Strengthening capacity for newborn screening, diagnosis and management of birth defects

WHO technical consultation on planning and implementing services integrated into national health systems in low- and middle-income countries

World Health Organization

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Foreword

Over the past two decades, remarkable progress has been made in reducing child mortality, particularly from infectious causes. As a result of this success, birth defects now account for an increasing proportion of neonatal and underfive deaths, as well as a substantial burden of lifelong disability, especially in low and middleincome countries. Addressing birth defects is therefore no longer optional; it is an essential component of equitable, peoplecentred health systems and of the global commitment to universal health coverage.

Recognizing this imperative, the World Health Organization convened a series of technical consultations to examine how countries can strengthen newborn screening, diagnosis, management and longterm care of birth defects, fully integrated into routine health services. This work is firmly aligned with the priorities of the WHO Fourteenth General Programme of Work, which emphasizes equity, impact and resilience across the life course.

This report synthesizes the collective experience of frontrunner countries that have initiated stateled programmes at scale. Their experiences underscore a clear and consistent lesson: newborn screening must be embedded within a functioning continuum of care. Early detection must be linked to timely diagnostic confirmation, effective treatment, structured followup, rehabilitation services and sustained support for families. Fragmented or standalone screening approaches are insufficient to deliver meaningful health gains.

Equity and human rights are central to this agenda. Every child, regardless of place of birth or socioeconomic circumstance, has the right to a healthy start in life. Integrating services for birth defects into national health systems helps reduce preventable mortality and disability, protects families from catastrophic health expenditure, and promotes social inclusion across the life course.

I would like to express my sincere appreciation to the ministries of health, experts, clinicians, researchers, civil society partners, WHO colleagues and, importantly, individuals and families with lived experience who contributed their time, knowledge and insights to these consultations. Their collective commitment has shaped a practical, countryinformed framework to support Member States in strengthening sustainable, inclusive and effective programmes – ensuring that all children not only survive but thrive.

Pascale Allotey

Director

Department of Sexual, Reproductive, Maternal,
Child and Adolescent Health and Ageing

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Abbreviations

CH	congenital hypothyroidism
CHD	congenital heart defect
DEIC	district early-intervention centre
IMR	infant mortality rate
LMIC	low- and middle-income countries
NBS	newborn screening
NGO	nongovernmental organization
PKU	phenylketonuria
SCD	sickle cell disease
RBSK	Rashtriya Bal Swasthya Karyakram
UMIC	upper-middle-income country
WHO	World Health Organization

Executive summary

As low- and middle-income countries (LMICs) succeed in reducing child mortality from infectious causes, birth defects now account for a proportionally growing share of deaths among children under five years of age. The 2024 estimates from the United Nations Inter-Agency Group for Child Mortality Estimation show that between 2000 and 2023 the proportion of underfive deaths attributable to birth defects increased from 1% to 4% in sub–Saharan Africa, and from 3% to 11% in South Asia. Beyond mortality, these conditions impose a substantial, often underrecognized, burden of morbidity that infants and children endure throughout childhood and into later life, along with broader impacts on individuals, families and communities.

Hearing aid fitting for a child with hearing impairment ©Starkey foundation

Integrating newborn screening (NBS), together with systems for the diagnosis and management of birth defects, into national health systems is essential for addressing birth defects, which are now an increasingly significant cause of mortality and morbidity among newborns, infants and children. It also upholds every child's right to a healthy start and ensures equity in access to care. This integration is an important component of universal health coverage.

Most high-income countries have well-established NBS programmes for birth defects, supported by well-defined diagnostic and management pathways. A few 'front-runner' LMICs have recently initiated large, state-led initiatives to strengthen screening, diagnosis and management of birth defects among newborns and children. It is important for health ministries in LMICs to begin prioritizing birth defects within their health systems, as 90% of children born with serious birth defects reside in LMICs, and 95% of birth defect related deaths occur in these contexts.

The increasingly high priority given to addressing birth defects is reflected in the recent Seventy-seventh World Health Assembly resolution, Agenda item 11.7 (WHA77.5), which calls for accelerating progress towards reducing maternal, newborn and child mortality in order to achieve Sustainable Development Goal targets 3.1 and 3.2. This resolution also invited Member States to consider implementing universal NBS, diagnosis, management and long-term care of children with birth defects.

Aligned with the priorities set out in this resolution, the World Health Organization (WHO) convened a series of technical consultations focused on state-led initiatives for NBS, diagnosis and management of birth defects integrated into routine health services in LMICs. These consultations were held over five rounds between September 2024 and April 2025.

This series of global consultations consolidated experiences from front-runner LMICs on their implementation of large national/subnational state-led initiatives for NBS, diagnosis and management of one or more birth defects. The purpose of these consultations was to examine and consolidate models established within LMIC health systems that address birth defects among newborns and children, and to support other interested LMICs in planning similar initiatives. Parents and persons affected by birth defects in LMICs also shared their experiences, participated in discussions, and provided views and suggestions for care in their settings. The consultations were also attended by clinicians, academics, social scientists, programme staff, epidemiologists and nongovernmental organizations (NGOs) working on birth defects.

Purpose of the consultations

The purpose of these consultations was to develop a practical, countryinformed operational framework – drawn from state-led models in frontrunner LMICs – to support programme managers and ministries of health in other LMICs in planning and implementing NBS, diagnosis, management and followup services for infants with birth defects within national health systems.

The specific objectives of the consultations were to:

(i) Drive technical exchange and consolidate learning on stateled models for NBS, diagnosis and management of priority birth defects within national health systems with ministries of health from front-runner low, middle, and selected uppermiddleincome countries.

(ii) Map and analyse the full continuum of care – from prioritizing conditions to screening, diagnostic confirmation, clinical management, follow-up and financing – and identify practical, scalable approaches that LMICs can adapt within their own health systems, including at community level.

(iii) Integrate lived experience into programme design by systematically capturing and applying insights from individuals with birth defects and their families, ensuring that emerging frameworks are grounded in realworld needs, barriers and opportunities for peoplecentred care.

(iv) Cocreate an operational framework that supports programme managers and ministries of health to plan, implement and strengthen stateled NBS, diagnosis, management and followup services within routine health systems in low and middleincome settings.

The consultations were attended by 107 participants, including representatives from 11 ministries of health – seven from LMICs and four from UMICs. Country representatives presented their initiatives and health system models of care for NBS, diagnosis and management of birth defects. They described when and why programmes were initiated, how initial screening conditions were selected, what preparations were made within their health system and how programmes were funded at scale. Presentations also covered the involvement of various components of the health system across all levels; strengths and challenges from public–private partnerships in managing these conditions; development of data systems; follow-up mechanisms; and state-led arrangements for long-term care, including rehabilitation services and support for families.

Discussions among experts covered various issues such as the appropriate time for an LMIC to start a programme to address birth defects in newborns and children; practical considerations for selecting priority conditions; necessary health system preparations for different tracer conditions; the role of champions; the need for simple data systems; NBS in context of early hospital discharge of the mother–baby pair, and long-term care in LMICs. Family members of those affected by birth defects, as well as the affected persons themselves, also shared their experiences with care and the challenges related to stigma.

The expert dialogues that followed helped distil common principles, feasible entry points and crosscutting system challenges – such as financing gaps, workforce shortages and followup constraints. These insights shaped a framework focused on feasibility, phased implementation and healthsystem strengthening. Perspectives from individuals with lived experience helped the consultations maintain a peoplecentred approach, emphasizing communication, family support and inclusive service design. Together, these contributions enabled the translation of diverse country practices into this report, which aims to support programme managers and ministries of health in LMICs in planning and implementing NBS, diagnostic and management services within national health systems.

When treatment for clubfoot is initiated during infancy, correction is usually achieved within six to eight weeks (Philippines) ©MiracleFeet

