

# Birth defects in the developing world, integrating screening, diagnosis and management into health systems



World Health  
Organization

**International Conference on birth defects in developing countries**

1<sup>st</sup> March 2023, Santiago, Chile

Dr Anshu Banerjee, Director, MCA Department, WHO/Geneva

# Sustainable Development Goals – on reducing neonatal and child mortality



Sustainable Development Goals (SDG) and WHO Global Program of Work 13

- Emphasizes equity and Universal Health Coverage
- SDG targets for 2030 are:
  - Reduce neonatal mortality to at least as low as 12 deaths per 1,000 live births;
  - Reduce under-5 mortality to at least as low as 25 deaths per 1,000 live births.

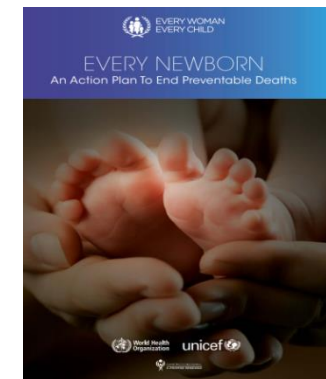


Every Newborn Action Plan (ENAP)

Global Strategy for women's children's adolescent's health: *Survive, thrive, transform.*

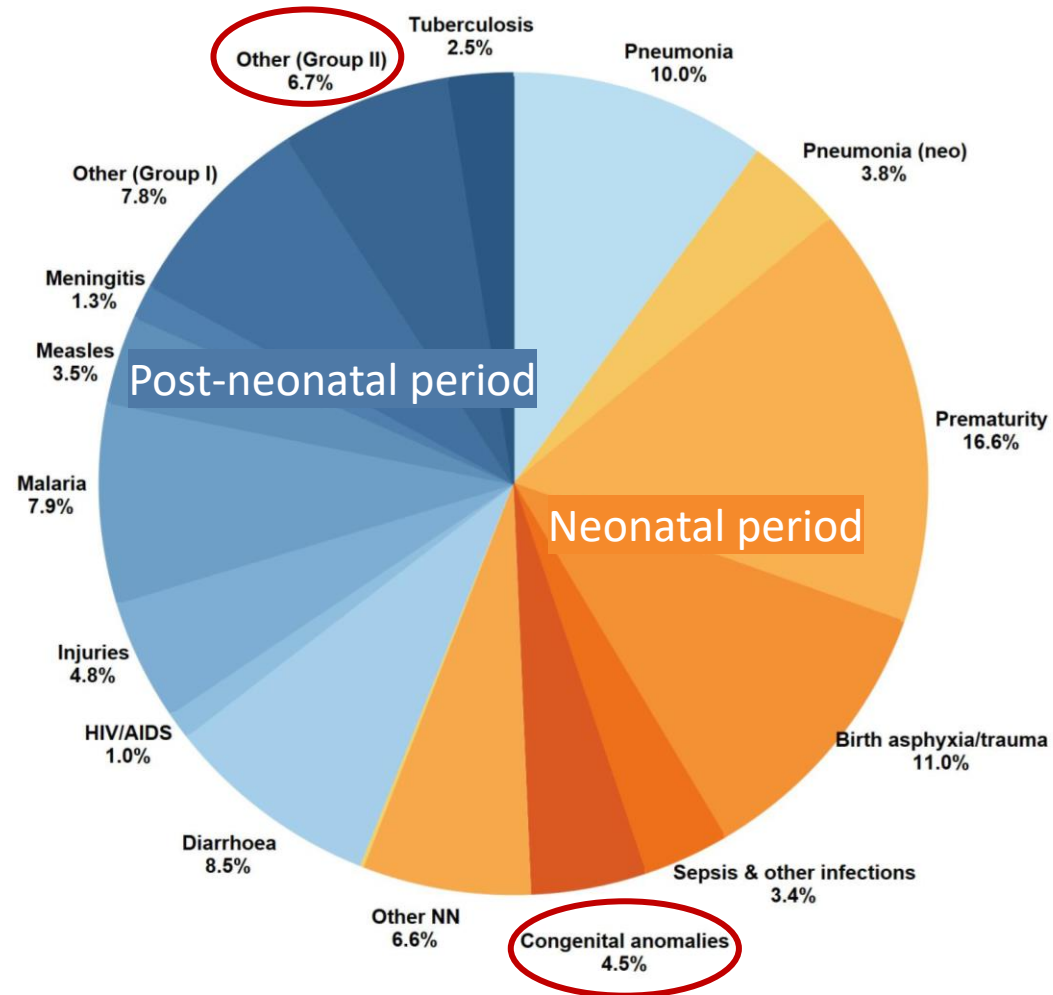
- *Targets align with the SDGs*

***Surveillance, prevention and providing care for children with birth defects are an important strategies to reach child survival targets***



# Birth defects contribute to around 9% of under-5 mortality

Number of death 1 to 59 months
Acute respiratory infections: 540 000
Diarrhoea: 450 000
Malaria: 420 000
Injuries: 253 671



Number of deaths first 28 days
Prematurity: 880 000
Birth asphyxia/trauma: 580 000
Congenital anomalies: 240 000
Neonatal sepsis: 200 000

The leading causes of death (% of total deaths) for newborns (first 28 days of life) and postneonatal children

