



The global challenges of surgical congenital anomalies: Evidence, models, and lessons

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ABSTRACT

The treatment of congenital anomalies (structural birth defects) is common to all the surgical sub-specialties dealing with children. Globally more than 90 % of all babies born with a congenital anomaly are born in middle-and-low-income countries where there is often limited access to needed surgical care. Challenges include lacks of early identification, registry and surveillance systems, missing referral pathways, shortage of trained surgical expertise and insufficient surgical infrastructure. Poverty, transportation logistics, financial constraints and social stigma are also serious barriers for families. There is, however, growing recognition of the priority to expand services, encouraged by the World Health Organization and other global players, and examples of successful models of care. Registry programs are growing, especially in Latin America. The Ponseti method of clubfoot care has been revolutionary on a global scale. The role of not-for-profit non-governmental-organizations has been instrumental in fundraising, training and logistical support as exemplified in the care of oro-facial clefts. Specialized “niche” hospitals are providing needed sub-specialist expertise. The way forward includes the need for effective partnerships, innovative methods to distribute care out from referral hospitals into the districts and the development of national plans embedded in national health policy.

Introduction

Congenital anomalies, or structural birth defects, constitute a significant component of the knowledge base and technical expertise of surgeons and anesthetists in all the subspecialties of pediatric surgery. The reality is that such sub-specialty expertise is severely under-represented in lower- and middle- income countries (LMIC's) where more than 90 % of all babies with birth defects are born and where access to even the most basic evaluation and surgical treatment services may be limited.¹ Birth defects account for almost 9 % of all under-5 mortality, worldwide² and those who survive often face a life of disability, stigma, isolation and even physical abuse. The resulting impact on the lives of children and families from premature death or lifetime disability can be profound. What can be done to bring equity in access to care and treatment to ensure that all children born with a birth defect, anywhere in the world, can reach their maximum health and life potential? What are the barriers to equitable care, and are there examples of potential solutions? The aim of this paper is to provide a discussion of the challenges posed by these questions followed by examples

of models and coalitions that are improving access to diagnosis and care globally.

Global context

Understanding the context of recent global developments will allow pediatric surgeons to make a case for commitment and allocation of funding and resources for not only surgical care of babies born with birth defects, but also for advancing birth defects surveillance, prevention and early detection. While many single gene defects manifest metabolically with progressive and permanent impact on healthy development that prove complex and difficult to treat medically, structural birth defects (e.g., clubfoot, oro-facial clefts and neural tube defects), if identified and treated surgically early in life, have potential to allow a child to enjoy complete and permanent cures.

The incidence of structural birth defects in different countries and regions varies significantly and our knowledge of the actual prevalence is complicated by lower resourced countries lacking comprehensive identification and reporting mechanisms and with significant numbers

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of births happening outside accredited health facilities. The presence or absence of causal factors including folate intake, maternal health and nutritional status, medication, vaccination, and maternal infection, vary drastically. A metanalysis of data from Sub-Saharan Africa showed the highest incidence in Southern Africa (43/1000) with musculoskeletal defects most prevalent.³

In 2010, the 63rd World Health Assembly (WHA) of the World Health Organization (WHO) passed a resolution on birth defects. The report by the WHO Secretariat to inform the WHA discussion stated, “Surgery is an important but largely unheralded component of the services required to treat children with birth defects” and named simple congenital heart defects, cleft lip and palate, clubfoot, congenital cataracts, and gastrointestinal and urogenital abnormalities as some of the conditions that are amenable to cost-effective surgical interventions.⁴ Meanwhile, the publications of the Lancet Commission on Global Surgery and the Disease Control Priorities of the World Bank on Essential Surgery in 2015 highlighted the fact that at least 1.7 billion children in the world do not have access to even basic surgical resources.⁵

The United Nations Sustainable Development Goals (SDGs), a set of 17 global goals for human growth and development to be achieved by 2030, also serve as a tool for advocating for surgery for birth defects.⁶ The third SDG (SDG3) aims to end preventable deaths of children under 5 years of age and aims to promote universal health coverage (UHC). Many birth defects are life threatening and universal access to surgical care for serious birth defects is needed to end both preventable childhood death and preventable disabling impairment by 2030.