Key messages from the consultations

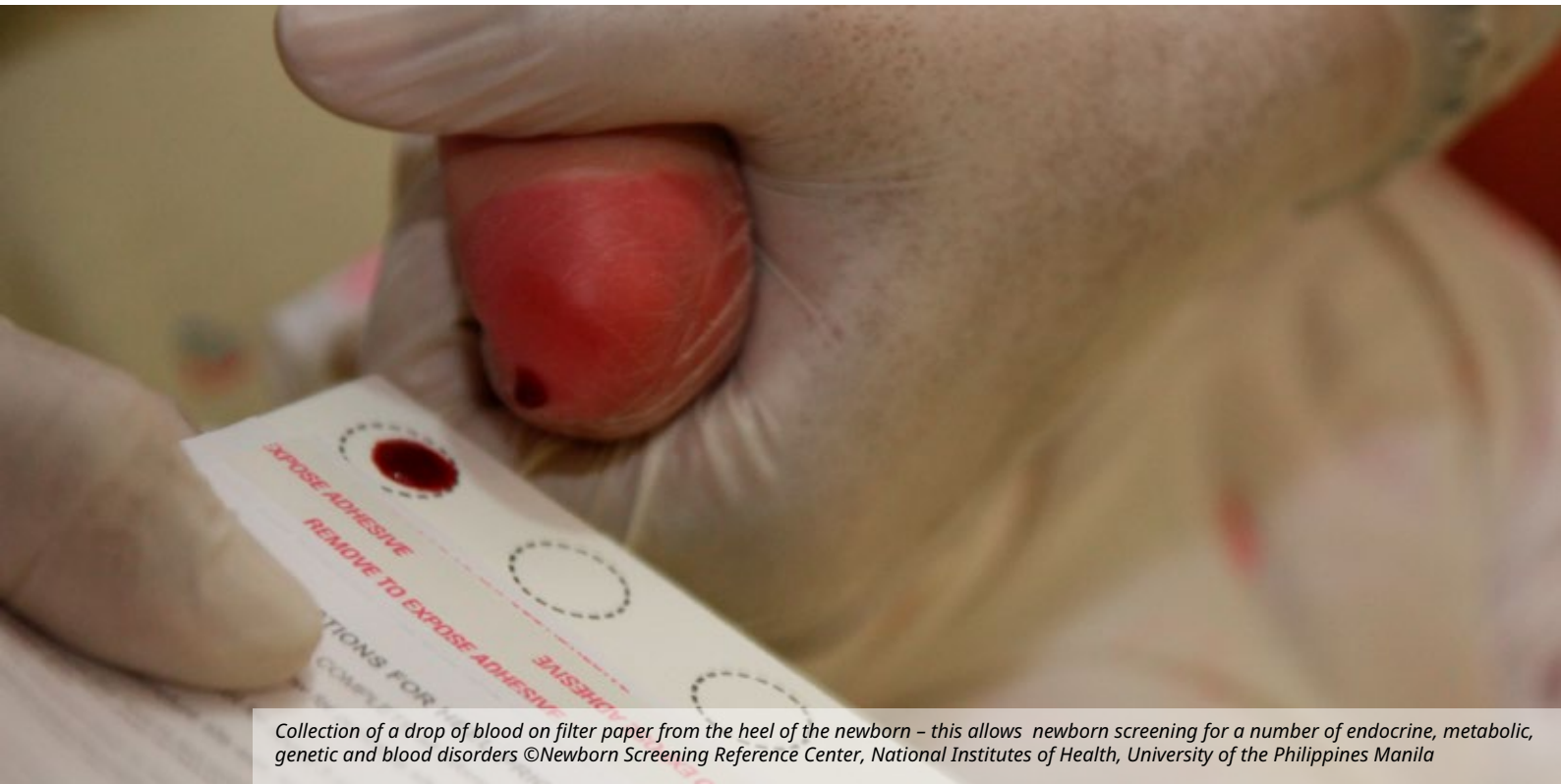
1. Integration of NBS, diagnosis and management of birth defects into national health systems has helped countries reduce preventable mortality and morbidity, advance equity and uphold a child's right to a healthy start. This aligns with recent World Health Assembly resolutions encouraging Member States to consider universal NBS and longterm care of children with birth defects.
2. As infectious causes of underfive mortality decline, birth defects represent a growing proportion of deaths and disability, particularly in LMICs. Countries have initiated programmes at different stages of socioeconomic development (of which infant mortality rate is a proxy measure), driven by academic pilots, champion-led efforts, professional advocacy and/or strong political commitment.
3. Birth defects are an extremely heterogeneous set of conditions. LMICs have adopted pragmatic entry points by limiting screening to a selected set of conditions that can be feasibly managed effectively within the country's health system. Successful programmes usually start with a limited set of conditions and expand as capacity grows. Screening is not an end in itself; a recurring principle that emerged from countries was that conditions were included only when treatment and care pathways existed in the country's health system. While a number of formal criteria to guide the initial selection of conditions emphasize (a) disease burden, (b) feasibility of screening, diagnosis and management within the health system, and (c) affordability, local realities – including advocacy, political interest and expert professional judgment – have significantly shaped the selection of conditions to prioritize in LMICs.
4. Successful LMIC programmes mapped the feasibility of the continuum of care for each prioritized condition to prepare the health system before national scale-up. For each condition, feasibility assessments covered screening modality, confirmatory diagnosis, referral pathways, treatment capacity (medical, surgical, assistive technology), and followup. An operational framework to review the alignment between health system capacity and initial condition selection was developed during the consultations (Fig. 1, page xiv).
5. Successful LMIC NBS programmes have institutionalized short and longterm followup. Shortterm followup ensures confirmatory testing, diagnosis and linkage to care; longterm followup secures linkages to disease-related services, and monitors treatment adherence, development and rehabilitation. Responsibilities are defined and shared between families, birth facilities, the NBS programme, primary care staff (including community health workers) and specialists.
6. Simple, purposeful data systems are established at programme inception and gain sophistication over time. These systems track newborns/children through screening, shortterm followup, diagnosis, management and longterm followup. Data systems are a key element of programme functioning to ensure newborns/children move through the continuum of care. They also support programme monitoring for quality and accountability. Some programmes have defined key performance indicators, such as coverage, recall rates, timetoconfirmation, treatment initiation, adherence, surgical outcomes. These data are used to optimize implementation, direct resources, redesign protocols and close gaps in the continuum of care.
7. Some LMICs planned and invested early in rehabilitation and inclusive services. For example, India's district early-intervention centres illustrate planned and funded services for early intervention and rehabilitation. Because children with disabilities face inequities and exclusion from routine services, the twintrack approach was emphasized, i.e. making routine services accessible to all children while providing specialized services to those children in need.
8. Emergency referral transport for urgent, life-threatening birth defects is critical – screening without safe referral, does not save lives. For lifethreatening defects (such as critical congenital heart defects), some LMIC programmes have invested in neonatalcapable ambulances, trained staff, standardized protocols and realtime monitoring.
9. Sustainable financing is central to universal NBS and care. Populationbased programmes for NBS are a core component of universal health coverage because they affect all newborns regardless of socioeconomic status. Strong programmes use national insurance or state

budgets to cover screening, diagnostic confirmation, treatment and followup, minimizing out-of-pocket payments. Reliance on outofpocket payments is inequitable and unsuitable for populationbased programmes. Donor support can complement government funding, but responsibilities of each partner must be clearly articulated and plans for transition to state ownership must be defined to ensure sustainability.

10. Legislation can help anchor national leadership. Some LMICs e.g. Philippines, have enacted laws mandating the provision of NBS at birthing facilities and embedding services into national insurance programmes. Legal mandates alone do not guarantee implementation; adequate financing, oversight and stakeholder engagement are also required. Several LMICs have also run successful programmes without legislation.
11. State-led multisectoral partnerships have been important in improving access to care and programme efficiency in many LMICs. Stateled coordination across public hospitals, private sector hospitals (in countries where this sector is large), NGOs, academia, parent groups, and other ministries, has accelerated scaleup and built sustainability. State-led public–private partnerships have helped expand specialized services (e.g. paediatric cardiac surgery), clarified costs, ensured the implementation of quality standards, and formalized data sharing. Integrated social support across health, social justice, education and local government has helped reduce fragmentation and strengthened inclusive care.
12. Affected persons and their families spoke at the consultations about facing stigma and social exclusion, as well as financial and psychological strain, with mothers disproportionately affected across countries. Addressing stigma and strengthening family support are integral components of birth defect programmes. Information, education and communication strategies, along with structured support services, enhance family resilience and improve outcomes for children.

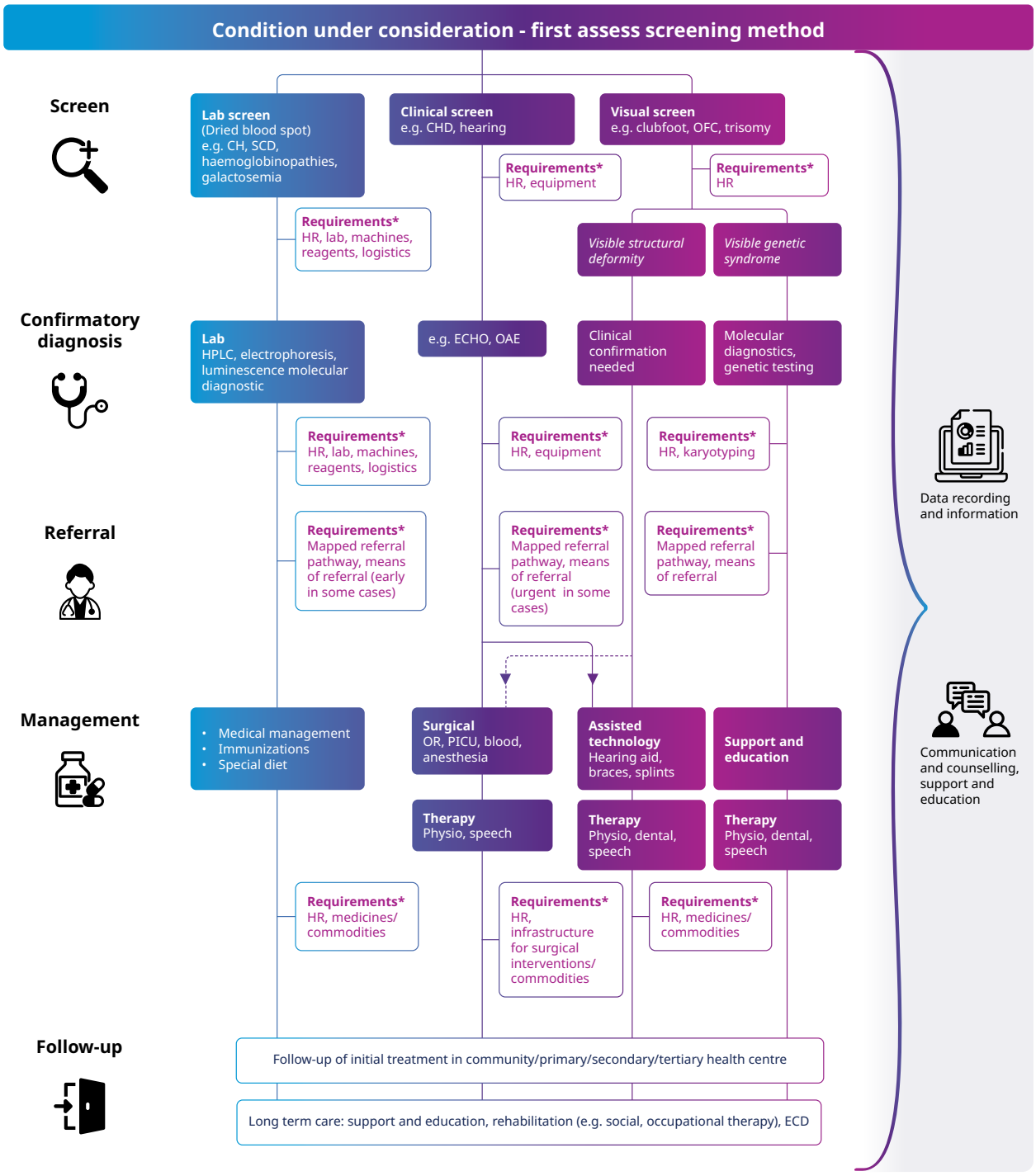
Operational framework derived from the consultations to assess health system feasibility and preparedness to provide services for newborn screening, diagnosis and management of one or more birth defects prioritized in a country

As one of the stated objectives, an operational framework was developed from the consultations, drawing on country programmes and insights synthesized from the country presentations and expert discussions across the five consultation rounds. These inputs provided realworld evidence on how frontrunner LMICs have introduced and scaled-up NBS, diagnosis, management and followup for birth defects. The framework provides a practical, visual screening algorithm (Fig. 1) to assess the feasibility of the health system in providing services for a particular birth defect.



