most important challenge in mmc is the fact that the majority of cases are potentially preventable through periconception folic acid (the concept of folate-preventable spina bifida, or SBA-F), either as a supplement or through food fortification. Despite overwhelming evidence, worldwide efforts in this regard have had less impact than one might expect.

Early antenatal diagnosis allows parents choice in determining whether or not to continue with a pregnancy, and in some countries the option of fetal surgery. The importance of genetic counselling in this regard cannot be overstated. For those babies born with an open mmc, the goal has to be surgical closure within 24-36 hours to prevent colonization of the neural placode and devastating CNS infection.

Around half of these babies will develop hydrocephalus requiring surgical treatment (most often a ventriculoperitoneal shunt), while other neurosurgical complications such as tethered spinal cord or symptomatic Chiari II malformation are rare.

Almost invariable however is the presence of a neurogenic bladder, but social continence can be achieved in most through the simple procedure of clean intermittent catheterization.

These children are best managed in a multidisciplinary clinic and greater attention is now paid to transition of care through adolescence, and sexuality.

It is reassuring that with early and appropriate treatment, and expert multidisciplinary care, a good quality of life can be achieved in many. A major limiting factor in advocating for these patients is inadequate epidemiological data, especially in regions where the condition is most common, hence a plea is made for more efforts in this regard, in keeping with the theme of this congress.

Keynote Lecture:
"So what?" Finding meaning and purpose in data

LD Botto, MD
Professor of Pediatrics
University of Utah School of Medicine
Salt Lake City, UT, United States

Email of corresponding Author: lorenzo.botto@hsc.utah.edu

Birth defects surveillance programs often face the question "so what?" from policymakers and funders – prevalence estimates are important but how does this improve outcomes or supports prevention? The triple surveillance framework recognizes this criticism and proposed to integrate causal exposures (folate status, maternal diabetes, smoking), disease occurrence (neural tube defects, congenital heart disease, orofacial clefts), and longitudinal outcomes (NTD mortality, cleft hospitalizations, cardiac anomaly burden) to generate actionable intelligence for prevention and policy change.

Resource limitations and implementation challenges are valid concerns. To maximize efficiency without sacrificing epidemiological rigor, implementation should leverage existing resources—hospital discharge databases, vital statistics, specialty clinic networks, and public health programs (smoking cessation, nutrition, diabetes surveillance). In addition, prioritizing high-impact conditions, which can include selected disorders beyond structural anomalies (e.g., metabolic diseases part of newborn screening)—and focusing on data quality over quantity can deliver significant community value.

Integrated surveillance of the causal chain of disease, from risk factor to outcome, expands the emphasis from description to action by revealing hidden patterns that can inform policy and clinical decisions. Ultimately, success hinges on strategic use of high-quality data, high-impact conditions, and collaboration with established health information systems. By broadening their perspective, birth defect programs, far from becoming redundant, can find novel meaning and purpose, demonstrate greater value, and play an increasingly vital role in improving community health.

Outcomes of Neural Tube Defects Among Children Under 15 Years in Eastern Ethiopia: A Descriptive Report

Dr Samrawit Tegene[1], Helina Heluf[1,2], Professor Nega Assefa[1], Dr Lola Madrid[1,2]
[1]Hararghe Health Research, Haramaya University, Harar, Ethiopia, [2]London School of Hygiene and Tropical Medicine, London, United Kingdom

Email of corresponding Author: sabebaw@hararghe.org

Background: Neural tube defects (NTDs) are severe congenital anomalies caused by the incomplete closure of the neural tube during early embryonic development, typically between 21 and 28 days post-conception. Globally, these birth defects, including spina bifida and anencephaly, are a significant cause of morbidity, lifelong disability, and mortality. The burden of NTDs is particularly high in low- and middle-income countries (LMICs) like Ethiopia, where affected children often face severe neurological impairments requiring complex and extensive medical management, including surgical interventions and long-term follow-up. However, limited access to appropriate and specialized care frequently leads to severe physical, social, and psychological challenges. Overall, mortality rates among children with NTDs are estimated to be tenfold higher than in non-affected children, with the highest rates observed in LMICs primarily due to these limitations in specialized medical and surgical care. Despite this critical need, comprehensive population-based data on NTD outcomes in high-mortality settings like Eastern Ethiopia remain scarce. This descriptive report, done as part of the SALT/NTD project, aims to highlight the outcome of children affected by NTDs who survive after birth identified through the NTD surveillance in the catchment area and the clinical surveillance at Hiwot Fana Comprehensive Specialized Hospital (HFCSH).
Methods: The study population comprised all children with NTDs identified through the ongoing NTD surveillance system and those enrolled in a cohort study at HFCSH. These children were subsequently followed in the Neurosurgery unit of HFCSH based on clinical indication. Data were collected using a standardized questionnaire, consistent with tools employed by other hospitals in Ethiopia collaborating with Reach Another Foundation for NTD follow-up. Data were recorded on standardized forms by trained research staff at HFCSH during each scheduled follow-up visit by the neurosurgeon and/or multidisciplinary team, primarily covering the first year of life for enrolled children.
Results: A total of 216 children with NTDs were enrolled in the study between June 1, 2023, and May 31, 2025. Of the total cases, 49% were females and 51% males. The observed distribution of neural tube defects was as follows: Myelomeningocele (MMC) accounted for 83 cases, MMC with Hydrocephalus for 66 cases, Meningocele for 41 cases, Encephalocele for 18 cases, and Lipomyelomeningocele for 8 cases. Regarding surgical intervention, 72.2% of the cases underwent surgery, while 27.8% did not. At the end of the study period, the follow-up status of the cohort indicated that 68 children were alive and actively on follow-up, 81 of them were lost to follow-up, 54 children died, and 13 were admitted to the hospital recently.
Conclusion: MMC is the predominant type of NTD observed in this cohort, with a high proportion of affected children undergoing surgical intervention, underscoring the significant demand for specialized neurosurgical services in Eastern Ethiopia. The observed high rates of loss to follow-up and mortality highlight persistent challenges in providing comprehensive and sustained care. These findings emphasize an urgent need for sustained surveillance, improved access to specialized medical care, and robust, easily accessible aftercare programs to improve the overall quality of life for children with NTDs. Furthermore, given the long-term nature of care, it is crucial to provide psychological, emotional, and financial support to parents. Expanding access to care in nearby areas is also essential to ensure timely and continuous support for these vulnerable children and their families.

Does previous antenatal care improve folic acid uptake in subsequent pregnancies? A systematic review and meta-analysis

Mr Utibe Ebong[1], Dr Jason Wilson[2], Dr Joanne Given[3], Professor Frank Casey[1], Dr Maria Loane[3], Professor Helen Dolk[1]
[1]School of Medicine, Ulster University, Belfast, United Kingdom, [2]School of Sport and Exercise Science, Ulster University, Belfast, United Kingdom, [3]Institute of Nursing and Health Research, Ulster University, Belfast, United Kingdom

Email of corresponding Author: ebong-u@ulster.ac.uk

Background & Objectives: During foetal development, the neural tube typically closes within the first 28 days after conception. If the closure is incomplete, it can result in a range of congenital anomalies known as neural tube defects (NTDs). Sufficient serum folate levels are crucial for the successful completion of this closure. However, for most women, serum folate levels are often insufficient. Thus, folic acid supplementation (FAS) is advised. In many countries, staple foods are often fortified with folate, either mandatorily or voluntarily. Health information on FAS has been reported to increase FAS uptake. Given their exposure to FAS-related information from previous pregnancies, multiparous women might be expected to have better FAS practices than primiparous pregnant women. The objective of this review was to examine whether folic acid (preconception, periconceptional and postconceptional) uptake differs between primiparous and multiparous women.
Method: The systematic review was conducted in accordance with the PRISMA guidelines. Four databases (Medline-OVID, CINAHL Ultimate, SCOPUS, and ProQuest Medical) were searched using a combination of Medical Subject Headings and text words, with input from a subject librarian. Search records were screened using a set of inclusion and exclusion criteria. Inclusion criteria stipulated publications from observational studies comparing folic acid use and non-use between primiparous and multiparous women, where FAS was measured as tablets taken before and/or during pregnancy. Given the introduction of FAS guidelines in the 1990s, the review included papers published in English from 1994 to 2024. Two reviewers independently screened search records. To ensure reliability and accuracy of the screening and selection process, three reviewers independently assessed a random subset (10%) of the publications. Unadjusted odds ratios (OR) and 95% confidence intervals were calculated using Stata 18 to estimate the effect of parity on supplementation. With the expectation of variability between studies, the random-effects method (DerSimonian and Laird) was used to account for heterogeneity in the measurement of folic acid intake, definitions of parity, as well as pre- and peri-conceptional periods, among other factors. Additional analyses were conducted to investigate potential sources of heterogeneity based on study characteristics.
Results: The search strategy returned 10,982 records from all four databases. Of the 10,982 records identified and screened, 69 studies were included in the review. Twenty-three of the studies were conducted in Europe, 19 in Asia, nine in North America, six in Africa, five in Australia, five in the Middle East, and two in South America. Multiparous women were consistently less likely to take folic acid before and during pregnancy than primiparous women. For preconceptional use, the odds were 35% lower among multiparous women (OR: 0.65; 95% CI: 0.59 – 0.72; n = 21 studies). The lower preconception use among multiparous women also applied to high-risk women (OR = 0.58, 95% CI: 0.47 – 0.71; n = 3 studies). For periconceptional use, the odds were 33% lower among multiparous women (OR: 0.67; 95% CI: 0.60 – 0.76; n = 26 studies). Postconception, the odds were 25% lower among multiparous women (OR: 0.75; 95% CI: 0.68 – 0.83; n = 29 studies); however, by the second trimester, there was no difference between the two parity groups (OR: 1.03; 95% CI: 0.94 – 1.12; n = 5 studies).
Discussion & Conclusion: Multiparous women almost universally have experience of antenatal care from their previous pregnancy(ies). However, the results of our review suggest that the knowledge of folic acid gained from antenatal care is failing to improve, or even maintain, levels of preconceptional FAS for future pregnancies. This was also applicable to pregnant women with a higher risk of giving birth to children with

NTDs. Behavioural theories, such as the Capability-Opportunity-Motivation Behaviour framework, stipulate that improvements in knowledge (psychological capacity) alone are insufficient to achieve behaviour change. Factors such as motivation and opportunity, in addition to capability, are required to achieve sustainable behaviour change. The role of parity in folic acid uptake requires further investigation, and more attention should be given to targeting interconceptional interventions at multiparous women.

Monday, 3rd November

Session 2 – Oral Presentations

Moderator: Cecilia Mellado

Burden of Birth Defects: Review of methods and results.
Kathleen Strong (Presented by Hannah Blenclowe)

The Sub-Saharan Congenital Anomalies Network (sSCAN).
Emma Kalk

Ubomi Buhle Project & Global Birth Defects App.
Diane Lavies

Burden of Birth Defects: Review of methods and results

Dr Kathleen Strong[1], Dr Jamie Perin, Dr Helen Malherbe (presented by Dr Hannah Blencowe)
[1]Who/HQ, Geneva, Switzerland, [2]Johns Hopkins University Bloomberg School of Global Health, Baltimore, USA, [3]Centre for Human Metabolomics, North-West University, Potchefstroom, South Africa

Email of corresponding Author: strongk@who.int

Background: An estimated 8 million newborns are born with a congenital anomaly each year, and of these an estimated 240,000 die worldwide in their first month of life, making birth defects a leading cause of neonatal deaths. Understanding the birth prevalence of these common birth defects and their geographic distribution allows for better planning and implementation of programs for screening, treatment, management and long-term care of those born with these conditions. WHO and partners last produced estimates for birth incidence of selected birth defects using a data repository called the Modell Global Database (MGDb). An updated version of the MGDb was used to produce estimates of congenital anomaly birth prevalence globally, regionally and for 201 countries. Assumptions are highlighted with a view towards making recommendations about how to routinely update estimates so that they become useful for national screening, treatment, management and long-term care programs for children born with congenital anomalies. Accurate and trustworthy information will provide governments, communities and families with the tools they need to support children born with congenital conditions.
Methods: The MGDb is a modeling tool designed to estimate the global and country-specific burden of severe, early-onset, endogenous congenital disorders (CDs) from birth through age five. These disorders originate primarily from genetic or chromosomal causes and are either fatal or severely disabling without intervention. Only endogenous congenital conditions that can cause premature death and/or disability in the absence of care and which manifest before the age of 10 years are included. The MGDb treats congenital disorders as a closed system, accounting for all outcomes of affected pregnancies including stillbirth, termination, death, cure, or survival with disability. New data come from birth defect registries and also from systematic reviews designed to retrieve studies from low- and middle-income countries which lack birth defect registries. We present our methods, models, data sources and assumptions for these results and discuss the implications for newborn screening, prevention, treatment and care for children born with these conditions. Mortality data were obtained from a different source. The WHO Global Health Estimates which use national civil registration and vital statistics (CRVS) data, as well as a range of alternative sources for countries lacking CRVS.
Results: The leading causes of mortality due to birth defects globally are congenital heart defects (CHDs) and neural tube defects (NTDs) [WHO GHE 2021]. These two preventable conditions account for over half of all neonatal deaths due to birth defects [WHO GHE 2021]. Other conditions include the trisomies of chromosomes 21, 13 and 18, Orofacial clefts, limb defects (Club foot), Sickle Cell Disease and Thalassemia.
Sickle cell disease (3.2 per 1,000 live births) has the highest global live birth incidence, followed by severe congenital heart defects (3.1 per 1,000 live births), oral facial clefts (1.3 per 1,000 live births), and neural tube defects (1.1 per 1,000 live births).
Discussion and Conclusions:
These estimates need further refinement based on a comprehensive understanding of country context. A major limitation is the inconsistent use of terminology with hampers comparisons of estimates produced from different institutions. Widely different estimates may reflect differences in assumptions, aggregated causes, or even points in time. Further investigation is needed to reconcile differences and make the estimates fully useful for program planning, monitoring and evaluation. Child under-5 survival has improved dramatically over the last 3 decades because action was taken to address the leading direct causes of under-5 death including, prematurity, perinatal conditions, pneumonia, malaria and diarrhoea. Congenital anomalies now account for an increasing proportion of under-5 deaths, up to 30% in low under-5 mortality settings. Nonetheless, these statistics hide the real burden of congenital anomalies to families and

communities. Those with congenital conditions are more often born prematurely and when they survive the neonatal period, are more susceptible to death from the infectious diseases of childhood, i.e., malaria, pneumonia, diarrhoea and malnutrition. Unfortunately, those who survive often face a lifetime of severe disability and stigma. With this in mind, burden of birth defects data are needed more than ever to advocate for more resources for screening, treatment and care for children born with these conditions.

The Sub-Saharan Congenital Anomalies Network (sSCAN).

Emma Kalk

Email of corresponding Author: emma.kalk@uct.ac.za

The sub-Saharan African Congenital Anomalies Network (sSCAN) was established in 2021 to address the fragmented research, surveillance and care programmes for children with congenital anomalies (CA) in the region. sSCAN established a community of practice with the long-term aim to promote the prevention and early diagnosis of CA and care for children and families. We have adopted a Triple Surveillance Approach, including both CA and rare diseases in our remit and expanding the concept of surveillance to include prevalence of conditions, of known causes and of health outcomes. The broad aims of the Network focus on important drivers of CA-related neonatal and under-5 morbidity and mortality in sub-Saharan Africa (SSA) and include 1. surveillance (including identification and prevention of risk factors); 2. improving survival and reducing disability in affected children (including diagnosis, medical and surgical treatment, rehabilitation); and 3. engagement of affected communities in developing the research agenda. Research activities in these areas remain isolated and uncoordinated.

sSCAN includes 16 projects across 12 countries, and other stakeholders in sub-Saharan Africa. Most projects are fully or jointly supported by donor partners with a specific disease/exposure focus: collaboration within Africa is uncommon. sSCAN has hosted 15 webinars and supported grant applications and publications. The platform can provide opportunities to build capacity, share expertise, and optimize data use towards contextually relevant solutions for CA.

sSCAN successfully establishes a much-needed community of practice to address clinical, surveillance and advocacy needs related to CA in SSA. The Network provides a centralized repository for resources in a fragmented and resource-constrained setting. As we work towards a more formal common data system to support regional analyses, we continue to expand and sustain clinical care, research and advocacy for children and families affected by CA in SSA.

Ubomi Buhle Project & Global Birth Defects App.

Diane Lavies

Email of corresponding Author: diane.lavies@uct.ac.za

The UBOMI BUHLE (Understanding Birth Outcomes from Mothers and Infants, Building Healthcare by Linking Exposures) project has established a Pregnancy Exposure Registry in collaboration with 5 research partners across 3 provinces in South Africa. Prospective enrolment of pregnant women takes place across 17 sentinel sites. Routine antenatal and perinatal data is collected at the antenatal and associated delivery facilities, emphasising accurate recording of gestational dating, fetal exposures and pregnancy/birth outcomes. Bespoke adaptation of the surveillance version of the Global Birth Defects App has aligned the data fields with the National Department of Health congenital disorder notification form, and allows remote review and diagnosis of congenital disorders by geneticists.

The project has strengthened service delivery at sentinel sites through nurse training, support of gestational ultrasound and genetics counselling services. Training modules include safer use of medicine in pregnancy, standardised procedures for neonatal surface examinations, recognition of common malformations, and bereavement counselling.

A key focus of the registry is the assessment of the safety of antiretrovirals for treatment and prevention of HIV. Additional data is collected to support risk analyses on other exposures including comorbid illnesses, vaccines and other medicines.

To date, almost 90 000 women have been enrolled with an outcome available in 80% of completed pregnancies. Anonymised data is routinely submitted to a central database, and datasets are requested through a Data Access Committee, guided by principles of equity, accountability and beneficence. Project governance structures include representation from the National and Provincial Department of Health.

UBOMI BUHLE is well-placed to collect robust background rates of congenital anomalies in South Africa while evaluating the safety of medicines and vaccines commonly used in pregnancy. The prospective approach with a high cohort retention rate and flexibility to assess numerous exposures, as well as a state-of-the-art data management system makes this an effective and agile surveillance platform for maternal and newborn health in South Africa.

Monday, 3rd November

Session 3 – Oral Presentations

Moderator: Florence Rouget

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Unmasking the burden of Neural Tube Defects in eastern Ethiopia, the SALT project

Ass Prof Lola Madrid[1], Samrawit Abebaw[2], Helina Heluf[1], Dr Merga Dheresa[2], Gezahegn Mengesha[2], Mohammed Abdulkadir[2], Faisel Abdi[2], Tadesse Dufera[2], Yenenesh Tilahun[2], Yunus Edris[1], Tadesse Gure[2], Prof Nega Assefa[2]
[1]London School of Hygiene and Tropical Medicine, London, United Kingdom, [2]College of Health and Medical Sciences, Haramaya University, Harar, Ethiopia

Email of corresponding Author: lola.madrid-castillo@lshtm.ac.uk

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Background and Objectives: Neural tube defects (NTDs) are serious birth defects from incomplete neural tube closure, causing brain and spinal issues, leading to death or disability. Despite advancements in prenatal care and medical technology, NTDs remain a significant public health issue, especially in low and middle-income countries (LMIC). In eastern Ethiopia, for example, a pooled prevalence of NTD as high as 63 per 10000 live births has been reported. However, these estimates lack precision because Ethiopia lacks comprehensive, population-based data on NTD burden and risk factors, due to weak birth defects surveillance, death registration, and their linkages. The 2016 Ethiopian Micronutrient Survey found 32% of non-pregnant women of reproductive age had RBC folate deficiency, highest at 54.84% in Harari. The small sample did not cover eastern Hararghe, where NTD cases among stillbirths and children aged below five years were reported by CHAMPS, with an adjusted NTD mortality rate of 104.0/10,000 births (95% CI: 94·3–116·4). After agreement with the national stakeholders, the CHAMPS-Ethiopia team developed a program called The SALT project "Spine bifida and Anencephaly in Ethiopia, foLate for prevention", to understand the prevalence of NTDs at the population level and determine folic acid insufficiency in WRA age and explore feasibility and acceptability of a potential intervention to reduce NTDs, such as a well-established mandatory food fortification program with folic acid in eastern Ethiopia.
Methods: this SALT project includes a quantitative component with different study designs: i) a cross-sectional study to determine the overall and birth prevalence (number of births affected with NTD among total number of births) and by types of NTDs in abortions, stillbirths, children and adults at population level identified through an ongoing pregnancy, mortality and clinical surveillance already established in the catchment area; ii) a cohort study of live-born babies affected with neural tube defects followed in Hiwot Fana Hospital for one year to determine the outcomes (death/disability); iii) a cross-sectional study to assess the prevalence and determinants of folate insufficiency of 450 women of reproductive age (WRA) selected by simple random sampling of fixed number of records under the demographic surveillance sites (DSS) in Kersa, Harar, and Haramaya and estimate the risk of NTD based on WHO threshold. It also has a qualitative component, incorporating observations, interviews, and focus group discussions to assess community perceptions of this condition, as well as the feasibility and acceptability of consuming fortified food.
Results: The SALT study started its NTD surveillance, follow-up components, and qualitative research on 1 June 2023. In the first year of the SALT project (from 1 June 2023 to 31 May 2024), we identified 330 NTD cases, of which 121 (37%) resulted in death. In 153 cases (46%), we were unable to confirm the vital status within the last three months. Among them, 129 were residents in the DSS, and three were excluded as they had birth defects other than NTD. Finally, 126 NTD cases were included in the analysis, yielding a birth prevalence of 98.7 (95% CI: 79.8-120.6) per 10,000 total births, being higher in rural areas (128.7, 95%CI: 103.9-157.6 per 10,000 births) than in urban (7.9, 95% CI: 9.9 – 28.5, p<0.01). The population-based prevalence was 1.8 (95%CI: 1.4-2.3) per 10,000 population. The folate survey started in June 2024 and was completed in May 2025. IT included 463 WRA, with a median age was 28 years (15 – 49), 74% were Muslims, 39% never attended school, 14% had history of abortion, 15% had a history of stillbirth and <1% had a previous case of NTD. The preliminary results of the first 194 cases analysed, 87 (45%) had folate red blood cell (RBC) folate deficiency and 188 (97%) had folate insufficiency. The community perceives the causes of NTD from a religious perspective but also to exposure to factors during pregnancy. Acceptability of a fortified salt could be good if aligned with health education on its benefits and feasibility will depend on affordability.

Discussion and conclusion: The first year of the SALT study revealed a concerning burden of NTD in eastern Ethiopia, with a birth prevalence of 98.7 per 10,000 births. This rate is significantly higher in rural areas compared to urban settings, highlighting disparities in maternal health and potential folate deficiency. The high case fatality rate (37%) and the large proportion of unconfirmed vital status (46%) reflect limitations in surveillance, follow-up systems, and healthcare access. The folate survey findings reveal critical nutritional gaps among women of reproductive age (WRA). Nearly half of the women assessed had RBC folate deficiency (45%), and the vast majority (97%) had folate insufficiency—well below the WHO threshold for NTD prevention. These deficiencies, alongside low education levels and previous poor pregnancy outcomes emphasise the urgent need for folate-based interventions. Additionally, cultural and religious beliefs influence community understanding of NTD causation, indicating the need for context-sensitive health education. Despite these challenges, the acceptability of fortified salt as a preventive intervention appears promising. Community receptiveness can be enhanced through targeted education that aligns biomedical explanations with local beliefs. However, the success of such an intervention depends on its affordability and consistent access, especially in rural regions where NTD prevalence is highest.
In conclusion, the findings underscore the urgent need for folate interventions, enhanced surveillance, and culturally sensitive education to address the substantial burden of NTDs and improve maternal and child health outcomes in Ethiopia.

Burden and Risk Profile of Neural Tube Defects in Colombia: Evidence from ECLAMC Surveillance, 1982–2022

Dra María Camila León-Sanabria[1], Dra Lina Ibañez[2], Dr. Nicolás Niederbacher[1], Dr. Santiago Andrade[1], **Dr Karen Sarmiento**[3], Ms Adriana Guaca[4], Dr Ignacio Zarante[1,5]
[1]Institute of Human Genetics, Faculty of Medicine, Pontificia Universidad Javeriana, Bogotá, Colombia, [2]Colombian Association of Medical Geneticists and Genomic Medicine (ACMGEN) , Bogotá, Colombia, [3]Department of Physiological Sciences, Faculty of Medicine, Pontificia Universidad Javeriana, Bogotá, Colombia, [4]District Health Secretariat, Subdirectorate of Public Health Surveillance, Lead Specialist for Birth Defects, Bogotá, Colombia, [5]Hospital Universitario San Ignacio, Bogotá, Colombia

Email of corresponding Author: kssarmiento@gmail.com

Introduction and Objectives: Neural tube defects (NTDs) occur during embryonic development in the fourth week of gestation, with a global prevalence ranging from 1 to 10 cases per 1,000 live births (LB). In Colombia, a prevalence of 13 per 10,000 LB has been reported, with more than 300,000 cases occurring worldwide each year. NTDs are characterized by diverse clinical manifestations and may present as isolated or syndromic conditions. The two most common forms are anencephaly and spina bifida, which can result in death within the first days of life or cause permanent disability. Currently, it has been demonstrated that up to 72% of NTDs can be prevented through the implementation of educational and informational campaigns aimed at raising public awareness about the importance of daily folate intake, periconceptional multivitamin supplementation, and food fortification with folic acid. Given this context, the objective of the present study is to determine the association between NTDs and multiple maternal, paternal, perinatal, and neonatal risk factors, as well as their occurrence in a cohort from Bogotá and Cali, Colombia, between 1982 and 2022.
Methodology: A case-control study was conducted using data from the Latin American Collaborative Study of Congenital Malformations (ECLAMC), collected between 1982 and 2022 from 25 hospitals in Bogotá and Cali, Colombia. Each newborn was examined by a qualified physician trained in standardized physical examination for the detection of birth defects (BDs). Information was obtained through direct interviews with the parents and recorded in a standardized ECLAMC form, including maternal history, perinatal conditions, and neonatal characteristics. Cases included LB or stillbirths (SB) weighing ≥500 g diagnosed with NTDs (ICD-10 codes Q050–Q059, Q760). Controls were newborns without BDs, selected immediately after each case in the same hospital, with a 1:4 ratio. Cases were classified as isolated (only NTD), polymalformed (with additional anomalies), complex when it includes defects of the nervous system or limb anomalies secondary to the NTD, or syndromic (associated with chromosomal abnormalities). Quantitative variables were summarized using medians and interquartile ranges, while qualitative variables were presented as absolute and relative frequencies. Prevalence was reported per 10,000 LB and analyzed annually to identify temporal trends. Comparisons between cases and controls were made based on maternal and paternal age (≥35 vs. <35 years), sex, birth weight (<2500 g vs. ≥2500 g), parity (≥3 vs. <3 pregnancies), and gestational age (<37 vs. ≥37 weeks). Odds ratios (OR) with 95% confidence intervals (CI) were calculated. Statistical significance was defined as $p \leq 0.05$. Analyses were performed using STATA v.14.
Results: A total of 198 cases of NTDs were identified between 1982 and 2022, with an adjusted prevalence rate of 0.52 per 10,000 LB (95% CI: 0.45–0.61). The trend displayed an oscillating pattern, with increases beginning in 2004 and a peak in 2006 (1.434 per 1,000 LB), likely related to the implementation of ECLAMC's active surveillance program. Among the cases, 51% were isolated NTDs, 29.8% were complex, 18.7% were polymalformed, and only one case presented with a chromosomal anomaly (Edwards syndrome). Among polymalformed cases (n=82), the most frequently affected systems were digestive (20.7%), musculoskeletal (19.5%), ear/face/neck (17.1%), and cardiovascular (14.6%), accounting for over 70% of the total anomalies. Cases had a significantly lower median birth weight compared to controls (2800 g vs. 3023 g; p <0.000001) and lower parental age (mothers: 25 vs. 28 years; fathers: 28 vs. 30 years; p <0.00004 and p=0.0015, respectively). A significant association was found between low birth weight (<2500 g) and NTDs (OR: 2.33;

95% CI: 1.61–3.34; p < 0.001). No significant associations were observed with advanced maternal or paternal age (>35 years), sex, parity >3 pregnancies, or gestational age <37 weeks. Parity showed no statistical association (p = 0.3377).