Challenges

The 2010 WHA Birth Defects Resolution urged member nations to accelerate birth defects care, prevention, screening and surveillance.⁴ While laudable, actual implementation of action for birth defects at national and global levels is fraught with lack of policy, resources and commitments.

As we write this paper, 13 years have passed since the above-mentioned WHA resolution and much work remains. Serious and stark disparities exist globally in terms of achieving universal access to surgical treatment for children born with birth defects. The surgical, anesthetic and nursing care expertise required to care for infants with serious birth defects are some of the most sophisticated skills to teach and master. In lower income environments the shortage of skilled healthcare workers and the lack of infrastructure and resources, such as technology, surgical instruments, supplies, operation theatres, intensive care units and rehabilitation facilities, mean that rare specialized surgical and anesthetic care personnel and facilities are largely located in urban centers, remote from the majority of the population.

A significant barrier to universal surgical access for birth defects is cost. Lower income countries lack robust universal healthcare coverage that supports surgical procedures that are considered expensive, and the private health insurance sector is underdeveloped. Ministries of Health have limited budgets to provide specialty services and concentrate their investment on the larger issues of communicable and common non-communicable diseases that are responsible for the large proportion of childhood mortality. Economically disadvantaged families seeking essential surgical and rehabilitative care for their children face the obstacles of distance, cost of transportation, and time away from work and domestic duties. They are then often required to pay for needed medical supplies such as gloves, needles and bandages. For many children and families, distance, hardship, and cost of accessing the specialized care for their children are prohibitive. Economic studies have also shown that paying for their healthcare further impoverishes the population.⁷ The result is that children who survive with structural birth defects are often left behind, facing stigma, abuse and isolation, leading to lives of further impoverishment and exclusion.

Prioritization of care for children born with structural birth impairments is hampered by lack of surveillance data. The development of



Fig. 1. Teenager with uncorrected clubfeet. Painful feet with ulcerations preventing walking and excluding school access. The deformity is now much more difficult to correct surgically with less effective result than with early intervention.

(Photo courtesy of Dr. S. Mannion).

birth defect registries is encouraged by the World Health Organization and is the mainstay of their genomics program and of epidemiological research. A Birth Defects Surveillance Toolkit has been published in collaboration with the US Centers for Disease Control and Prevention.⁸ To date, however, few low-income countries have robust reporting systems for birth defects with many births occurring in the community outside hospital services hampering data collection.³ Attempts to utilize registries to inform referral and treatment are a relatively new undertaking but have proven problematic even in developed countries because of barriers of confidentiality and resource support.⁹ The barriers to implementation of population based surveillance systems in lower resource countries are likely to be even more problematic.

When the challenges of early identification, referral and treatment are not met, the result is increased mortality from the primary impairment but also from vulnerability to intercurrent illnesses, infections and malnutrition. Physical impairments become more entrenched resulting in protracted disability impacting functioning, daily lives and psychologic well-being. Families face economic burdens as they attempt to access health resources. Social stigma and discrimination result in exclusion from community activities and education exacerbating the disability/poverty cycle.¹⁰ If surgical services are eventually sourced late in the day the surgery is more complex with higher complication rates and less efficacious end result (Fig. 1).

Successful models

While there are numerous barriers and challenges are there examples of successful implementation strategies of surveillance and care of birth impairments?

Surveillance and early detection

An example of a successful registry is the Latin American network of congenital malformation surveillance: ReLAMC. This network of six countries in Latin America has supported development of national registries of birth defects, combining their data to allow comparative frequencies in similar populations.¹¹ This has resulted in surveillance of more than 9 million births in 3 years with identification of more than 100,000 babies with congenital anomalies. Data from this network has been powerful for both advocacy and implementation of national strategic plans.