Collection of a drop of blood on filter paper from the heel of the newborn – this allows newborn screening for a number of endocrine, metabolic, genetic and blood disorders ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

Fig. 1. An operational framework to assess health system preparedness for newborn screening, diagnosis and management of a selected birth defect



CH: congenital hypothyroidism; ECD: early childhood development; ECHO: echocardiography; HPLC: highperformance liquid chromatography; HR: human resources; OAE: otoacoustic emissions; OFC: orofacial clefts; OR: operating room; PICU: paediatric intensive care unit; SCD: sickle cell disease. (requirements listed are not comprehensive, but indicative)*

As a starting point, the method of screening for a particular condition often indicates the requirements for pathways to diagnose and manage that condition within the health system. Broadly, three screening methods – (i) biochemical screening, (ii) clinical screening, and (iii) visual examination at birth for a selected birth defect of interest – help generate pathways that highlight areas where capacities need to be strengthened or integrated into the system and stimulate discussions on how such strengthening can be facilitated.

Programme managers can use the algorithm for any condition under consideration for prioritization. It enables them to identify and detail the specific capacities required in the health system for each part of the continuum of care, i.e. screening, diagnosis, referral, management follow-up and long-term care, for the condition of interest.

In summary, the experiences shared by front-runner LMICs highlight that meaningful progress in addressing birth defects requires sustained political commitment, careful prioritization and systemwide strengthening. Stateled, equityoriented initiatives – anchored in feasible care pathways, robust data systems, inclusive services and strong multisectoral partnerships – demonstrate that front-runner LMICs show improvements are possible, even in resource-constrained settings.

As more LMICs embark on or expand programmes for NBS, diagnosis, management and followup, these collective lessons from front-runner LMICs provide practical experience to support national efforts, promote South–South learning, and ensure that every child has the opportunity for a healthy start and the chance to reach their full potential.

A mother wishing her child well before surgery for his orofacial cleft, India ©Operation Smile

During the last stage of clubfoot treatment, children wear a foot abduction brace to maintain the feet in the proper position and prevent relapse (Senegal) ©MiracleFeet

1. Background

Birth defects – also known as congenital anomalies, congenital disorders or congenital conditions – are structural or functional abnormalities that develop during intrauterine life and may be identified prenatally, at birth, or later in life. Worldwide, birth defects affect an estimated eight million newborns, (3–6% of global births), and account for almost 0.4–0.5 million deaths annually in children under five years of age *(1–3)*. However, beyond the high mortality caused by these conditions is an even greater and largely uncounted burden of morbidity. Infants and children often face lifelong impairments when these conditions are not detected early and managed effectively. This burden and mortality are highest in low- and middle-income countries (LMICs), where nine out of ten children born with a serious congenital disorder reside *(4)*.

Most high-income countries and upper-middle-income countries (UMICs) have well developed newborn screening (NBS) programmes, with clearly defined pathways for confirmatory diagnosis, management and long-term care of children with birth defects. The situation in LMICs has been somewhat different, with the primary focus of the last few decades being on reducing infant and child mortality from infectious causes by increasing vaccination coverage, improving water and sanitation, and enabling better access to care. These investments in maternal and child health services have contributed to a 52% decline in global under-five mortalities between 2000 and 2023 *(5)*. As LMICs succeed in reducing child mortality, birth defects now account for an increasingly larger proportion of child deaths. The 2024 estimates from the United Nations Inter-Agency Group for Child Mortality Estimation show that proportional mortality among children under five years of age due to birth defects increased from 1% to 4% in sub-Saharan Africa, and from 3% to 11% in South Asia between 2000 and 2023 *(5)*. This increase in proportional mortality also implies a rise in the substantial, but unmeasured burden of morbidity that infants and children bear during childhood and later in life.

In recognition of the growing importance of screening for diagnosing and managing birth defects as early as possible, a number of LMICs have initiated NBS services within their health systems over the past decade. These initiatives, in 'front-runner' LMICs (such as Egypt, India, Sri Lanka, the Philippines, Uganda) are led by national health ministries and operate at a national (or regional) scale, with pathways from screening, diagnosis, management to longer term care clearly articulated for a set of prioritized birth defects.

The rising priority given to addressing birth defects is reflected in the recent Seventy-seventh World Health Assembly resolution (WHA77.5, Agenda item 11.7) *(6),* which aims to accelerate progress towards reducing maternal, newborn and child mortality to achieve Sustainable Development Goal targets 3.1 and 3.2. The resolution also included an invitation to Member States to consider implementing universal NBS, diagnosis, management and long-term care of children with birth defects.

In response, WHO convened this series of global consultations to consolidate experiences from front-runner LMICs on the initiation and implementation of large national and subnational state-led initiatives for NBS, diagnosis and management of one or more birth effects. The goal was to support LMICs in planning, implementing and integrating state-led initiatives for NBS, diagnosis and management of one or more birth defects, into routine health systems. Besides health ministry representatives from LMICs (and some UMICs), individuals living with birth defects and their families, also shared experiences and provided suggestions for interventions to support their care. These contributions help to inform LMICs as they plan health system responses focused on NBS, diagnosis and management of one or more birth defects.

1.1 Consultation objectives

The **purpose** of these consultations was to develop a practical, countryinformed operational framework – drawn from stateled models in frontrunner LMICs – to support programme managers and ministries of health in planning and implementing NBS, diagnosis, management and followup services for infants with birth defects within national health systems.

The specific objectives of the consultations were to:

(i) Drive technical exchange and consolidate learning on stateled models for NBS, diagnosis and management of priority birth defects within national health systems with ministries of health from front-runner low, middle, and selected uppermiddleincome countries.

(ii) Map and analyse the full continuum of care – from prioritizing conditions to screening, diagnostic confirmation, clinical management, follow-up and financing – and identify practical, scalable approaches that LMICs can adapt within their own health systems, including at community level.

(iii) Integrate lived experience into programme design by systematically capturing and applying insights from individuals with birth defects and their families, ensuring that emerging frameworks are grounded in realworld needs, barriers and opportunities for peoplecentred care.

(iv) Cocreate an operational framework that supports programme managers and ministries of health to plan, implement and strengthen stateled NBS, diagnosis, management and followup services within routine health systems in low and middleincome settings.

Newborn screening for congenital hearing impairment using an otoacoustic emission test ©Otto Mejia

1.2 Target audience

This document is primarily intended for programme managers in ministries of health in LMICs who are designing and implementing services for NBS, diagnosis and management of birth defects, integrated into national health systems.

In addition, certain aspects of this report are relevant to other stakeholders at the country level, including nongovernmental organizations (NGOs), professional associations, the private sector and development partners.

1.3 Format of the consultation

This series of consultations was hosted over five virtual sessions (rounds) beginning in September 2024. Each round comprised two consecutive day-long sessions of four hours each.

Participants included representatives from 11 ministries of health across all WHO regions, persons affected by a birth defect, parents/caregivers, NGO representatives, clinicians (neonatology, paediatrics, paediatric surgery, physiotherapists, speech therapists), social scientists, epidemiologists and academia. A total of 107 experts from 33 countries contributed their experience and expertise. A detailed list of speakers and participants is provided in Annex 1.

There was also representation from WHO regional offices and related departments. In accordance with WHO policy, all participants submitted declarations of interest and confidentiality agreements before the consultation.

The broad focus of each round of the consultation is presented in Table 1.

Table 1. Focus areas for each round of the consultations

Round 1 **16–17 September 2024**	Focus: Presentations from different front-runner LMICs on services and programmes currently implemented in the country
Round 2 **28–29 October 2024**	Focus: Considerations while selecting initial target conditions for NBS, diagnosis and management in LMICs – burden and feasibility
Round 3 **2–3 December 2024**	Focus: Preparing health systems in LMICs based on the selected priority condition(s) for NBS, diagnosis and management, public–private partnerships, data systems, referral mechanisms, involving different levels of the health system, ensuring sustainability
Round 4 **10–11 February 2025**	Focus (day 1): Payment mechanisms for NBS, diagnosis and management of key birth defects in LMICs. State financing, national insurance, donor funding and sustainability risks Focus (day 2): Closing the loop – short- and long-term follow up
Round 5 **16–17 April 2025**	Focus (day 1): Rehabilitation services and improving access to health care for children with a disability Focus (day 2): Family support, stigma and advocacy