Discussion: According to EUROCAT, the global prevalence of NTDs during the same period of our study was 3.07 (2.99–3.66) per 10,000 LB, reflecting a lower prevalence in our city. This discrepancy could be attributed to limitations in local surveillance programs and underreporting or low detection rates. Nonetheless, the decline in prevalence observed in certain periods aligns with public health interventions, notably the mandatory fortification of flour with folic acid since 1996—an approach that has proven effective in reducing NTDs across several countries. Additional contributing factors may include the gradual adoption of prenatal multivitamin supplementation with folic acid, improvements in prenatal care, and expanded health coverage during the 2000s. The most common NTD observed was spina bifida without hydrocephaly, followed by spina bifida with hydrocephaly and anencephaly. In contrast, anencephaly is the most frequently reported NTD worldwide. This difference may be related to pregnancy terminations and early miscarriages, considering the high lethality associated with anencephaly. The lower median birth weight observed among affected cases supports the previously documented association between low birth weight and NTDs, possibly indicating a link with intrauterine growth restriction or adverse perinatal conditions related to structural defects of the CNS. Moreover, the proportion of polymalformed cases underscores the potential role of multifactorial etiological mechanisms, likely involving complex interactions among genetic, nutritional, and environmental factors that remain incompletely understood.These findings highlight the importance of strengthening surveillance systems and promoting preventive strategies, while also underscoring the need for further research to better understand the complex etiological factors underlying NTDs in diverse populations.

The Prevalence of Neural Tube Defects in Eastern Ethiopia, a Population-Based Surveillance

Ms Helina Heluf[1,2], Dr. Hannah Blencowe[1], Dr. Kedir Teji Roba[2], Dr. Samrawit Abebaw[2], Dr. Merga Dheresa[2], Mr. Gezahegn Mengesha[2], Mr. Mohammed Jemal Mohammed[2], Mr. Mohammed Abdulkadir Omere[2], Dr. Faisel Abdi Hassen[2], Mr. Tadesse Dufera[2], Mrs. Yenenesh Tilahun[2], Dr. Yunus Edris[1,2], Dr. Tadesse Gure[2], Dr. Nega Assefa[2], Dr Lola Madrid[1,2]

[1]Department of Infectious Disease Epidemiology and International Health, London School of Hygiene & Tropical Medicine, London, United Kingdom, [2]College of Health and Medical Sciences, Haramaya University, Harar, Ethiopia

Email of corresponding Author: Helina.Gebrekidan@lshtm.ac.uk

Background: Neural tube defects (NTDs) are congenital anomalies strongly linked to stillbirth, morbidity, disability, and premature death. To date, all studies conducted in Ethiopia are hospital-based, which can lead to overestimating NTD prevalence due to referral of complicated cases but also underestimating it, as they often exclude fatal outcomes such as miscarriages and stillbirths, which may account for up to 50% of NTD cases. In eastern Ethiopia, NTD-related mortality was also reported to be high among stillbirths and children aged below five years, with an adjusted mortality rate attributed to NTDs of 104.0/10,000 births (95% CI: 94·3–116·4). The absence of population-level data hampers the planning of prevention and intervention strategies in Ethiopia. Understanding the burden of NTDs at the population level will inform investments and health policies aimed at prevention and better care for affected individuals. This study aims to determine the birth and population-level prevalences of NTDs in three health and demographic surveillance systems (HDSSs) located in eastern Ethiopia.

Methods: A population-based cross-sectional study was conducted in the HDSSs located in Harar (urban), Haramaya, and Kersa (both rural), eastern Ethiopia, from June 1, 2023 to May 31, 2024. All individuals diagnosed with NTDs (spina bifida, craniorachischisis, anencephaly, encephalocele, and iniencephaly), including foetuses in utero, children, and adults who were currently alive or deceased (spontaneous pregnancy losses: <28 weeks of gestation (miscarriages), ≥28 weeks (stillbirths), or deaths at any age), were eligible for inclusion. NTD cases were identified through routine home visits by HDSS data collectors in all catchment areas, or by midwives and nurses via ongoing pregnancy, mortality, and hospital clinical surveillance systems within the catchment area. A paediatrician confirmed the diagnosis and classification of NTDs using all information collected for cases including case photographs. Incidence (birth prevalence) and population-based prevalence of NTDs were computed overall and by NTD type, sex, age groups, and residence using the HDSS and pregnancy surveillance denominators. Pearson chi-square analysis was conducted to determine if there was a statistically significant difference in prevalence among the various categories. Kaplan–Meier survival analysis was performed to examine the time-to-event for NTD-affected pregnancies resulting in in-utero foetal demise and compared to foetal loss patterns for unaffected pregnancies within the catchment area.

Results: Overall, 126 NTD cases were included in the analysis, yielding a birth prevalence of 98.7 (95% CI: 79.8-120.6) per 10,000 births. The population-based prevalence was 1.8 (95%CI: 1.4-2.3) per 10,000 population. Among the 266 stillbirths recorded at the three HDSS sites during our study period, 57 (21.4%) were associated with NTDs, resulting in a birth prevalence of 59.8 (95% CI: 45.4-77.5) per 10,000 births. Prevalence varied by site, being significantly highest in Kersa HDSS (birth prevalence 140.6 (95%CI: 106.4-181.3) per 10,000 births; population-based prevalence 2.2 (95%CI: 1.5-3.0) per 10,000 population)) and rural areas (birth prevalence 128.7 (95%CI: 103.9-157.6) per 10,000 births; population-based prevalence 1.9 (95%CI: 1.4-2.5) per 10,000 population). Spina bifida accounted for 51.6% of the total cases, (birth prevalence: 49.3 (95%CI: 36.3-65.6) per 10,000 births; population-based prevalence 1.5 (95%CI: 1.1-1.9) per 10,000 population) and 25.4% of all NTD cases were craniorachischises with a birth prevalence of 24.1 (95%CI: 15.3-36.2) per 10,000 births. Of 126 enrolled, 78 (62%) died during the study period (68 in-utero demises and 10 child deaths). In the Kaplan–Meier survival analysis, a dynamic pattern of pregnancy loss was observed across

pregnancies with and without NTDs, characterised by an initial stable period followed by a gradual increase in loss after 28 weeks of gestation.

Discussion and conclusion: Our findings indicate a substantial burden of NTDs, particularly with many cases of spina bifida and craniorachischisis. The high rates of stillbirths and low survival among affected live births resulted in a low population-based prevalence of NTDs, contrasting with the elevated birth prevalence observed in the study area, especially in rural regions. This emphasises the critical need for intervention with periconceptional folate supplementation. Mandatory folic acid food fortification of a widely consumed staple food, proven to be both successful and cost-effective in many countries, offers a sustainable approach to preventing NTDs. This report presents the first population-level prevalence data from Ethiopia before the full implementation of a food fortification program. Therefore, it will act as a vital baseline to evaluate the effects of the forthcoming mandatory folic acid food fortification initiatives planned by the Ethiopian Ministry of Health. Prevention measures such as preconception dietary counselling, screening, medical follow-up, and the use of folic acid supplements before and during pregnancy can also help reduce the prevalence of NTDs, disabilities, and related mortalities. As a result, community-based health education promoting preconception care and early ANC is essential for raising awareness, potentially preventing NTDs and other birth defects, and enhancing maternal and child outcomes.

Enhancing Rare Birth Defect Surveillance: Using Unaffected Birth Intervals to Detect Shifts in Microcephaly Occurrence

Mr Robert Serunjogi[1], Dr. Daniel Mumpe-Mwanja[1], Ms Florence Namalinzi[1], Ms Joyce Namale-Matovu[1], Mr. Denis Kimbugwe[1], Dr. Irene Lubega[1], Dr. Emily Namara Lugolobi[2], Prof. Philippa Musoke[1,3]
[1]Makerere University - Johns Hopkins University Research Collaboration, Kampala, Uganda, [2]Division of Global HIV & TB, U.S. Centers for Disease Control and Prevention, Kampala, Uganda, [3]Department of Paediatrics and Child Health, College of Health Sciences, Makerere University, Kampala, Uganda

Email of corresponding Author: rserunjogi@mujhu.org

Background & Objectives: Microcephaly is a rare birth defect in which an infant's head circumference is significantly smaller than expected for sex and gestational age, often reflecting underlying abnormal brain development. This condition is associated with a wide spectrum of neurodevelopmental impairments, including intellectual disability, motor dysfunction, and sensory deficits, which can persist throughout life. Detecting shifts in the incidence of microcephaly over time is essential for informing and guiding public health interventions. However, due to the low baseline frequency of microencephaly, traditional surveillance methods, typically based on crude counts, rates, or proportions over time, lack the sensitivity to identify emerging trends early, especially in low-resource settings. Subtle increases in case frequency may be dismissed as random variations. This limitation was evident during the 2015–2016 Zika virus outbreak, when delayed recognition of rising microcephaly cases hindered timely response efforts. Strengthening surveillance systems to improve sensitivity to such rare but major occurrences is therefore crucial. Our objective was to evaluate whether interval-based statistical process control (SPC) methods could improve early detection of non-random changes in the frequency of microcephaly cases compared to traditional prevalence-based approaches using hospital-based birth surveillance data.
Methods: We used retrospective data from a hospital-based birth defects surveillance dataset focusing on microcephaly cases reported across four major hospitals (Kawempe National Referral, Lubaga, St. Francis Nsambya, and Mengo Hospital) in Kampala. All informative live births and fetal losses were examined for defects by surveillance midwives and confirmed by physicians, and review by expert birth defects specialists. A total of 43 microcephaly cases met the inclusion criteria, providing a sufficient sample that exceeded the recommended minimum of 20–25 cases for meaningful SPC analysis. We employed the Geometric chart (G-Chart), which models the number of non-events between rare events under a geometric distribution, to visualize variation in inter-case intervals over time, helping detect unusually frequent or infrequent events.
We plotted SPC charts to visualize temporal trends using two key rules for shift detection: (1) the "same-side rule," which flags sequences of points consistently above or below the mean; and (2) the "1-sigma zone rule," for two out of three consecutive points beyond one standard deviation (SD) from the mean on the same side. Control limits were derived using the empirical mean and SD of the interval distribution. We also implemented a Cumulative Sum (CUSUM) control chart to detect small shifts over time. The chart was parameterized using the observed data to define the center and SD, with a decision interval (h) of 1 to enhance sensitivity to moderate shifts, and a shift detection threshold of 1 SD. All visualizations were generated using R statistical software with relevant quality control packages.
Results: Of 238,461 births from August 2015 to December 2022, 43 cases of microcephaly were identified, corresponding to an overall period prevalence of 1.8 per 10,000 births (95% CI: 1.3–2.4). The prevalence showed a declining trend, falling from 4.8 per 10,000 between April-June 2021 to 1.24 per 10,000 between October-December 2022. The mean interval between microcephaly cases was 5,458.42 (SD=5,457.9) unaffected births, with a broad range (under 500 to over 15,000 births), displaying a wide variability. The chart identified six intervals exceeding the upper control limit, suggesting sporadic but significant delays between cases. Two violations were observed out of three consecutive intervals of the "1-sigma" rule (Cases 22–24) signaling emerging non-random variation.Eight consecutive intervals observed between cases 36 and 43 fell below the mean, satisfying the "same-side" rule and indicating a sustained increase in microcephaly

frequency, indicating a significant downward shift. The CUSUM chart revealed small but persistent changes in microcephaly frequency. A clear upward deviation began at Case 18, reflecting longer intervals and suggesting a temporary reduction in microcephaly occurrence. By Case 36, the cumulative sum crossed below the center line marking the start of a sustained downward trend. Although the lower decision threshold was not yet breached, the consistent decline supported a gradual increase in case frequency over time.

Discussion & Conclusion: Our analysis shows that interval-based SPC utilizing the G-chart and CUSUM chart effectively detected non-random shifts in microcephaly frequency that were not evident in prevalence charts. By focusing on the number of unaffected births between cases, the interval-based approach enhances sensitivity to temporal variation in rare events. A prolonged run of intervals below the mean and high-interval outliers violated standard SPC rules, indicating potential clustering and shifts in incidence. The CUSUM chart confirmed these findings, identifying both momentary reductions and sustained increases in case frequency through cumulative deviations beyond decision thresholds or crossing the expected process average. This method offers advantages over rate-based surveillance, particularly for rare conditions. However, its interpretation is not straightforward, as rising incidence leads to fewer non-cases, making low values more indicative of concern. While the prevalence rate decreased, the interval-based chart showed shorter gaps between cases, signaling increased microcephaly frequency from a case-to-case perspective. This inverse relationship may result in a floor effect where control limits approach zero, complicating signal detection and reducing sensitivity in low-prevalence settings. Additional factors such as improved diagnostics, surveillance intensity, or changes in reporting could influence trends independently of the true incidence. Temporal clustering might also reflect environmental or infectious exposures. Gap-based monitoring represents a promising complementary tool for early detection of rare events like microcephaly. Its integration into existing birth defect surveillance could enable faster public health responses to emerging threats. Future work can validate this approach across other rare outcomes and assess its scalability within national monitoring systems.

Epidemiology of Rare Diseases in Brazil: the Brazilian Rare Diseases Network perspective

Miss Claudia Fernandes Lorea[1,2,3], Dra Bibiana Mello de Oliveira[2,3], Miss Isadora Viegas[2,3], Miss Carolina Peçaibes[3], Dr Filipe Bernardi[2], Dr Angelina X. Acosta[2], Dr Anete Sevciovic Grumach[2], Dr Carlos Eduardo Steiner[2], Dr Chong Ae Kim[2], Dr Debora Gusmão Melo[2], Dr Ellaine Doris Fernandes Carvalho[2], Dr Erlane Marques[2], Dr Faradiba Sarquis Serpa[2], Dr Juan Clinton Llerena Junior[2], Dra Isabella Monlleó[2], Dr Luiz Carlos Santana da Silva[2], Dra Mara Lucia Schmitz Ferreira Santos[2], Dra Marcela Câmara Machado Costa[2], Dra Marcia Maria Costa Giacon Giusti[2], Dr Marcial Francis Galera[2], Dra Maria Teresinha de Oliveira Cardoso[2], Dra Marilaine Matos de Menezes Ferreira[2], Dr Ney Cristian Amaral Boa Sorte[2], Dra Paula Frassinetti Vasconcelos de Medeiros[2], Dr Paulo Gazzola Zen[2], Dra Raquel Tavares Boy Da Silva[2], Dra Rayana Elias Maia[2], Dra Solange Oliveira Rodrigues Vale[2], Dra Tatiana Amorim[2], Miss Vania Mesquita Gadelha Prazeres[2], Mr Victor Cassão[2], Dr Victor Evangelista de Faria Ferraz[2], Dra Ida Vanessa Doederlein Schwartz[1,2], Dr Domingos Alves[2], Dra Têmis Maria Félix[2,3]

[1]Universidade Federal do Rio Grande do Sul - UFRGS, Porto Alegre, Brazil, [2]National Network for Rare Diseases - RARAS Network Project, , Brazil, [3]Hospital de Clínicas de Porto Alegre, Porto Alegre, Brazil

Epidemiology of Rare Diseases in Brazil: the Brazilian Rare Diseases Network perspective

Email of corresponding Author: claudialorea@hotmail.com

Background & Objectives: Rare diseases (RD) collectively affect approximately 10% of the Brazilian population, yet national epidemiological data remain fragmented and primarily limited to a few isolated conditions. In 2014, the Brazilian Policy for Comprehensive Care for People with Rare Diseases was established to improve diagnosis, treatment, and quality of life. To operationalize this policy and generate structured epidemiological data, the Brazilian Rare Diseases Network (RARAS Network) was launched in 2020. This national initiative encompasses 36 specialized centers, including university hospitals, Rare Disease Reference Services (RDRS), and Newborn Screening Reference Services (NSRS), distributed across all five Brazilian regions. The network is structured as a multicenter, ambispective cohort study designed to collect and analyze comprehensive RD data to support national policy and service planning.

Methods: Data collection occurred in two phases: a retrospective phase (cases seen between 2018–2019, registered between 2020–2021) and an ongoing prospective phase (initiated in 2022). Clinical and sociodemographic data were collected using a standardized form, designed specifically for this project and hosted on the REDCap platform. Data elements include patient demographics, clinical history, family history, consanguinity, age at first symptoms, diagnostic methods, diagnostic status, Human Phenotype Ontology terms (HPO), treatment modalities, hospitalizations, and comorbidities. Each participant received a unique identifier, to enable longitudinal tracking across study phases. Ethics approval was obtained from all participating centers, with data governance and quality monitoring centrally coordinated by the University of São Paulo.

Results: As of June 2025, 19,059 cases were registered in the network database. The median age of participants was 13.2 years (mean, 19.8 years; SD, 18.5), with females comprising 51.2% of the cases. Geographical distribution reflected both population density and access to specialized services: Northeast (37.5%), Southeast (31.9%), South (13.8%), Center-West (11.6%), and North (5.2%). Consanguinity was reported in 8.3% of cases, with the highest prevalence in the Northeast region (12.3%). Regarding diagnostic status, 65.6% of cases received a confirmed diagnosis, 63.63% of these confirmed through etiological methods: 42.4% by molecular analysis, 32.4% by biochemical tests, 13.6% by cytogenetic analysis, and 7.58% by anatomopathological methods. Clinical criteria alone confirmed 31.7% of diagnoses. Diagnosis remained suspected in 17.8% and undiagnosed in 16.6% of cases. Familial recurrence was reported in 26.3%. The mean diagnostic odyssey, defined as the time from first symptoms to definitive diagnosis, was 6.3 years. Notably, the average interval between first visit at a specialized reference center and the diagnosis was approximately 2.4 years, indicating that restricted and delayed access to specialized centers significantly contributes to the overall diagnostic delay. In the retrospective phase, 44.5% of patients had at least one hospitalization; in the

prospective phase, 39.8%. During the prospective phase, patients had a mean of 2.2 hospitalizations (SD 3.5), median of 1 (range 1–66). Median hospitalization duration was 12 days.

Discussion and conclusion: The RARAS Network is the first coordinated national initiative to systematically map RD in Brazil. These preliminary findings reveal significant diagnostic delays, largely driven by limited access to specialized centers, alongside regional disparities in care. These insights underscore the urgent need to expand early access strategies to reduce the diagnostic odyssey and improve RD outcomes.

Breaking Down the Burden of Rare Diseases in South Africa: A National Point Prevalence Study Using Orphanet Methodology

Ass Prof Helen Malherbe[1]
[1]Centre For Human Metabolomics, Desmond Tutu Medical School, Faculty of Health Sciences, North-west University, Johannesburg, South Africa

Email of corresponding Author: helen.malherbe@nwu.ac.za

Background & Objectives: Rare diseases (RDs) are individually rare but collectively common and significant, affecting an estimated 6–8% of the global population, or >350 million people. In South Africa (SA), the RD burden remains poorly understood due to a lack of national surveillance data, poor diagnostic infrastructure, and fragmented implementation of policy and service frameworks. Estimates suggest >4 million South Africans are affected by RD, but without sufficient granularity for effective planning or intervention. Anecdotal evidence shows most RDs are genetic, begin in childhood, often lead to early mortality, and are frequently associated with lifelong disability. Health systems, particularly in low- and middle-income countries (LMICs) like SA, often deprioritize RDs due to limited data, perceived costs, and competing health priorities. This study was commissioned by the Rare Disease Access Initiative (RDAI) and funded by the Innovative Pharmaceutical Association of South Africa (IPASA), following a request from the National Department of Health, to quantify the RD burden in SA more precisely.
The objectives were to:
1. Estimate national point prevalence using internationally recognized data and local population inputs;
2. Disaggregate the RD population by age of onset, inheritance, medical domain, and disability;
3. Estimate the subset requiring high-cost therapies;
4. Provide evidence to inform research priorities, national surveillance, workforce planning, and equitable access to diagnostics and care in alignment with World Health Assembly (WHA) resolutions and Sustainable Development Goals (SDGs).

This study presents SA's first comprehensive national point prevalence estimate of RDs using Orphanet methodology—supporting policy-level planning and improved outcomes for persons living with a rare disease (PLWRD).

Method: This desktop study utilized the Orphanet database (July 2024 release), a globally recognized resource on RDs including epidemiological indicators, natural history, inheritance patterns, disability, and medical domains. Four curated datasets were extracted: epidemiology, natural history, functional consequences (disability), and linearisation (medical domains). Filters excluded non-relevant conditions (e.g., rare cancers, poisonings, infectious diseases), resulting in a refined list of 3,728 unique RD entities—representing 58% of the total Orphanet database.
Two types of prevalence data were analysed:
1. Direct Point Prevalence—conditions with population-level data, described by class and/or value;
2. Indirect Point Prevalence—conditions described only by individual case or family reports, extrapolated globally and applied proportionally to SA's 2024 population (63 million).

Microsoft Power BI and Excel were used to link datasets via ORPHAcode identifiers and integrate local population data to calculate national burden estimates. Additional variables analysed included age of onset, inheritance patterns, and disability (activity limitation/participation restriction).
A manual list of 60 RD with globally approved high-cost therapies was compiled via literature review and expert input, and validated by national and international specialists. Regulatory approval and availability of these therapies in SA were also indicated. The study upheld ethical principles of transparency, data sharing, and replicability, with engagement from government, civil society, and global stakeholders to ensure methodological and contextual validity.

Results: The mean point prevalence of RDs in SA was calculated at 4.8%, ranging from 3.7% to 5.9%, equating to approximately 3.03 million affected individuals across 3,728 RD entities. Key findings, which align with previous Orphanet studies, include:

- 404 diseases (11%) account for 98% of all PLWRD in SA;
- 77% present in childhood, 7% antenatally, and 11% in adulthood;
- 42% follow autosomal recessive and 27% autosomal dominant inheritance patterns;
- Over 1.3 million individuals live with RD-related functional disability in SA.

Only 5% of RD patients require high-cost therapies. Of the 60 RD identified with globally approved high-cost treatments included in the study, only 21 are approved in SA, with limited access reported.
Prevalence distribution revealed:

- 78% of the burden arises from RD with a prevalence of 1–5 per 10,000;
- The rarest diseases (<1 per 1,000,000) account for 84% of RD but <1% of patients.

Top Orphanet medical domains include:

- Developmental defects during embryogenesis (37%);
- Neurological conditions (20%);
- Inborn errors of metabolism (8%).

This analysis provides granular insight into the scale, complexity, and health system demands associated with rare diseases in South Africa, laying the groundwork for improved surveillance and response strategies.
Discussion & Conclusion: This study provides the first nationally stratified, evidence-based estimate of the RD burden in South Africa using internationally validated methodology. Findings support the urgent prioritization of RDs within national health planning—particularly since most affected individuals are children, who face diagnostic delays, limited care, and early mortality. This higher-resolution mapping offers visibility into the scale and characteristics of PLWRD in SA and can inform research, surveillance, workforce development, newborn screening, and access strategies. The concentration of 98% of patients within 404 conditions provides a clear strategic entry point for policy makers and planners. The finding that over 1.3 million people live with RD-related disability underscores the need for rehabilitation, respite, and integrated care services. Importantly, the study challenges the perception that RD care is universally unaffordable: only 5% of patients require high-cost treatment. Many conditions are manageable through early diagnosis and low-cost interventions, including dietary and hormone therapies. This highlights the need to strengthen early diagnostic capacity, particularly in paediatric and newborn care. The estimates serve as a replicable framework for South Africa and may guide other LMICs lacking empiric data but seeking to develop national RD strategies. Findings align with the 2025 WHA resolution on RDs and the global Universal Health Coverage (UHC) and equity agenda. Integrating this evidence into national planning is essential to achieving SDG 3 and upholding the rights of PLWRD across the life course.