An efficient early detection and referral system is critical for ensuring

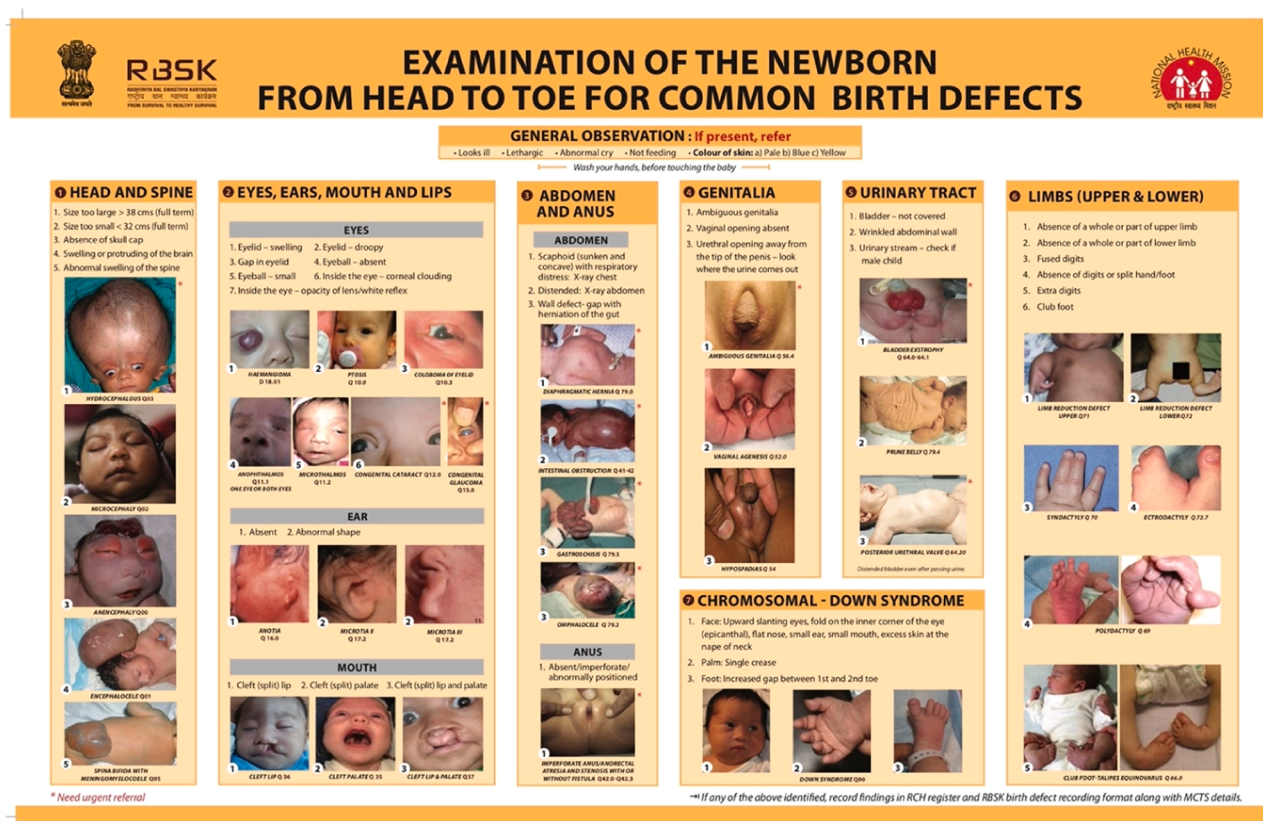


Fig. 2. The birth defects illustrated sheet used by ASHA's in the RBSK program.

timely treatment of birth defects that can be corrected through surgical interventions. The Rashtriya Bal Swasthya Karyakram (RBSK) program of the National Health Mission in India is ostensibly the best example of a grassroots national initiative with the potential to impact the identification, referral and treatment of children with birth defects in a lower resource country. This program trains community health workers, called ASHAs, to identify children in their communities with birth defects, disabilities, diseases and developmental delays, the 4D's. The program seeks to bring awareness and identification at community level, and facilitate referral to early intervention centers. A specific national curriculum has been developed, with visual materials, guiding community workers in the identification of common identifiable anomalies (Fig. 2). One study of RBSK from Ahmedabad, India, included a cohort of 102 children identified of which 43 % had birth defects.¹² The most common defects were congenital heart defects, clubfoot, cleft lip and palate, congenital deafness and Down syndrome. Appropriate cases were referred for surgical treatment including cochlear implants. While the program aims are laudable, the logistics of implementation in a country as populous and vast as India has been problematic.¹³ The program, however, is optimistic and can potentially be used as a template for national initiatives in other countries.