1.4 Consultative process and synthesis

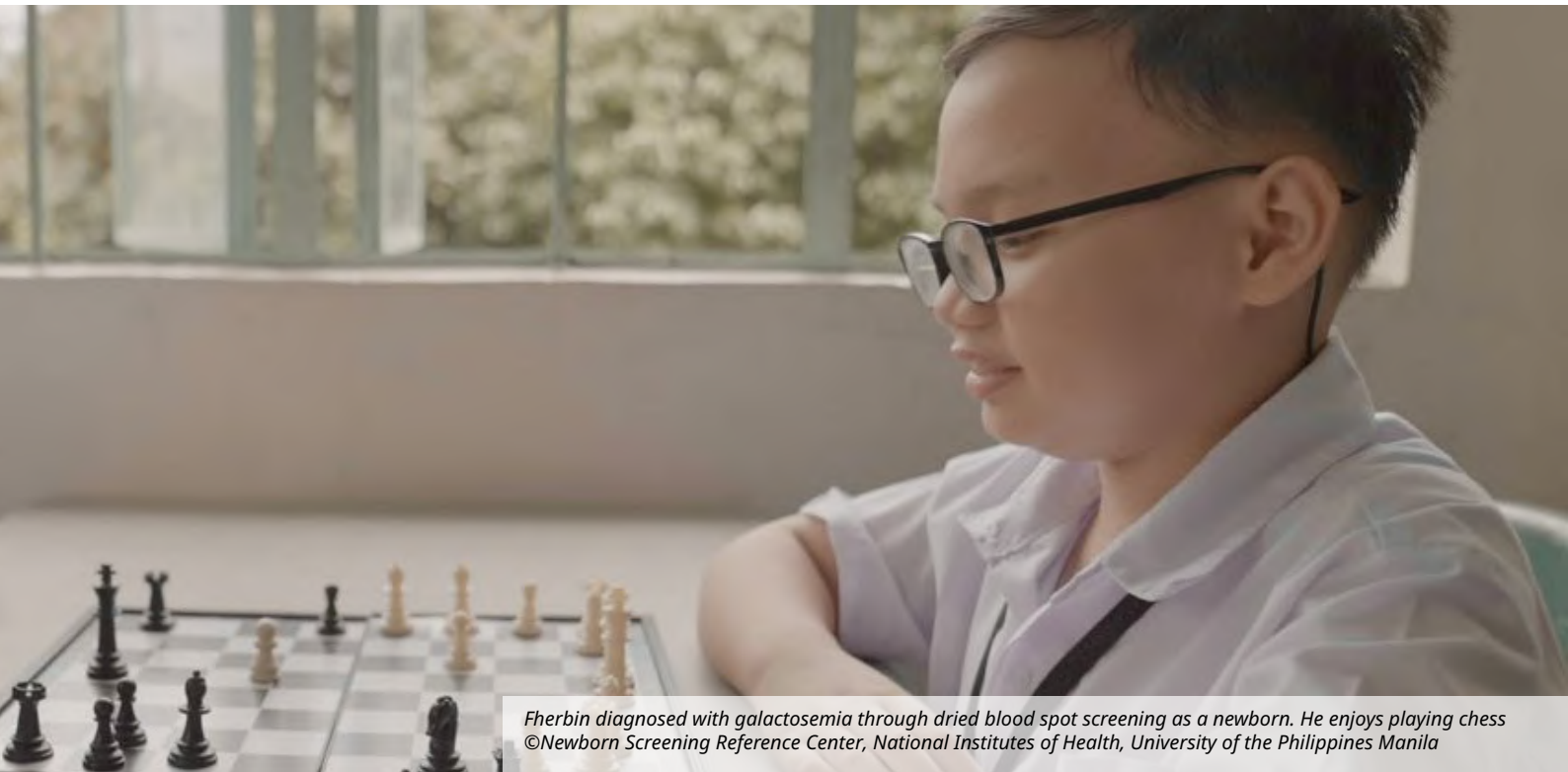
The operational framework presented in this document was shaped directly by practical programme experiences and insights consolidated from country presentations and expert discussions across the five rounds of consultations. These inputs served as the empirical foundation for the framework's structure, and key learnings documented here.

First, country presentations provided realworld models of implementation, describing how ministries of health in frontrunner LMICs introduced and scaled NBS, diagnosis, management and followup services for one or more birth defects. These presentations offered details on programme triggers, initial condition selection, preparedness assessments, governance arrangements, financing models, referral pathways, data systems and longterm followup mechanisms. By comparing diverse national approaches – from biochemical screening panels to clinical and visual examinationbased screening – the consultations generated a consolidated understanding of feasible entry points and strategies in varied resource contexts.

Second, the expert discussions that followed each presentation distilled common principles and operational lessons across countries. These exchanges examined the factors that enabled programme initiation, the ways countries addressed health system constraints, and the role of multisectoral partnerships in strengthening sustainability. The dialogue also identified crosscutting challenges – including financing gaps, human resource shortages, disparities in coverage and weaknesses in followup systems – which informed the framework's emphasis on feasibility assessment, phased implementation and systems strengthening rather than diseasespecific verticalization.

Third, perspectives from individuals with lived experience and families contributed critical insights into stigma, communication needs, and the burden of navigating fragmented systems. These testimonies helped maintain a peoplecentred orientation throughout the consultations, underscoring the importance of counselling, family support, inclusive service design, and longterm continuity of care.

Finally, the synthesis of these inputs enabled the translation of diverse country experiences into an operational framework to support health managers in LMICs planning the implementation of NBS, diagnosis and management of one or more birth defects within national health systems. The developed framework draws on the patterns, requirements and decision points repeatedly observed across settings – such as the need to begin with a small set of conditions, the alignment of screening, diagnostic and management modalities with healthsystem capacity, the centrality of referral and data systems, and the integration of rehabilitation and disabilityinclusive services.



Fherbin diagnosed with galactosemia through dried blood spot screening as a newborn. He enjoys playing chess
©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

2. Setting the scene

Dr Anshu Banerjee, Director of the WHO Department of Maternal, Newborn, Child and Adolescent Health and Ageing, underscored the need for stronger prioritization of birth defects in LMICs, aligning with the Seventyseventh World Health Assembly resolution encouraging countries to consider universal NBS, diagnosis, management and longterm care of children with birth defects to advance Sustainable Development Goal targets, 3.1 and 3.2. He emphasized that screening must be embedded within a full continuum of care, including surgical and nonsurgical treatment, longterm management and followup. Although addressing birth defects comprehensively requires a lifecourse approach – preconception, pregnancy and newborn/child interventions – this consultation focuses on supporting countries to plan and integrate NBS, diagnosis and management into routine health systems.

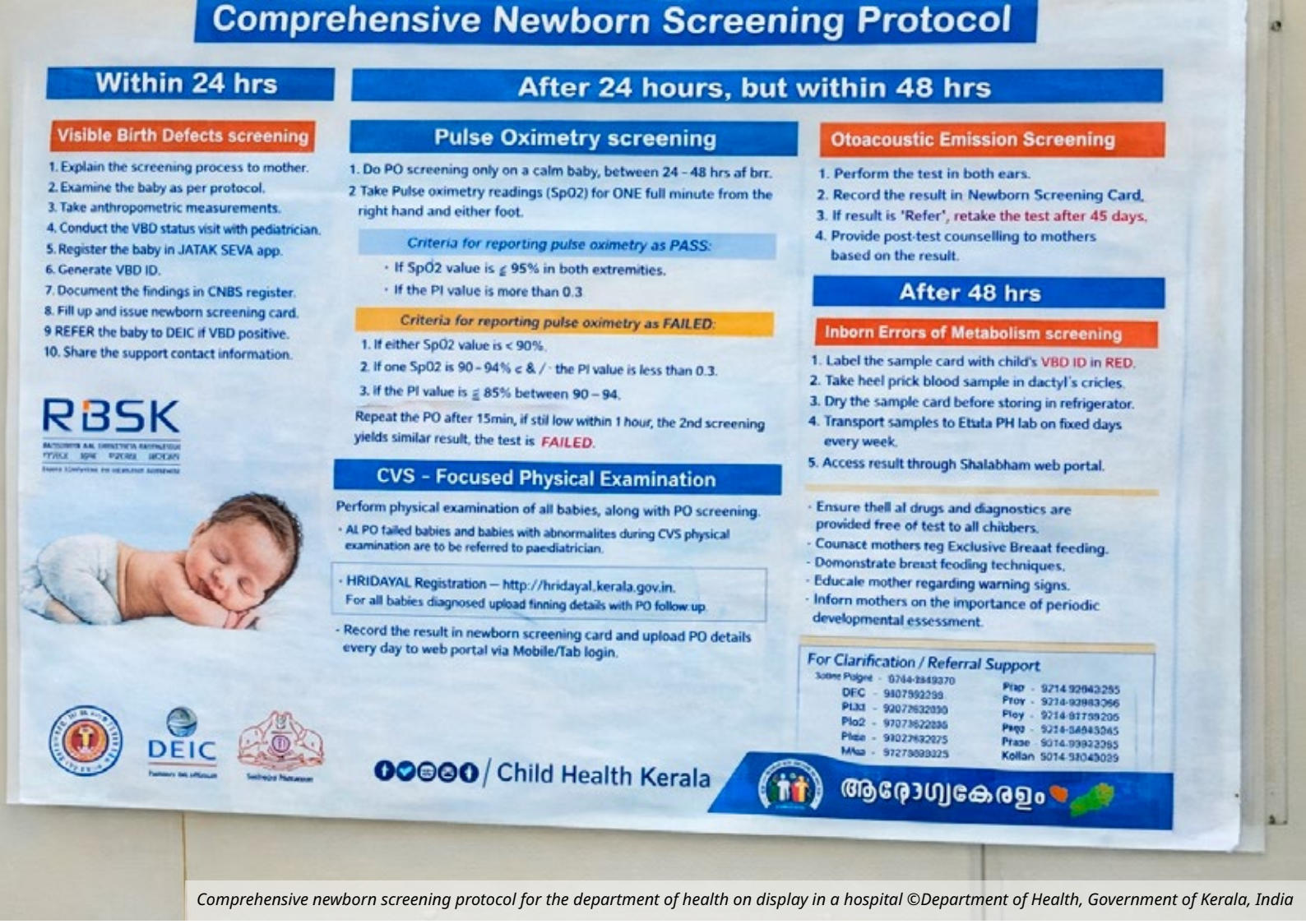
2.1 Framing the consultation

Dr Ayesha De Costa, Scientist at the same department framed the consultation. In addition to the Seventyseventh World Health Assembly resolution encouraging Member States to consider implementing comprehensive NBS programmes, focusing on diagnosis, management and long-term care of children with birth defects, an earlier resolution was passed by the World Health Assembly in 2010 *(7)*. That resolution urged WHO to support Member States with initiating and monitoring screening programmes, integrating approaches for the prevention and care of birth defects into primary health care, identifying successful models of care that can be applied in LMICs, promoting international cooperation and building expertise and capacity in newborn and child health.

The aim of these consultations was to bring together representatives from ministries of health in front-runner LMICs (and a few UMICs) to present their state-led models for NBS, diagnosis and management of one or more birth defects, integrated into routine health systems. The consultations also sought to understand these initiatives in terms of drivers, preparations, implementation, achievements and challenges, and to discuss difficulties and possible solutions relevant for LMICs.