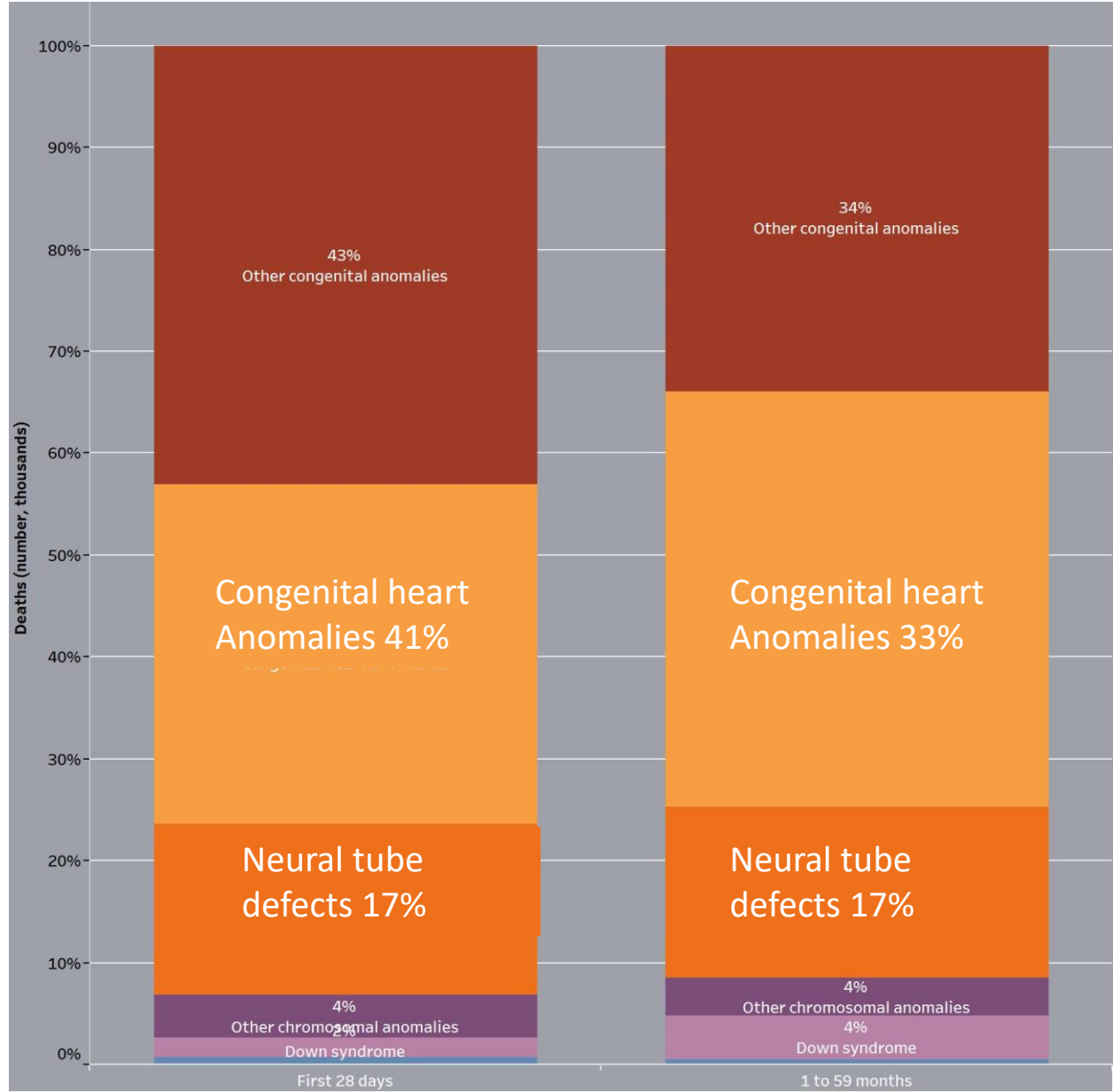
## Definition of other cause categories

Other NN (neonatal): other infectious causes, HIV/AIDS, pertussis, measles, meningitis, malaria, diarrhoea, injuries and other NCDS

Other (group I) (Post-neonatal): Other infectious, tetanus, neonatal sepsis, prematurity, birth asphyxia

Other (group II) (Post-neonatal): congenital anomalies, other NCDs

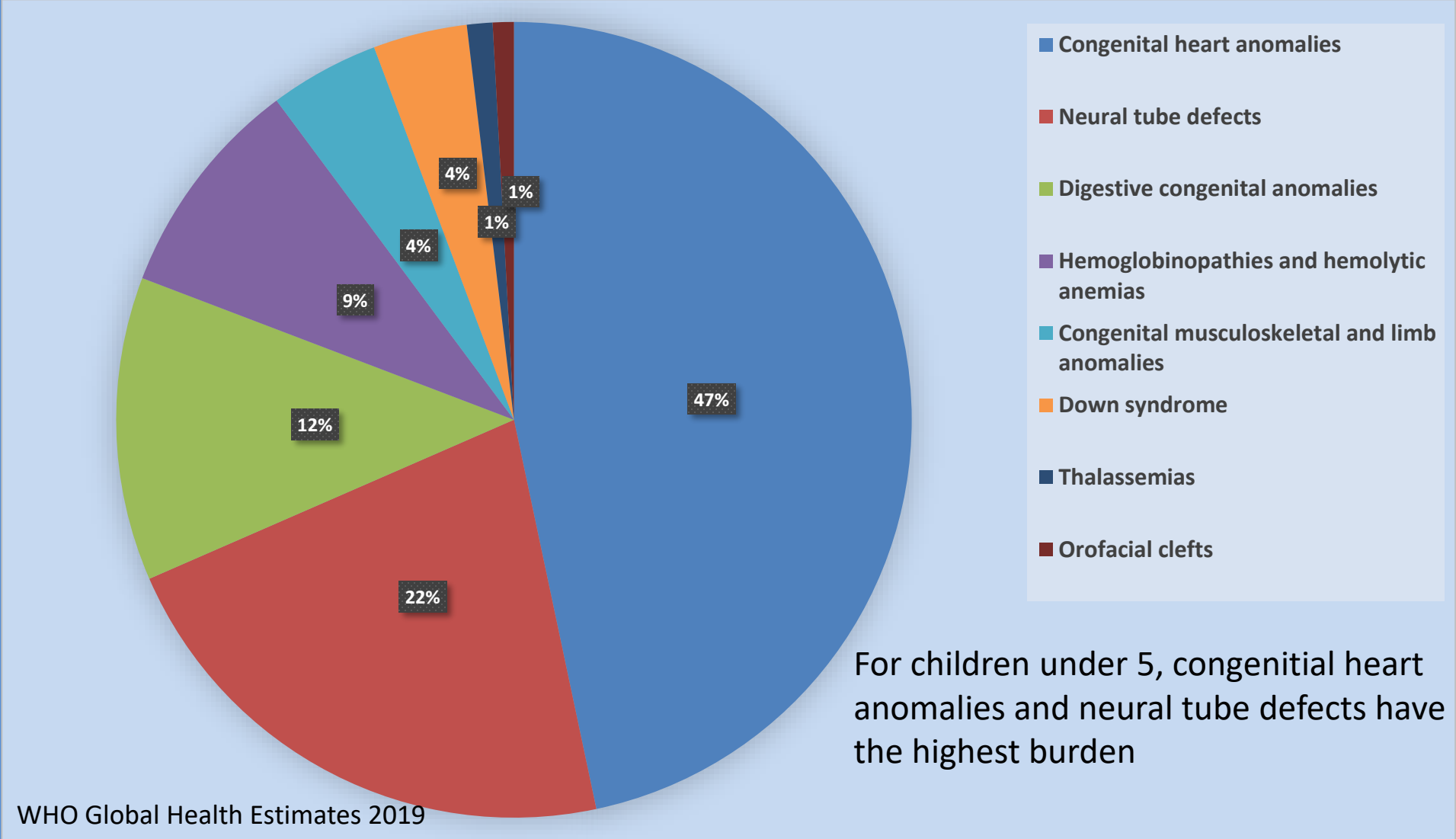
# Global cause-specific mortality, estimates from WHO Global Health Estimates for 2019