Correlation of population health and economic indicators with presence of congenital anomaly registries – a global secondary data analysis

Ms Martina Schatz[1], Dr Felicity Vidya Mehendale[2]
[1]University Of Edinburgh Medical School, Edinburgh, United Kingdom, [2]Centre for Global Health Research, Usher Institute, University of Edinburgh, Edinburgh, United Kingdom

Email of corresponding Author: smartina312@gmail.com

Background and Objective: Congenital anomalies (CA) are structural or functional changes that occur during intrauterine life. According to the World Health Organisation (WHO), every year, about 295,000 babies die within the first 4 weeks of birth due to CA. It has been estimated that the vast majority (94%) of severe CA occur in LMICs. Understanding the epidemiological profile of CA, by means of information systems and monitoring with national coverage, provides countries with an opportunity to evaluate the impact on the population. This knowledge also produces useful information that can inform health policy to ensure equitable access to timely and high-quality care, thus reducing morbidity and mortality. While national congenital anomaly registers (CARs) can help improve quality and access to timely healthcare for congenital anomalies, many countries do not have CARs and a number of factors may present barriers to the development of national CARs. Similarly, UNICEF reports that 4 in 10 children under age 5, around 166 milllion worldwide, are living without being registered, with most being from Asia and Sub-Saharan Africa. Without this, there is no establishment of children's existence or legal proof if their identity. These children are more likely to miss out on public services that help secure their fundamental rights such as healthcare and education and are in greater danger of exploitation.
This study aims to evaluate the correlation between birth registration rates (BRR), both in children under 1 and under 5 years of age, and existence of national and/or regional CARs. Additionally, we explore the correlation with the World Bank (WB) country classification by income as well as gross domestic product (GDP), health expenditure and infant mortality rate (IMR).
Method: A secondary data analysis was performed using data from UNICEF and the WB. The WB data included 217 countries and UNICEF included 202 countries. In order to include indicators from both the WB and UNICEF we included only the 196 countries that were present in both datasets. All data analysis and correlations were performed on R version 4.3.
UNICEF Data: Birth registration rate data came from UNICEF and we used rates under 1 year and under 5 years from August 2024.
The World Bank Data: Three relevant population health and economic indicators were selected and countries were categorised by the WB classification by income and region. The three indicators selected were average GDP per capita (in USD), average health expenditure (% of GDP) and infant mortality rate per 1000 births. The most recent data for the first two indicators were from 2023, however infant mortality had incomplete data for 2023 so we decided to analyse 2022 data for IMR.
Congenital Anomaly Registers: Data regarding the existence of national and regional CARs worldwide were collected by cross-referencing the following sources; Modell's (MGDb) dataset, ICBDSR, EUROCAT (for Europe), ReLAMC (for South America), SEAR-NBBD (for southeast asia) and sSCAN (for sub-Saharan Africa).
The comprehensiveness of CARs in terms of the range of conditions included was not specified consistently. We therefore recorded only the existence of a CAR, irrespective of the conditions registered. Countries were classified according to the CAR status into those that had (1) a national CAR with or without regional CAR, (2) only a regional CAR and (3) no CAR data available.
Results: Countries lacking a national CARs tend to have lower GDPs, increased infant mortality rates and lower birth registration rates. There is a strong correlation between a higher income status and the likelihood of having a national CAR.

30/196 countries had a national CAR, 19/196 had a regional CAR and 147/196 had no CAR. Data on regional registeries were variable, with CARs existing only in selected regions of individual countries. Analysis by country income group showed that national CARs existed for 35% of HICs, 13% of UMICs and 0% of LMICs and LICs. The percentage of countries with no registries were: HIC: 46.2%, UMIC: 75.5%, LMIC 98% and LIC 100%. In terms of birth registration rates, there was a significant positive correlation between higher income group and BRR for under 1 and under 5 years. In all country income groups, the under 1 register had fewer data available compared to the under 5 register. 85/196 countries are missing a register for under 1 year, while 15/196 are missing a register for under 5 years. BRR in LICs in the under 1 group was only 53.7%.
Whilst there was a wide variation in GDP between different income groups (GDP per capita ranged from USD 793 – USD 44,969), the percentage of health care spending showed less variation across countries. LMICs spent the least on healthcare (5.6%), followed by LICs (6.6%) and UMICs (6.8%). HICs spent 8.1% (only 1.5% more than LICs). We found no correlation between CAR status and % health care expenditure.
Discussion and Conclusion: While the importance of CARs to improve globatl health is widely recognised, most LMICs and LICs still do not report having a national CAR. Our study suggests that countries without CARs have broader, more complex health systems challenges as reflected by lower BRR and higher IMR. Crucially, the most striking pattern was the impact of country income status, and this despite similar % of GDP spent on healthcare. It is important to stress that it is not possible to look at the existence of these CARs in isolation and to consider the impact of broader economic and health systems variables, in the development and maintenance of CARs. This is reflected by sustainable development goal (SDG) 16.9 which emphasises the importance of registration and a legal identity for all newborns, which is vital if we are to achieve SDG 3, good health and well-being, by 2030.

Monday, 3rd November

Session 4 – Oral Presentations

Moderator: Sarah Stevens

Emerging Oropouche Virus Infection in Pregnancy: Vertical Transmission and Perinatal Outcomes in Brazil, 2023–2025.
Lavinia Schuler-Faccini

Prevalence and types of congenital anomalies in eight villages in Pune district, India.
Anita Kar

Cleft Lip and Palate in Colombia: Trends, Birth Outcomes, and Perinatal Determinants from a Four-Decade Case-Control Study.
Karen Sarmiento

Evaluating the data quality of the birth defects surveillance system in Tshwane district, Gauteng, South Africa 2023.
Vuyiswa Lebese

A Decade of Clinic-Integrated Clubfoot Surveillance in South Africa: A Locally Adapted Model for Data Collection and Service Strengthening in Resource-Constrained Public Health Systems.
Karen Moss

Developing and Validating a Neonatal Screening Tool for Congenital Anomalies to be used in Low- and Middle-Income Country Settings.
Hana Gebreselassie

Integration and Sustainability of Birth Defect Surveillance in Participating Hospitals: Summary of Consultative Meetings Held with Frontline Health Workers in Kampala, Uganda.
Joyce Namale-Matovu

Racial and Ethnic Differences in the Prevalence of Congenital Heart Defects: A Scoping Review.
Wendy Nembhard

Emerging Oropouche Virus Infection in Pregnancy: Vertical Transmission and Perinatal Outcomes in Brazil, 2023–2025

Prof Lavinia Schuler-Faccini[1,2], Dr Claudia Fernandes Lorea[2]
[1]Hospital De Clinicas De Porto Alegre, Porto Alegre, Brazil, [2]Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil

Email of corresponding Author: lschuler@hcpa.edu.br

Background & Objectives: Oropouche virus (OROV), an Orthobunyavirus of the Peribunyaviridae family, is an emerging arbovirus transmitted primarily by Culicoides paraensis midges. Since 2023, OROV transmission has expanded in Brazil, spreading from endemic Amazonian regions to southeastern and northeastern states, with over 25,000 confirmed human infections as of mid-2025. While typically associated with febrile illness, recent evidence implicates OROV in vertical transmission and severe perinatal outcomes, including congenital anomalies and fetal deaths. Detection of OROV RNA in fetal tissues, placenta, cerebrospinal fluid, and neonatal serum confirms transplacental infection. Despite these emerging findings, systematic data on OROV's impact during pregnancy are scarce. This study aims to describe maternal clinical characteristics, timing of infection, evidence of vertical transmission, and detailed perinatal outcomes in pregnancies exposed to OROV in Brazil between 2023 and 2025.
Method: A preliminary descriptive review of suspected and confirmed maternal OROV infections was conducted using data aggregated from published case reports, state surveillance databases, and institutional hospital registries across five Brazilian states: Acre, Amazonas, Ceará, Espírito Santo, and Pernambuco. Inclusion criteria comprised laboratory-confirmed maternal OROV infection (RT-PCR or serology), or neonatal/fetal detection of OROV RNA in tissue samples. Maternal clinical data, gestational age at the time of infection, diagnostic methods, and pregnancy outcomes were recorded. Evidence of vertical transmission was based on the direct detection of viral RNA in fetal or neonatal specimens.
Additionally, one case from São Paulo, involving first-trimester infection and a newborn with CNS anomalies, is under ongoing investigation for differential diagnosis with genetic syndromes; exome sequencing has been performed. Cases were summarized descriptively to characterize maternal infection timing, outcomes, and evidence of vertical transmission.
Results:A total of 26 pregnancies with confirmed OROV exposure were analyzed. Among these, 25 pregnancies were fully evaluated, and one case (São Paulo) remains under investigation. Most infections (60%) occurred during the third trimester, while 16% occurred in the first trimester and 16% in the second trimester. Adverse pregnancy outcomes were recorded in 6 of 25 evaluated cases (24%), including three intrauterine fetal demises (IFD), two miscarriages, and one stillbirth. Among the 19 live births, five neonates (26%) presented major congenital anomalies, predominantly involving the central nervous system (CNS). Documented anomalies included microcephaly (three cases), corpus callosum dysgenesis (two cases), pachygyria (one case), ventriculomegaly (two cases), and chorioretinal scarring in at least two neonates.
Vertical transmission was confirmed in two neonates via detection of OROV RNA in cerebrospinal fluid and serum. An additional four cases of probable vertical transmission were inferred through RT-PCR positivity in placental tissues. The 26th case from São Paulo involves a first-trimester suspected but unconfirmed maternal OROV infection with a newborn with CNS anomalies.
Discussion & Conclusion: Oropouche virus infection is potentially associated with central nervous system congenital anomalies and poor fetal outcomes. Given the recent OROV spread out of the Amazonian region and the widespread circulation of its reassortant lineage, the teratogenic potential of this virus must be urgently investigated through active surveillance and pathogenesis studies. Prospective cohort studies are essential for determining the timing and mechanisms of transmission, elucidating teratogenic risks, and guiding prevention strategies.

Intensified vector control and improvements in diagnostic and clinical management guidelines are critical for protecting maternal-fetal health amid the ongoing spread of OROV.

Prevalence and types of congenital anomalies in eight villages in Pune district, India

Dr Anita Pal[1], Ms Bhagyashree Radhakrishnan
[1]Birth Defects Research Foundation, Pune, India

Email of corresponding Author: dranitakar@gmail.com

Background: In India, services for congenital anomalies are offered through an ongoing child screening service, the RBSK (Rashtriya Bal Swasthya Karyakram). Screening occurs at community health and educational facilities for children in the 0 to 18-year age group. Newborn screening is conducted by nurse midwives at government health facilities. Post-discharge, community health (ASHA) workers are trained to identify selected visible anomalies among infants. For children at government pre-schools (2 to 5 years), and schools (6-18 years), mobile health teams are responsible for conducting health checkups, which include screening for selected congenital anomalies. Screening protocols and referral instructions are available for all levels of the service. District Early Intervention Centers are responsible for further diagnosis, medical care and rehabilitation. Children in the 0 to 5 year age group are eligible for free-of-cost care for selected conditions. While data on birth prevalence of congenital anomalies in India is available, the population-prevalence of congenital anomalies, diagnosis, access to medical care and rehabilitation therapies, and educational enrollment of children with congenital anomalies is unavailable.
Objectives: The purpose of the study was to identify (1) the community prevalence and types of congenital anomalies among children under five years of age, diagnosis, access to care, educational enrollment, prevalence of disability/functional impairment, (2) maternal risk factors, and prevalence of prenatal diagnosis, and (3) the number of children with congenital anomalies in the service area of a Primary Health Centre (PHC).
Methods: An observational study among children under five years of age was conducted in eight villages of a PHC (population 42 495, population under five years 3652) located in Pune district, India. Data on congenital anomalies among children under five years of age was collected using a survey, and supplemented with data from health staff and community health workers. Congenital anomalies were documented from clinical records available with caregivers. A pictorial tool was used for probing for undiagnosed cases. The tool included a sketch of a baby and a list of prevalent congenital anomalies. The tool was intended to assist data collectors to specifically point out anatomical regions to mothers, and probe about the presence of an anomaly (eg undescended testicles). A structured questionnaire was used to collect data on termination of pregnancy, child demographics, child health status, medical consultation, treatment, and rehabilitation care, and preschool enrolment. Disability data was collected using the Washington Group/UNICEF module on Child Functioning (WG-CF). Total congenital anomaly rate (living and deceased cases) and congenital anomaly survivor rate was calculated per 10 000 children below the age of five years. System wise prevalence of congenital anomalies was calculated by categorizing the disorders using the International Classification of Diseases 10th revision (ICD-10) guidelines. Disability was reported as per the WG-CF analysis guidelines.
Results: There were 33/3652 children with congenital anomalies in the PHC service area. Total congenital anomaly rate and congenital anomaly survivor rate was 90 per 10 000 (95%CI 62.2-126.8), and 79 per 10 000 (95%CI 53.1-113.9) children under five years. Less severe anomalies such as anomalies of the musculoskeletal system (30 per 10 000 U5 children) and anomalies of the eye, ear, face and neck (27 per 10 000 U5 children) were more prevalent than congenital malformations of the circulatory system (16.4 per 10 000 U5). A clinical diagnosis was reported for about half (52%) of the cases. Only 41% of caregivers followed advised treatment. Treatment was obtained through out-of-pocket expenditure due to lack of awareness about available RBSK services. Congenital anomalies were associated with 13% of childhood disability. Maternal risk factors identified a history of rubella infection during pregnancy, abortion pill usage without medical supervision, and prematurity. Termination of pregnancy due to a fetal anomaly was reported in 12% cases in five years preceding the survey. Four out of ten eligible children (untreated clubfoot, microphthalmus, strabismus, abnormal head shape) were not attending preschool.

Discussion and Conclusions: The study reemphasizes the need for birth defects surveillance, and for strengthening services to ensure diagnosis and optimal care, reduce out of pocket expenditure, correct avertable functional impairments, and ensure preschool enrolment. The study findings have been used for developing a framework of public health interventions for birth defects, including surveillance and reporting, newborn screening and case finding, appropriate treatment and rehabilitation, prevention, capacity building and most importantly, including awareness about congenital anomalies as a routine component of maternal and child health services.

Cleft Lip and Palate in Colombia: Trends, Birth Outcomes, and Perinatal Determinants from a Four-Decade Case-Control Study

Dr. Nicolás Niederbacher[1], Dra Maria Camila León-Sanabria[1], Dr. Santiago Andrade[1], Dra Lina Ibañez[2], Dra Karen Sarmiento[3], Mrs Adriana Guaca[4], Dr Ignacio Zarante[1,5]
[1]Institute of Human Genetics, Faculty of Medicine, Pontificia Universidad Javeriana, Bogotá, Colombia, [2]Colombian Association of Medical Geneticists and Genomic Medicine (ACMGen), Bogotá, Colombia, [3]Department of Physiological Sciences, Faculty of Medicine, Pontificia Universidad Javeriana, Bogotá, Colombia, [4]District Health Secretariat, Subdirectorate of Public Health Surveillance, Lead Specialist for Birth Defects, Bogotá, Colombia, [5]Hospital Universitario San Ignacio, Bogotá, Colombia

Email of corresponding Author: kssarmiento@gmail.com

Introduction and Objectives

Cleft lip and palate (CLP) is the most common congenital craniofacial malformation, resulting from a failure in the fusion of facial processes during critical stages of embryonic development. It is classified based on the anatomical structures involved—lip, alveolar process, hard and/or soft palate—and may present in isolated or syndromic forms, as well as unilaterally or bilaterally. The global prevalence of CLP is estimated to range between 14.3 and 20 cases per 10,000 live births.The consequences of CLP have significant impacts on essential functions such as feeding, nasal breathing, craniofacial growth, speech production, hearing, and dental development. Its etiology is recognized as multifactorial, involving both genetic predisposition and environmental exposures. Commonly described maternal risk factors include tobacco use, alcohol consumption, and nutritional deficiencies—particularly folic acid—as well as exposure to certain medications. Additionally, maternal and paternal age has been discussed in the literature as a potential contributing factor in the development of CLP. Given these considerations, the objective of this study was to determine the association between maternal, perinatal, and neonatal risk factors and the occurrence of CLP in live births and stillbirths diagnosed at 25 hospitals in Bogotá, Colombia. The data were obtained from the Latin American Collaborative Study of Congenital Malformations (ECLAMC) and span the period from 1982 to 2022. This research aims to provide population-based evidence to contribute to the etiological understanding of CLP in the local context, thereby supporting future prevention and public health strategies.

Methods

A case-control study was conducted using data from the Latin American Collaborative Study of Congenital Malformations (ECLAMC), collected between 1982 and 2022 in 25 hospitals in Bogotá and Cali, Colombia. Each newborn was examined by a qualified physician trained in standardized physical examination for the detection of congenital defects (CD). Information was obtained through direct interviews with parents and recorded using the standardized ECLAMC form, including variables on maternal history, perinatal conditions, and neonatal characteristics. Cases were live births or stillbirths weighing ≥500 g diagnosed with cleft lip with or without cleft palate (CL±P) or isolated cleft palate (CP), according to ICD-10 codes Q35.0–Q35.9; Q36.0, Q36.1, Q36.9; and Q37.0–Q37.9. Controls were newborns without congenital defects, selected immediately after each case in the same hospital at a 1:4 ratio. Anomalies were classified as: isolated; polymalformed, when an additional anomaly in another organ/system was present; or syndromic, when associated with a chromosomal abnormality. Clinical classification distinguished CP cases (incomplete or complete cleft palate, other palatal anomalies, gum notch) and CL±P cases (cleft lip alone, cleft lip with cleft palate, incomplete or complete cleft lip, other lip anomalies, microform cleft lip).

Quantitative variables were summarized using medians and interquartile ranges; qualitative variables with absolute and relative frequencies. Prevalence was reported per 10,000 live births and analyzed annually. Associations were assessed using odds ratios (OR) and 95% confidence intervals. Statistical significance was set at $p \leq 0.05$. Analyses were performed using STATA v.14.

Results

A total of 519 cases of cleft lip and/or palate (CL/P) were identified during the study period. The majority were classified as isolated cases (n=317; 61%), followed by polymalformed (n=193; 37%) and syndromic cases (n=9; 2%). When categorized by cleft type, CL±P was more frequent (77.8%) than isolated cleft palate (CP) (22.2%). Regarding sex distribution, a higher proportion was observed among male newborns (n=284; 54.7%), with a prevalence rate of 0.75 per 10,000 live births (95% CI: 0.66–0.84), compared to 0.61 in females (95% CI: 0.53–0.69). The overall adjusted prevalence was 1.37 per 10,000 live births (95% CI: 1.26–1.49).

The temporal trend in CL/P prevalence showed considerable variability between 1989 and 2022. During the early years (1989–2003), rates remained consistently low, all below 0.5 per 10,000 live births. A marked increase was observed in 2004, peaking in 2006 (1.434 per 10,000), coinciding with the expansion of the active surveillance system. In subsequent years, despite fluctuations, prevalence remained mostly above 0.5, with another notable peak in 2012 (1.304), suggesting sustained improvements in case detection.

Clinically, 6.6% of the cases corresponded to stillbirths (n=34/519; 95% CI: 4.7%–9.1%), with a higher proportion in the cleft palate (CP) group. When comparing perinatal and reproductive characteristics between cases and controls, statistically significant differences ($p < 0.05$) were found in birth weight (median: 2844 g in cases vs. 3116 g in controls), gestational age (38 vs. 39 weeks), number of previous pregnancies (median: 2 vs. 1–2), and maternal age (26 vs. 27 years). No statistically significant difference was observed in paternal age.

Discussion and conclusions

Over a 40-year period of active surveillance in Bogotá and Cali, the adjusted prevalence of cleft lip and/or palate (CL/P) was 1.37 per 10,000 live births—significantly lower than the global estimates of 14–20 per 10,000 reported by EUROCAT. In Colombia, previous ECLAMC-based studies have documented a national prevalence closer to 3.37 per 10,000, suggesting that our lower regional rate may reflect differences in institutional coverage and case capture. Nonetheless, the increasing trend observed since 2004 supports the notion that long-term surveillance enhances both case detection and data quality.

The significant associations between CL/P and low birth weight, prematurity, and high parity are consistent with international findings, including EUROCAT data, which highlight adverse perinatal conditions as indirect markers of vulnerability during craniofacial embryogenesis. Similarly, high parity has been identified as a risk factor in prior Latin American studies from ECLAMC. No significant associations were found with advanced parental age or infant sex, aligning with literature showing no consensus on these demographic variables.

Altogether, our findings reinforce the role of perinatal rather than strictly genetic or demographic factors in CL/P etiology. They also underscore the value of high-quality, long-term surveillance systems in generating reliable evidence to inform public health strategies.

In conclusion, this study provides strong support for improving prenatal care, maternal nutrition, and targeted interventions for high-risk pregnancies, while also encouraging future research into epigenetic mechanisms that may mediate the interplay between genetic predisposition and adverse perinatal environments.

Evaluating the data quality of the birth defects surveillance system in Tshwane district, Gauteng, 2023

Ms Vuyiswa Lebese[1,2], Ms Thembekile Zwane[2], Dr Manala Makua[3]
[1]National Department of Health, Pretoria, South Africa, [2]South Africa Field Epidemiology Training Programme, National Institute for Communicable Diseases a Division of the National Health Laboratory Service, Johannesburg, South Africa, [3]University of Pretoria, Faculty of Health Sciences, Pretoria, South Africa

Email of corresponding Author: vuyiswa.lebese@health.gov.za

Introduction: Birth defects (BDs) are structural or functional abnormalities, including metabolic disorders, present at or before birth. Some BDs are visible at birth, while others appear later. They range from minor to major in severity. Major BDs can be life-threatening or impact a child's development and health if untreated, while minor BDs usually have no significant medical or social consequences. Individually rare, BDs collectively cause significant illness and death in children, especially in populations where infectious diseases and nutritional deficiencies are under control. In Africa, under-5 mortality due to BDs is rising across all age groups. In 2019, BD related mortality among children under 5 in low- and middle-income countries (LMICs) was 3 per 1000 live births, compared to 1.3 per 1000 in high-income countries. In South Africa, BDs contributed to 5.9% of under-5 deaths in 2015. Among neonates over 1000g between 2012 and 2016, BDs were the fourth leading cause of death, responsible for 11.8% of neonatal deaths. Accurate surveillance of BDs is important for understanding their impact on child mortality and for reaching child health targets. BD surveillance in South Africa began in 1980 and was revised in 2001, 2006, and 2021 following various national policy updates. However, the current system has never been formally evaluated. This study aims to describe how the birth defects surveillance system (BDSS) operates, calculate the proportion of BDs reported, and assess the quality of its data in Tshwane district in 2023.
Method: A cross-sectional study was conducted using secondary data collected through the BDSS in Tshwane District, Gauteng, in 2023. The system was evaluated using the European Centre for Disease Prevention and Control guidelines for data quality monitoring and surveillance system evaluation. Tshwane district has 16 delivering facilities, all expected to report birth defects. Reporting is guided by a case definition that includes any structural or functional abnormality identified during pregnancy, at birth, or later in life, excluding cerebral palsy and autism spectrum disorders. The study population included all cases reported to the BDSS in 2023 that met this definition. A desktop review was conducted to map the system's operations and notification process from birth to report submission at the National Department of Health (NDOH). Data were cleaned and analysed using Stata version 18. Maternal, neonatal, and epidemiological characteristics were summarised using frequencies and percentages. The proportion of specific BDs was calculated by dividing the number of each BD by the total reported cases. Data quality was evaluated based on internal completeness and validity, and timeliness. Completeness was assessed by the percentage of missing or unknown fields across ten priority variables. It was rated as good if ≥ 90%, moderate if 70–89%, and poor if < 70%. Internal validity was assessed through the percentage of cases with accurate use and application of ICD-10 codes for diagnosis. Timeliness was evaluated by calculating the time between birth, notification, and receipt of the report at the NDOH, with 40 days set as the standard.
Results: The operation of the BDSS is guided by National Guidelines, which require a system that captures complete and accurate data to support the planning and delivery of genetic services. All children are examined through different health service points, and any identified BD is reported using the BD Notification Tool (BDNT). If a diagnosis is not available immediately, a preliminary diagnosis is recorded and coded and updated later. Notification follows a passive case ascertainment approach. Facilities submit completed BDNT forms to district and provincial coordinators, who forward them to the NDOH. Zero-notification forms are submitted in months without BD cases. In 2023, 634 BDs were reported. Most mothers were aged 20–34 years (426; 67.2%), multigravida (269; 42.4%), and multiparous (241; 38.0%). Common maternal conditions included HIV (82; 12.9%) and hypertension (23; 3.6%). The majority of mothers began antenatal care after 20 weeks (294; 46.4%) and had 4–7 visits (290; 45.7%). Among the neonates, 587 (92.6%) were live births; 160

(25.2%) had low birth weight, and 158 (24.9%) were preterm. Musculoskeletal defects were most common (281; 44.32%), followed by genetic defects (94; 14.83%). Priority variables had a moderate average completion rate of 77.6%. Variables with good completion included neonate's gestational age, birth weight, pregnancy outcome, mother's first antenatal visit, and age. There are differences in internal validity across facility types, with district hospitals (61.7%) showing better adherence to ICD-10 coding than academic hospitals (34.4%). Academic and specialized hospitals reported timeliness above 60 days from birth to notification receipt by the NDOH.

Discussion and conclusion: Maternal age in the study aligns with national birth trends. The diagnosis of HIV and hypertension as the most common maternal conditions highlights the impact of maternal comorbidities on BD prevalence. Early BANC booking is a recognised intervention to prevent BDs as it allows for early identification and intervention of women at increased risk. Consistent attendance of ANC, in line with the WHO BANC Plus strategy adopted by the country, allows for women to be monitored throughout pregnancy, helping prevent poor outcomes such as stillbirths, low birth weight, and preterm deliveries, which are associated with an increased risk of BDs. Only proportions could be calculated due to the life course approach in reporting, which limits birth prevalence estimates. Musculoskeletal defects were most common due to the inclusion of minor defects. Neural tube defect rates were low, possibly due to public health interventions. Data completeness varied, with only five of the ten priority variables showing good completeness. Variables related to diagnosis, e.g., ICD-10 codes, type of investigation, and genetic counselling, had the lowest completeness, indicating gaps in training and reporting amongst others. Internal validity was inconsistent across facility types. District and tertiary hospitals had the highest accuracy in ICD-10 coding, while specialist and academic hospitals showed poorer performance, possibly due to diagnostic complexity. Timeliness of reporting also varied, with PHCs and district hospitals performing better than specialist facilities. Improving data accuracy, completeness, and reporting timelines is critical for effective surveillance and reducing birth defect-related mortality.