The rationale for addressing birth defects in LMICs: As child survival improves in LMICs through better control of infectious diseases and their associated mortality, birth defects account for an increasingly larger proportion of under-five mortalities, as well as unmeasured morbidity. The 2024 report by United Nations Inter-Agency Group for Child Mortality Estimation *(5)* suggest that proportional mortality among children under-five years of age from birth defects, has increased from 1% to 4% in sub-Saharan Africa, and from 3% to 11% in South Asia between 2000 and 2023 *(5)*. LMICs also face a unique set of challenges including a ‘double burden’ of childhood disease (being in the epidemiological transition), competing health priorities, limited resources and nascent health insurance systems to support universal health coverage. Additionally, birth defects are an extremely heterogeneous set of conditions that do not easily lend themselves to a conventional disease programme structure.

Despite these challenges, addressing NBS, diagnosis and management of birth defects within national health systems is increasingly important given that in 2006, 90% of children born with serious birth defects are born in LMICs and most deaths of children with birth defects also occur in these settings *(4)*. Mortality for the same birth defect is manifold times higher in LMICs compared to high-income settings largely because of underprepared and resource constrained health systems *(8)*. Maternal undernutrition or suboptimal rubella immunization rates, still seen in some LMICs, are important contributory factors for birth defects in the newborn. Consanguineous marriages are also more common (>20%) in some settings in parts of Asia and North Africa *(9)*.



Comprehensive newborn screening protocol for the department of health on display in a hospital ©Department of Health, Government of Kerala, India

Besides the affected individual, the family of a child born with a birth defect is likely to experience loss of income and face social stigma from an uninformed community. It is therefore important for health ministries in LMICs to address birth defects in children.

While the challenges mentioned above remain, some LMICs have already recognized the importance of identifying and managing birth defects in their youngest citizens. These countries have embarked upon state-led initiatives at scale, to address one or more birth defects that the country has considered a priority. This WHO convened consultation included a sharing of such initiatives, largely by LMICs in the global South, as a means of promoting South–South learning, as the challenges faced by LMICs in addressing birth defects within their health systems are likely to be broadly similar. Representatives from national and regional programmes from Argentina, Armenia, Brazil, Cuba, Egypt, India (national Rashtriya Bal Swasthya Karyakram; Kerala Hridyam programme), the Philippines, Sri Lanka, Uganda and United Republic of Tanzania shared their experiences of implementing programmes within national health systems, expanding them, and the successes and challenges faced. Integrating screening, diagnosis and management of one or more birth defects into routine services in LMICs is key to the sustainability of these initiatives. Several countries have initiated programmes in varied ways, often beginning with one or more conditions depending on the context. They have expanded initiatives at different rates, engaged nongovernmental stakeholders to varying extents, financed initiatives through diverse approaches, faced a range of challenges, and achieved differing levels of success.

These consultations provided a platform for LMICs (and a few UMICs) with national initiatives for NBS, diagnosis and management of birth defects, to present their experiences with national initiatives, including when and why state-led programmes were started, which conditions were prioritized and why, how programmes were funded, and how diagnostic, referral and treatment pathways were established. They also described mechanisms for follow-up, partnerships with nongovernmental stakeholders, programme expansion, and the successes and challenges encountered.

Importantly, young people affected by birth defects and families of affected persons living in LMICs provided livedexperience perspectives that informed discussions on improving care.

These consultations have NBS for birth defects as a starting point, as early detection and treatment offer the best possibility for survival (e.g. gastroschisis, anorectal malformations), prevent irreversible damage (e.g. congenital hypothyroidism, hearing impairment), and minimize physical (e.g. clubfoot) and intellectual impairment (e.g. inborn errors of metabolism). Newborn screening at a system level identifies children early, allowing early entry into diagnostic and subsequent care pathways as soon as possible. However, while some countries screen children later (e.g. at school entry), it is important to build national NBS programmes as early detection and management allows for the best outcomes for the child.

As part of universal health coverage, populationbased NBS promotes equity, reduces longterm impairment, lowers lifetime care costs, supports families and strengthens health systems. It ensures every child has the right to have a healthy start in life.

A note on terminology:

- A range of terms – *birth defects, congenital disorders, and congenital anomalies* – are widely used, but no globally agreed terminology exists. Stakeholders and families have highlighted the need for language that is accurate, respectful and nonstigmatizing. Although discussions have taken place, consensus has not yet been reached. In the absence of a universally accepted term, this document uses birth defects, consistent with terminology in the Sixtyseventh and Seventyseventh World Health Assembly resolutions.
- In the context of NBS, the term *screening* is used to denote physical examinations (e.g. visible birth defects), clinical tests (e.g. pulse oximetry for critical CHD, hearing screening, red reflex for congenital cataract), or blood spot screening (e.g. endocrine, metabolic and haematological conditions).

Shyam in India, with his parents before surgery ©Operation Smile

Shyam in his village with his friends post-surgery ©Operation Smile

3. State-led national/regional initiatives for newborn screening, diagnosis and management of one or more birth defects in health systems

3.1 Country programmes

The consultation opened with a narrative review of the status of screening programmes for selected birth defects in LMICs. The review of publicly available information indicated that very few LMICs had initiatives for NBS for birth defects at scale (i.e. >50% of all births screened). This was followed by presentations from national health ministries in front-runner LMICs (and a few UMICs) that already have state-led national or regional initiatives for NBS, diagnosis and management of one or birth defects in national health systems. A short summary of each country programme is presented here, with further details provided in Annex 2.

This set of country programme descriptions[1] is followed by lessons learned. These lessons incorporate expert discussions on each country's initiatives and the models implemented. They are presented in a Q&A format to consolidate information from different country experiences into similar topical areas, making it more accessible for stakeholders.

3.1.1 The Philippines: newborn screening programme

The Philippines began its national NBS in 1996 through a pilot in 24 hospitals, targeting five metabolic and endocrine disorders using dried bloodspot testing. Following successful implementation, the programme expanded nationally and now screens for 29 conditions (all using dried bloodspot) across more than 7000 facilities, supported by seven accredited laboratories and 33 continuity clinics. Screening is covered under the national health insurance scheme as mandated by the 2004 Newborn Screening Act, which also requires all birthing facilities to provide NBS. Strengths of the programme include strong legislation, insurance coverage and quality assurance. Challenges persist around early discharge, followup and system resilience. The programme is now expanding beyond conditions screened by bloodspots to include selected visible anomalies and functional defects.

3.1.2 India: national Rashtriya Bal Swasthya Karyakram for birth defects

India's Rashtriya Bal Swasthya Karyakram (RBSK), launched in 2013, is a national programme for early identification and intervention in children with birth defects, diseases, deficiencies and developmental delays. This programme for birth defects began with NBS for nine priority conditions selected on the basis of prevalence, feasibility of diagnosis and availability of treatment, and later expanded to include additional conditions as statelevel readiness improved.

Screening is delivered through over 22 000 birthing facilities, 11 800 mobile health teams, and 430 district early-intervention centres (DEICs) offering diagnostic and multidisciplinary services. Tertiary centres manage complex and genetic conditions. The programme aims to minimize outofpocket expenditure through national insurance schemes, particularly for lowincome families. Over three years, 28.2 million children were screened and 0.9 million identified with a birth defect. Key challenges include workforce shortages, infrastructure gaps, incomplete national data systems, and followup constraints. Monitoring relies on routine reporting, reviews and defined performance indicators. Continued investments in financing, capacity and data systems are being made for longterm sustainability.

[1] The statistics in the country presentations in this section are reported by the respective countries and do not represent official WHO statistics.

3.1.3 Sri Lanka: approach to newborn screening and birth defect management

Sri Lanka has achieved low infant mortality, yet birth defects contribute to 40% of infant deaths, prompting the development of a National Strategic Plan on Birth Defects Surveillance, Prevention and Care (*2024–2030*). Newborn screening is integrated into routine examinations, covering visible anomalies, congenital hypothyroidism, eye abnormalities and critical congenital heart defects, with hearing screening (in some centres). Congenital hypothyroidism screening now reaches 80% of births, supported by a central laboratory system and defined follow-up. The Plan faces constraints in financing, equipment, specialist availability, surgical capacity and data systems. Despite these challenges, Sri Lanka continues to strengthen screening, follow-up and information systems to improve outcomes for affected children.

3.1.4 Uganda: national sickle cell disease programme

Uganda's national sickle cell disease (SCD) programme targets highburden areas by screening newborns and children up to two years and providing care through dedicated sickle cell clinics. The programme was built on the national early infant HIV diagnostic infrastructure, enabling a 2014 survey that tested 100 000 infants and mapped national SCD prevalence. Fourteen highburden districts became the focus of targeted screening. Key components include screening at maternity and immunization sites, trained health workers, a hubandspoke sample transport system, dedicated laboratories, 100 clinics, hydroxyurea provision, data systems and national guidelines. Major challenges include limited coverage, fragmented financing and the absence of a national registry.

3.1.5 India (Kerala): Hridyam programme for congenital heart defects

Hridyam is a stateled programme designed to strengthen the screening, diagnosis and management of congenital heart defects (CHDs) in children. Launched in response to stagnant infant mortality (12 per 1000 live births for a decade since 2006), with evidence of CHD as a major contributor, the programme adopts a coordinated, systemwide approach.

The core components include statewide NBS using pulse oximetry, expanded echocardiography capacity, pooled surgical slots and centralized followup. A digital casemanagement platform registers all children, allocates them to accredited centres, tracks clinical pathways and supports family choice. Public–private partnerships have expanded specialist capacity, improved intensive care unit (ICU) services and strengthened referral and transport systems. Cashless treatment is provided in public and empanelled private hospitals through joint state and national financing. Hridyam's strengths lie in strategic prioritization, strong government leadership, comprehensive system strengthening and transparent monitoring. Since its launch, the programme has improved equity, reduced delays, supported over 9000 children, and completed more than 12 000 surgeries. Infant mortality rate (IMR) dropped from 12 to 6 per 1000 live births in Kerala. Challenges include sustaining adequate funding, engaging private-sector partners and managing workforce turnover.