Congenital heart anomalies and neural tube defects account for most deaths due to congenital anomalies under-five years.

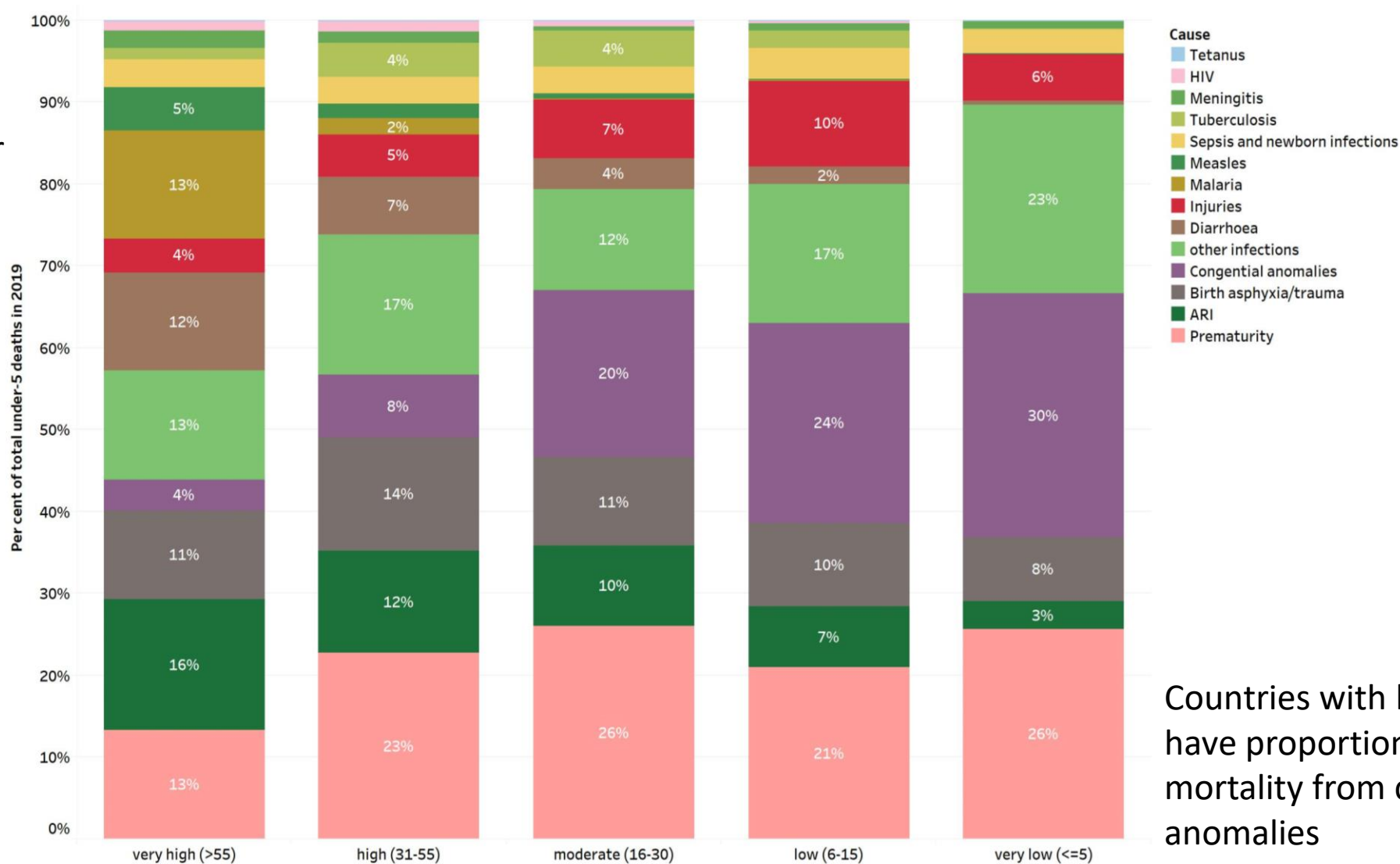
58% for neonates and 50% for postneonatal under-5s