A Decade of Clinic-Integrated Clubfoot Surveillance in South Africa: A Locally Adapted Model for Data Collection and Service Strengthening in Resource-Constrained Public Health Systems

Ms Karen Moss[1], Alan Millar[1]
[1]Steps Charity Npc, Cape Town, South Africa

Email of corresponding Author: karen@steps.org.za

Background & Objectives:
Clubfoot is a common congenital birth defect affecting one in every 500–1,000 live births globally. Without treatment, it results in lifelong physical disability, social exclusion, and limited access to education and economic opportunity. While clubfoot is fully correctable using early intervention and a non-invasive treatment method, many children in low and middle-income countries do not receive complete care due to under-resourced public health systems and a lack of treatment tracking infrastructure. In South Africa, the national Department of Health does not operate an effective surveillance system for birth defects. This absence of data impacts planning, funding, and the ability to improve patient outcomes. Steps Clubfoot Care, a nonprofit organisation supporting a national network of clubfoot clinics in South Africa, identified the urgent need for consistent and locally appropriate data collection to support programme improvement, impact measurement, and healthcare advocacy. This abstract presents a 10-year overview (2015–2025) of Steps' development of a clinic-integrated, anonymised clubfoot surveillance model. The system has allowed for real-time monitoring of treatment trends, resource allocation, and data-informed advocacy. The objectives of the model were to develop a surveillance system that was ethical, compliant with national data protection laws, feasible in clinics with limited connectivity, and able to empower clinic teams to contribute consistently to a growing national dataset. The model is currently implemented in 48 public clinics across all nine provinces of South Africa, contributing significantly to national disability prevention efforts and potentially offering a scalable template for other congenital birth conditions.
Method: Steps initially trialled international tools such as the Iowa Clubfoot Registry and cloud-based platforms like Salesforce to record patient data from clinics. However, these systems presented challenges for South African implementation, as they did not support anonymisation and stored data outside of the country, failing to comply with South Africa's Protection of Personal Information Act (POPIA). Additionally, many partner clinics lacked reliable Wi-Fi, electricity, or dedicated IT support, making cloud-based systems impractical for daily use. Steps developed a simplified data model tailored to the realities of public healthcare clinics in South Africa. The current model was built using accessible free tools from Google, including spreadsheets and cloud storage, enabling offline functionality and local hosting. Data is anonymised and collected weekly through standardised forms designed for ease of use by clinic staff. Clinic-based data coordinators employed by Steps collect over 60% of the data, with the remaining collected by clinic staff using the Steps Clinic Coordination Toolkit. Key indicators include number of patients enrolled, gender, diagnosis (unilateral or bilateral), weekly attendance, and patients currently in treatment. A new in-house mobile data collection application, developed using Google AppSheet, is in trial phase. Designed to work in low-connectivity environments, it enhances data capture and aligns with privacy protocols. Real-time dashboards using Google Looker Studio are currently in development to support internal feedback.
Results: Between 2015 and 2025, Steps' data system was implemented in more than 48 partner clinics across all nine provinces of South Africa. More than 12,000 children have been recorded through the system to date, with year-on-year improvements in reporting rates and data accuracy. In 2024, data collection recorded 17,662 patient visits, 1,127 new patients were enrolled, and an estimated 4,592 active patients in treatment nationally. The model has proven effective in providing detailed insights into service gaps and strengths across the public health clubfoot network. Clinics with complete and accurate data have been able to use the statistics to motivate for additional staff, expanded treatment space, and ongoing inclusion in provincial budgets. Longitudinal data trends have supported internal programme planning, external donor impact reporting, and national advocacy to strengthen early childhood disability services. Clinics using the toolkit and

supported by trained coordinators consistently outperformed unsupported clinics in data accuracy and timeliness. The trend from 2017 to 2024 shows a 263% percent improvement in patient visits recorded, coinciding with the introduction of Steps clinic coordination and targeted engagement strategies. Data has also informed the strategic placement of new clinics, the expansion of training programmes, and the integration of brace supply logistics into treatment tracking. Steps is piloting an app-based data collection system to improve functionality, while continuing to ensure patient privacy. This system has laid the groundwork for a national, ethical, and practical approach to surveillance for congenital conditions in resource-constrained environments.

Discussion & Conclusion: Steps Clubfoot Care's ten-year experience in developing and maintaining a clinic-integrated data system illustrates the power of local innovation in public health surveillance. Unlike top-down digital systems that often fail in low-resource contexts, the Steps model is grounded in clinic-level realities - poor connectivity, limited staffing, and inconsistent record-keeping. It works within these limitations to deliver useful, ethical, and actionable data. The model demonstrates that reliable surveillance can be achieved through local ownership, tailored tools, and ongoing support, even without sophisticated infrastructure. Key enablers of success include investment in training, ethical compliance under South Africa's POPIA law, and a flexible data model designed around the needs of clinic staff and programme requirements. Steps created its system using readily available, no-cost tools adapted from Google's suite of applications. This approach enabled a cost-effective and scalable model that met privacy requirements and functioned in resource-limited settings. This innovation allowed Steps to create a surveillance framework that is unique to the South African context but potentially transferable to other congenital conditions. As the model evolves, it offers a replicable template for public health surveillance in low and middle-income countries. It highlights how ethical, collaborative, and locally adapted solutions can support early diagnosis, disability prevention and health equity, especially in contexts where government surveillance systems are absent or underdeveloped.

Developing and Validating a Neonatal Screening Tool for Congenital Anomalies to be used in Low- and Middle-Income Country Settings

Ass Prof Hana Gebreselassie[1], Professor Kokila Lakhoo
[1]St. Paul's Hospital Millennium Medical College, Addis Ababa, Ethiopia

Email of corresponding Author: hana.abebe@sphmmc.edu.et

Background: Congenital anomalies are among the common health problems faced by children in low and middle-income countries, contributing substantially to infant mortality rates. Due to limited access to newborn screening programs in most of the resource-limited settings, many congenital anomalies go undiagnosed and untreated, leading to adverse outcomes. This study aimed to develop and validate a newborn screening tool for congenital anomalies to be used in resource-limited settings.
Methods: A draft screening tool that uses clinical examination findings only was developed through literature review and underwent two rounds of Delphi consensus. Tool validation was conducted by applying it to a representative sample of neonates who were delivered and/or admitted to the neonatal intensive care unit at St. Paul's Hospital Millennium Medical College. Data was collected using Kobo Collect and then exported to Microsoft Excel and Statistical Package for Social Sciences (SPSS) version 26 for analysis. The sensitivity and specificity of the screening tool were calculated to assess its validity.
Results: A total of 1160 neonates were screened for congenital anomalies, of which 673(58%) were males. The mean age of the newborns was 26.9±33 hours. The prevalence of congenital anomalies in our series was 5.7%. The central nervous system was the most frequently involved body system (33.7%), followed by genitourinary (18.5%), gastrointestinal (10%), and musculoskeletal (10%) systems. More than one anomaly was diagnosed in 11(13.6%) neonates. The sensitivity and specificity of the screening tool were 86.4% and 97.6% respectively. Furthermore, the positive and negative predictive values of the screening tool were found to be 70.4% and 97.6%, respectively.
Conclusion: Congenital anomalies are not rare findings in our hospital. The newly developed neonatal screening tool in this study has demonstrated significant validity and is both cost-effective and easily deployable, including in rural environments.

Integration and Sustainability of Birth Defect Surveillance in Participating Hospitals: Summary of Consultative Meetings Held with Frontline Health Workers in Kampala, Uganda.

Ms Joyce Namale-Matovu[1]
[1]Makerere University - Johns Hopkins University Research Collaboration, Kampala, Uganda

Email of corresponding Author: jmatovu@mujhu.org

Background & Objectives: The Birth Defect (BD) Surveillance Project has been implemented across four hospitals in Kampala, Uganda: Mulago Hospital (a government facility) and three faith-based hospitals—Mengo, Lubaga, and Nsambya. The primary objective of the project is to determine the prevalence of birth defects and examine possible associations between maternal use of antiretroviral therapy (ART) and common medications during early pregnancy and the occurrence of birth defects. As the project nears its conclusion, there is a critical need to ensure a smooth transition and the long-term sustainability of its core activities within routine labour ward services. Key activities include: reviewing medical records and interviewing mothers using Android-based tablets, examining all births for the presence or absence of birth defects, confirming specific defects of interest, counselling mothers of affected newborns and linking them to appropriate care and obtaining informed consent from mothers for participation in a case-control study. Additional tasks encompass photographing eligible newborns, enrolling participants into the surveillance project, collecting and submitting data, performing quality control checks, and compiling reports.
To support the transition process, a series of informal consultative meetings were conducted with midwives, both those directly involved in BD surveillance and others supporting project activities. The main objective of these meetings was to gather insights on how to effectively integrate and sustain BD surveillance activities within existing labour ward services. Discussions focused on potential strategies for sustainability, perceived sustainability benefits, challenges, and recommendations for ensuring continued implementation. This document presents the key findings and outcomes of those meetings.
Method: Informal consultative meetings were held in March and April 2025 in both government and faith-based hospitals; Participants included 12 independent surveillance midwives from the Makerere University–Johns Hopkins University (MU-JHU) research collaboration who support the implementation of BD surveillance activities at the government hospital, 20 midwives from the faith-based hospitals that are involved in BD surveillance, 16 government hospital midwives who are not formally part of the surveillance project but deliver hospital mothers and work alongside the MU-JHU independent surveillance midwives.
Meetings took place on-site at the participating hospitals and were conducted as focused brainstorming sessions led by the BD core team members. The consultative sessions involved 4–5 midwives per session working across different shifts. In-charges were briefed on the upcoming closure of the long-running BD Surveillance project and supported during the consultative sessions.
After the meetings, reports were prepared summarizing key discussion points, decisions, and agreed upon actions and recommendations to guide the integration and sustainability of BD surveillance within routine existing labour ward services.
Results: The meetings revealed strong support in both government and faith-based midwives for the integration and long-term sustainability of BD surveillance within existing labour ward services. The key strategies identified included: securing buy-in from hospital leadership and other key stakeholders, building the capacity of midwives, simplifying existing BD systems and processes, particularly within the government hospital including the current reporting structure, data collection tools, submission procedures, quality control checks and lengthy and time-consuming consent process for mothers. On perceived benefits, midwives across all hospitals identified several including: improved knowledge, skills, and competence in conducting newborn examinations, early detection and management of BDs, strengthened data collection processes, enhanced counselling for parents affected and timely follow-up care and referrals for infants with BDs. Despite the strong support and perceived benefits, several challenges were reported. Government midwives cited increased workload and time-consuming documentation requirements and difficulties in

collecting complete and accurate data were raised. Other concerns were staff motivation/retention, unreliable internet connectivity, communication gaps and being less familiar with BD surveillance procedures. Nonetheless, most of them expressed a willingness to take on BD responsibilities as part of their routine duties without expecting additional incentives. Faith-based midwives were worried about motivation and indicated a preference for salary increases to support BD activities. General recommendations from all midwives were to get support from the BD core team for an extended period as they adopt to the new roles. **Discussion & Conclusion:** In many low-resource settings, donor-funded health initiatives are time-limited thus posing challenges for long-term sustainability. The integration and sustainability of BD surveillance activities will be prioritized through continuous stakeholder engagement - Ministry of Health officials, hospital administrators, ward in-charges, information and technology heads, and records officers through structured kick-off meetings, site sensitization visits, and collaborative planning sessions. This will enhance ownership and alignment with existing health systems. To ensure quality implementation, we will strengthen midwives' and doctors' capacity to build competence and confidence among the frontline providers through continuous medical education, intensive refresher trainings, on-the-job mentorship, and coaching. Midwives in faith-based hospitals already perform BD-related activities, making sustainability more feasible in these settings. However, government hospital midwives with limited prior involvement will require additional time and support and this will be provided by local experts. To enhance sustainability, midwives from both hospital types will be assigned routine BD responsibilities. We also shall review and simplify processes across facilities, developing user-friendly data tools to enhance accuracy and understanding. In conclusion, integration and sustainability of BD surveillance is feasible with deliberate planning and flexibility. Aligning with hospital specific policies and supporting a gradual transition through local expert teams may allow activities maintenance beyond the project's timeframe. This model may offer a replicable approach for similar health system strengthening efforts.

Racial and Ethnic Differences in the Prevalence of Congenital Heart Defects: A Scoping Review

Dr Maria D. Politis[1], Dr Jenil Patel[1,2], Dr Elijah Bolin[3], **Dr Wendy Nembhard**[1,4]
[1]Arkansas Center for Birth Defects Research and Prevention, University of Arkansas for Medical Sciences, Little Rock, United States, [2]Department of Epidemiology, Human Genetics and Environmental Sciences, University of Texas Health Science Center at Houston School of Public Health, Dallas, United States, [3]Department of Pediatrics, College of Medicine, University of Arkansas for Medical Sciences, Little Rock, United States, [4]Department of Epidemiology, University of Arkansas for Medical Sciences, Little Rock, United States

Email of corresponding Author: wnnembhard@uams.edu

Background and Objectives: Congenital heart defects (CHDs) are the most common birth defects, and the leading cause of birth defect associated morbidity and mortality. As methods for detecting CHDs have improved, the reported prevalence of CHDs has increased over time. However, current research suggests the prevalence of CHDs varies by race and ethnicity. We conducted a scoping review to determine the global prevalence of CHDs by race and ethnicity.
Methods: We searched PubMed and Web of Science for English-language, empirical studies published from 1980 to 2019 that reported on the birth prevalence of CHDs in individuals aged 18 years or younger, disaggregated by race and ethnicity. Two reviewers (MDP and JP) independently screened titles and abstracts, performed full-text reviews, and extracted data using Covidence. Disagreements were resolved through consensus. Studies were excluded if they lacked age-specific data, cumulative CHD estimates, or sufficient detail for synthesis. We summarized prevalence ranges reported across studies.
Results: This scoping review identified 58 studies focused specifically on the prevalence or incidence of CHDs. Most studies were observational or cross-sectional and primarily conducted in the United States, with some from international settings. Reported CHD prevalence varied significantly by race and ethnicity. For total CHDs, rates ranged from 1.9 to 14.4 per 1,000 live births among Non-Hispanic White children, 1.7 to 12.8 among Non-Hispanic Black children, 2.1 to 13.3 among Hispanic children, and 9.45 to 10.2 among Asian children. Higher prevalence was reported among Aboriginal (1.3 to 19.0 per 1,000 live births) and American Indian populations (10.1 to 13.69 per 1,000 live births). Seven studies also addressed CHD complexity. Isolated CHD rates were consistent across groups, while complex CHDs were less common, with lower prevalence across all racial and ethnic groups. Conotruncal defects, particularly tetralogy of Fallot, were widely reported, with variation among racial and ethnic groups. Prevalence among Non-Hispanic Black children reached up to 9.49 per 1,000. Thirty-two studies each also examined left (LVOTO) and right ventricular outflow tract defects, showing the highest LVOTO prevalence among Inuit populations. For hypoplastic left heart syndrome, rates ranged from 1.9 to 3.9 per 1,000 live births among Non-Hispanic White children, 2.2 to 3.1 among Non-Hispanic Black children, and 1.2 to 2.7 among Hispanic children.
Discussion and Conclusion: Racial and ethnic disparities in the prevalence of CHDs among infants and children have been consistently observed across numerous studies. These differences may be influenced by a combination of genetic, environmental, socioeconomic, and healthcare access factors, as well as differences in stillbirth and termination rates and prenatal care, yet the specific mechanisms of these disparities remain unclear. Further research is needed to explore the underlying etiologic pathways, including the role of social determinants of health, to better understand and reduce inequities in CHD occurrence.

Tuesday, 4th November

Session 5 – Oral Presentations

Moderator: Karen Sarmiento

Estimating the burden of risk factors and the potential for prevention: the PEACE tool.
Lorenzo Botto

Using congenital anomaly registration to monitor the evaluative roll out of non-invasive prenatal testing (NIPT) for trisomy 21, 18 and 13 in England as part of a national screening programme.
Sarah Stevens

NIPT for sexual chromosome abnormalities; case series.
Lucian Pop

Neonatal Jaundice-Associated Mortality as a Sentinel Indicator of G6PD Deficiency Burden in Brazil.
Lavinia Schuler-Faccini

Prevalence and risk factors associated with major congenital disorders identified through the UBOMI BUHLE pregnancy exposure registry in Soweto, South Africa.
Dorothy Sefara

Enhancing screening of congenital anomalies in low resource settings: Insights from the MiMBa Pregnancy Exposure Registry in Kenya.
Kennedy Weseka

Newborn Screening in South Africa: Current Status, Challenges, and Opportunities for Expansion.
Chris Vorster

Merging clinic-based research with population-based surveillance: the Ponseti clubfoot network.
Paul Romitti

Estimating the burden of risk factors and the potential for prevention: the PEACE tool

Dr Lorenzo Botto[1], Brit Warner[1], Dr. Matthew Reeder[1], Sergey Krikov[1], Dr Marcia Feldkamp[1]
[1]University Of Utah, Dpt Pediatrics / Medical Genetics, Salt Lake City, United States

Email of corresponding Author: Lorenzo.Botto@hsc.utah.edu

Background and Objectives: Although most birth defects have complex or unknown causes, a variable but substantial number are associated with modifiable risk factors and are therefore potentially preventable. However, beyond well-known examples such as folic acid supplementation, awareness of primary prevention remains limited among health professionals and policymakers. Greater impact could be achieved if stakeholders had access to locally relevant estimates of the number of cases attributable to preventable exposures. A key barrier is that evidence on risk factors—such as maternal smoking or pregestational diabetes—is dispersed across the literature, limiting accessibility and practical application. Another challenge is translating relative risk into credible, population-specific estimates of preventable cases. This requires high-quality, current data on both risk magnitude and exposure prevalence, as well as careful assessment of potential biases. The PEACE tool (Population Estimate of Attributable Cases for Exposure) was developed to address this gap. It offers a transparent, evidence-based method to estimate the number of birth defects and adverse pregnancy outcomes attributable to selected risk factors in specific populations. It also models potential reductions in case counts under varying prevention scenarios. The objective of this study is to demonstrate the utility of the PEACE tool, using maternal smoking vs. orofacial clefts and maternal diabetes vs. tetralogy of Fallot as examples. We present attributable burden estimates across multiple geographic and epidemiologic settings. These examples highlight how translating risk into attributable cases can inform locally tailored prevention strategies and strengthen the case for public health investment in modifiable risk factors.

Methods: We analyzed data from two examples. In the first, we generated estimates for selected countries participating in the 2025 Birth Defect Report of the International Clearinghouse for Birth Defects Surveillance and Research (www.icbdsr.org), which provided data on the prevalence of orofacial clefts and tetralogy of Fallot. Country-level estimates of smoking and diabetes prevalence among women of childbearing age were obtained from public sources, including the Institute for Health Metrics and Evaluation (IHME; healthdata.org) and peer-reviewed publications. Odds ratios (ORs) for associations between risk factors and specific birth defects were derived from the literature: smoking was associated with an OR of 1.22 for cleft palate and 1.34 for cleft lip with or without cleft palate (Fell et al., 2022), while pregestational diabetes was associated with an OR of 4.89 for tetralogy of Fallot (Correa et al., 2008). The second example focused on the United States to demonstrate the use of more granular, state-level data and to estimate the potential economic impact of prevention. State-specific smoking prevalence among women aged 18–44 was obtained from the Behavioral Risk Factor Surveillance System (CDC, 2020–2021), and smoking during pregnancy from the National Center for Health Statistics (2016–2021). Economic cost estimates were derived from CDC data, including a mean additional annual cost of $13,405 per child aged 0–10 with an orofacial cleft and a lifetime cost of $101,000 (in 2007 U.S. dollars; Boulet et al., 2009).

Results. In the ICBDSR sample, maternal smoking prevalence ranged from 5.9% to 25.7%. The estimated number of orofacial cleft cases (cleft palate and cleft lip with or without cleft palate) attributable to smoking increased with smoking rates, from 25 to 153 cases per million births. Cleft lip with or without cleft palate accounted for 77.1% of these cases. Pregestational diabetes prevalence ranged from 0.5% to 4.9%, corresponding to 6 to 78 cases of tetralogy of Fallot attributable to maternal diabetes per million births. In the United States example, an estimated 5,881 infants are born annually with orofacial clefts. Of these, 186 cases were attributable to maternal smoking, based on state-specific smoking prevalence estimates among women of childbearing age. These smoking rates varied widely by state. Three states accounted for 22% of smoking-attributable cases, and nine states accounted for 50% of such cases nationwide. Using CDC cost

estimates, eliminating maternal smoking could result in approximately $18.6 million in lifetime healthcare cost savings associated with orofacial clefts for a single-year birth cohort.

Conclusions. These findings highlight the ongoing global burden of preventable birth defects, such as orofacial clefts and tetralogy of Fallot, associated with maternal smoking and pregestational diabetes. The strong association between risk factor prevalence and estimated case counts underscores the potential impact of prevention strategies targeting modifiable maternal health risks. Country- and subnational-level data are essential for identifying high-burden areas and tailoring interventions to local needs. For policymakers, access to granular, region-specific information supports evidence-based decision-making, efficient resource allocation, and targeted public health action. However, estimates should be interpreted with caution due to variability in surveillance quality, uncertainty in prevalence data, and limitations of modeling assumptions. These findings also emphasize the urgent need to sustain and strengthen public health data systems—especially amid ongoing threats to surveillance infrastructure in many regions. Continued investment in robust maternal and child health surveillance, including data on key risk factors, is critical for effective monitoring, priority setting, and prevention. Finally, by integrating diverse data sources and enabling real-time estimation and visualization of the preventable birth defect burden, tools like the PEACE platform can support surveillance programs in translating scientific evidence into concise, actionable insights for policymakers. This enhances their capacity to advocate for prevention, design targeted interventions, and inform data-driven policy decisions that improve maternal and child health outcomes worldwide.

Using congenital anomaly registration to monitor the evaluative roll out of non-invasive prenatal testing (NIPT) for trisomy 21, 18 and 13 in England as part of a national screening programme.

Mrs Sarah Stevens[1], Jennifer Broughan[1], Elizabeth Tempest[1], Maisie Blyth[1], Nadia Permalloo[1], Gina Brondel, Kate Fleming[1], Corinne Mallinson[1], Anna Springett[1], Robin Glover[1], Ann Tonks[1], Rebecca Till[1], Denise Williams[1], Steven Hardy[1], Kathryn Johnson[1]
[1]Nhs England, London, United Kingdom

Email of corresponding Author: sarah.stevens1@nhs.net

Background: Non-invasive prenatal testing (NIPT) for Down's syndrome, Edwards' syndrome and Patau's syndrome has become increasingly well-established within antenatal screening worldwide. Within England the evaluative roll out of NIPT began as part of the National Health Service (NHS) Fetal Anomaly Screening Programme (FASP) pathway in June 2021 and ran until March 2024. NIPT is offered to all pregnant individuals with a higher chance result (>1 in 150) NHS combined or NHS quadruple test.
Methods: The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) part of the National Disease Registration Service (NDRS) linked data from first-line NHS screening tests, NIPT tests, cytogenetic tests and congenital anomaly registration data. These linked data were used to describe the eligible population, those that had the test and monitor test characteristics and turnaround times. The evaluative roll out approach gave the opportunity to collect evidence and make necessary changes to service delivery in real time.
Results: This is the largest cohort of pregnant individuals (N=43,215) where first line screening higher chance results (>1 in 150) are linked to NIPT screening test for Down's syndrome, Edwards' syndrome and Patau's syndrome. Test failure rates and turnaround times are provided for a national programme over a three-year time period. During the rollout, the total number of singleton pregnancies with a higher chance first-line screen (eligible population) was 43,215 of which a majority (70.3% N=30,367) had NIPT. Of those having NIPT 98.5% received a reportable result, and 87.2% of samples were reported to the maternity service within 5 days of being received in the laboratory. Adjustments in service delivery were made in response to insight from data analysis.
Conclusion: This evaluative rollout demonstrates the feasibility of providing NIPT in a large population. the study provides further information for pregnant individuals in a timely way to better support and enable a personal informed choice to be made about screening. Linkage and integration of laboratory data is a compelling approach in monitoring aspects of performance of a screening test in a real-world setting. The evaluative roll out approach taken will inform in-service evaluations for future expansion of other national antenatal and newborn screening programmes.

NIPT for sexual chromosome abnormalities; case series.

Dr Lucian Pop[1], Dr Nicolae Bacalbasa[1], Dr Irina Balescu[1]
[1]Carol Davila University Of Medicine -POLIZU HOSPITAL, Bucharest, Romania

Email of corresponding Author: popluciangh@icloud.com

Background: Technological advances and the commercialisation of science have made Non-invasive prenatal testing (NIPT) generally available outside the clinical setting and without medical recommendation. The foremost question that mothers address is related to foetal gender.
Method: We did a retrospective study over three years regarding our cases of sexual chromosome abnormalities and the outcome of the pregnancy. We had 8 cases of sexual chromosome abnormalities involving NIPT. All tests were self-funded, and all were formed as a first-line screening test.
Results: Noninvasive prenatal testing was positive for Monosomy X in 5 cases but it was truly positive in only two of them. Our case series results are similar to most studies published in the literature, NIPT picking up monosomy X cases with fetal abnormalities while falsely labelling three other cases. One case of Triple X syndrome and one of diploid Y were correctly diagnosed through NIPT.
Discussion: It is a matter of time and cost until cfDNA becomes the primary screening method for chromosomal abnormalities. As alluded to, placental mosaicism can yield false-positive results; therefore, amniocentesis is preferred, particularly in cases with a normal scan. A thorough scanning evaluation is necessary before or after a positive outcome in abnormal NIPT results. Our cases and the descriptive review show that this influences the accuracy of DNA. Placenta's complex biology has not been fully understood yet and only a comprehensive and deep knowledge of its process can provide a clear and fully benefit to the patient.