Pulse oximetry screening for congenital heart defects in a public sector hospital ©Department of Health, Government of Kerala, India

Perspective
Harikrishnan, a 26-year-old male engineer from Kerala, India, born with a complex congenital heart defect

I was born in rural south India as the only child of a middle-class family. On the twentieth day of my life, I was diagnosed with cyanotic CHD. At that time, paediatric cardiology was just emerging in India, with only a few treatment centres in major cities. My parents struggled to find a clear roadmap for my care – awareness among primary care physicians was low, and getting appropriate advice was nearly impossible, even though my parents took me to many doctors and hospitals, always wanting the best for me.

As a child, I lacked the stamina and vigour of my peers. I couldn't play outside with friends and was mostly confined indoors. Even simple activities such as climbing stairs left me breathless, so I missed out on the games and play that other children enjoyed. My schooling was frequently interrupted by health issues, which affected me academically and socially. Books and the Internet became my main sources of learning and intellectual growth. I developed a passion for electronics and technology, setting up a mini workshop at home.

A breakthrough for my condition came in 2014, when I was sixteen. I underwent cardiac magnetic resonance imaging in Kerala, which, although new for the time yet somewhat inadequate, allowed doctors to create a 3D-printed life-size model of my heart using emerging prototyping technology. This innovation was a game changer, giving doctors the insight needed to plan and perform the surgery that finally let me experience what normal oxygen levels feel like.

Inspired by how technology helped me, I now focus my career on 3D modelling for CHD, pursuing a PhD in this area and working with 3D models to help others like me in a tertiary hospital in Kerala. This is my way of giving back.

3.1.6 Egypt: national neonatal screening and birth defect initiatives

Egypt's neonatal screening initiatives began in 1999 with congenital hypothyroidism and phenylketonuria, later expanding in 2021 to 19 genetic disorders prioritized by a scientific committee. The Ministry of Health and Population leads the initiatives with support from the national insurance, the Egyptian Center for Disease Control and Prevention for genetic confirmation, and university hospitals for treatment and followup. Services are free and delivered through 317 screening points nationwide. Since 2021, 500 000 newborns have been screened, identifying 3785 cases, mostly with glucose-6-phosphate dehydrogenase deficiency. A national hearing screening programme launched in 2020 has screened 7 million children, while specialized services for cleft lip and palate have begun to expand.

3.1.7 India: National Sickle Cell Anaemia Elimination Mission

India's sickle cell mission, launched in 2023, is a national, multisectoral initiative targeting the high burden of SCD, particularly among tribal populations. India accounts for nearly 50 000 of the estimated 500 000 SCD births globally each year, underscoring the scale and public health significance of the disease in the country. The mission focuses on prevention, early detection and comprehensive management through coordinated action at all levels of the health system. Screening begins at community and primary care levels through door-to-door campaigns, health camps and institutional screening, with community health workers playing a central role. District hospitals provide confirmatory diagnosis, ongoing care and prophylactic therapy, while tertiary Centres of Competence offer advanced diagnostics, prenatal testing, surgery and bone marrow transplantation. Key strategies include dedicated financing, standardized guidelines, reliable supplies of hydroxyurea and other essential medicines, an SCD registry and telemedicine support. Public–private partnerships strengthen affordable screening and supply chains. Multisectoral collaboration across health, tribal development, education, research and social welfare is central to achieving the goal of eliminating SCD as a public health problem by 2047.

3.1.8 Argentina: national programme for newborn screening

Argentina has developed a comprehensive national approach to NBS and CHD management, grounded in legislation and strong federal–provincial coordination. The national NBS programme, launched in 2006 after identifying low coverage, was reinforced by a 2007 law mandating screening for six priority conditions. Under a shared financing model, the national government supplies laboratory inputs while provinces manage service delivery. Coverage has increased from 52% to 98%, supported by 20 laboratories, structured diagnostic pathways, training and initial treatment coverage. The programme benefits from mandatory reporting and the capacity to add new conditions based on scientific evidence, though disparities in resources and specialist availability remain.

Recognizing CHD as a leading cause of birth defect-related mortality, Argentina built a staged national CHD programme. Early phases established registries, financing and hospital networks, followed by integration into a resultsbased financing scheme and strengthened infrastructure. The Federal Network for the Care of Congenital Heart Defects has expanded diagnosis and treatment capacity, significantly reduced waiting lists, and improved infant outcomes, despite persistent regional inequities.

3.1.9 Cuba: national programme for birth defects and genetic disorders

Cuba established a national programme for the diagnosis, management and prevention of genetic disorders and birth defects in 1981, supported by the National Medical Genetics Network, which integrates clinical services and 38 laboratories across all levels of the health system. Capacity building has been central, with specialist training in medical genetics and genetic counselling incorporated into university curricula, and more than 900 counsellors trained. The National Center for Medical Genetics oversees the Network and maintains a national registry for birth defects. Newborn screening covers six metabolic and endocrine conditions, using locally developed technology, with testing decentralized to the municipal level. Cardiac surgery is provided in two national institutes, and pulse oximetry is planned for inclusion in screening.

3.1.10 Brazil: National Neonatal Screening Program

Brazil's national NBS programme, launched in 2001, has expanded in phased stages to include phenylketonuria, congenital hypothyroidism, SCD, haemoglobinopathies, cystic fibrosis, congenital adrenal hyperplasia, biotinidase deficiency, and most recently, congenital toxoplasmosis. Coverage has remained high, reaching 80–84% of births, with samples collected at more than 23 000 facilities. Hearing screening became mandatory in 2010, though coverage remains at 43% and followup challenges persist, particularly in remote areas. Brazil also conducts surveillance for visible birth defects and introduced nationwide pulse oximetry screening for critical CHD in 2017, although surgical capacity remains insufficient despite ongoing expansion efforts.

Perspective
Ruth Nankanja, teacher, entrepreneur and advocate from Uganda – mother of Lillian born with osteogenesis imperfecta

When Lillian was born, we didn't know she had a birth defect. She cried persistently and refused to feed, and after several days in the hospital, we took her home. While bathing her at home, my mother discovered a fracture in her arm. We took her back and further x-rays revealed more fractures. She was treated with casts that were available at the local club foot clinic, but the deformities kept recurring whenever the casts were removed. Immunizations were painful, as her casts had to be cut and replaced each time which were very painful for her, and for us to see her in pain. At age two, we visited an orthopaedic surgeon who explained her condition and began corrective surgeries. Today, Lillian has insisted on going to boarding school with her siblings.

Early diagnosis would have helped her suffer less and perhaps have fewer physical difficulties. For families like us, financial support is crucial, as repeated hospital visits, surgeries and assistive devices are expensive. Stock-outs of medical supplies increased our burden. My husband has been supportive but, in many families, men need to be better engaged to share responsibilities with mothers in these situations.

3.1.11 Armenia: universal newborn screening programme

Armenia's NBS programme is a statefunded, universal programme integrated across public and private providers. Initiated with Swiss support, the programme transitioned to full government financing within five years and now covers congenital hypothyroidism, hearing impairment, phenylketonuria, developmental dysplasia of the hip, severe CHDs and congenital adrenal hyperplasia. Quality assurance is led by the National Screening and Referral Laboratory with international oversight. Training for laboratory and maternity staff has shifted from external to national programmes, supported by ongoing awareness campaigns. Electronic data systems enable coordinated screening, referral and followup, although resource constraints limit expansion to additional conditions.

3.2 Key lessons learned from state-led LMIC initiatives for newborn screening, diagnosis and management of one or more birth defects

3.2.1 When is it appropriate for an LMIC to start a programme to screen, diagnose and manage one or more birth defects?

Infant mortality rate can be regarded as a broad proxy for both socioeconomic development and health system performance. From the consultations, it was clear that countries have initiated national programmes at different stages of development, and there is no consistent association between a country's IMR – as an indicator of its socioeconomic context – and the timing of the implementation of national initiatives addressing NBS and the management of birth defects. Argentina, Brazil, Cuba, India and the Philippines began NBS at varying IMR. In practice, programmes were triggered by advocacy, disease burden, donor support, academic pilots or political commitment, rather than considerations of IMR. Participants emphasized that NBS is grounded in equity and social justice, ensuring that all children receive timely diagnosis and care regardless of national levels of socioeconomic development.

Newborn screening for congenital hearing impairment ©Otto Mejia

3.2.2 How did programmes begin in LMICs? What have been triggers?

Countries have initiated NBS programmes through diverse pathways shaped by local priorities, resources and health system readiness. Key triggers include advocacy by clinicians, patient support groups/families; champions (the Philippines), professional organizations (Sri Lanka), high disease burden (Sri Lanka, Uganda); donor-supported pilots (the Philippines); academic evidence; and/or strong political commitment (India). The presentations highlighted that while some programmes began as small pilots which subsequently expanded, others were launched at state or national levels from the outset.

In the Philippines, champions both inside and outside its Department of Health played a pivotal role in launching and scaling up the programme. In Sri Lanka, the hearing screening initiative was advocated for by the College of Otorhinolaryngologists of Sri Lanka. Uganda's national SCD programme was initiated in response to the high prevalence of SCD (burden), with advocacy from affected families and support from donor-funded projects. Donor resources and country interest often converge, as seen in Uganda and other countries, enabling pilot projects or targeted initiatives that later expand into national programmes. Kerala's Hridyam programme for CHDs was launched after the state's Department of Health recognized CHD as a significant contributor to infant mortality.

Discussions revealed that academic pilots have also served as catalysts. In the Philippines, a small-scale academic initiative provided critical data and operational experience, which convinced policymakers to support a nationwide rollout backed by legislation and national insurance. Similarly, strong political commitment from the highest levels has been a key factor in countries like Egypt and India, where government leadership has successfully institutionalized and sustained NBS programmes.

Programmes may start as small projects in restricted geographic areas, allowing for focused implementation and learning before scaling up. For instance, pilot phases in Uganda and the Philippines helped map disease burden and refine systems, while Kerala (India) launched statewide initiatives from the outset.

Ultimately, country experiences demonstrated that the success of NBS programmes are shaped by local needs, available resources, political leadership and the capacity to build partnerships and adapt to changing circumstances. Leadership from the ministry of health and integration into national health systems have been essential for sustainability.