Taking advantage of the dramatic uptake of mobile phone technology in developing countries, the ZikaPLAN Project, consisting of a collaborative international expert group formed as an outcome of the Zika virus epidemic, has produced an innovative mobile app for surveillance with an atlas of 98 congenital anomalies identified at birth.¹⁴ It is anticipated that its use by fieldworkers will improve identification and surveillance of birth defects.

Better identification and surveillance, however, must lead to better care to be considered efficacious and even ethical. A "triple surveillance" approach has been advocated, integrating public health with surveillance, in the interests of enhancing clinical care.¹⁵ A particular challenge is how to identify structural birth defects not immediately evident by

visual inspection. Developmental dislocation of the hip, for example, is a common birth defect that results in serious permanent impairment if not identified and treated in the neonatal period but requires expertise in specific clinical tests (Barlow and Ortolani maneuvers) that are difficult tests not easily taught and not easily mastered by non-specialists. Universal screening of infants for congenital deafness requires availability of automated auditory brainstem response and otoacoustic emissions technology with associated training. In India the need for congenital deafness surveillance has been recognized and a conceptual framework developed and advocated, yet there is still no universal screening program in place.¹⁶

Care initiatives

Examples of successful efforts in the treatment of structural birth defects include treatment of talipes equino-varus (clubfoot) using the Ponseti technique, surgical treatment of oro-facial clefts, congenital hydrocephalus, congenital cataract, and congenital heart defects amongst others. Congenital talipes equino-varus is the most common birth defect causing locomotor impairment and has resulted in disability for millions of children globally. In recent years clubfoot treatment programs utilizing the Ponseti principles have become the gold standard of how to implement awareness and capacity building on a national scale using public health principles. The Uganda Clubfoot Project and Uganda Sustainable Clubfoot Care Project were developed and implemented at the turn of the millennium and consisted of an intense awareness program, curriculum development, and training.¹⁷ Task shifting to non-physician providers facilitated implementation at community level. The result was a national plan embedded in the national health policy of Uganda. The principles of the Uganda projects have now been implemented in numerous countries worldwide with great success and have led to development of the Global Clubfoot Initiative, a collaboration of more than 35 organizations.¹⁸ Key principles include