Neonatal Jaundice-Associated Mortality as a Sentinel Indicator of G6PD Deficiency Burden in Brazil

Mr Ayoade Desmond Babalola[2], Mr Ricardo Rohweder[2], Dr Thayne Woycinck Kowalski[1,2], Dr Suzy Alves Camey[1], **Prof Lavinia Schuler-Faccini[1,2]**, Dr Giovanna Giudicelli[1,2]
[1]Hospital De Clinicas De Porto Alegre, Porto Alegre, Brazil, [2]Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil

Email of corresponding Author: lschuler@hcpa.edu.br

Introduction: Glucose-6-phosphate dehydrogenase (G6PD) is a critical enzyme in the pentose phosphate pathway, essential for producing nicotinamide adenine dinucleotide phosphate (NADPH), which maintains cellular redox balance. G6PD is encoded by the homonymous gene on chromosome X (Xq28), a highly polymorphic region, making the gene susceptible to high variability. Variants of clinical relevance result in reduced enzyme activity at various levels and are collectively referred to as G6PD deficiency. This X-linked disorder exhibits low penetrance and variable severity, affecting over 500 million people globally, with a high prevalence in Africa, the Middle East, and Southeast Asia. In neonates, G6PD deficiency often manifests as haemolytic anaemia and jaundice, particularly under oxidative stress. Brazil, with a population of over 200 million and a substantial proportion of African ancestry, has a strong surveillance system for both live births and mortality. Notwithstanding, G6PD deficiency remains underrecognized. The poor characterisation of this condition may have masked a significant burden of G6PD deficiency on Brazil's neonatal mortality. Thus, this study sought to characterize demographic and regional patterns of neonatal mortality jaundice due to excessive haemolysis (ICD-10 code P558) as a proxy for undiagnosed G6PD deficiency.
Materials and Methods: We used data from Brazil's national health systems (2009-2023), including the live birth registry (SINASC – Sistema de Informações sobre Nascidos Vivos) and the mortality registry (SIM – Sistema de Informação sobre Mortalidade). Key variables included maternal age, maternal education, birth weight, gestational age, sex, mode of delivery, race/ethnicity, and geographic region. Concerning race/ethnicity, it is described in Brazil as follows: Black, referring to individuals with African ancestry; Brown, consisting of mixed ancestry; Yellow, for Asian individuals; and Indigenous-Amerindians. Descriptive statistics, such as frequency counts, were used to profile cases. Time trend analysis was used to assess changes in P58-related mortality. Poisson regression models estimated crude and adjusted rate ratios using live births as offsets. A hierarchical framework was applied to adjust the crude rate risks, grouping variables into blocks using epidemiological proximity: Block 1 (maternal education), Block 2 (maternal age and race/ethnicity), Block 3 (neonatal sex). Only variables with no significant association (Chi-squared test) were included in the hierarchical model. Due to small sample size limitations, all non-white racial/ethnicity categories (Black, Brown, Yellow, and Indigenous) were combined into a composite group (BBYI) for statistical analysis. All analyses were conducted using R (version 4.3.3).
Results: Over 15 years, 42.5 million live births and 547,538 infant deaths were recorded, including 354 neonatal deaths attributed to P58-coded causes. In the demographic statistics, the deaths disproportionately affected males, infants of black and brown, and indigenous race/ethnicities, vaginal deliveries, and those born in the North and Northeast, deviating from the overall infant mortality patterns. P58-related deaths declined by 3.7% over the study period. Bivariate analysis showed significantly elevated risks associated with maternal age <20 years (Rate Ratio (RR): 2.05; 95% CI: 1.58, 2.63), low maternal education (RR: 3.46; 95% CI: 2.75, 4.36), male sex (RR: 2.16; 95% CI: 1.73, 2.72), BBYI ethnicity (RR: 1.91; 95% CI: 1.48, 2.48), preterm birth <32 weeks (RR: 21.79; 95% CI: 16.11, 29.08), low birth weight <1500g (RR: 17.28; 95% CI: 12.58, 23.32), vaginal delivery (RR: 3.18; 95% CI: 2.50, 4.06), and being born in the Northeast region (RR: 5.80; 95% CI: 4.07, 8.52) or North (RR: 13.56; 95% CI: 9.46, 19.98). Adjusted models in the hierarchical analysis confirmed increased risks for young maternal age <20 (RR: 1.56; 95% CI: 1.17, 2.05), low maternal education (RR: 3.06; 95% CI: 2.38, 3.92), BBYI race/ethnicity (RR: 1.46; 95% CI: 1.10, 1.95), and male sex (RR: 1.98; 95% CI: 1.54, 2.58).
Discussion and Conclusion: These findings reveal a distinct pattern of P58-associated neonatal mortality, alluding to G6PD deficiency as a likely contributor. The marked male vulnerability found in this study aligns

with the X-linked inheritance of G6PD deficiency. Furthermore, regional disparities, particularly in Brazil's North and Northeast, reflect structural inequalities and potential underdiagnosis compounded by limited access to optimal healthcare. The increased risk linked to vaginal delivery may indicate oxidative stress triggered by metabolic stress or vertical transmission of infections to G6PD-deficient neonates, who have been previously shown to exhibit vulnerabilities to sepsis. Risks linked to low birth weight and preterm deliveries align with known vulnerabilities among neonates. Our hierarchical modeling approach confirmed that both distal and proximal factors jointly influence the risk of infant mortality, with demographic factors exerting the strongest effect. This study supports the use of ICD code P58 as a sentinel marker for G6PD deficiency in regions with little or no data on this condition. Furthermore, an urgent need for neonatal screening and equitable access to healthcare in underserved areas is advocated. Further research, including G6PD activity testing, is essential to confirm these patterns and inform interventions.

Prevalence and risk factors associated with major congenital disorders identified through the UBOMI BUHLE pregnancy exposure registry in Soweto, South Africa.

Ms Dorothy Sefara[1], Professor Michelle Groome[1], Dr Iubomi Levies[2], Mrs Ashleigh Fritz[1], Mr Bongani Ntimani[1], Professor Ushma Mehta[2], Professor Lee Fairlie[3]
[1]South African Medical Research Council Vaccines and Infectious Diseases Analytics Research Unit, Faculty of Health Sciences, University of the Witwatersrand., Johannesburg, South Africa, [2]Division of Clinical Pharmacology, Department of Medicine, Faculty of Health Sciences, University of Cape Town, South Africa. , Johannesburg, South Africa, [3]Wits RHI, University of the Witwatersrand, Johannesburg, Gauteng, South Africa. , Johannesburg, South Africa

Email of corresponding Author: dorothy.sefara@wits-vida.org

Background and Objectives: Congenital disorders (CD) are defined as structural or functional abnormalities that develop during the intrauterine period and can be identified prenatally, at birth, or sometimes only later in life. Causes of CD are multifactorial and may include endogenous causes or exposure to an abnormal foetal environment. Globally, about eight million neonates are born with a CD annually. CD is a leading cause of under-5 mortality and neonatal deaths globally. Most children born with a major CD live in low-and middle-income countries, where more than 95% of CD-related deaths occur. There are limited data on the prevalence of CD across South Africa. A report using data from the Western Cape Pregnancy Exposure Registry showed a prevalence of 7.2 per 1000 births for major CD identified on surface examination at birth. A prospective, hospital-based, observational study in Soweto, Gauteng Province attributed 6/243 (2%) of stillbirths (>22 weeks gestation; ≥500g) to foetal, genetic, or structural abnormalities. The UBOMI BUHLE Pregnancy Exposure Registry (UB PER) prospectively enrols women attending antenatal care at first antenatal visit at 17 selected sentinel sites (primary care obstetric facilities) in three provinces in South Africa and tracks them through to pregnancy outcome, collecting data on the neonate at birth. We used data collected through UB PER sites in Soweto, Gauteng to estimate the prevalence of major CD among live- and stillbirths and investigate the risk factors associated with major CD.

Method: The study was conducted at four Midwifery and Obstetric Units (Chiawelo, Itireleng, Lilian Ngoyi, Mofolo) and two hospitals (Bheki Mlangeni and Chris Hani Baragwanath Academic Hospitals) in Soweto. Eligible women were consented and enrolled prospectively and followed up to pregnancy outcome. Neonates were followed up until discharge from delivery facility. Inclusion criteria included pregnant women receiving antenatal care at one of the study sites with a known pregnancy outcome at a gestational age of at least 22 weeks or baby weighing at least 500g at birth. Data on maternal demographics, medical history, pregnancy details, exposure information (medication and lifestyle factors), and delivery outcomes were collected by trained study staff. CD were identified by neonatal surface examination, review of medical records and case review by specialists, where required. Major CD were defined as structural anomalies with an impact on physical, intellectual or social wellbeing. Data from participants enrolled from July 2021 and with a documented birth outcome to June 2025 were included in the analysis. Descriptive statistics were used to describe the demographic and obstetric characteristics of pregnant women giving birth to infants with a major CD compared to those without any identified CD. Continuous variables are described using mean and standard deviation or median and interquartile range depending on whether data were normally distributed or not. Categorical variables are presented as proportions/frequencies. Chi square or Fishers exact test was used to compare the two groups, as appropriate.

Results: A total of 8530 babies were born to 8401 women, of which 8359 (98.0%) were born alive and 171 (2.0%) were stillborn. There were 122 babies (1.4%) born with any CD identified by surface examination at birth. Fifty-one babies (0.6%) had at least one major CD, 53 babies had at least one minor and no major CD, and 18 had unclassified CDs. Of the babies with a major CD, most had one major CD (86.3%) while 7 babies had >1 major CD. The most common major CD were talipes equinovarus (n=26), cleft lip and/or cleft palate (n=7), hydrocephalus (n=3), albinism (n=3), gastroschisis (n=3) and trisomy 21 (n=2). Of those with major CD,

5/51 (9.8%) were stillborn, 21 (41.2%) had a birthweight <2500g and 11 (32.4%) were born at <37 weeks gestation. There was a significantly higher prevalence of major CD among stillborn babies (5/171, 2.9%) compared to those born alive (46/8359, 0.6%; p=0.004). Babies with a major CD were more likely to be born <37 weeks gestation (odds ratio (OR) 2.6 (95% CI: 1.3-5.3) and more likely to have a birthweight <2500g (OR 3.6; 95% CI: 2.1-6.3) compared to babies born without a CD. When comparing women who gave birth to a baby with a major CD to those without any CD, there was no significant difference in maternal age (27.1 vs. 28.3 years, respectively), HIV prevalence (20.0% vs. 21.1%, respectively), use of illicit substances, herbal medicines, alcohol or tobacco, or history of a previous baby born with a CD.

Discussion and conclusion: This study provides estimates of the prevalence of major CD among live- and stillbirths in Soweto. It is reassuring that no difference in the prevalence of major CD was observed in women living with HIV compared to women without HIV. Importantly, identification of significant risk factors for major CD might have been limited by the small sample size as this preliminary analysis only included data from sites in Soweto. Inclusion of data from all the UB PER sites across three provinces will increase the sample size and enable a more robust and generalisable analysis of prevalence and risk factors for CD. The UB PER project has a strong capacity building and health systems strengthening focus, resulting in strengthened clinical care and record keeping for pregnant women and their newborns at sentinel sites which will benefit mothers and babies. Training of health care workers on identification and documentation of CD was provided through UB PER which will improve identification, classification, notification and correct referral for babies born with CD. The study is limited to CD identified through surface examination at birth so underestimates the prevalence of total CD including functional CD and those manifesting or identified after birth. These findings can be used to assist policy makers in resource allocation and planning for comprehensive care. In addition, these data provide baseline rates of CD, so any deviation from normal rates can provide an early warning and trigger investigation of possible pregnancy exposures.

Enhancing screening of congenital anomalies in low resource settings: Insights from the MiMBa Pregnancy Exposure Registry in Kenya

Mr Kennedy Wekesa[1], Dr Bernard Omondi[1], Dr Caroline Osoro[1], Mr Michael Alaw[1], Mr Bernard Asuke[1], Mr Titus Ochieng[1], Ms Mercy Chepkirui[1,2], Dr Martina Oneko[1], Prof Feiko terKuile[1,2], Dr Hellen Barsosio[1,2], Dr Stephanie Dellicour[2]
[1]Kenya Medical Research Institute, 1578 Kisumu, Kenya, [2]Liverpool School of Tropical Medicine, Liverpool, United Kingdom

Email of corresponding Author: kennedy.maube@lstmed.ac.uk

Background and objectives: Congenital anomalies (CAs) represent a significant global health burden, with over 90% of related deaths occurring in low- and middle-income countries (LMICs). In these settings, fragmented healthcare systems, limited diagnostic capacity, and inadequate surveillance programs contribute to underreporting and inaccurate prevalence estimates, hindering effective public health interventions. Lack or inaccurate prevalence estimates of CAs in many LMICs, mainly due to limited diagnostic capacity and inadequate surveillance, significantly hamper research efforts, particularly in pharmacovigilance studies of therapeutics in pregnancy, as it becomes challenging to determine whether the observed rates of anomalies in exposed populations differ from that of the general population. It also complicates study planning by limiting the ability to accurately calculate required sample size. The World Health Organization (WHO) is advocating for improved CA prevention and management through integrated screening, timely diagnosis, and comprehensive care. In this presentation, we share lessons learnt from the MiMBa Pregnancy Exposure Registry on CA screening and surveillance in Kenya.
Method: The MiMBa pregnancy exposure registry is a multi-centre prospective cohort study in Homa Bay County, Kenya, assessing antimalarial safety during pregnancy. Women of childbearing age were enrolled by community health promoters, screened periodically for pregnancy, and referred to routine antenatal care upon confirmation. Study clinicians offered ultrasound scans for gestational dating and fetal viability. Healthcare workers across 42 sentinel facilities received training to systematically screen newborns for CAs using the Global Birth Defect Description and Coding (GBDDC) application. Major CAs were defined as those requiring medical/surgical intervention or significantly affecting quality of life while minor CAs were those with minimal functional impact. Infants were examined at delivery and during the six-week postnatal visit. Suspected CAs were recorded in study-specific case report forms and uploaded to the GBDDC app. Major CA cases were reviewed anonymously by experts via the GBDDC platform for diagnosis confirmation and inclusion in safety analyses. Critical cases were immediately referred for emergency care, while noncritical cases were scheduled for pediatric review. For Congenital Heart Defect (CHD) screening, healthcare workers performed SpO2 measurements on fingers and toes; infants with SPO2<95% or differences greater than 2% were flagged for further assessment and echocardiography. A subset of infants was followed up at 6 and 12 months for neurodevelopmental assessment, with repeated CA screening. Descriptive analysis assessed the prevalence and timing of minor and major CAs and diagnostic consistency across healthcare levels. The ICBDSR Data Quality Indicator tool was used for assessing data quality and completeness.
Results: Between February 2021 and May 2025, a total of 11,703 pregnancies were recorded in Kenya, of which 10,039 (86%) had recorded outcomes including 9,270 livebirths, 190 stillbirths, 571 miscarriages and eight ectopic pregnancies. Among the livebirths and stillbirths, 1,050 (11%) suspected CAs were identified by healthcare practitioners, 936 (89%) minor and 124 (12%) major. The timing of detection for major anomaly varied with 73 (59%) detected at delivery, 47 (38%) at week six postnatally, 2 (2%) at six months, and 2 (2%) at 12 months. Of the 124 suspected cases of major CAs diagnosed by healthcare workers, 78 were confirmed as major by the expert panel. Nine (12%) major CA were identified in stillborn infants and 69 (88%) in livebirths. Diagnosis code selected by the healthcare worker was corrected in 5% of cases after expert review. Examination by a local paediatrician occurred in 80 (65%) cases with 10 (8%) evaluated by pediatric cardiologist. Thirthy-four (27%) not reviewed by pediatrician were stillbirths or critically ill neonates referred

immediately after birth. Among the 78 major CAs confirmed by the expert panel, talipes and limb deformities were the most common accounting for 15% (n=19), followed by CHDs at 8% (n=10). Prenatal detection of major CA was rare with only 8% (n=6) cases identified via ultrasound. Using the Data Quality Indicator tool, ventral abdominal wall defects, talipes, and cleft lip and palate conditions met minimum prevalence thresholds, while neural tube defects, critical CHDs, and Down syndrome didn't meet the target.

Discussion and Conclusion: The MiMBa study highlights important gaps and opportunities in CA screening in low-resource settings. An overall prevalence rate of 1% among live births falls at the lower end of the global range (1-3%), whereas the 5% prevalence among stillbirths is notably lower compared to global estimates that can reach up to 23% especially in high income countries. This discrepancy may reflect limited diagnostic capacity, as the majority of major CAs were identified only within the first six weeks of life, primarily through basic physical surface examinations and CHD screening, rather than through comprehensive diagnostic imaging or genetic testing. Despite systematic screening, CHD underdiagnosis likely from subtle presentations and limited diagnostic access. The lower prevalence of neural tube defects may be a true reflection of the prevalence in this population, as they are identified by surface examination. These findings emphasize the need to strengthen prenatal and postnatal CA surveillance with standardised screening protocols, improved diagnostic access such as echocardiography, enhanced training, and establishment of a national congenital anomaly registry to guide prevention and care strategies.

Newborn Screening in South Africa: Current Status, Challenges, and Opportunities for Expansion

Ass Prof Chris Vorster[1], Ass Prof Helen Malherbe[1]
[1]Centre For Human Metabolomics, North-west University, Johannesburg, South Africa

Email of corresponding Author: Chris.Vorster@nwu.ac.za

Background & Objectives: Biochemical newborn screening (NBS) is a proven public health intervention that enables early detection and treatment of selected rare diseases (RD), reducing preventable morbidity, mortality, and lifelong disability. Globally, NBS is widely implemented in high-income countries, but progress in low- and middle-income countries (LMIC), including South Africa (SA), has been limited. SA's RD burden—estimated to affect 1 in 15 citizens, equating to over four million people, half of whom are children—is compounded by delayed diagnoses and poor access to treatment, with many patients remaining undiagnosed. While pilot programmes and research initiatives have demonstrated the feasibility of NBS in SA, systematic national implementation is lacking. In SA, biochemical NBS is currently limited to opportunistic screening in private healthcare and selected public-sector pilot programmes, primarily for congenital hypothyroidism (CH), covering less than 0.5% of annual births. Timely diagnosis of RD through NBS has been identified as a strategic priority, requiring specific national actions by recent World Health Assembly Resolutions and in the context of Sustainable Development Goal 3.2 to reduce preventable newborn and under-5 mortality. This study aims to synthesise current knowledge on NBS in SA, describe existing service delivery models and screening coverage, identify barriers to expansion, and outline strategic opportunities for developing an equitable, sustainable national NBS programme.
Method: A desktop review of the South African NBS landscape was conducted, drawing on peer-reviewed and grey literature, national policy documents and livebirth reports, global NBS reviews, and workshop outputs. Service mapping included sectoral provision (public vs private), NBS panel composition, laboratory capacity, and referral pathways. This review was descriptive in nature and did not involve new patient data collection; findings were synthesised to inform policy recommendations and identify feasible, high-impact next steps.
Results: There is no national, publicly funded NBS programme in SA. The Centre for Human Metabolomics (CHM) at North West University (NWU) provides a 22-condition dried blood spot panel to both healthcare sectors on a user-pay basis, with most uptake from the private sector. Public-sector NBS is restricted to limited CH screening in some Cape Town facilities. Between January 1998 and September 2023, 125,888 newborn specimens were tested across both sectors, representing ≈0.44% of ~28.7 million births over this period. Early postpartum discharge (~6 hours for healthy dyads) in the public sector hinders the ideal 24–48-hour collection window, and weak tracking systems contribute to significant loss-to-follow-up. The February 2023 National NBS Workshop—attended by all relevant stakeholders—agreed on CH as a strategic starting point for public-sector implementation and recommended a demonstration project in two contrasting provinces. Plans are underway to implement pilot studies in the Limpopo and North West Provinces. The review found substantial technical capacity, including at CHM and regional laboratories, but no national coordination, standardised protocols, or sustainable financing. International comparators also suggest CH NBS is feasible, cost-effective, and a strong foundation for progressive expansion to additional conditions. Recently secured funding for a CH NBS Cost Utility Analysis (CUA) will quantify cost per health outcome, budget impact, and opportunity costs, addressing Department of Health requirements for financial evidence before adopting new, and expanding existing services. Subsequent studies are planned to quantify the cost of expanding the NBS panel, following once-off costs for CH NBS.
Discussion & Conclusion: South Africa's NBS landscape is characterised by technical readiness but minimal public-sector reach. Historical coverage of ~0.44% of births over 25 years underscores a missed public health opportunity. The 2023 NBS in SA workshop provided consensus on CH as an entry-point, aligning with WHO guidance for prioritising conditions with high prevalence, significant morbidity, and cost-effective, available and accessible treatment. The forthcoming CH CUA study will provide essential evidence for policymakers,

quantifying value-for-money, budget impact, health outcomes and opportunity costs within a resource-constrained system. The dual health system in SA—public (~84% of the population) and private (~16%)—requires a design that leverages CHM's existing infrastructure while embedding governance, financing, and service delivery under public stewardship to ensure equity. Implementation challenges include aligning NBS sampling with postpartum care practices, developing robust tracking systems to reduce loss-to-follow-up, and integrating NBS into maternal and child health workflows and aligning with existing programmes. Early wins could be achieved by synchronising sample collection with existing touchpoints (e.g., HIV/AIDS programmes, immunisation visits etc) and using established laboratory networks and in infrastructure for logistics. As CH NBS demonstrates feasibility and cost-effectiveness, the programme could be expanded to include other high-yield conditions such as congenital adrenal hyperplasia and hemoglobinopathies, guided by local burden and fiscal space. Strategic investment now could transform SA's NBS from a fragmented, privately driven service to a coordinated, publicly financed national programme, yielding substantial health and economic returns and reducing preventable morbidity and mortality in South African newborns.

Merging clinic-based research with population-based surveillance: the Ponseti Clubfoot network

Dr Paul Romitti[1,2], Dr Jose Morcuende[3,4], Dr Val Sheffield[4,5], Dr Kristin Conway[1], Dr Marilize Burger[6], Ms Karen Moss[7], Dr Ushma Mehta[8], Dr Joan Morris[9]
[1]Department of Epidemiology, College of Public Health, University of Iowa, Iowa City, United States, [2]Iowa Registry for Congenital and Inherited Disorders, Iowa City, United States, [3]Departmentof Orthopaedics and Rehabilitation, Carver College of Medicine, University of Iowa, Iowa City, United States, [4]Stead Family Department of Pediatrics, Carver College of Medicine, University of Iowa, Iowa City, United States, [5]Department of Ophthalmology, Carver College of Medicine, University of Iowa, Iowa City, United States, [6]Division of Orthopaedic Surgery, Department of Surgical Sciences, Faculty of Medicine and Health Sciences, Stellenbosch University, Cape Town, South Africa, [7]Steps Charity NPC, Cape Town, South Africa, [8]Division of Clinical Pharmacology, Department of Medicine, University of Cape Town, Cape Town, South Africa, [9]School of Health & Medical Sciences, City St George's University of London, London, United Kingdom

Email of corresponding Author: paul-romitti@uiowa.edu

Background and Objectives: Investigating the etiopathogenesis of nonsyndromic birth defects requires comprehensive examination of environmental and genetic factors. These studies often leverage population-based surveillance programs to identify and recruit subjects, typically using mail and telephone correspondence. Our experience with two United States (US) case-control studies from 1997-present shows a marked decline in participation rates over the study period, threatening external validity of findings. Although the Iowa Registry for Congenital and Inherited Disorders (IRCID) has maintained some of the highest participation rates in each study, our evaluation suggests that this decline can be attributed to technology preferences and additional factors. Addressing these factors can improve participation rates, including biological specimen collection, which is critical to improve understanding of the etiopathogenesis of defects with strong genetic predispositions. One such defect, nonsyndromic (isolated) clubfoot, is the most common musculoskeletal defect among newborns, affecting 1 in 750 live births worldwide, with 80% of affected children born in low- and middle-income countries (LMICs). Few high throughput genomic studies have been conducted for clubfoot with little consensus in associated variants identified. Similarly, few comprehensive studies of environmental exposures have been conducted; except for cigarette smoking, findings are equivocal. Addressing the challenges encountered in the US case-control studies, we used the resources of the University of Iowa Ponseti Clubfoot Treatment and Research Center (PCTRC) and IRCID to establish the Ponseti Clubfoot Network (PCN), a research network to comprehensively examine clubfoot etiopathogenesis. Using the resources of the Ponseti International Association (PIA), we are expanding our work to LMICs.
Methods: Previously, we compared collecting exposure data and biological specimens using web-based surveys and consent documents with telephone interviews and paper-based consent documents and showed increased participation rates for mothers using a web-based mode. For PCN, we extended this work to conduct clinic-based recruitment evaluating maternal factors to improve participation rates, specifically integrating clinical providers familiar to mothers as principal research team members. Mothers whose children previously received or are currently receiving treatment from PCTRC are assigned to a retrospective (2010-2024) or prospective (2025-present) cohort; follow-up uses our previously developed and tested protocols. Data collection (reimbursement) for the retrospective cohort includes completion of a web-based family history questionnaire by mothers ($10) and at-home saliva specimen collection by the child-parent trio ($10 per family member); the prospective cohort also includes completion of a web-based pregnancy exposure questionnaire by mothers ($20). We plan to recruit 250 mothers from our retrospective cohort and 50 from our prospective cohort to collect sufficient specimens for whole genome sequencing and reduce recall time between pregnancy and completion of the exposure questionnaire. Participation is estimated for each cohort with rates stratified by selected characteristics (child birth year; maternal age, educational attainment, race/ethnicity, parity). Questionnaire items will be used to evaluate mothers' experiences with

response choice, frequencies, patterns, and breakoffs. Also, cost per family to enroll, complete questionnaires, and provide saliva specimens will be estimated. To evaluate external validity, proportions and selected characteristics of enrollees are compared with those of all nonsyndromic clubfoot cases ascertained by IRCID.