3.2.3 Did all newborn screening, diagnosis and management programmes have a pilot phase?

Deliberations noted that not all LMICs initiate NBS or birth defect programmes with a pilot phase; some, such as Kerala's Hridyam programme on CHDs, were launched statewide from the outset. However, most countries used pilot projects to generate essential evidence and strengthen system readiness. Pilots helped to map disease burden and identify priority conditions, as seen in Uganda's SCD programme. They also provided a controlled environment to test and refine screening, diagnostic, management and followup models – as illustrated by the Philippines, where pilot learning informed national protocols, logistics (sample transport, laboratory capacity) and communication systems (patient recall).

Pilot phases supported the definition of care pathways, clarified workforce roles, and established referral and treatment networks before national scaleup. In several countries, pilots provided data that helped persuade policymakers and funders, enabling transition to national programmes. Regardless of whether a pilot is used, expert exchanges underscored the importance of strong government leadership, integration into routine health systems, sustainable financing and continuous monitoring are essential for longterm impact and scaleup.

Newborn screening filter cards for dried blood sport collection ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

3.2.4 How were initial conditions selected for inclusion in national newborn screening initiatives?

Countries have used different approaches to select initial conditions for NBS, but all began with a small, feasible set of conditions rather than attempting to address all birth defects. Most programmes began with either one highpriority condition, such as SCD in Uganda, or a limited panel of four to eight conditions, as seen in Brazil, Egypt, India and the Philippines. Selection has been guided by national priorities, disease burden, feasibility and available resources.

Some countries chose metabolic conditions detectable by bloodspot testing, while others prioritized visible birth defects requiring minimal screening infrastructure. The Philippines began their national programme with endocrine and metabolic disorders influenced by professional champions who were clinical geneticists. Structural birth defects are being introduced now, more than a decade after the national NBS programme was launched. Conversely, India focused on nine major birth defects (visible or detected by clinical screen), selected based on prevalence, ease of diagnosis, treatment availability and affordability within the health system. The approach was to start small with visible birth defects, as these were relatively easy to detect at birth with low infrastructure needs, and then scale up in a phased manner to include functional defects, such as CHD, followed by metabolic defects in some states, depending on the availability of funds and health system readiness.

Decisions were typically led by ministries of health, informed by clinicians, scientific experts and professional bodies, particularly where local epidemiological data were limited. From the consultations, a consistent principle emerged across LMICs: the prioritizing of conditions for which effective treatment is available. Establishing downstream pathways for confirmatory diagnosis, clinical management and longterm followup proved essential, supported in some cases by public–private partnerships and specialist training to address system gaps.

Clubfoot can be detected at birth by a simple visual examination. Treatment is most effective if started soon after birth when the tendons and ligaments are at their most elastic (Ecuador) ©MiracleFeet

3.2.5 How have countries expanded newborn screening programmes?

In most cases, countries began NBS programmes with a small number of conditions that were prevalent and could be feasibly diagnosed and managed within existing health systems. Screening served as an entry point for expansion, with country programmes prioritizing conditions for which treatment was available, and care pathways were well defined.

The discussions highlighted that expansion typically occurs incrementally as system capacity grows, guided by scientific evidence, expert reviews and national priorities. Initial conditions vary: Brazil, Egypt and the Philippines began with endocrine and metabolic disorders, later adding visible anomalies and hearing impairment, while India initially focused on visible birth defects before adding metabolic conditions in selected states. Programmes periodically removed conditions with limited benefit (e.g. the Philippines), highlighting the importance of continuous evaluation. The pace and scope of expansion appear to be closely tied to health system capacity. Countries with robust infrastructure, trained personnel and sustainable financing could expand more rapidly, while those facing resource constraints adopted a more cautious and considered approach.

Scientific mechanisms – such as national technical working groups and expert commissions – supported decisions on adding or removing conditions (e.g. Argentina, the Philippines). It was noted that overall, programme expansion is a dynamic, contextspecific process that balances scientific evidence, feasibility and stakeholder input.

3.2.6 Have LMICs planned care pathways for newborn screening initiatives from the outset?

Discussions revealed that programmes approached followup for NBS in different ways. In the Philippines, early implementation focused primarily on screening, with shortterm followup continuing until diagnostic confirmation. Over time, the need for structured longterm followup including treatment and monitoring, became clear, and these services were integrated a decade later. In contrast, Kerala's Hridyam programme for CHDs adopted a full continuumofcare

model from the outset – spanning screening, diagnosis, surgical management and long-term care. Stabilization of sick newborns, emergency transport and referral pathways were central components. Detailed protocols were developed for each stage, and healthsystem strengthening was aligned with the requirements of the care pathway. This comprehensive approach was later adapted for additional congenital conditions, such as clubfoot.

Country experiences across programmes underscored that screening alone was insufficient. It must function as the entry point to a comprehensive continuum of care. Countries began with a limited set of priority conditions, ensuring that pathways for diagnosis, referral and management were clear, feasible and sustainable.

3.2.7 What has been the role of legislation in LMICs in scaling up newborn screening and integrating it into national health systems?

The Philippines' Newborn Screening Act of 2004 established a legal and operational framework that formalized national NBS, designated the Department of Health as the lead agency, integrated NBS into the health insurance system, and required all birthing facilities to provide screening as a condition of licensure. This legislative mandate enabled rapid expansion of laboratories and continuity clinics nationwide. However, experience shared during the consultations highlighted that legislation alone is neither necessary nor sufficient for achieving universal coverage. In several countries, strong ministry of health leadership, effective policy implementation and sustainable financing – rather than legal mandates – were the primary drivers of high NBS coverage. Deliberations indicated that legislation alone was insufficient unless linked to financing and stakeholder engagement to ensure longterm sustainability.

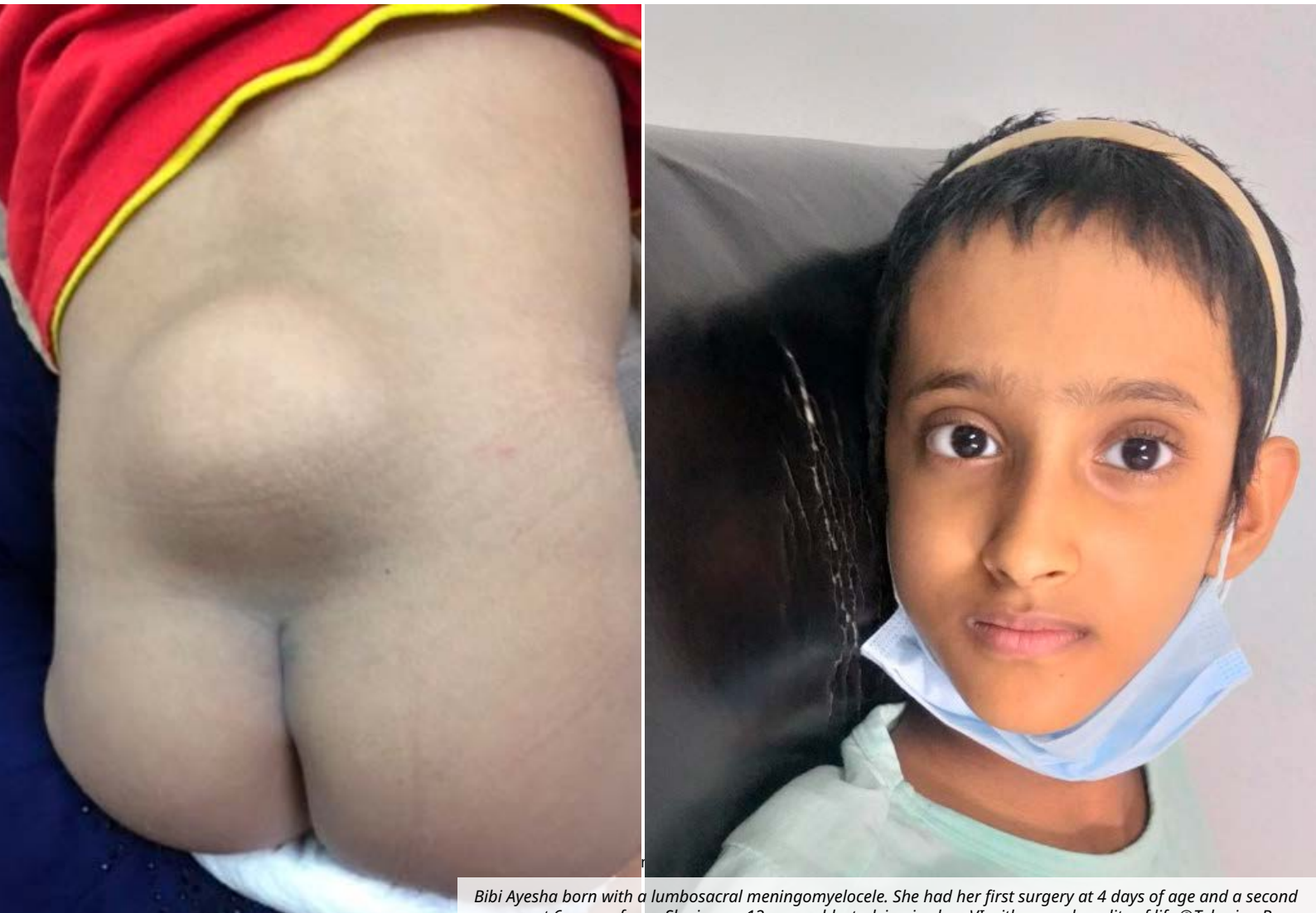
3.2.8 What are the key facilitators for implementing initiatives in LMICs to address one or more birth defects at scale within national health systems?

Consultations indicated that:

- Strong political commitment and leadership are central to establishing and expanding NBS programmes, with advocacy from health professionals, as well as champions and/or families often catalysing action. In India and the Philippines, government leadership and policy prioritization enabled the launch and expansion of NBS programmes.
- Sustainable financing through national insurance or government-funded packages ensures equity and minimizes out-of-pocket costs (e.g. Brazil, India).
- Multisectoral partnerships – including NGOs, academia, international agencies and private providers – strengthen technical capacity, provide supplemental funding and build capacity. Public–private partnerships are especially important for expanding access to specialized care (e.g. India's RBSK and Hridyam programmes).
- Establishing simple, functional data systems for tracking screening, diagnosis and short- and long-term follow-up enables effective programme monitoring and evaluation.
- Continuous education of parents, providers and communities improves awareness and uptake of NBS.
- Legislative and policy frameworks, such as the Philippines' NBS Act, creates enabling conditions, though effective implementation and ministry engagement remain critical, even in the absence of formal laws. A number of LMICs have implemented successful programs in the absence of formal legislation.