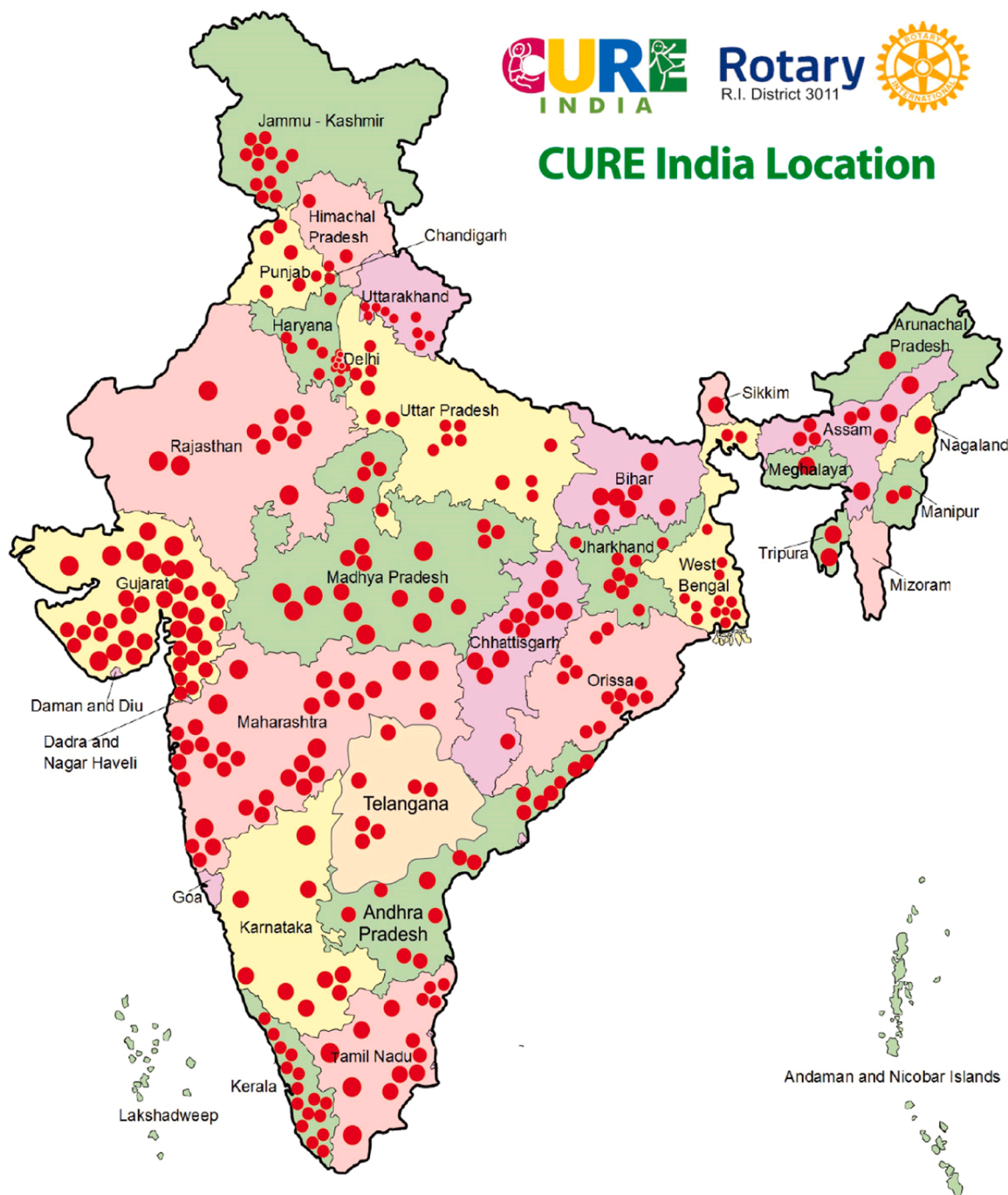


Fig. 3. Program map from Cure India Clubfoot Trust showing clubfoot treatment clinics distributed in all Indian states.

the development of a standardized curriculum that can be implemented throughout the health sector, with capacity building of health personnel from birth attendants to primary care practitioners to subspecialized orthopedic surgeons. The methodology has proven to be effective in many low resource countries.¹⁹ India has implemented the program on a statewide basis (Fig. 3). Data from a supporting non-governmental organization (NGO), Cure Clubfoot India Trust, shows more than 90,000 children have been treated in India alone by this one organization since 2009.²⁰ Another NGO, MiracleFeet has enrolled over 80,000 children in clubfoot treatment in over 30 LMICs since its inception in 2010.²¹

The surgical treatment of oro-facial clefts is another example of a successful strategy for treating a structural birth defect. Two models of care have been widely implemented, the drop-in model and local capacity model; well-resourced missions of expatriate expertise working

with local partners providing brief high volume surgical camps while other organizations provide funding on a per-case basis to local surgeons and support local infrastructure for surgical care.²² One collaborating NGO reports having supported cleft care for more than 1.5 million children.²³

Common to these successes, and others, is the involvement of not-for-profit NGOs, bringing in training, technical and financial resources from the global north to the global south in the spirit of collaboration. This includes academic institutions and professional organizations that have also actively influenced the global agenda for birth defects care, prevention and surveillance. The Global Initiative for Children's Surgery, a coalition of pediatric surgical care providers from around the world, has developed a guideline document, Optimal Resources for Children's Surgery, in the interest of improving the delivery of surgical

care for children in LMICs.²⁴ The biannual International Conference on Birth Defects and Disabilities in the Developing World (ICBD), with support from the March of Dimes and other global partners, brings together pediatric, genetic and surgical care leaders, as well as policy professional and parent groups, focusing on improving surveillance, diagnosis and pathways of care for children born with birth defects.²⁵

Discussion

Equitable access to safe and timely surgical care without financial hardships for all children born with a structural birth defect anywhere in the world, is an aspirational goal. This can only be accomplished through consensus and collaboration between national governments, professional and academic institutions, NGOs, and global influencers.

The success in improving access to effective treatment programs for clubfoot and oro-facial clefts demonstrates that it is possible to make surgical care widely accessible through a public health system model in collaboration with national Ministries of Health, that includes not just specialist referral centers but district hospitals and community health units, with strong support from communities themselves and with significant logistical and financial support from NGOs. Task shifting to train healthcare workers in districts and communities to perform specialized procedures, whenever possible, is key to improving access to care.