# Years Lived with Disability (DALYS) for children under-5 with congenital anomalies



# Birth defects are increasingly a cause of under-5 deaths as under-5 mortality rates decline

Countries with high U5MR have proportionally higher mortality from infectious diseases



Countries with low U5MR have proportionally higher mortality from congenital anomalies

Levels of under-5 mortality rate per 1000

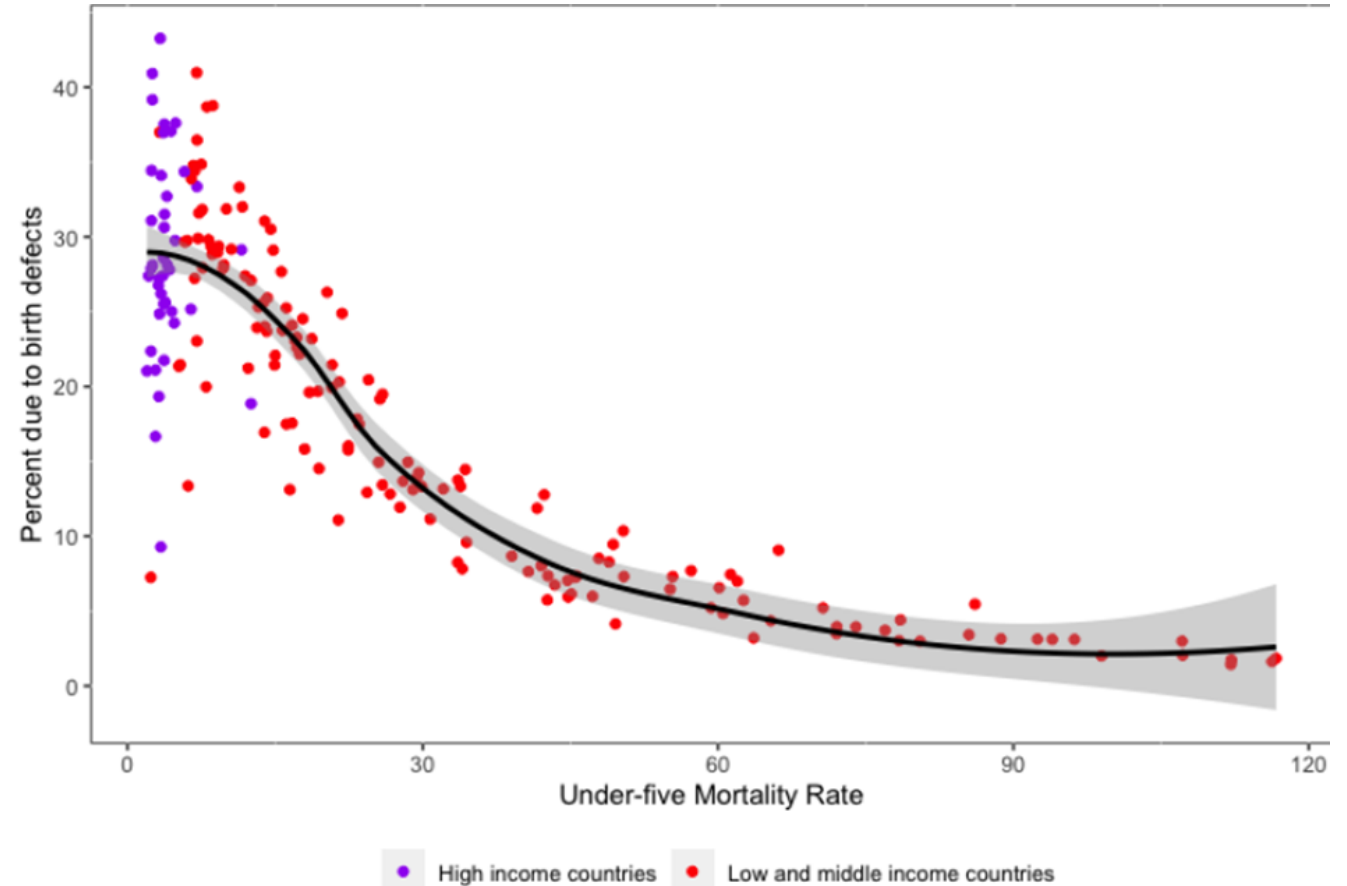


# Epidemiological transitions in LMICS - implications for child mortality from birth defects

**As neonatal and under-5 mortality rates decline in LMICs, birth defects become a larger proportion of the cause of neonatal and under-5 deaths**

Important to note that even if the proportions in LMICs look relatively small compared to HIC setting, in absolute numbers these are much higher!

In 2019 – mortality from birth defects in HIC settings was 14,300, where as in LMIC it was 389,600



# Why is it important for LMICs to invest in prevention, diagnosis and management of birth defects?



- Treating children with birth defects early gives the affected child the best the possible chance to lead as healthy a life as possible.
- Children with disability from birth defects are exposed to multiple inequities – they have more limitations in functioning, poorer health and shorter lives
- Birth defects presently responsible for 9% of deaths in children under 5. Proportion likely to increase
- 90% of these deaths occur in LMICs. Local burdens of birth defects are likely to be higher but uncertain as they are poorly captured
- By including birth defects in global and national priorities we can fulfil the SDG’s commitment to “leaving no one behind”, improving the situation of millions of affected children and their families



Important to prepare systems for response now!