Results: To date for our retrospective cohort, we have enrolled 118 children diagnosed with nonsyndromic clubfoot treated at the University of Iowa PTCRC. Of these, 86 (73%) birth mothers of these children have completed the family health history questionnaire, and families of 62 (53%) mothers who completed the family health history questionnaire provided saliva specimens; 55 (89%) of families providing saliva specimens were complete child-parent trios. Our preliminary questionnaire response rate is higher than that (67%) for questionnaire data collection in the US National Birth Defects Prevention Study (NBDPS; 1997-2011) and well-exceeds that (44%) for the current US Birth Defects Study to Evaluate Pregnancy exposureS (BD-STEPS; 2014-present). Collection of both questionnaire and biological specimens in PCN also well-exceeds that (37%) from the US NBDPS (biological specimen collection from child-parent trios is not included in the US BD-STEPS protocol). As data collection progresses, we continue to evaluate participation rates by selected characteristics and maternal experience with questionnaire items. Data collection for the prospective cohort (2025-present) was recently commenced. When data collection concludes, we will estimate cost per family and evaluate external validity of our participation. Additionally, we have established a partnership with Steps Clubfoot Care, a nonprofit organization supporting a national network of clubfoot clinics in South Africa and investigators at Stellenbosch University in Cape Town, South Africa to expand PCN to the Western Cape by means of their established National Congenital Talipes Equino-Varus (CTEV) Database. We are also exploring expansion of PCN to other South African provinces.

Discussion and Conclusions: In Iowa, use of web-based questionnaires for studies of nonsyndromic birth defects produces participation rates exceeding those achieved using telephone interviews. This finding supports our previous hypothesis that web-based data collection approaches are increasingly preferred by mothers given the migration from landline to smartphone use. Our use of clinic-based ascertainment integrating clinical providers familiar to the study subjects appears to improve participation rates over those obtained from recruitment and consenting by surveillance program staff typically unfamiliar to subjects. Combining web-based approaches with clinic-based recruitment facilitates implementation of data collection activities in orthopedic clinics in LMICs and can be applied to conduct comprehensive investigations of other surgically-treated birth defects in LMICs. Our retrospective cohort provides a time-efficient approach to establish a repository of biological specimens to facilitate identifying causal variants across diverse populations. Our prospective cohort facilitates building a repository of epidemiological data to evaluate the environmental exposures in the presence or absence of causal variants identified. In summary, clubfoot occurs in every country around the world with no economic, social, or cultural influences. Together, these cohorts also offer opportunity for life course surveillance of patient experiences. Untreated, clubfoot persists as a rigid, unsightly deformity, being one of the most common causes of physical disability. Even with correction in infancy, varying rates of relapse have been observed in childhood and adolescence, providing additional evidence of its heterogenous etiopathogenesis. Improved understanding of this etiopathogenesis can guide development of precision treatments specific to an underlying pathogenicity and improve patient outcomes.

Tuesday, 4th November

Session 6 – Round Table Session

Moderators:
Mary Ann Thomas & Anke Rissman

Round Table Session
Panel Discussion and Action Plan of Topics Pre-selected by Birth Defect Surveillance Networks (EUROCAT/NBDPN/ICBDSR/sSCAN/Ubomi Buhle/ReLAMC)

ICBDSR New Report Format
Lorenzo Botto

Round Table Session

Panel Discussion and Action Plan of Topics Pre-selected by Birth Defect Surveillance Networks (EUROCAT/NBDPN/ICBDSR/sSCAN/Ubomi Buhle/RELAMC)

ICBDSR New Report Format

Lorenzo Botto

Wednesday, 5th November

Session 7 – Oral Presentations

Moderator: Wendy Nembhard

A systematic review of the long-term outcomes of neural tube defects in the Eastern African region.
Hope Mwangudzah

Descriptive Analysis of Congenital Anomalies Among Admitted Newborns across hospitals implementing with the NEST360 Alliance.
Keisuke Ogasawara

Privacy-Enhancing Technologies for International Registry Collaboration: An England-US Pilot
Sarah Stevens

Biobanks as Catalysts for Birth Defect and Rare Disease Programs: Linking Diagnostics, Treatment, Research, and Awareness in LMIC.
Engela Helena Condradie

Missed Opportunities: Addressing Diagnostic Delays in Structural Birth Defects Among Ethiopian Children Through Screening and System Strengthening.
Hiwote Assefa

Preliminary declaration for publication on ICBDSR annual meeting
Helen Malherbe & Emma Kalk

A systematic review of the long-term outcomes of neural tube defects in the Eastern African region

Ms Hope Mwangudzah[1]
[1]University Of Bristol, Bristol, United Kingdom

Email of corresponding Author: hope.mwangudzah@bristol.ac.uk

Background and Objectives: High childhood mortality and morbidity from neural tube defects (NTDs) in low and middle-income countries (LMICs) represent a complex public health challenge. Whilst folic acid supplementation and food fortification have been shown to reduce the occurrence of NTDs, current prevention efforts in LMICs have achieved limited success, resulting in a high burden of NTDs. This is primarily due to inadequate preconception care, including unplanned pregnancies, insufficient family planning, and low health literacy levels. With advances in care, high-income countries (HICs) have enhanced survival and quality of life for children with NTDs. In contrast, progress in LMICs remains slow; therefore, improving quality of life for these children must be prioritized by addressing immediate healthcare needs. We aimed to describe the long-term outcomes of NTDs in the Eastern African (EA) region among individuals up to 18 years across four areas through:
1. Survival rates from birth to 18 years
2. Physical complications include paralysis, bladder/bowel dysfunction and hydrocephalus
3. Educational outcomes, including school attendance and special education needs (SEN)
4. Neurodevelopmental delays in vision, cognition, speech, hearing, and motor function

Methods: PubMed Central, PubMed/Medline, the Cochrane Library, Web of Science, and Africa Index Medicus were systematically searched from the beginning to April 4, 2025, with restrictions to English-language publications. We included observational studies that reported long-term outcomes of NTDs in the EA region. The Risk of Bias In Non-Randomised Studies-Of Exposure (ROBINS-E) tool was used to assess study quality. Outcome measures included risk of specific conditions: mortality, hydrocephalus, paralysis, urine and bowel dysfunction, neurodevelopmental delays, school attendance and SEN.

Results: We identified 19 studies comprising data from 4784 study participants. Myelomeningocele (MMC) was the most common NTD subtype studied with respect to long-term outcomes, followed by encephalocele. The probability of 10-year survival after defect repair was 51%. These children frequently experienced hydrocephalus and urinary dysfunction, which were reported across six countries in the EA region. Most children with urinary dysfunction also had lower limb paralysis; however, it was difficult to evaluate the impact of paralysis on bowel dysfunction since only two studies reported on this outcome. School attendance ranged from 36% to 56% in studies reporting this, with limited data on children with SEN and neurodevelopmental delays. Only two studies separately reported on hearing and cognition development. One study examined cognition by comparing siblings of children with spina bifida and the children with spina bifida who attended and did not attend school. Better cognition was observed in children attending school, although it was less pronounced than that of their siblings. No studies on long-term NTD outcomes were identified in the majority (71%) of the countries in the EA region.

Discussion and Summary: This systematic review offers crucial insights into the long-term outcomes of NTDs in the EA region, highlighting a remarkable disease burden and notable gaps in research. The 51% ten-year survival rate sharply contrasts with figures from high-income countries (85-90%), reflecting limited healthcare infrastructure, delayed diagnosis, and inadequate post-operative care. The high mortality underscores broader healthcare system challenges in managing complex conditions, including the absence of structured follow-up. Consistent with the literature, MMC was the most studied subtype and the most common form of NTD. Hydrocephalus and urinary dysfunction, also known as neurogenic bladder, were reported across six countries, aligning with global patterns. The strong link between urinary dysfunction and lower limb paralysis indicates severe spinal cord involvement, likely due to delayed surgical intervention. Conversely, its relationship with bowel dysfunction could not be inferred due to the lack of studies that reported this outcome.The school attendance rates, ranging from 36 to 56% highlight remarkable educational disparities

for children affected by NTDs. Furthermore, the limited data available on neurodevelopmental delays present a challenge in determining the true extent of cognitive, speech and hearing impairments among this population. Notably, the lack of research in 71% of EA countries greatly restricts our understanding of the disease burden in the region and hinders the development of evidence-based strategies. These findings emphasise the urgent need for comprehensive NTD management programmes that go beyond surgical intervention to include long-term multidisciplinary care, improved prevention strategies, and expanded research initiatives across the region.

Descriptive Analysis of Congenital Anomalies Among Admitted Newborns across hospitals implementing with the NEST360 Alliance.

Mr Keisuke Ogasawara[1], Dr. Kristina Shemwell[1], Dr. Hannah Blencowe[1], Dr. James Cross[1], Dr. Joy Lawn[1]
[1]LSHTM, London, United Kingdom

Email of corresponding Author: guts2keiogasawara@gmail.com

Background & Objectives: Congenital anomalies (CAs) are a major contributor to neonatal mortality, affecting an estimated 3–6% of births globally. This burden is unequally distributed, with over 90% of cases occurring in low- and middle-income countries, where CAs are a leading cause of neonatal death. In these settings, early identification and management are often constrained by limited access to diagnostic tools, varied clinical training, and weak health system capacities. Consequently, the epidemiological understanding of CAs remains limited, particularly in sub-Saharan Africa, where many anomalies go undetected. Improved data are needed to support evidence-based policy, resource allocation, and clinical training in this region. The Newborn Essential Solutions and Technologies (NEST360) Alliance provides a unique platform to address this data gap. As an initiative strengthening neonatal care through technology, clinical education, and standardized data systems, NEST360 systematically collects clinical data from a large network of hospitals across sub-Saharan Africa. Therefore, this study aims to describe the prevalence and characteristics of CAs among admitted neonates using data from the NEST360 Neonatal Inpatient Dataset.
Method: This multi-site, cross-sectional study utilized secondary data from the Neonatal Inpatient Dataset, a prospective clinical registry established by the NEST360 Alliance. The dataset includes neonatal admissions from 66 hospitals, ranging from district-level to tertiary national facilities, across four sub-Saharan African countries: Malawi, Kenya, Tanzania, and Nigeria. This analysis included all neonates with a diagnosed congenital anomaly admitted between January 1, 2021, and December 31, 2024. Descriptive analyses were performed on key demographic and clinical characteristics, including sex, country of birth, birth weight, gestational age, and survival to discharge. Following the NEST360 classification protocol, anomalies were grouped into predefined categories, including neural tube defects, abdominal defects, congenital heart disease, orofacial clefts, talipes and chromosomal disease. All analyses were conducted using Stata. Ethical approvals were obtained from the London School of Hygiene & Tropical Medicine and from local institutional review boards in each participating country.
Results: A total of 638,690 neonatal admissions were recorded, of which 391,876 met the inclusion criteria. Among the 388,649 neonates with a final diagnosis, 14,836 (3.81%) were diagnosed with at least one CA. By anomaly type, abdominal defects (27%) were the most frequently reported, followed by congenital heart disease (19%) and neural tube defects (18%). The prevalence of CAs varied significantly by country. No significant association was observed with sex. A dose-response relationship was found between the odds of a CA diagnosis and both birth weight and gestational age. Compared with infants weighing <1000 g, the crude odds of diagnosis were highest among those weighing >4000 g. Similarly, compared with infants born at <28 weeks' gestation, term neonates (37–41 weeks) had the highest crude odds of diagnosis. The mortality rate before discharge was substantially higher for neonates with CAs (23.6%) than for those without (13.3%), corresponding to significantly higher odds of death (OR: 2.02, 95% CI: 1.94–2.10). Mortality risk also varied by anomaly type, with abdominal defects (31.4%), chromosomal diseases (30.9%), and heart defects (28.0%) associated with the highest mortality rates.
Discussion & Conclusion: This study, the largest of its kind in sub-Saharan Africa, highlights that CAs are a significant factor in neonatal admissions across health facilities in sub-Saharan Africa and are strongly associated with an increased risk of in-patient mortality. The higher prevalence of CAs observed in admitted term infants compared to their preterm counterparts likely reflects admission bias: preterm infants are more likely to be admitted for supportive care associated with prematurity alone, whereas term infants are admitted more selectively for specific medical conditions. Furthermore, the predominance of externally visible anomalies over internal defects may reflect the challenges of detecting conditions that require

specialized diagnostic tools like echocardiography, or lack of facilities required for autopsy following neonatal death. Therefore, this study may underestimate the overall burden of CAs amongst admitted infants. Nevertheless, this study provides a crucial foundation for understanding the burden of CAs in this region by analysing the current situation and identifying key challenges within this unprecedentedly large cohort. Therefore, strengthening diagnostic capacity and leveraging existing hospital networks to establish sustainable, multi-site surveillance systems will be critical to accurately quantify the burden and guide targeted public health interventions.

Privacy-Enhancing Technologies for International Registry Collaboration: An England-US Pilot

Bardia Gueval[1], Lucy Irvine[1], James Thomas[1], Brian Rous[1], Serban Negoita[2], Lynne Penberthy[3], John Smith[4], **Mrs Sarah Stevens**[1]
[1]NHS England, London, United Kingdom, [2]National Cancer Institute, , United States, [3]Westat, Rockville, United States, [4]Department for Innovation, Science and Technology, London, United Kingdom

Email of corresponding Author: sarah.stevens1@nhs.net

Lay Summary: Privacy-Enhancing Technologies (PETs) offer innovative solutions for international data collaboration without sharing sensitive information. Our proof-of-concept pilot assesses how these emerging technologies could enable valuable cross-border research on rare diseases while maintaining robust privacy protections.

Background: Childhood cancers are a group of rare and ultra-rare diseases. The low number of patients hampers the research required to contribute to advances in diagnosis, treatment, and outcome. Therefore, large-scale international data collaborations in childhood cancer are crucial. However, international data sharing processes face long-standing challenges from privacy regulations and resource-intensive governance processes with uncertain outcomes. We hypothesised that a layered PET architecture could offer a promising solution to epidemiological research by enabling cross-country analysis while keeping sensitive data secure.

Objective: To develop and evaluate a PET-enabled federated querying system that facilitates secure international analysis of rare paediatric cancer data between national disease registries while maintaining privacy.

Methods: We implemented a federated analytics infrastructure connecting two major cancer registries: the US Surveillance, Epidemiology, and End Results Program (SEER) at the National Cancer Institute (NCI) and the National Disease Registration Service (NDRS) NHS England. We harmonised core demographic, clinical and outcome data across registries by aligning coding structures. Our analysis focused on seven rare paediatric cancers in children aged 0-19 years (2013-2022). A federated querying system was developed to enable cross-country analyses by aggregating data held in local nodes. Novel PETs and privacy thresholds were applied to mitigate the risk of individuals being reidentified:

1. Federated Analytics (FA): Keeps data in its original location while enabling collaborative analysis across distributed sources.
2. Trusted Execution Environment (TEE): Provides an encrypted processing enclave for secure data aggregation.
3. Differential Privacy (DP): A mathematical privacy framework whereby controlled noise is added to aggregated analysis results, ensuring individuals' data cannot be reliably inferred.

We conducted comprehensive testing to validate the system's core functionality, privacy features and analytical accuracy with initial testing conducted on synthetic data.

Results: A federated system was developed that analyses cross-border England-US datasets, while applying multiple PETs and maintaining compliance with each jurisdiction's data governance requirements. Initial testing with England registry data demonstrates successful data harmonisation and query execution, with early verification against existing summary statistics showing analytical accuracy. Learnings from this pilot indicate that using PETs could enhance international epidemiological research, while supporting population-level analysis that would lack statistical power in single registry studies.

Conclusion: Our work demonstrates how PETs can streamline international health data collaboration while preserving privacy and regulatory compliance. This approach has potential scalable value for rare disease research where international data collaboration is needed. The system's architecture can be expanded to include additional registries and accommodate more complex research questions, representing an important advance in balancing data utility with privacy protection for cross-border health research.

Biobanks as Catalysts for Birth Defect and Rare Disease Programs: Linking Diagnostics, Treatment, Research, and Awareness in Low- and Middle-Income Countries

Dr Engela Helena Conradie[1], Prof Ilse Du Preez[1], Prof Helen Malherbe[1], Prof Christian Johannes Hendriksz[1,2], Prof Barend Christiaan Vorster[1]
[1]Centre for Human Metabolomics, Desmond Tutu School of Medicine, Faculty of Health Sciences, North-West University, Potchefstroom, South Africa, [2]A Rare Cause, England and Wales registered Charity nr:1198461, 6 The Green, Ilkley LS29 7FF, United Kingdom

Email of corresponding Author: Elne.Conradie@nwu.ac.za

Background & Objectives: Birth defects and rare diseases (RDs) collectively affect millions globally, yet in low- and middle-income countries (LMICs), diagnostic and treatment pathways remain fragmented. Limited access to specialised testing, scattered infrastructure, and minimal integration of clinical data severely constrain early detection, coordinated care, and research capacity. Biobanks—repositories that store biological samples alongside rich clinical and demographic data—are uniquely positioned to bridge these gaps by linking biospecimens to diagnosis, treatment, research, and public awareness initiatives. This abstract showcases how a South African rare disease biobank has evolved beyond a traditional repository to become a strategic connector across diagnostic, public health, and community engagement programs in LMIC contexts. Four interlinked initiatives illustrate this role: (1) the FYMCA pilot, which created a multinational clinician training and dried blood spot (DBS) diagnostic network for lysosomal storage disorders; (2) The Babylog concept, a proposed integrated clinical data platform for longitudinal child health monitoring and early intervention; (3) The Nngwe initiative, which unifies genomics infrastructure to improve rare disease diagnosis efficiency and equity; and (4) Collaboration with the Rare Diseases South Africa (RDSA) patient registry to align biospecimen collection with patient-reported outcomes. The study objective is to demonstrate how integrating biobanking with clinical data systems, cross-border diagnostics, and patient advocacy networks can: (a) Improve early and accurate diagnosis; (b) Facilitate research translation; and (c) Build sustained awareness and policy relevance for rare diseases and birth defects in LMICs. This integrated approach offers a scalable, adaptable model for similar settings worldwide.
Method: The biobank's integrative approach leveraged four complementary strategies:
1. Multinational Diagnostic Pilot (FYMCA Project)
A collaboration between FYMCA Medical Ltd., the International Gaucher Alliance, and the Centre for Human Metabolomics established a clinician training program and DBS-based diagnostic pipeline for six lysosomal storage disorders across eight Sub-Saharan African countries. Samples were shipped to South Africa for analysis, while training sessions blended theoretical and practical components.
2. Child Health Data Integration (Babylog)
Conceptualised as a national, secure, interoperable database, Babylog aims to longitudinally capture maternal, neonatal, and child health indicators—including newborn screening results and developmental assessments—linking these data to biobanks for future research integration. Stakeholder mapping, needs assessment, and ethical framework development formed the preparatory phase.
3. Omics Infrastructure Integration (Nngwe Initiative)
Coordinated under DIPLOMICS and the Department of Science and Innovation, Nngwe created a shared national workflow for disease-agnostic genomics, metabolomics and proteomics testing, consolidating existing omics facilities, data-sharing agreements, and bioinformatics capacity to address diagnostic inequities for rare diseases.
4. Patient Registry Alignment (RDSA Collaboration)
Partnership with RDSA ensured that biospecimen collection protocols captured data aligned with patient-reported outcomes, registry variables, and community priorities.
These strategies shared core principles: community engagement at design stage, adherence to ethical and governance standards, and alignment with existing health system capacities. Data and sample flows were

designed to be bidirectional—feeding back results to clinicians and patients while informing research and public health strategies.

Results: The integrative model delivered tangible outputs across the four initiatives:

- FYMCA Project: Trained clinicians from eight countries registered 56 patient samples, and achieved a high diagnostic yield for Gaucher disease and mucopolysaccharidosis type II. Variants identified aligned with global patterns, while Fabry disease diagnoses were rare—prompting calls for targeted awareness. Key barriers included sample transport delays and gaps in referral networks.
- Babylog: Stakeholder consultations confirmed widespread support for integrated data capture, highlighting gaps in newborn screening follow-up and early developmental monitoring. The proposed architecture was endorsed for pilot implementation in the North West province, with potential linkage to national birth defect surveillance and biobanks.
- Nngwe Initiative: Mapped and integrated existing omics platforms into a coordinated workflow, reducing turnaround times for certain rare disease diagnoses and enabling cross-institutional case discussions. Early-stage pilots demonstrated feasibility for multi-site sample processing and data sharing.
- RDSA Collaboration: Established protocols for harmonising biobank data with patient registry records, ensuring community priorities—such as timely return of results and informed consent in multiple languages—were embedded in practice.

Across initiatives, the biobank facilitated connections between laboratory analysis, clinical care, data platforms, and advocacy. This generated cross-sector visibility, influenced health policy discussions on newborn screening, and positioned the biobank as a trusted partner in both research and patient communities.

Discussion & Conclusion: These results underscore the potential for biobanks in LMICs to act as active, integrative agents in the rare disease and birth defect ecosystem—moving beyond passive sample storage towards dynamic facilitation of diagnosis, care, and advocacy. By embedding biobanking within multinational diagnostic networks (FYMCA), health information systems (Babylog), genomics infrastructure (Nngwe), and patient registries (RDSA), the model addressed critical gaps: fragmented diagnostic pathways, underutilised infrastructure, and disconnection between research and patient needs. The high diagnostic yield in the FYMCA pilot, combined with the strong stakeholder engagement for Babylog, demonstrates that coordinated, ethically grounded integration of biospecimens and clinical data can accelerate early detection and improve health system responsiveness. Challenges—such as logistics in sample transport, variable regulatory environments, and sustainability of cross-border collaborations—highlight the need for robust governance frameworks and capacity building. The alignment of registry and biobank data illustrates how patient voices can directly inform research priorities, enhancing both scientific relevance and trust. For LMICs, where resources are limited but the burden of undiagnosed conditions is high, this approach offers a scalable blueprint. Future directions include expanding newborn screening linkages, integrating additional omics data, and embedding these networks within national health policy. Ultimately, the rare disease biobank's evolution into a hub connecting biospecimens, clinical data, and communities demonstrates how such institutions can catalyse systemic improvements—transforming individual diagnostic journeys while strengthening public health and research ecosystems.

Missed Opportunities: Addressing Diagnostic Delays in Structural Birth Defects Among Ethiopian Children Through Screening and System Strengthening

Dr Hiwote Assefa[1], Dr Hana Gebreselassie[1], Dr Ephrem Kerego[1]
[1]Saint Paul's Hospital Millennium Medical College, Addis Ababa, Ethiopia

Email of corresponding Author: hiwote.girma@sphmmc.edu.et

Background and objective: Congenital birth defects are among the common health problems faced by children in the global south, contributing substantially to infant mortality rates. This study intends to describe the timing of diagnosis of children with structural birth defects and the reasons for delayed diagnosis in those who presented at a later age.
Methods: A cross-sectional study design was utilized to assess diagnostic delays in children with structural birth defects. The study included all pediatric patients evaluated for structural birth defects at three public hospitals in Addis Ababa during the designated study period. Data were collected through structured interviews with parents or legal guardians, supplemented by medical chart reviews. Data entry and analysis were performed using the Statistical Package for Social Sciences (SPSS), version 26. A multivariate logistic regression was conducted to identify associations between delayed diagnosis and selected independent variables.
Results: A total of 423 children with structural birth defects were enrolled in the study, of whom 358 (84.6%) were male. The mean age of the participants was 3.4 ± 3.5 years. The three most frequently observed structural birth defects were hypospadias (133 cases, 31.4%), undescended testes (95 cases, 22.4%), and anorectal malformations (38 cases, 8.9%). Despite 97.2% of mothers (441) having attended antenatal care, antenatal diagnosis was achieved in only 32 children (7.6%). The health facility delivery rate was high (94.6%), yet among infants born with visible or physically detectable birth defects, only 25% were diagnosed prior to discharge. Overall, delayed diagnosis of structural birth defects was observed in 274 children (64.8%). The most commonly reported reasons for diagnostic delay included lack of parental or provider awareness, delayed referral, and reassurance by health professionals. Multivariable logistic regression analysis identified several factors significantly associated with delayed diagnosis: male sex ($P = 0.035$; AOR = 1.81, 95% CI: 1.043–3.139), parental education below college level ($P = 0.049$; AOR = 1.52, 95% CI: 1.002–2.311), and delivery at a health center rather than a hospital ($P < 0.001$; AOR = 2.17, 95% CI: 1.405–3.325).
Discussion and Conclusion: This study examined the timing and factors contributing to delayed diagnosis of structural birth defects in 423 children. Despite high antenatal follow-up and health facility deliveries, prenatal detection was critically low, and three-quarters of overt anomalies were missed at birth. Male predominance was consistent with global trends, and genitourinary anomalies were most common, similar to findings from Brazil. Diagnosis delays mirrored patterns seen across sub-Saharan Africa, driven by socioeconomic barriers, limited access to specialized care, and a shortage of trained professionals. Notably, delayed referral and false reassurance from healthcare providers were more prominent causes of missed diagnoses than financial or access-related issues. This highlights the urgent need for capacity-building among health workers to improve early recognition and management of birth defects. Compared to global data, antenatal detection rates in this study were significantly lower, underscoring the importance of integrating prenatal ultrasound screening into routine care. In conclusion, there is a significant delay in diagnosing structural birth defects during both the antenatal and postnatal periods. These missed opportunities can be addressed through comprehensive screening programs, capacity-building sessions for healthcare workers, and raising public awareness on structural birth defects. Crucially, implementation of these strategies should prioritize empowering primary health care units, particularly health centers, to facilitate the early diagnosis of structural birth defects.