The proven efficacy of the Ponseti method of clubfoot treatment spawned a plethora of philanthropic NGOs (e.g., MiracleFeet) supporting and sustaining treatment in under-resourced countries where national health systems did not have capacity. The success of wide use of the Ponseti method lies in the fact that clubfoot treatment with this method can be performed successfully by various levels of specifically trained non-specialist providers at district level in a distributive model of care and complex surgical procedures are not required. Clubfoot treatment, therefore, is a “low hanging fruit” in the context of structural birth defects. The challenge of task shifting with most other structural birth defects is whether more general practitioners can be trained to do some of the required complex surgical procedures at district level. A significant limiting factor in making surgical treatments more accessible for some birth defects at district level is limited access to safe anesthetic services. As such, specialized facilities with adequate resources and trained staff would need to be established in health facilities other than the main academic centers.

While such facilities should ideally be established within the public health delivery systems, the contribution of not-for-profit “niche” hospitals, faith-based mission hospitals and other so called “charitable platforms” must be recognized.²⁶ Specific examples are the reconstructive orthopedic, plastic and neurosurgical hospitals represented by the CURE partnership, CoRSU Hospital in Uganda, the Foundation of Orthopedics and Complex Spine (FOCOS) in Ghana and the Hospital and Rehabilitation Center for Disabled Children (HRDC) in Nepal amongst others. These well-staffed and well-resourced hospital organizations, with their focused mandate, can undertake sophisticated reconstruction procedures for congenital limb deformities and other birth defects which would not be possible in overwhelmed public hospitals. The specialized children’s neurosurgical center in Uganda, for example, has performed large volumes of procedures for congenital hydrocephalus and myelodysplasia, proving the efficacy of endoscopic third ventriculostomy procedures in the African context.²⁷

An existing challenge is that some of the successful programs are vertical in nature, focusing on one impairment. It seems obvious that what is needed is comprehensive training of birth attendants in the recognition of all structural birth defects with appropriate comprehensive referral and treatment pathways. At the heart of a public health approach to care of congenital anomalies is the challenge to shift emphasis out of tertiary institutions and into the community. Innovative solutions are needed to reach and treat children and families in rural and remote communities with both technical and financial support.

Furthermore, a multi-disciplinary and multi-dimensional approach is needed to ensure that a child and his or her family have socio-economic and rehabilitation support before and after the surgical intervention to enable them to reach their maximum potential.²⁸

Conclusion

Structural birth defects are a health issue that is common to all the sub-specialty disciplines of pediatric surgery and anesthesia. Birth defects are common and account for significant childhood mortality and disability. Achieving global equity in birth defect surveillance, prevention and care is a major challenge with daunting barriers to overcome. Individual successful efforts have been established and could serve as templates for future programs. Collaborative multi-disciplinary efforts through public-private partnerships are needed above all there needs to be a shift towards a public health approach, moving out from tertiary institutions into the community and integrating prevention, identification, surveillance, referral and treatment of birth defects into the maternal newborn health agenda of the national health plans.