To improve survival and wellbeing of children with birth defects, screening, diagnosis and management need to be integrated into routine health systems





# World Health Assembly Resolution on Birth Defects

The Sixty-third World Health Assembly (2010) passed resolution 63.17 promoting primary prevention and improving the health of children with birth defects by:

- initiation and monitoring of population-screening programmes such as **screening of newborn infants**
- **integration** of approaches to the prevention and care of birth defects into primary health care, with an emphasis on maternal and child health
- Identify successful **models of care** that can be applied in low- and middle-income countries.
- Developing **expertise and building capacity**
- Promoting **international cooperation**

# WHO global response

## ➤ Surveillance and estimates of burden:

- Birth defects surveillance toolkit and case definitions
- Development of country level estimates of birth defects

## ➤ Prevention:

- Folic acid fortification and supplementation
- Elimination of Congenital rubella syndrome and Congenital syphilis
- Antenatal and postnatal care

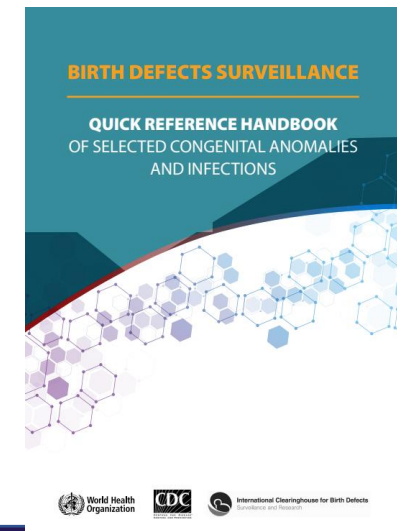
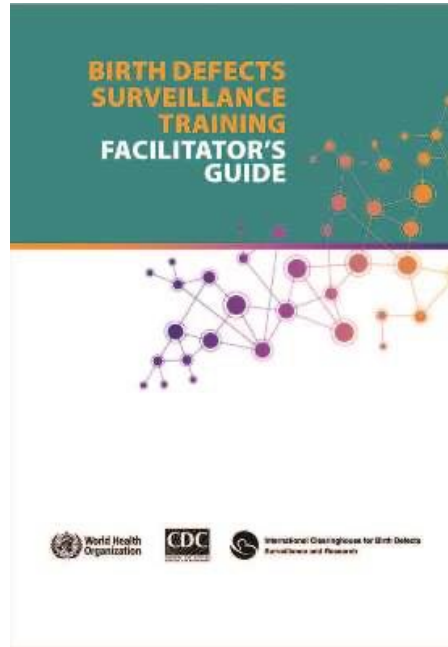
## ➤ Screening, diagnosis and management

- Plan the development of guidelines for the screening and management of important birth defects in LMIC settings.
- Support integration of screening, diagnosis and management into national maternal, newborn and child health programmes



# Birth Defects Surveillance toolkit

- Aligns with WHA 2010 Resolution
- A joint effort of WHO, CDC & ICBDSR
- Copyright: WHO
- Three components:
  - A Manual for Program Managers
    - 2<sup>nd</sup> edition 2020
  - Quick Reference Handbook 2020
    - Replaced the Atlas (2015)
  - Facilitator's Guide
    - 2015
    - Revised in 2022





# Revision of Global Burden of Birth Defects by WHO and partners

- ❑ Working with partners -CDC, March of Dimes and academic groups from LSHTM and Johns Hopkins University, Rare Diseases SA; all groups with data welcome to contribute
  
- ❑ Technical working group 27 members, representation across all regions and LMICs
  - ❖ 4 task teams tackling aspects of the revision including scope, standard terminology, model and data sources
  - ❖ First product is a peer-reviewed journal article (BMJ Open) on mortality from congenital anomalies; additional paper forthcoming on standardizing terminology
  - ❖ Revision is expected early in 2024

# Prevention: Vaccination against Rubella

- WHO member states using rubella containing vaccines in their national programs is increasing (173/194 states in 2019)
- Congenital rubella syndrome – falling overall, but relatively high in some LMICs
- WHO encourages member states to incorporate rubella vaccination as quickly as possible into national immunization programs.



# Congenital rubella syndrome



## Disclaimer

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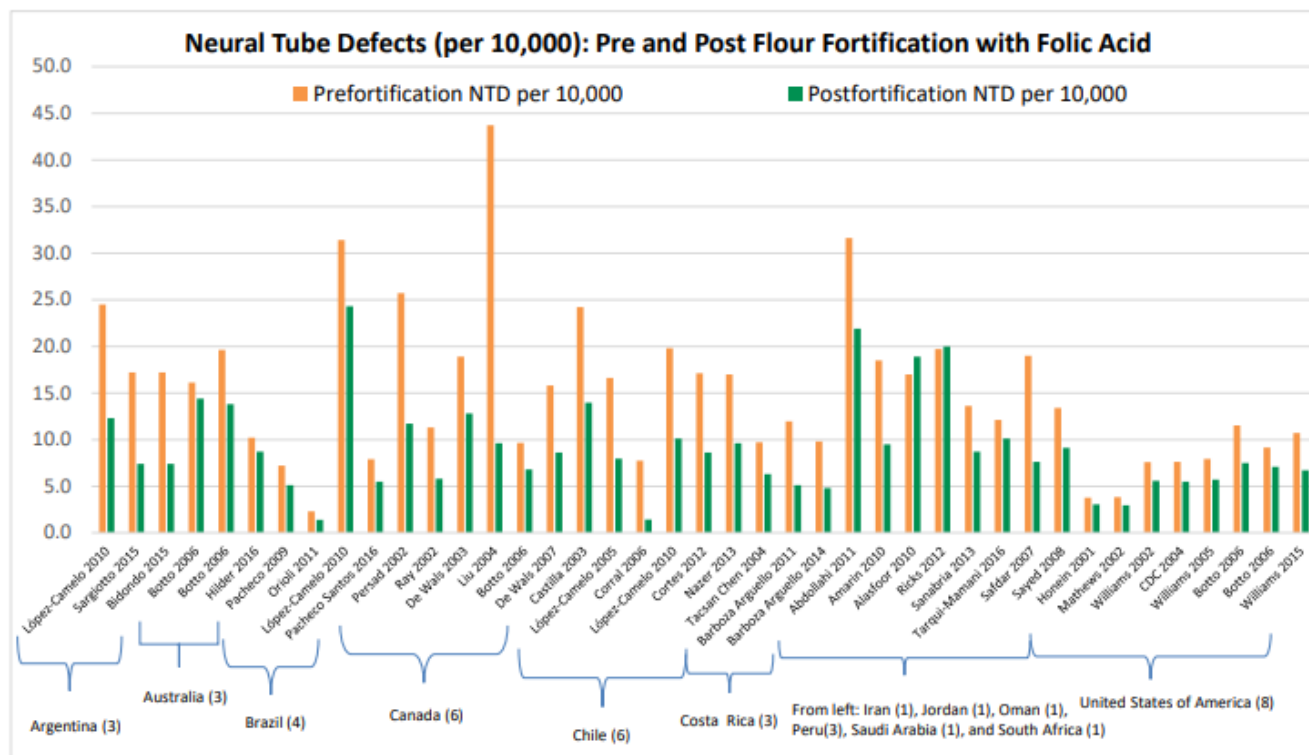