Preliminary declaration for publication on ICBDSR annual meeting 2025

Helen Malherbe & Emma Kalk

Posters

(Alphabetical order by corresponding author's last name)

The posters will be shown during all breaks in the Conference Foyer

Impact Evaluation of the Global Birth Defects Description and Coding Mobile Application - *Utibe Ebong*

Major Congenital Anomalies: 23 Years of Prevalence and Postnatal Outcomes in Very-Low-Birth-Weight Infants in the NEOCOSUR Neonatal Network - Ivonne D'Apremont

Is knowledge enough? Investigating the role of parity and the missed opportunities for folic acid supplementation among women in Northern Ireland - *Utibe Ebong*

Rare Diseases in an Indigenous Brazilian Population in Southern Brazil: Insights from a Field Surveillance Initiative – *Claudia Fernandes Lorea*

Outcome of prenatally diagnosed Neural Tube Defects – implications for counselling - *Karen Fieggen*

Clinical Patterns and Outcomes of Prenatally Diagnosed Hydronephrosis in Children: A Four-Year Review at a Tertiary Hospital in Addis Ababa, Ethiopia - *Ephrem Kerego*

Early Findings on an Artificial Intelligence Cardiac View Classification Model for Neonatal Congenital Heart Disease Screening in Sub-Saharan Africa - *Aminkeng Leke*

New Public Health Strategies and Infant Mortality Reduction in Chile - *Cecilia Mellado*

Congenital diaphragmatic hernia and its genetic etiologies: 11 years of experience in a high-complexity center - *Allisson Müller Delgado*

Congenital anomaly mortality in India : Analysis of data from two national mortality surveillance datasets - *Anita Pal*

The Alarming Increase in Birth Defects at the Expense of Congenital Heart Diseases: The Maternal Risk Paradox Phenomenon - *Karen Sarmiento*

Mapping isolates with a high frequency of congenital anomalies and rare diseases: CENISO - Brazil - *Lavinia Schuler-Faccini*

MRI assisted Neurosonography (MAN) two case series that demonstrate efficiency of Fusion Multimodality Technology in Fetal neuroradiology diagnostics. MALFOVAR (Malformation and variable anatomy registry) update 2025 - *Tidhar Steiner*

Major Congenital Anomalies: 23 Years of Prevalence and Postnatal Outcomes in Very-Low-Birth-Weight Infants in the NEOCOSUR Neonatal Network

Dr Ivonne D'Apremont, Dr Alberto Toso, Statistics Claudia Musalem, Dr. Paulina Tosos, Dr. Maria Eugenia Hubner, Dr. Cecilia Mellado
[1]Pontificia Universidad Católica de Chile, SANTIAGO, Chile

Email of corresponding Author: ebong-u@ulster.ac.uk

Background and Objectives
Major congenital anomalies (MCAs) are an important yet often underestimated cause of infant mortality, morbidity, and disability. The WHO estimates that approximately 8 million newborns worldwide are born with a congenital anomaly each year; however, regional registries remain scarce.
The aim of this study was to describe the prevalence, characteristics, and postnatal outcomes of very-low-birth-weight (VLBW) newborns diagnosed with MCAs within the NEOCOSUR Neonatal Network between 2001 and 2023.
Methods
We conducted a retrospective cohort study based on prospectively collected data from infants born between 23 and 36 weeks' gestation with birth weights of 400–1500 g. Data were obtained from 40 South American Neonatal Intensive Care Units (NICUs) participating in the NEOCOSUR Network (Argentina, Chile, Paraguay, Peru, and Uruguay) from 2001 to 2023. MCA prevalence was analyzed according to gestational age, birth weight, and affected organ system. Overall mortality, place of death (delivery room vs. post-admission), and postnatal age at death were described. Kaplan–Meier methods were used to analyze survival.
Results
During the study period, 33,274 VLBW newborns were registered in NEOCOSUR centers. Of these, 1,810 (5.5%) were diagnosed with MCAs. The proportion of infants with MCAs increased with higher gestational age, while no significant differences were observed by birth weight. Among MCA subtypes (data available since 2012), congenital heart defects were the most frequent (20%), followed by central nervous system defects (18%), chromosomal abnormalities (17%), and digestive system defects (14%). Overall mortality was 61.4% (n = 1,111); 35% of deaths occurred in the delivery room and 65% after NICU admission. The median age at death was 9 days. Survivors had prolonged hospitalizations, with a median length of stay of 66 days.
Discussion and Conclusion
Although the prevalence of MCAs in the NEOCOSUR Network is relatively low (5.5%), mortality among affected VLBW infants is high, with 35% of deaths occurring in the delivery room and 65% after admission. Only 38.6% of cases were discharged home. Consistent with previous reports, more than half of the MCAs were congenital heart defects, central nervous system malformations, or chromosomal anomalies. The NEOCOSUR Network will continue to strengthen registries on primary causes of death and the appropriateness of therapeutic interventions, aiming to highlight the burden of MCAs in VLBW newborns and to guide strategies to improve care in the Region.

Impact Evaluation of the Global Birth Defects Description and Coding Mobile Application

Professor Helen Dolk[1], **Mr Utibe Ebong**[1]
[1]School of Medicine, Ulster University, Belfast, United Kingdom

Email of corresponding Author: ebong-u@ulster.ac.uk

Background & Objectives: As part of the ZikaPLAN project, a Zika research response consortium funded by the EUHorizon from 2016 to 2020, the Global Birth Defects Description and Coding (GBDDC) mobile app was developed. This mobile application is available on the Apple App Store and Google Play Store and has been translated into French, Spanish, and Portuguese. Until recently, user support was funded by the Ubomi Buhle project. The app is available in a basic and surveillance version. The latter collects data, including photos, which are uploaded to a server from which they can be downloaded to the project database. Specific projects have implemented modifications, such as an online review platform funded by the MiMBA project, which allows data uploaded from the app to be viewed and commented on by birth defect experts before being downloaded into the project database. This impact evaluation project will document its current use globally and highlight specific current and past use cases.
Method: The evaluation consisted of registration statistics, a survey sent to registrants from 2022 to 2025, online interviews, feedback questionnaires and meetings. As the app is mainly used offline, pop-up evaluations are difficult. A validation study is in progress in three countries, comparing data recorded using the app with expert diagnoses.
Results: To date, there are 3,642 registrants of the app, with 85.6% using the basic version and 14.4% using the surveillance version. Registrants were mostly nurses (41%), doctors (20%), Researchers (6%) and Paediatricians (6%). Registrants were spread across 77 countries, with the majority of registrations coming from Brazil (n = 2,362). Data on registrations also showed that a majority (41%) of the registrations were in 2024. The survey was sent to 2,182 users who registered in the last three years. The response rate was low (2%), but respondents typically used the app for research (55%), clinical work (34%), public health surveillance (29%), and teaching health professionals (24%). Eighty-six per cent found the app useful. The surveillance version has been used by four projects investigating risk factors for birth defects, including antiretrovirals, antimalarials, Zika, and Oropouche. Other research and surveillance projects are commencing their use, starting with the exploration of the sandbox surveillance version. Validation data to date suggest high correspondence with expert diagnosis, but with a tendency to include details of multiple anomalies in the text rather than in code. Current barriers to use include projects that require an app to collect information on all births, rather than only babies with birth defects, despite the app being designed as a complementary tool.
Discussion & Conclusion: Recently, the World Health Organisation published its Digital Adaptation Kit (DAK) for birth defects surveillance, highlighting the GBDDC app's role in instigating this initiative. The app illustrates the potential of mHealth interventions in low-resource areas of low and middle-income countries. However, it also highlights problems in sustainability, such as funding for version updates and user support, which are needed. Opportunities for AI assistance are being explored due to the rapid developments in this area.

Is knowledge enough? Investigating the role of parity and the missed opportunities for folic acid supplementation among women in Northern Ireland.

Mr Utibe Ebong[1], Dr Joanne Given[2], Professor Frank Casey[1], Dr Maria Loane[2], Professor Helen Dolk[1]
[1]School of Medicine, Ulster University, Belfast, United Kingdom, [2]Institute of Nursing and Health Research, Ulster University, Belfast, United Kingdom

Email of corresponding Author: ebong-u@ulster.ac.uk

Background & Objectives:
Early initiation of folic acid is vital to prevent NTDs. In the United Kingdom (UK), women are advised to take 400 micrograms of folic acid starting three months before conception, or 5 milligrams for high-risk women. Healthcare professionals play a crucial role in educating women about the importance of folic acid. Despite awareness of the importance of folic acid, its intake remains low across populations. Studies have identified unplanned pregnancies as one of the key reasons for low folic acid intake. Unplanned pregnancies account for approximately 40% globally and between 34% and 45% of pregnancies in the UK. This study examined the relationship between parity, pregnancy planning and folic acid intake. It also investigated the role of pregnancy-related events, such as gestational week when women suspected pregnancy, confirmed pregnancy, and contacted health services, and their influence on supplement use.
Method: This study was a secondary analysis of the Northern Ireland (NI) Baby Hearts study. The NI Baby Hearts study sampled 286 women with children who had congenital heart disease as cases and 966 controls recruited during clinical screening at approximately 20 gestational weeks from 2014 to 2016. The sample was representative of the women in NI during the study period. The NI Baby Hearts study linked data collected via a questionnaire with maternal health records from the NI Maternity System, prescription data from the Enhanced Prescribing Database, and area-based deprivation data from the NI Statistics and Research Agency. This study used data from the control group. Logistic regression analyses were computed to identify determinants of periconceptional folic acid intake. A directed acyclic graph (DAG) was used to visualise causal pathways to identify possible confounding variables. Literature review identified covariates and their relationships with each other for the DAG. The DAG identified eight confounding factors to be adjusted for in the analysis: (i) Age, (ii) Deprivation, (iii) Education, (iv) Marital status, (v) Occupation, (vi) Pregnancy planning, (vii) Previous loss/Abortion, and (viii) Race/Ethnicity/Nationality. However, ethnicity/race/nationality could not be adjusted for due to missing information about this covariate in the NI Baby Hearts database. Odds ratios (ORs) were reported with 95% confidence intervals. Analyses were conducted using Stata version 18.
Results: Of the 966 women, 40.8% initiated folic acid periconceptionally, 47.2% initiated intake postconception, and 12.0% did not take folic acid. The likelihood of folic acid intake within the periconceptional period was lower among women who did not plan for the pregnancy compared to those who planned (aOR 0.08 (95% CI 0.05 – 0.12)). Parity was another significant predictor of folic acid intake as multiparous women were less likely than primiparous women to initiate intake during the periconceptional period (aOR 0.48 (95% CI 0.33 – 0.69)). Women who lived in the most deprived areas were less likely than those living in the least deprived areas to initiate intake during the periconceptional period (aOR 0.48 (95% CI 0.27 – 0.85)). Similarly, women who had poor dietary habits were less likely than those with a healthy dietary habit to take folic acid periconceptionally (aOR 0.44 (95% CI 0.28 – 0.69)). On the other hand, having two or more previous pregnancy losses significantly increased the likelihood of initiating folic acid periconceptionally (aOR 2.04 (95% CI 1.15 – 3.62)). Pregnancy-related events had a remarkable influence on folic acid intake. The disparity in folic acid intake between primiparous and multiparous women with a planned pregnancy diminished after the median gestational week when women suspected they were pregnant (4 weeks). By the fifth gestational week when women confirmed they were pregnant, intake among primiparous and multiparous women with an unplanned pregnancy increased substantially. And following women's contact with their General Practitioner (GP) at the seventh gestational week, the disparity in intake

between women with a planned and unplanned pregnancy diminished steadily until there was no disparity between these groups of women.

Discussion & Conclusion: In the UK, including NI, there is no mandatory food fortification, so prevention of NTD depends on FAS. These findings highlight important disparities in folic acid supplementation among pregnant women in NI. Considering that folic acid intake sharply increased after suspecting pregnancy, our findings suggest that motivation rather than knowledge is the major driver of supplementation behaviour. Behaviour change theories such as the Capability-Opportunity-Motivation (COM-B) Behaviour framework and the Teachable Moment theory corroborate this perspective. Our findings show that health service contact is useful in bridging the disparity in intake between women with planned and unplanned pregnancies. However, this is too late an intervention as folic acid intake should be initiated before pregnancy. It is therefore important that GPs and midwives leverage preconception care to reinforce women's motivation for a positive pregnancy outcome and create opportunities for adequate supplementation, in addition to providing knowledge on the importance of folic acid.

Rare Diseases in an Indigenous Brazilian Population in Southern Brazil: Insights from a Field Surveillance Initiative

Miss Claudia Fernandes Lorea[1], Paulyana Corecco-Moura[1], Laércio Moreira Cardoso-Júnior[1], Emilly de Jesus Garcia Ataíde[1,3], Luciane Flach[4], Regina Reggiori[4], Dra Simone de Menezes Karam[2], Dra Maria Teresa Vieira Sanseverino[3], Dra Lavínia Schuler-Faccini[1]

[1]Universidade Federal do Rio Grande do Sul - UFRGS, Porto Alegre, Brazil, [2]Fundação Universidade de Rio Grande (FURG), Rio Grande, Brazil, [3]Hospital de Clínicas de Porto Alegre, Porto Alegre, Brazil, [4]Hospital Santo Antônio, Tenente Portela, Brazil

Email of corresponding Author: claudialorea@hotmail.com

Background & Objectives: Indigenous populations in Brazil, including the Kaingang community in Southern Brazil, face significant barriers to accessing specialized medical care due to long road distances from referral centers and logistical challenges limiting access to consultations. These barriers contribute to underdiagnosis of congenital anomalies and genetic disorders. To address these gaps, Brazil's project for congenital anomalies surveillance incorporated a targeted field initiative aimed at improving diagnostic coverage in this population. This study reports the results of direct field assessments by medical geneticists conducted within the Kaingang territory, with the objective of reducing the diagnostic odyssey regarding rare diseases. This field initiative was motivated by two current projects that are assessing the local high prevalence of albinism and galactosemia in this region, the aim is to establish accurate epidemiological data while providing equitable diagnostic care.

Method: Medical geneticists conducted clinical evaluations during a field visit to Tenente Portela, approximately 470 kilometers by road from Porto Alegre, the state's primary referral center. Patients from the Kaingang community were referred by local health professionals and evaluated following ethical standards for indigenous populations. Detailed phenotypic assessments were combined with molecular testing using multigene panel and exome sequencing to achieve definitive diagnoses. In cases of pathogenic findings, parental testing was recommended to confirm segregation patterns, especially in newly identified or complex diagnoses.

Results: Five confirmed genetic diagnoses were established among the individuals assessed. A mother and daughter were diagnosed with hypochondroplasia, each carrying a heterozygous pathogenic variant in the FGFR3 gene. A 9-year-old girl presented with cerebellar ataxia and recurrent infections, leading to a diagnosis of ataxia-telangiectasia following the identification of biallelic pathogenic variants in the ATM gene. In a newborn who died within one hour after birth due to nonimmune hydrops fetalis, molecular analysis identified two pathogenic variants: a frameshift mutation in PIEZO1 (p.Arg1868Hisfs*), consistent with an autosomal recessive congenital lymphatic malformation, and a stop-gain variant in COL11A2 (p.Arg1379*), associated with autosomal dominant connective tissue disorders. Both parents of the affected newborn were asymptomatic, and segregation testing is currently underway to confirm inheritance patterns. Additionally, unrelated children from the same territory, a boy and a girl, currently under care in the reference center for rare diseases were diagnosed with an inborn error of immunity, with molecular testing revealing the same biallelic pathogenic variants in DOCK8. Both children exhibited recurrent respiratory infections and cutaneous manifestations. All families received online follow-up consults, including genetic counselling by medical geneticists. Support for the local healthcare team was also provided. These findings demonstrate the presence of rare genetic conditions involving both autosomal dominant and recessive inheritance patterns within the Kaingang population.

Discussion & Conclusion:

The results of this field surveillance initiative underscore the necessity of expanding access to genetic diagnostic services for remote and indigenous populations in Brazil. The detection of both established and complex dual genetic diagnoses, such as the co-occurrence of PIEZO1-related lymphatic malformation and COL11A2-associated connective tissue disorder in a single newborn, highlights the clinical utility of

comprehensive molecular testing. Furthermore, the identification of inborn errors of immunity, as well as skeletal dysplasia and DNA repair disorders, reflects a broad spectrum of genetic diseases affecting this community. The known high prevalence of albinism and galactosemia among the Kaingang and related populations, which is currently being systematically evaluated, further reinforces the importance of targeted genetic screening strategies to quantify local disease burden and guide preventive interventions. Geographical isolation, long travel distances, and limited availability of specialized services exacerbate health disparities faced by indigenous populations, necessitating field-based approaches such as this initiative. Integration of indigenous communities into national congenital anomalies surveillance systems is essential to improve diagnostic accuracy, inform public health policy, and promote health equity. Integration of online follow-up consultations further demonstrated the potential of distance-based approaches to support ongoing care, improve diagnostic follow-up, and provide sustained genetic counseling in indigenous and other geographically isolated communities.

Outcome of prenatally diagnosed Neural Tube Defects – implications for counselling.

Ass Prof Karen Fieggen[1], Dr Alison Boutall[2], Dr Ayesha Osman[2], Ms Vhuthu Mamapa[3], Ms Mignon Raath[3]
[1]Department of Medicine, University Of Cape Town, Cape Town, South Africa, [2]Department of Obstetrics and Gynaecology, University of Cape Town, Cape Town, South Africa, [3]University of Cape Town, Cape Town, South Africa

Email of corresponding Author: karen.fieggen@uct.ac.za

Background and objective: Neural tube defects (NTD's) are one of the most common structural congenital abnormalities worldwide. Prenatal diagnosis by high resolution fetal ultrasound has improved the early detection of NTDs and allows for pregnant women to make choices regarding management of their pregnancies within the context of the law. Accurate, evidence-based counselling is central to informed choice. The aim of this retrospective record review is to document the ultrasound findings, postnatal findings and both short- and medium-term outcomes in fetal NTDs diagnosed at the Groote Schuur Hospital (GSH) fetal medicine unit.
Method: The review includes fetal diagnoses of NTD between January 2015 and December 2019 with a minimum follow up time of 5 years in survivors where the findings were made or confirmed at the GSH fetal medicine unit. Cases are ascertained form the unit's database which is considered a complete record. Where pregnancies continued, delivery and neonatal records were retrospectively reviewed as were the hospital records that were available for children referred for surgery and follow up.
Results: 107 cases of NTD diagnosed on ultrasound at GSH within the review period have been identified. Information on serial ultrasound findings in the fetus, maternal teratogen exposure, decision regarding further management of the pregnancy as well as postnatal outcome including surgical procedures are being collected which along with analysis is still underway.
Discussion and conclusion: With no access to fetal surgery but good neonatal and neurosurgical care as well as multidisciplinary follow up, it is envisaged that this real-world experience within the South African health care context will inform accurate counselling of women with affected pregnancies and have implications for service delivery both locally and in other middle-income countries.

Clinical Patterns and Outcomes of Prenatally Diagnosed Hydronephrosis in Children: A Four-Year Review at a Tertiary Hospital in Addis Ababa, Ethiopia

Dr Ephrem Kerego[1], Dr Hana Gebreselassie[1], Dr Hiwote Assefa[1]
[1]Saint Paul's Hospital Millennium Medical College, Addis Ababa, Ethiopia

Email of corresponding Author: ephremnk2014@gmail.com

Background and Objective: Antenatal hydronephrosis (ANH) refers to the dilatation of the fetal urinary tract identified during prenatal imaging. The detection of genitourinary anomalies in utero has increased significantly with the widespread use of routine anatomical ultrasound examinations during pregnancy. Among the diagnostic parameters, the anteroposterior diameter of the renal pelvis is one of the most extensively studied. A commonly accepted threshold of ≥7 mm in the third trimester is used to identify fetuses who warrant postnatal evaluation. The primary clinical challenge lies in differentiating children at risk of progressive renal impairment from those with transient or non-significant dilatation. Early identification is critical, as some patients may require further diagnostic testing and timely medical or surgical intervention before the onset of symptoms or irreversible renal damage. While many cases can be managed conservatively with observation, others necessitate active treatment to prevent long-term complications. The study objective is to evaluate the clinical management practices and follow-up outcomes of children with prenatally diagnosed hydronephrosis in low- and middle-income countries, with a focus on identifying gaps in care and opportunities for improving early intervention strategies.
Method: This was a four-year retrospective study conducted at a tertiary care hospital, utilizing a cross-sectional design. The study population included all patients with a documented diagnosis of antenatal hydronephrosis during the study period. Ethical approval was obtained from the institutional review board prior to data collection. A structured questionnaire was developed to capture relevant clinical and demographic variables. Data were extracted from patient records, systematically compiled, and analyzed to interpret patterns in diagnosis, management, and outcomes.
Results: A total of 52 patients with ANH were included in the study, with 90.4% diagnosed during the third trimester. Postnatally, the majority (71.2%) presented within the first week of life. The male-to-female ratio was 2.71:1. The left kidney was affected in 43.1% of cases, while bilateral involvement was observed in 41.2%. Prenatal ureteral dilatation was identified in only 6.3% of patients. The most common underlying cause of ANH was pelviureteric junction obstruction (28.8%). Urinary tract infections (UTIs) were documented in 25% of patients, of whom 28.8% received prophylactic antibiotics. Associated anomalies were present in 11.5% of cases, with undescended testes being the most frequently reported. Spontaneous resolution occurred in 34.6% of patients, whereas 42.3% required surgical intervention. At one-year follow-up, 51.9% of patients demonstrated clinical improvement, while 9.6% showed deterioration in renal outcomes
Discussion and Conclusion: This study highlights the clinical course of 52 ANH patients, showing male predominance and third trimester diagnosis, similar to findings from Saudi Arabia and Iran. Left-sided and unilateral involvement were common, with undescended testes as the leading associated anomaly. PUJO was the most frequent cause, while VUR was rare compared to European data. One-third had spontaneous resolution, mostly unilateral. UTIs were more frequent than in other studies, and prophylactic antibiotics were primarily used for VUR. Surgical intervention was needed in 42.3%, a higher rate than reported elsewhere. Outcome comparisons were limited due to inconsistent grading and APD documentation across patients. The significant spontaneous and surgical improvements observed in this study underscore the importance of antenatal diagnosis and consistent follow-up.

Early Findings on an Artificial Intelligence Cardiac View Classification Model for Neonatal Congenital Heart Disease Screening in Sub-Saharan Africa

Dr Aminkeng Leke[1], Dr. Lionel Landry Sop Deffo[1], Mr. Sabastian Wirisiy Yunkavi[1], Dr. Thomas Aldersley[2], Dr. Thomas Day[3,4], Dr. Andrew P. King[3], Dr. Patrick McAllister[5], Dr. Michel N Maboh[1], Prof. John Lawrenson[2,7], Dr. Cabral Tantchou[1,8], Dr. Bernhard Kainz[9], Dr. Frank Casey[10], Dr. Raymond Bond[5], Dr. Finlay Dewar[5], Mr. Kelson Tchinda Ngoe[1], Mr. Ndeh Mugri Frunwi[1], Dr. Liesl Zühlke[2,6], Prof. Helen Dolk[10]
[1]Digital Technology and Innovation Hub, Health Research Foundation Buea, Buea, Cameroon, [2]Department of Paediatrics and Child Health, Division of Paediatric Cardiology, University of Cape Town, Cape Town, South Africa, [3]School of Biomedical Engineering and Imaging Sciences, King's College London, London, United Kingdom, [4]Department of Congenital Cardiology, Evelina London Children's Healthcare, Guy's and St Thomas' NHS Foundation Trust, London, UK, [5]School of Engineering, Belfast Campus, Ulster University, UK, Belfast, [6]South African Medical Research Council, ZA , South Africa, [7]Department of Paediatrics, Division of Paediatric Cardiology, Stellenbosch University, Stellenbosch, South Africa, [8]Shisong cardiac centre / St Padre Pio hospital, Akwa Nord. Douala, Cameroon, Douala, Cameroon, [9]Department of Computing, South Kensington Campus, Imperial College London, London, United Kingdom, [10]School of Medicine, Ulster University, United Kingdom

Email of corresponding author: leke@hrfbuea.org

Background & Objectives: Congenital heart disease (CHD) is the most common serious congenital anomaly and a major contributor to preventable neonatal mortality. Each year, it accounts for an estimated 181,000 infant deaths worldwide, with 25% of these deaths occurring in Sub-Saharan Africa (SSA). In high-income settings, advances in pulse oximetry, clinical examination, and confirmatory echocardiography have significantly reduced CHD-related mortality by enabling earlier detection and timely intervention. In contrast, outcomes in SSA remain poor due to systemic barriers that limit screening and diagnosis. These include a severe shortage of paediatric cardiologists, concerns regarding the reduced accuracy of pulse oximetry in dark-skinned neonates, and the geographic centralization of the few paediatric cardiac centres to major cities, leaving peripheral maternity units underserved. Consequently, families are often required to travel hundreds of kilometers for confirmatory scans, many of which ultimately rule out CHD, imposing avoidable costs, delays, and burdens on healthcare systems. These persistent inequities highlight the urgent need for scalable, affordable, and context-appropriate solutions. Advances in portable ultrasound devices, combined with telemedicine and artificial intelligence (AI), provide a unique opportunity to reframe neonatal CHD screening in resource-limited settings. Funded by the US NIH, under the Data Science Innovation for Africa (DSI-Africa) initiative, this study aims to develop an AI-assisted echocardiography model that can be incorporated into mobile ultrasound systems to enable non-experts such as nurses, midwifes, sonographers and medical doctors to perform basic cardiac ultrasound sweeps on neonates suspected of CHD and extract accurate cardiac images that can be sent to a remote paediatric cardiologist for interpretation.
Methods: Based on the 2024 American Society of Echocardiography (ASE) guidelines, our expert paediatric cardiologist panel identified eight key cardiac views for screening: subcostal long axis (SCLA), apical four chamber (A4C), apical five chamber (A5C), parasternal long axis (PLA), parasternal short axis at the aortic valve (PSA-AV), mitral valve (PSA-MV), papillary muscle (PSA-PM), and suprasternal long axis (SSLA). Data collection is ongoing at St Padre Pio Hospital (Cameroon) and the Red Cross War Memorial Children's Hospital (South Africa), enrolling infants aged 0–2 years with and without CHD. Two complementary datasets are being built: a retrospective set of short clips from ~500 routine echocardiograms and a prospective set of ~1,000 full studies designed to capture real-world variability and background noise typical of non-expert acquisition. All videos are processed through a custom anonymization pipeline (automated de-identification, pseudonymization, metadata preservation) before being uploaded to Labelbox for structured annotation by cardiologists, including both target and non-target/background views. Model development is conducted in PyTorch using EfficientNetV2 backbones pre-trained on ImageNet. A two-phase fine-tuning approach is

employed: (1) training classification heads with frozen backbones, and (2) full-network fine-tuning at reduced learning rates. To enhance robustness, stratified 3-fold cross-validation and domain-specific augmentations (rotations, scaling, shear, translations simulating probe motion) are applied. Two models are under development: Model A for cardiac view classification and Model B for image quality assessment. Performance is evaluated using accuracy, sensitivity (recall), precision, and F1-score, reported per cardiac view and at aggregate macro-averages.