References

- Christianson A, Howson C, Modell B. *March of Dimes Global Report on Birth Defects: The Hidden Toll of Dying and Disabled Children*. White Plains, NY: March of Dimes Birth Defects Foundation; 2006.
- Perin J, et al. Global, regional, and national causes of under-5 mortality in 2000–19: an updated systematic analysis with implications for the sustainable development goals. *The Lancet Child and Adolescent Health*. 2022;6(2):106–115. [https://doi.org/10.1016/S2352-4642\(21\)00311-4](https://doi.org/10.1016/S2352-4642(21)00311-4).
- Adane F, Afework M, Seyoum G, Gebrie A. Prevalence and associated factors of birth defects among newborns in sub-Saharan African countries: a systematic review and meta-analysis. *Pan Afr Med J*. 2020;36:19.
- World Health Organization. Sixty-third world health assembly (A63/10). April 2010. https://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_10-en.pdf.
- Mullapudi B, et al. Estimates of number of children and adolescents without access to surgical care. *Bull World Health Organ*. 2019;97:254–258.
- United Nations: department of economic and social affairs. <https://sdgs.un.org/goals>. Accessed November 21, 2023.
- McIntyre D, Thiede M, Dahlgren G, Whitehead M. What are the economic consequences for households of illness and of paying for health care in low- and middle-income country contexts? *Soc Sci Med*. 2006;62(4), 2005.07.001. Epub 2005 Aug 15.
- World Health Organization. 2023 <https://www.who.int/news/item/02-12-2020-new-toolkit-aims-to-improve-global-birth-defects-surveillance>. Accessed April 3, 2023.
- Farel A, Meyer R, Hicken M, Edmonds L. Registry to referral: using birth defects registries to refer infants and toddlers for early intervention services. *Birth Defects Res A Clin Mol Teratol*. 2003;67(9):647–650.
- Shetty N, et al. Unravelling the challenges: a critical review of congenital malformations in low socioeconomic strata of developing countries. *Cureus*. 2023;15(7):e41800.
- Orioli IM, et al. The Latin American network for congenital malformation surveillance: reLAMC. *Am J Med Genet*. 2020;184C:1078–1091.
- Sharma R, et al. Profile, quality of life and client satisfaction of beneficiaries registered under Rashtriya Bal Swasthya Karyakram (RBSK) at an urban primary health center, Ahmedabad: a retrospective cohort study. *Indian J Community Med*. 2022;47:567–570.
- Chakraborty S, Chakraborty A, Mitra S, Gupta S, Lahiri A, Banerjee N. Evaluation of the rashtriya bal swasthya karyakram (RBSK): a national children healthcare program in a health district of West Bengal, India. *Indian J Public Health*. 2022;66:307–312.
- Dolk H, et al. Global birth defects app: an innovative tool for describing and coding congenital anomalies at birth in low resource settings. *Birth Defects Res*. 2021;113(14):1057–1073.
- Botto LD, Mastroiacovo P. From cause to care: triple surveillance for better outcomes in birth defects and rare diseases. *Eur J Med Genet*. 2018;61(9):551–555.
- Ansari MS. Implementation of national screening for hearing programs in India: challenges, opportunities and suggested framework. *Indian J Otolaryngol Head Neck Surg*. 2022;74(Suppl 3):4150–4158. Dec.
- Pirani S, Naddumba E, Mathias R, et al. Towards effective ponseti clubfoot care the Uganda sustainable clubfoot care project. *Clin Orthop Relat Res*. 2009;467:1154–1163.
- Owen RM, Penny JN, Mayo A, Morcuende J, Lavy C. A collaborative public health approach to clubfoot intervention in 10 low-income and middle-income countries: 2-year outcomes and lessons learnt. *J Pediatr Orthop B*. 2012;21(4):361–365.
- Shabtai L, Specht S, Herzenberg J. Worldwide spread of the Ponseti method for clubfoot. *World J Orthop*. 2014;5(5):585–590.
- Cure clubfoot India trust. <https://clubfootindia.in>. Accessed March 20, 2023.

21. MiracleFeet (2022). <https://www.miraclefeet.org/about-us/our-model>. Accessed November 21, 2023.
22. Smerica A, Hamilton S, Dibbs R, Ferry A, Hollier L. Smile train: a sustainable approach to global cleft care. *J Craniofac Surg*. 2022;33(2):409–412.
23. SmileTrain: <https://www.smiletrain.org/about-us>. Accessed April 2, 2023.
24. Goodman LF, et al. The global initiative for children's surgery: optimal resources for improving care. *Eur J Pediatr Surg*. 2018;28:51–59.
25. ICBD <https://www.marchofdimes.org/our-work/icbd-conference>. Accessed June 16, 2023.
26. Shrim M, Sleemi A, Ravilla T. Charitable platforms in global surgery: a systematic review of their effectiveness, cost-effectiveness, sustainability, and role training. *World J Surg*. 2015;39:10–20.
27. Warf B, Wright E, Kulkarni A. Factors affecting survival of infants with myelomeningocele in Southeastern Uganda. *J Neurosurg Pediatrics*. 2011;7, 000–000.
28. Smythe T, et al. Provision of rehabilitation for congenital anomalies. *Bull World Health Organ*. 2022;100:717–725.