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# Prevention: Fortification with folic acid

A review of 41 documents that reported the prevalence of NTDs per 10,000 births\* before and after the initiation of flour fortification with folic acid in 13 countries



- Reduction post fortification varies between 11-82% depending on baseline prevalence and quality implementation. Average: ~41%
- There are not established risks for adverse consequences resulting from the provision of folic acid from RCTs or fortification programmes.

1: <https://www.fjinetwork.org/savelives> | 2: [https://www.thelancet.com/journals/langlo/article/PIIS2214-109X\(22\)00213-3/fulltext](https://www.thelancet.com/journals/langlo/article/PIIS2214-109X(22)00213-3/fulltext) | 3: Field and Stover 2017



# Salt iodization

1986. WHA 39.31. Prevention and control of iodine disorders.

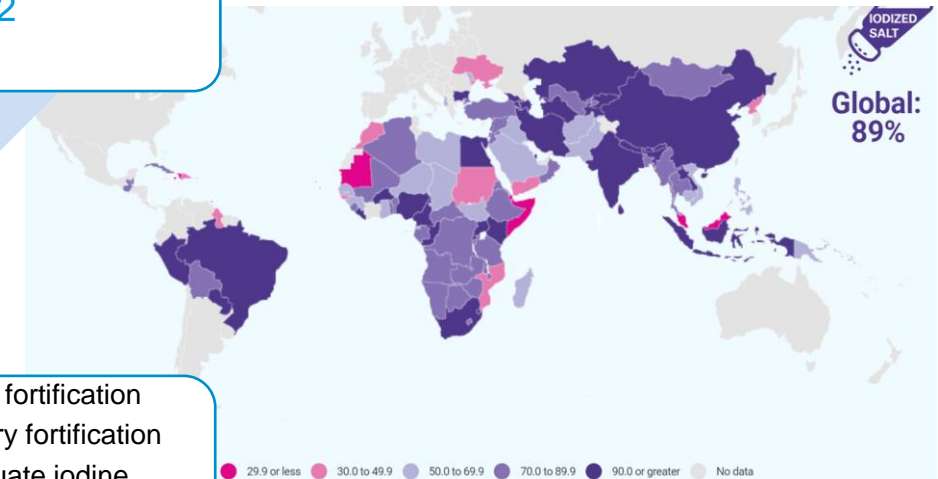
2003. WHA 58.24. Sustaining the elimination of iodine deficiency disorders

2020-22

17 countries mandatory fortification  
3 countries had voluntary fortification

82 countries mandatory fortification  
11 countries had voluntary fortification  
67 Countries with adequate iodine intake

126 countries mandatory fortification  
11 countries had voluntary fortification  
118 Countries with adequate iodine intake  
155 WHO countries with data



Recommended  
resolution for  
adoption at the  
76<sup>th</sup> World Health  
Assembly , 2023



EXECUTIVE BOARD  
152nd session  
Agenda item 18

EB152(13)  
3 February 2023

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**Accelerating efforts for preventing micronutrient  
deficiencies and their consequences, including spina  
bifida and other neural tube defects, through safe  
and effective food fortification**

The Executive Board, having considered the report by the Director-General,<sup>1</sup> highlighting the need to accelerate progress in safe and effective food fortification,<sup>2</sup>

Decided to recommend to the Seventy-sixth World Health Assembly the adoption of the following resolution:

The Seventy-sixth World Health Assembly,

Recalling resolutions WHA39.31 (1986) on prevention and control of iodine disorders; WHA45.33 (1992) on national strategies for prevention and control of micronutrient malnutrition; WHA58.24 (2005) on sustaining the elimination of iodine deficiency disorders; WHA65.6 (2012) on comprehensive implementation plan on maternal, infant and young child nutrition; and WHA68.19 (2015) on outcome of the Second International Conference on Nutrition, which promote food fortification as a mechanism to prevent micronutrient deficiencies and birth defects associated with nutritional deficiencies;