Results: This interim analysis focuses on Model A (cardiac view classification) trained on a retrospective dataset. A total of 6177 anonymized echocardiography clips from 309 infants were included, with class distribution showing marked imbalance: SCLA (13.5%), A4C (12.4%), A5C (6.2%), PLA (6.8%), PSA-AV (16.6%), PSA-MV (2.2%), PSA-PM (1.1%), SSLA (9.5%), "other cardiac views" (28.7%), and non-cardiac/background frames (3.0%). Using stratified 3-fold cross-validation, the model achieved an overall accuracy of 63.0%, with 62.3% precision, 63.0% sensitivity, and an F1-score of 62.5% across all classes. Performance varied substantially between views: sensitivity values ranged from 4.2% (PSA-PM) to 74.3% (PLA), precision values from 22.22% (PSA-PM) to 76.6% (SSLA), and F1-scores between 14.02% (PSA-PM) and 73.2% (A4C).

Discussion & Conclusion: These preliminary findings demonstrate proof-of-concept feasibility for an AI-assisted echocardiography model to support neonatal CHD screening in SSA, while also exposing key limitations that must be addressed before clinical deployment. Performance outcomes were highest for well-represented views such as PLA, A4C, and SSLA, whereas sensitivity for sparsely represented views like PSA-PM and PSA-MV was markedly lower, underscoring the dependence of model performance on balanced training data. The limited inclusion of non-cardiac backgrounds (3% of clips) further constrained evaluation under realistic noisy conditions, highlighting the need for prospective datasets that better reflect operational variability when used by non-expert operators. Importantly, our reliance on video analysis instead of still images represents an innovative strength: although methodologically more complex, this design captures temporal dynamics and probe-motion artifacts inherent in real-world scanning, thereby improving ecological validity compared with still frame–based studies. Future model refinement will incorporate the larger prospective dataset (~1,000 infants) to improve balance, variability, and generalizability, and may adopt temporal deep learning architectures (such as ConvLSTMs or Transformer-based methods) to better handle sequential video data. By embedding the AI tool into portable ultrasound devices, the project ultimately aims to empower frontline health workers—including nurses and midwives—to acquire diagnostic-quality sweeps, automate cardiac view identification, and transmit interpretable scans to paediatric cardiologists for remote confirmation. In conclusion, once fully trained and validated, this system has the potential to bridge critical gaps in CHD detection in SSA, improving access, reducing avoidable mortality, and strengthening neonatal cardiac care in resource-limited regions.

New Public Health Strategies and Infant Mortality Reduction in Chile

Dr Cecilia Mellado[1,2], Dr. Rosa Pardo[2,3], Paulina Leighton[2], Dr. Jorge Pacheco[2]
[1]Genetics Section and Inborn Errors of Metabolism, Pediatrics Division, School of Medicine, Pontificia Universidad Catolica de Chile, Santiago, Chile, [2]Department of Health Statistics (DEIS), Ministry of Health of Chile, Santiago, Chile, [3]Genetics Section and Neonatology Unit, Hospital Clinico Universidad de Chile, Santiago, Chile

Email of corresponding author: cmellads@uc.cl

Background & Objectives: Chile has achieved significant improvements in child health over the past three to four decades, associated with various public health strategies aimed at reducing preventable morbidity and mortality in early life. As of 2022, the infant mortality rate was 5.86 per 1,000 live births, although acute respiratory infections are no longer the leading cause of infant death, lower respiratory tract infections (LRTIs), particularly from respiratory syncytial virus (RSV), remain a relevant cause of preventable morbidity and mortality in infants.
In April 2024, the Ministry of Health implemented universal RSV immunization with nirsevimab, a monoclonal antibody administered to all neonates and infants prior to the beginning of the RSV season in Chile. The aim of this study was to assess the impact of this strategy on LRTI-related mortality in infants under one year of age.
Methods: We reviewed national data from the Department of Health Statistics (DEIS), Ministry of Health of Chile, on infant hospitalizations and deaths, comparing 2023 (pre-implementation) with 2024 and early 2025.
Results: Following the implementation of the nirsevimab, all-cause hospitalizations in infants decreased by 48%, RSV-related hospitalizations dropped by 76%, and RSV-related Intensive Care Unit (ICU) admission by 85% in 2024. Among immunized infants, there was a significant reduction in the duration of hospital stays and ICU bed-days. Regarding mortality due to LRTIs (ICD-10 codes J09-J18 and J20-J22), it declined by 69% between 2023 and 2024 (16 vs 5), and no LRTI-related deaths have been reported during the current RSV season.
Conclusion: Although not the leading cause of infant mortality, LRTIs remain a relevant public health challenge. The introduction of nirsevimab has shown a significant impact on RSV-related outcomes in Chile, supporting its potential implementation in other countries in the region.

Congenital diaphragmatic hernia and its genetic etiologies: 11 years of experience in a high-complexity center

Dr. Allisson Müller Delgado[1], Dr. Guillermo Lay-Son[1], Dr. Cecilia Mellado[1], Dr. Mariana Aracena[1], Dr. Matias Luco[1]
[1]Sección de Genética y Errores Innatos del Metabolismo, División de Pediatría, Escuela de Medicina, Pontificia Universidad Católica De Chile, Diagonal Paraguay 362, Chile

Email of corresponding Author: amullerd@gmail.com

Background & Objectives: Congenital diaphragmatic hernia (CDH) has an estimated prevalence of approximately 1 in 3,000 live births and represents a major cause of neonatal morbidity and mortality. While most cases are isolated, a significant proportion are associated with additional congenital anomalies or genetic syndromes, which are linked with worse outcomes. The prevalence of syndromic CDH is estimated at 3–4%, although this is likely underdiagnosed. Early recognition of a genetic etiology can influence clinical decision-making and family counseling. The aim of this study was to characterize newborns with CDH and a suspected or confirmed genetic syndrome and compare them to those presenting isolated CDH, focusing on clinical features, in-hospital outcomes, and the diagnostic yield of genetic testing.
Method: We conducted a retrospective cohort study at a high-complexity neonatal center in Chile, including liveborn neonates with CDH from 2013 to 2024. Patients were included if they were born at or referred to our center during the neonatal period. CDH cases were categorized as isolated, non-isolated (associated anomalies, without a suspected genetic syndrome), or syndromic (with a suspected or confirmed genetic diagnosis). We collected data on demographics, birth defect characteristics, timing of diagnosis, interventions (including Extra Corporeal Membrane Oxigenation-ECMO), in-hospital outcomes, and results from genetic testing (karyotype, chromosomal microarray, whole-exome sequencing). Comparative analyses between the syndromic and isolated groups were performed using odds ratios with 95% confidence intervals and significance testing ($p<0.05$).
Results: Among 366 neonates with CDH, 193 (52.7%) had isolated CDH and 78 (21.3%) were classified as syndromic; 28 of these had a confirmed genetic diagnosis. The most frequent etiologies were Down syndrome (n=4), Turner syndrome (n=2), mosaic trisomy 9 (n=2), Emanuel syndrome (n=2), and Wolf-Hirschhorn syndrome (n=2). Syndromic CDH was associated with significantly higher rates of prematurity (OR 4.37, $p<0.001$), right-sided defects (OR 2.21, $p=0.020$), and earlier prenatal diagnosis (mean 24.4 weeks, $p=0.037$). Although lower observed-to-expected lung-to-head ratios (LHR O/E) were noted in syndromic cases, the difference was not statitically significant ($p=0.077$). Use of ECMO was less frequent among syndromic cases (OR 0.52, $p=0.041$), while rates of surgical repair and postoperative survival did not differ significantly between groups. Hospital survival was significantly lower in syndromic CDH cases (OR 0.43, $p=0.006$), and length of hospital stay was longer ($p<0.001$). Congenital heart defects were the most common associated anomalies. Genetic testing showed variable diagnostic yields: 5.0% for karyotype, 45.5% for chromosomal microarray, and 66.7% for whole-exome sequencing.
Discussion & Conclusion: This study highlights important clinical differences between syndromic and isolated CDH. Syndromic cases showed higher rates of prematurity, right-sided defects, and mortality, underscoring their greater clinical complexity and poorer prognosis. Although surgical and postoperative outcomes were similar, syndromic patients had longer hospitalizations and were less likely to receive ECMO, potentially reflecting clinical decision-making influenced by comorbidities or prognosis. The high diagnostic yield of chromosomal microarray and whole-exome sequencing supports their use in CDH cases with associated anomalies or suspected syndromic features. However, only 36% of syndromic cases received a confirmed genetic diagnosis during hospitalization, suggesting that current diagnostic approaches may be insufficient or delayed. Routine genetic evaluation in newborns with CDH, particularly when additional anomalies are present, is essential to guide prognosis, optimize care strategies, and provide accurate counseling to families.

Incorporating comprehensive genetic testing earlier in the clinical workflow may improve diagnostic rates and inform individualized management plans.

Congenital anomaly mortality in India: Analysis of data from two national mortality surveillance datasets

Dr Anita Pal[1], Ms Samruddhi Gopal, Ms Dhanshri Kalokhe, Ms Prerna Narendra
[1]Birth Defects Research Foundation, Pune, India, [2]School of Health Sciences, Savitribai Phule Pune University, Pune, India

Email of corresponding Author: dranitakar@gmail.com

Background: Data on causes of mortality in India is available through two surveillance systems. Around 20% of deaths are medically certified, mostly at urban hospitals. The data is made available through the Medical Certification of Causes of Death. The Sample Registration System collects data from randomly sampled units located across urban and rural areas in India. Trained data collectors document mortality data as a narrative written in the local language. The assignment of cause of death is done independently by two trained physicians. Both systems use ICD-10 classification.
Objectives: Mortality caused by congenital anomalies is not reported through routine health information systems in India. The objective of this study was to describe congenital anomaly mortality in India using secondary data from the Medical Certification of Causes of Death 2021 (MCCD) and the Sample Registration System 2019-2020 (SRS).
Methods: Congenital anomaly mortality data was extracted from the MCCD and SRS and analysed. The MCCD included 18,11,688 medically certified deaths, occurring mostly at urban hospitals, and representing 22.5 per cent of total registered deaths in India in 2020. The SRS mortality data for the year 2019-21 was reported from 8841 sample units (4,958 rural and 3,883urban units) with approximately 8.4 million population across the country. Descriptive statistics was used to report the proportion and trends of congenital anomaly mortality in India.
Results: Congenital anomaly mortality constituted 0.5% (MCCD) and 0.3% (SRS) of all-cause mortality among all age groups. MCCD data identified that over half of congenital anomaly mortality occurred during infancy (54.5%). Among newborns, congenital anomalies were the fourth major group of conditions accounting for 5.2 percent deaths. Within this group, congenital malformations of the circulatory system accounted for nearly one third of deaths (28.6%), cleft lip and palate and spina bifida accounted for 15% and 2% congenital anomaly mortality, while a category labelled as 'other conditions' accounted for over half of mortality. Comparative trends indicated that major perinatal causes like prematurity and birth asphyxia have halved between 2013 and 2020 (8.1 % to 4.1 %), but the decline in congenital anomaly mortality was nearly stagnant (0.9% to 0.5 %). SRS data indicated higher mortality in urban (6.5%) than rural (5.0%) areas, and higher mortality in states with lower neonatal mortality (8.4%) as compared to states with higher neonatal mortality (3.5%).
Discussion and Conclusions: Data from these two surveillance systems are indicative of transition in causes of mortality. Rapid decline in other causes of child mortality but negligible reduction in congenital anomaly mortality, higher proportions of congenital anomaly mortality in urban areas and in low neonatal mortality states of the country is indicative of increase in these disorders as causes of infant mortality. Surveillance and data reporting systems need to be strengthened to improve the quality of data on congenital anomalies in India. Congenital anomaly mortality data should be incorporated into maternal and child health reporting systems in India.

The Alarming Increase in Birth Defects at the Expense of Congenital Heart Diseases: The Maternal Risk Paradox Phenomenon

Dr. Santiago Andrade[1], Dra Maria Camila León-Sanabria[1], Ms Adriana Guaca[2], **Dra Karen Sarmiento**[3], Dr. Ignacio Zarante[1,4]
[1]Institute of Human Genetics, Faculty of Medicine, Pontificia Universidad Javeriana, Bogotá, Colombia, [2]District Health Secretariat, Subdirectorate of Public Health Surveillance, Lead Specialist for Birth Defects, Bogotá, Colombia, [3]Department of Physiologic Sciences, Faculty of Medicine, Pontificia Universidad Javeriana, Bogotá, Colombia, [4]Hospital Universitario San Ignacio, Bogotá, Colombia

Email of corresponding Author: kssarmiento@gmail.com

Background & Objectives: In countries undergoing epidemiological transition, birth defects (BDs) have emerged as a leading cause of infant morbidity and mortality, especially in children under five years of age. In Colombia, all major BDs diagnosed in children under one year must be reported to the Public Health Surveillance System (SIVIGILA). Since 2010, BDs have become the primary cause of death among infants under one year old. This trend is particularly alarming in Bogotá, where a steady rise in prevalence mainly driven by congenital heart defects (CHDs) has been observed. This situation reflects what we describe as the "maternal risk paradox" where despite an increasing prevalence of maternal and neonatal risk factors—such as advanced maternal age, prematurity, and low birth weight—there has been no corresponding improvement in detection or outcomes. This study aimed to conduct an epidemiological analysis of the rising prevalence of BDs in Bogotá between 2020 and 2024 based on data reported to SIVIGILA. Our objectives were to characterize the most common BDs, identify associated maternal and perinatal risk factors, and uncover gaps in prenatal diagnosis. The findings aim to inform public health strategies to reduce preventable morbidity and mortality through enhanced surveillance, earlier detection, and targeted prevention efforts. Additionally, this study contextualizes Bogotá's data within broader Latin American and global trends, highlighting the need for policy action to mitigate the growing burden of BDs and to improve both child survival and long-term health outcomes.
Methods: A descriptive observational study was conducted using data from Bogotá's Public Health Surveillance database from 2020 to 2024. All reported cases of major BD in live births (LB) and stillbirths (SB) within the city were included. Collected variables included sex, maternal age, gestational age (categorized as extremely preterm <28 weeks, very preterm 28–31 weeks, moderate preterm 32–33 weeks, late preterm 34–36 weeks, and term ≥37 weeks), birth weight, and timing of diagnosis (prenatal vs postnatal). BDs were coded using ICD-10 (Q00–Q99), including select metabolic and sensory conditions. Annual trends were analyzed using absolute and relative frequencies by BD type and affected organ system. Prenatal detection rates, sex distribution, and associations with maternal and perinatal risk factors—such as advanced maternal age (≥35 years), prematurity, and low birth weight—were assessed. Descriptive statistics and trend analyses were performed. Data quality was ensured by removing duplicates and cross-verifying with local health reports. Ethical approval was not required, as the study utilized anonymized secondary data from a public health surveillance system.
Results: A total of 10,424 cases of BDs were reported in Bogotá from 2020 to 2024, reflecting a 151% increase from 1,265 cases in 2020 to 3,175 in 2024. However, during this period the number of LBs in Bogotá showed a steady decline, dropping from 79,322 in 2020 to 56,552 in 2024—a reduction of nearly 30%. This downward trend may reflect broader demographic shifts, including declining fertility rates or changes in access to reproductive health services. Regarding BD cases sex distribution showed a slight male predominance (51%). Median maternal age was 29 years (IQR: 23–34), 22% cases occurring in mothers aged ≥35 years. Nearly 25% of affected newborns were preterm, mainly late preterms (34–36 weeks: ~16%), and 7% born before <32 weeks of gestation. Multiple non-syndromic anomalies accounted for 5.6% of cases, while 2.5% were associated with recognized genetic syndromes—primarily chromosomal anomalies such as trisomies, and ~5% had complex or sequential anomalies. Only 34% cases were diagnosed prenatally, underscoring gaps in

screening coverage and early detection. CHDs were the most frequent (~35%), led by ventricular septal defect (n=1,153), atrial septal defect (n=899), patent ductus arteriosus (n=641) and aortic coarctation (n=204). Among non-cardiac defects, the most common included congenital hydronephrosis (n=703), polydactyly (n=478), clubfoot (n=370), Down syndrome (n=494), microcephaly (n=338), and microtia (n=282). Neural tube defects accounted for a combined of 1.6% of cases, with 91 cases of anencephaly and 71 of spina bifida. ~538 cases of orofacial clefts were reported.

Discussion & Conclusion: The sustained increase in BDs in Bogotá between 2020 and 2024 underscores their growing contribution to infant morbidity and mortality. The marked rise—particularly in CHDs—mirrors global trends but suggests a comparatively higher local burden. Septal defects emerged as the most prevalent anomalies, followed by musculoskeletal and urogenital malformations. The high prevalence of advanced maternal age, prematurity, and low birth weight supports the notion of the maternal risk paradox. Although the distribution of affected organ systems aligns with international patterns, the elevated prevalence observed in Bogotá may reflect enhanced surveillance efforts and lower rates of pregnancy termination following prenatal diagnosis—a known feature of the Colombian context. Notably, the limited prenatal detection rate (34%) highlights persistent barriers in access to screening and specialized fetal imaging. The predominance of isolated anomalies not linked to recognized syndromes reinforces the need to improve early identification and prevention strategies for non-syndromic defects. These findings point to critical opportunities to strengthen BD surveillance systems, improve data completeness, and integrate both primary prevention (e.g., nutritional supplementation, chronic disease management, avoidance of teratogenic exposures) and secondary prevention (e.g., prenatal screening, timely referral, and perinatal care). A coordinated and robust public health response is essential to reduce the preventable burden of BDs and to improve survival and long-term outcomes for affected children.

Mapping isolates with a high frequency of congenital anomalies and rare diseases: CENISO - Brazil

Prof Lavinia Schuler-Faccini[1,2,3], Dr Augusto Cesar Cardoso-dos-Santos[3], Vinicius Barreto Nolibos[2,3], Bruno Canabarro Delgado Coelho[2,3], Julia Rei Pires[2], MSc Paulyana Moura Corecco-Moura[2,3], MD Laercio Moreira Cardoso-Junior[2,3]

[1]Hospital De Clinicas De Porto Alegre, Porto Alegre, Brazil, [2]Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil, [3]INAGEMP - Brazilian National Institute of Population Medical Genetics, Porto Alegre, Brazil

Email of corresponding Author: lschuler@hcpa.edu.br

Background and Objectives: Clusters of rare diseases and congenital anomalies are described in the literature in various population isolates around the world. Mapping and investigating these clusters have significantly contributed to understanding the biology of genetic and congenital disorders. Furthermore, this approach serves as a crucial public health strategy for the surveillance and care of rare diseases, congenital anomalies, and for promoting health equity. The presence of numerous cases of uncommon, chronic, and complex health conditions presents considerable challenges for surveillance and appropriate care, particularly in Latin America. Here, such clusters are often geographically remote and isolated, making them difficult to recognize and access. In this work, we aim to report the strategy employed for the Brazilian population since 2009, which enables the spatial geolocation of clusters of congenital anomalies and genetic diseases. This strategy was recently extended and applied across the South American continent.

Methods: The Brazilian Census of Isolated Populations (CENISO) is a protocol designed for the mapping and investigation of genetic isolates within populations, typically identifying geographic clusters of genetic or multifactorial disorders. This protocol is based on the systematic search for "rumors." A rumor, in this context, refers to any account, whether oral or written, of the unusual occurrence of a suspected genetic disorder or congenital anomaly within a specific geographic area or sub-population. Complementary to the rumor-based strategy, a systematic literature search is also conducted. Rumors are meticulously recorded and subsequently validated through a progressive process involving pre-established steps. This classification system is structured into four ordered phases: Phase 1 involves the registration of a rumor without robust supporting evidence. Conference proceedings and expert reports are categorized as Phase 2 rumors. Phases 3 and 4 entail confirmed clusters that have been verified through demographic research. Research and healthcare strategies are then developed and implemented during these latter phases. Additionally, we maintain a classification system for "alarms," which denote a sudden temporal variation in the frequency of a specific congenital anomaly, thereby suggesting a potential environmental teratogen. A notable example of an alarm is the 2015-16 outbreak of microcephaly, primarily in Brazil and subsequently in other Latin American countries, which was secondary to maternal infection by the Zika virus during pregnancy. More recently, we have applied a Brazilian-inspired strategy to track similar clusters across the entire South American continent.

Results: Since its inception in 2009, CENISO has identified 170 confirmed clusters of congenital anomalies and rare diseases. The distribution of these clusters across Brazil is uneven, with a notable predominance in the Northeast and South regions, and a lower frequency observed in the North and Center-West regions. In numerous instances, the suspicion or confirmed identification of a cluster has directly led to scientific investigation and the provision of healthcare services for the affected communities. This includes crucial aspects such as diagnosis, genetic counseling, appropriate treatment, and, where feasible, molecular investigation. Furthermore, collaborative work with these communities has yielded significant social implications, including the dispelling of myths, a reduction in stigma, and the promotion of informed family planning. It is also worth noting that engagement with isolated communities frequently uncovers valuable historical, sociocultural, and demographic insights into the vast Brazilian population.

Discussion & Conclusion: The epidemiological landscape of these clusters strongly reflects the processes of population admixture, territorial occupation, and the sociodemographic profile of a country's population. In Brazil, the higher numbers observed in the Northeast region can be attributed to elevated levels of

consanguinity, which significantly contributes to a higher occurrence of recessive genetic diseases. Additionally, the underreporting of clusters in other macro-regions and the uneven distribution of healthcare professionals capable of recognizing these diseases, who are more concentrated in the South and Southeast regions, also play a role. The application of the CENISO strategy to South America allowed us to infer that similar risk factors are prevalent throughout the continent. The identification and study of genetic isolates and clusters represent a crucial tool for planning health priorities concerning rare diseases in low- and middle-income countries characterized by large populations. The CENISO strategy has consistently proven to be a cost-effective and efficient alternative within the Latin American context. By effectively identifying populations affected by congenital anomalies and rare diseases, and by centralizing available information on these patients, it becomes possible to provide accurate diagnoses and promote comprehensive medical care for affected individuals and their families.

MRI assisted Neurosonography (MAN) two case series that demonstrate efficiency of Fusion Multimodality Technology in Fetal neuroradiology diagnostics. MALFOVAR (Malformation and variable anatomy registry) update 2025.

Dr Tidhar Steiner[1], Dr. Gyorgy Balazs, Dr. Belics Zoran, Dr. Gabor Rudas
[1]MALFOVAR , Budapest Hungary, Hungary, [2]Semmelweis University , Budapest, Hungary

Email of corresponding Author: drsteiner@live.com

Background and objectives: Our goal is to demonstrate the feasibility and superiority of Multimodality fusion (US and MRI) MAN (MRI assisted neurosonography) compared to monomodality (US and MRI separately).
In order to achieve such goal we approached the challenge in two phases:
1. Demonstration of normal fetal neuroanatomy while using MAN.
2. Application of MAN (MRI assisted neurosonography) in fetal neuroradiological diagnostics.
Methods: We performed 2 separate case series which included altogether 15 cases.
The first case series was performed during the years 2021-2022 which included 8 cases of Fetal MAN. In this series we used a Fusion capable ultrasound machine GE Logiq E10 and MRI 3T Philips Ingenia and 3T GE Signa.
The second case series was preformed during the years 2024-2025 we demonstrated 7 cases of fetal MAN (MRI assisted neurosonography). In the second case series we tested a Fusion capable ultrasound machine of Sonoscape S80 Fenix and MRI 3T Philips Ingenia and 3T GE Signa.
Results: Altogether 15 MAN examination were performed with 15 fetuses.
Out of 15 cases we performed 1 case of post processing (Ultrasound and MRI) simultaneous synchronous examinations. In our experience, the 3D reconstruction images were not suffice in quality in order to use for fusion therefore we opted for postprocessing fetal MRI and live Manual MAN. The rest of the later 14 cases were performed with live Manual MAN. There were 4 cases which demonstrated normal neuroanatomay of the fetus. In this group the first case was done by post processing (Ultrasound and MRI) simultaneous synchronous examination while the further 3 cases were performed by live Manual MAN. We also performed 11 cases Fetal / placental malformation which included: Corpus callosum agenesis, two cases of Corpus callosum dysgenesis, Dandy walker malformation, Arnold Chiari malformation, Blakes pouch. MCD gyrification sulcation anomaly, Hydrocephalus and placental malformation. We also examined 2 cases of CNS bleedings which included Cerebellar bleeding and intraventricular choroid plexus bleeding.
Discussion and Conclusion: Our conclusion is that MAN (MRI assisted neurosonography) gives superior capabilities as a multimodality system compared to monomodality examinations. We demonstrated fetal fusion in neuroradiology using MAN technology with different ultrasound machines of different vendors (GE and Sonoscape). The efficacy of this technology was demonstrated to assist Normal anatomy and recognize further 8 types of variety of fetal neurological malformations. The two CNS bleedings cases demonstrate that such important events during pregnancy which may cause potentially serious damage to fetal brain can be detected effectively and more accurately with the help of MAN technology. The usage of ultrasound in fetal neuroradiology while using MAN technology (observing post-acquisition MRI) enables for the physician to detect lesions which may not be detected or vaguely observed with use of Monomodality (ultrasound and MRI separately). These 2 case series may enable a breakthrough in the field of fetal neuroradiology diagnostics were as further studies are needed to validate such technologies. The Combination of MRI and ultrasound with fusion technology in fetal diagnostics with the use of AI may assist physicians in the future to better understand and detect fetal malformations.

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