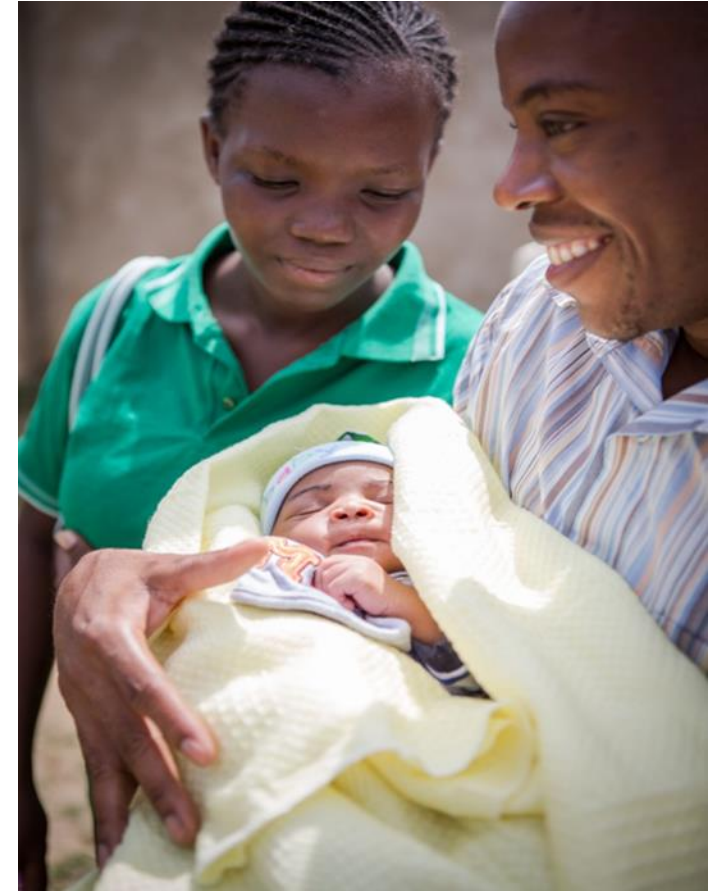
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# Newborn screening –for early diagnosis and management

## Embedding universal newborn screening in LMICs

### Why is this important for LMICs now?

- *Cannot be left unaddressed. To maximize life chances for children born today, important that congenital defects are recognized early, so that treatment can be started early and impairment/disability minimized. Common conditions like hearing impairment, congenital hypothyroidism – if routinely screened for and picked up early, make a large impact to the life chances of the affected child*
- *Newborn screening an increasingly important public health priority for MoH in LMICs*
- *WHO encourages members states, especially in LMIC settings, to consider universal newborn screening programs to screen for a few context relevant birth defects.*
- *Not useful to screen for conditions for which there are no back up services*



# Newborn Screening – clinical and biochemical

Clinical and biochemical

## Clinical screening:

- Visual birth defect screening at birth for structural defects (club feet, orofacial clefts)
- Emphasis now on hearing screen for newborns, particularly preterms/ LBW
- Screening for defects of sight
- Pulseoximetry based screening for critical congenital heart disease (in setting where further investigations and treatment pathways are prepared)

## Biochemical screening:

- E.g. Congenital hypothyroidism, hemoglobinopathies



# Clinical NBS in the context of WHO recommendation on post natal care

Released 30<sup>th</sup> March 2022

Recommendation for screening of specific birth defects in the newborn

- Universal screening for abnormalities of the eye (Recommendation 26)
- Universal screening for hearing impairment (recommendation 27)



# Initiating newborn screening

## *Considerations*

Consider conditions which are

- (i) Important in the context
- (ii) can be accurately diagnosed in country (if not presently, capacity can be feasibly built up)
- (iii) treatment is feasible (or can be built up reasonably) in the context of health systems
- (iv) makes a significant impact on the life of the infant/child
- (v) the health system can reasonably bear the cost



# Examples of NBS different LMICS (1)

Many LMICs do not have national NBS programs

Asia Pacific – Philippines –national health insurance supported NBS – covers 70% of all births – screening for 6-7 conditions. IAEA supported network helped countries undertake more widespread screening largely for congenital hypothyroidism. Led to government supported programs. Still young programs, but govt support to screening for low-income families in Korea raised screening uptake rates to 70%. Some patchy expansion into more expanded NBS – including for metabolic conditions. CCHD screening increasing in the region

China – national screening program covers >90% of newborns – CH and PKU universally screened for. Expanded NBS in some centers – mostly to inform the national program – other metabolic conditions emerging as important candidates, CCHD screening in some areas – cost effective in areas with good back up services

India: Rashtriya Bal Swasthya Karyakram (RBSK or National healthy child program) – recommends facility-based and community-based NBS, specifically listing CH, sickle cell disease, congenital hearing loss and congenital heart disease. Not implemented universally across country as yet. Indian Council for Medical research established a nation wide study to establish prevalences to inform NBS programs. CH and CAH are important, G6PD in some parts of the country

## Examples of NBS different LMICS (2)

Middle East and North Africa: Low income countries have had some support from IAEA – for CH. Saudi Arabia (high income) – screens for 16 conditions – CH, CAH and some metabolic disorders relatively higher prevalence.

Sub-Saharan Africa - 25% of all births, likely commensurate burden of birth defects. Local donor funded projects on NBS for SCD. Need for MoH stewardship to increase coverage of NBS in the population

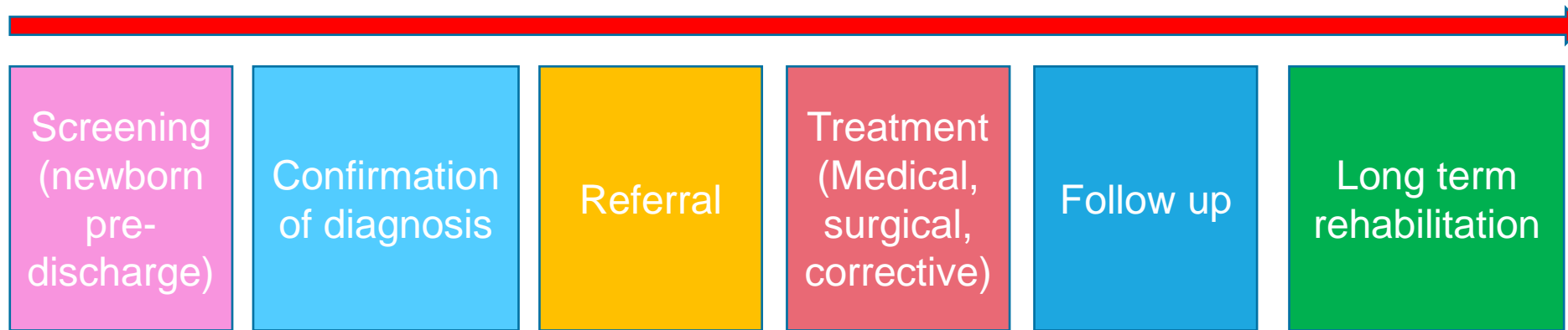
Latin America: NBS programs in almost all countries with variations in conditions screened and coverage

Lessons learned across these regions: gaps in back up services, follow up and treatment, phased introduction of conditions while generating evidence of burden at the same time to inform ongoing NBS initiatives



# From screening through to management

*The continuum of care*



For program managers - for each important congenital defect prioritized, consider: service delivery – levels at which each of the above occur? Human resource needs at each level? Referral mechanisms? Regionalization of specialist treatment? Devices/ medical products/ lab services needed? Financing mechanisms?

**“Surgical capacity is an essential part of universal health coverage”**

— WHO Director General, Dr. Tedros

Planning for  
pediatric  
surgery

## **Congenital Anomalies in Low- and Middle-Income Countries: The Unborn Child of Global Surgery**

Nicole A. Sitkin · Doruk Ozgediz · Peter Donkor ·  
Diana L. Farmer

> J Pak Med Assoc. 2019 Feb;69(Suppl 1)(1):S108-S111.

Journal of Pediatric Surgery 54 (2019) 845–853

Contents lists available at ScienceDirect

Journal of Pediatric Surgery

journal homepage: [www.elsevier.com/locate/jped surg](http://www.elsevier.com/locate/jped surg)

## **Global perspective of paediatric surgery in low and middle income countries**

Saqib Hamid Qazi <sup>1</sup>, Sohail Asghar Dogar <sup>1</sup>, Samie Asghar Dogar <sup>1</sup>, Tamara Fitzgerald <sup>1</sup>,  
Ayesha Saleem <sup>1</sup>, Jai K Das <sup>1</sup>

Delayed access to care and unmet burden of pediatric surgical disease in resource-constrained African countries

Yasmine Yousef <sup>a,\*</sup>, Angela Lee <sup>a</sup>, Frehun Ayele <sup>b</sup>, Dan Poenaru <sup>a</sup>

# WHO plan to develop guidelines on newborn screening, diagnosis and management for a few key conditions

- Support LMICs to initiate newborn screening, diagnosis and management of a few important conditions initially, through the development of a guideline
- Plan to initiate work in 2023, after funding is secured
- Guideline to focus on 5-6 priority conditions initially that LMICs could begin universal screening for and put in place diagnostic and management capacities for.



# Going Forward: getting birth defects on the agenda in all countries

## Challenge

- (i) Birth defects not prioritized in many LMIC health policies – seen as NCDs that don't have simple cures that can be operationalized through targeted programs
- (ii) Collectively may be relevant as a group, but individual disorders are rare

For the first time, the UHC compendium will include birth defects – to encourage ministries to start looking into systems to support the diagnosis and management of birth defects





# Stigma towards the child and family

תאריך פרסום: 16 במאי 2016

Advance Access Publication Date: 16 May 2016

Original Research Article

ARTICLE • Cad. Saúde Pública 38 (4) • 2022 • <https://doi.org/10.1590/0102-311X00104221>

## Stigmas of congenital Zika syndrome: family perspective

Estigmas da síndrome da Zika congênita: perspectivas das famílias

Estigmas del síndrome Zika congénito: perspectivas familiares

Martha Cristina Nunes Moreira Corina Helena Figueira Mendes Marcos Antonio Ferreira do Nascimento

Humanities & Social Sciences  
Communications



ARTICLE



<https://doi.org/10.1057/s41599-020-00677-7> OPEN

## Social stigma and cultural beliefs associated with cleft lip and/or palate: parental perceptions of their experience in Kenya

Stephen Gichuhi Kimotho <sup>1</sup> & Fiona Nduta Macharia <sup>2</sup>

## Stigma in Medical Settings As Reported Retrospectively by Women With Congenital Adrenal Hyperplasia (CAH) for Their Childhood and Adolescence

Heino F. L. Meyer-Bahlburg,<sup>1,2</sup> Dr. rer. nat., Jananne Khuri,<sup>1,2</sup> PhD, Jazmin Reyes-Portillo,<sup>1,2</sup> PhD, and Maria I. New<sup>3</sup>, MD

<sup>1</sup>NYS Psychiatric Institute, <sup>2</sup>Department of Psychiatry, College of Physicians and Surgeons of Columbia University, and <sup>3</sup>Department of Pediatrics, Mount Sinai School of Medicine

## Cleft lip and palate: Parental experiences of stigma, discrimination, and social/structural inequalities

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Website:  
[www.amsjournal.com](http://www.amsjournal.com)

DOI:  
10.4103/2231-0746.200336

Quick Response Code:



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# How can the health system support children with disability from a birth defects?



- Address stigma and discrimination by advocacy in the community, health workers training
- Design models of care that include services that reach people close to where they live, by reducing barriers to accessing health services
- Develop financing packages that include health services for children with birth defects
- Ensure that health sector partners understand that birth defects are not disabilities, but that they are health conditions that contribute to disability

## In Summary.....

We have a challenging agenda in front of us with regard to birth defects, the proportion is only rising. This is becoming an increasing issue of relevance for LMICs

The global health community (including donors) need to recognize and acknowledge the magnitude of the contribution of birth defects relative to other common causes of under 5 mortality

We are all in this together though different stakeholders focus on different birth defects

But we do need a joined up strategic partnerships to accelerate progress in this area so that countries have an integrated approach forward.

We look forward to moving together with the community going forwards.



**Thank you!**