3.2.9 What are the key challenges faced by LMICs in implementing initiatives for one or more birth defects at scale within their national health systems?

- Discussions on challenges highlighted that LMICs have often faced limited financial, human and infrastructural resources. Funding for screening, confirmatory diagnosis, treatment and follow-up was frequently inadequate given competing priorities. While engagement with nongovernment stakeholders may help alleviate certain resource constraints, continued reliance on donor support poses a risk to longterm sustainability. Programmes that began as pilots or donor-funded projects needed political commitment to eventually transition into stable, nationally supported initiatives.
- Establishing robust data systems for tracking screened newborns and ensuring those with positive results proceed to diagnosis, and long-term management (including follow-up) has been demanding as reported by a number of countries at the consultation. Besides good data systems for tracking, maintaining short- and long-term follow-up for affected children requires clearly defined referral pathways and multidisciplinary teams in some settings.
- Ensuring universal coverage has proven complex, especially in remote or underserved areas, where out-of-pocket payments for screening or treatment tend to exclude vulnerable families, and thereby undermine equity goals. Addressing these barriers has highlighted the need for long-term sustainability, requiring government commitment, investment and integration into the public health system.



Bibi Ayesha born with a lumbosacral meningomyelocele. She had her first surgery at 4 days of age and a second surgery at 6 years of age. She is now 12 years old, studying in class VI with a good quality of life ©Tahmina Banu

3.2.10 What are the broad components for newborn screening, diagnosis and management within a health system?

A comprehensive NBS programme as a system designed to optimize a child's development through timely detection, accurate diagnostic confirmation and appropriate management, supported by access to sustained long-term care. Key components identified as part of this structure include:

- Screening: This may involve external examinations for visible defects, clinical screening tests (such as pulse oximetry for CHDs, or hearing screens), or laboratory-based tests for endocrine/metabolic/blood disorders or genetic tests.
- Diagnosis: Specialist assessments and laboratory tests to confirm conditions and link families to appropriate management pathways.
- Management: Interventions may include surgical care (e.g. for CHDs in Argentina, Kerala, Sri Lanka), medical management (e.g. thyroxine for congenital hypothyroidism in Sri Lanka), special diets for metabolic conditions (in Egypt, the Philippines), provision of prostheses (e.g. hearing aids in Sri Lanka, foot abduction braces for clubfoot in Sri Lanka and the United Republic of Tanzania), and long-term follow-up with supportive services (e.g. speech therapy in Nepal).
- Follow-up: Organized processes and data systems are essential to track infants with positive screens, ensure families receive results and facilitate timely diagnostic testing (short-term follow-up). They also support management when required (medical, surgical, assisted technologies and rehabilitation services) including long-term follow up.
- Evaluation: Ongoing assessment and quality assurance have helped identify areas for improvement. Economic evaluations can be conducted once the programme has been implemented.
- Education: Targeted education for parents, health professionals and policymakers ties the system together.

The consultations concluded that implementing NBS programmes also strengthens broader health systems by enhancing service delivery across all levels of care, building multidisciplinary teams, reinforcing infrastructure, referral networks and data systems, and strengthening financing.

The position of the feet (above) is the only medical obstacle preventing children with clubfoot from living productive, active, and healthy lives. With proper treatment, more than 95% of babies born with idiopathic clubfoot achieve complete correction and full mobility (below) within the first year of life ©MiracleFeet

4. Selection of initial conditions in a programme for newborn screening, diagnosis and management of one or more birth defects

4.1 Principles to aid the selection of initial conditions

At the consultation, three sets of formally published principles or criteria for selecting a condition for inclusion in a NBS programme were discussed. Countries have adapted these principles, applied them pragmatically and/or made choices based on other factors.

(i) **Wilson and Jungner principles** (Box 1): Developed in 1968 for population screening, these have historically been used as a reference framework. Although the original principles focused on adult population screening programmes they have been used widely in the NBS context.

Box 1. Wilson and Jungner principles for population screening *(10)*

1. The condition should be an important health problem.
2. The natural history of the condition, including development from latent to declared disease, should be adequately understood.
3. There should be a recognizable latent or early symptomatic stage.
4. There should be a suitable test or examination.
5. The test should be acceptable to the population.
6. There should be an agreed policy on whom to treat as patients.
7. There should be an accepted treatment for patients with recognized disease.
8. Facilities for diagnosis and treatment should be available.
9. The cost of case-finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care.
10. Case-finding should be a continuing process and not a “once and for all” project

These principles have limitations including subjective elements, limited relevance for newborns and families, and the assumption of a one-condition–one-test model (which is no longer the case). A number of efforts have been made to review and amend these *(11)*, although they remain an important reference.

(ii) **Advisory Committee on Heritable Disorders in Newborns and Children** (Box 2) has used a structured approach to make recommendations for conditions to be included in NBS, based on seven essential considerations.

Box 2. Advisory Committee on Heritable Disorders in Newborns and Children *(12)*

1. Is there direct evidence that screening at birth leads to improved outcomes for the infant or for the child's family?
2. Is there a case definition that can be applied uniformly and reliably? What are the clinical history and disease spectrum, including the impact of timely recognition and treatment?
3. Is there a screening test or algorithm that has sufficient analytic validity?
4. Has the clinical validity of the screening test or algorithm, in combination with the diagnostic test or test algorithm, been determined, and is that validity adequate?
5. What is the clinical utility of the screening test or algorithm?
6. How cost-effective are screening, diagnosis and treatment for this disorder compared with usual clinical case detection and treatment?
7. What is the impact on the public health system? How prepared is the system to add the test(s)?

(iii) **Disease control priorities** third edition *(13)* aimed to develop an evidence-based essential package of health interventions to promote universal health coverage in LMICs, with components specifically addressing congenital and genetic disorders. For inclusion in this essential package, interventions were required to meet three characteristics, as shown in Box 3. Initiatives in LMICs have often begun with a condition that attracts interest from key stakeholders (whether political or through professional champions) and where the health system has existing – or reasonably attainable – capacity to screen, diagnose and manage the condition of interest. Nevertheless, when well implemented, such initiatives have provided a nidus to which other conditions have subsequently been added.

Box 3. Disease control priorities

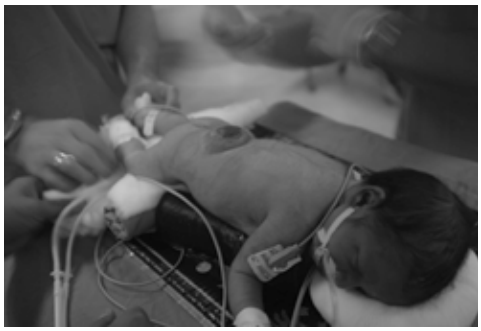
1. Addresses a significant disease burden
2. Feasible to implement in LMICs
3. Good value for money

Across these frameworks, three core criteria consistently emerge: **disease burden, feasibility within the health system, and affordability for sustainable implementation**. These are discussed below.

4.1.1 Disease burden

The selection of conditions for NBS programmes in LMICs is shaped by the need to maximize the impact of limited resources. Although it is important to prioritize conditions with significant disease burden, most LMICs lack high-quality, robust registry data to guide these choices.

Ministries of health, therefore, often rely on pragmatic decisionmaking informed by expert opinion, pilot projects, or clearly recognized highburden conditions, such as SCD in parts of subSaharan Africa. Political commitment can also shape priorities, where evidence is limited. For example, in Punjab, Pakistan, government interest in establishing the burden of adult cardiac disease was leveraged by clinician champions to secure funding for CHD and led to the creation of a CHD registry.



A newborn with spina Bifida, just a couple of hours after birth and already under general anaesthesia, the neurosurgeon's hands are visible, reflecting the intention to make a difference to the quality of life of this child ©2012 International Federation for Spina Bifida and Hydrocephalus. IF Photo Catalogue "Unfold"

While broad principles (which include burden of the condition) exist to guide the selection of initial conditions for inclusion in NBS programmes, country experiences suggest varying levels of adherence to these principles. In some countries evidence of burden of the conditions of interest from pilot projects is required, whereas in others expert advice from clinicians, professional bodies and academia has guided ministries of health in selecting the initial conditions. Typically, more information becomes available once a programme with a robust data system is in place.

4.1.2 Feasibility within the health system

What does it take to manage conditions in the health system – experiences of selected conditions from the frontline in LMICs

Besides disease burden, another important criterion for prioritizing a condition is the feasibility of screening, diagnosing and managing the condition within the existing health system. Clinical experts from various LMICs, working with different birth defects (including orofacial clefts, SCD, clubfoot, CHD, anorectal malformations) provided insights into what is required to establish systems for screening, diagnosis and management of that specific condition. Five conditions summarized here were presented at the consultation. Noteworthy across conditions were the involvement of all levels of the health system, the importance of multidisciplinary teams and the key role of the health ministry in ensuring sustainability.

Orofacial clefts *(Nepal):* The speaker from Nepal emphasized that effective management depended on strong government–NGO collaboration, a multidisciplinary workforce (including orthodontists, surgeons, nurses, speech therapists, nutritionists and audiologists), and task shifting to nurses. Nurses are central to the process, taking on responsibilities from screening and nutrition support to pre- and post-operative care, speech therapy and community outreach. Care spans all levels of the healthsystem, supported by wellequipped surgical services, communitybased speech camps and robust information systems. Longterm followup and sustainable financing, including public–private partnerships, remain essential.

Sickle cell disease *(Ghana):* Effective SCD services in Ghana require integration across all healthsystem levels, strong Ministry of Health leadership, and a trained workforce. Trained staff are essential for newborn screening, early intervention, complication management and family support. Care spans community education, primary care (screening, penicillin and malaria prophylaxis, pain management), referral hospitals (hydroxyurea therapy, acute complication management) and tertiary clinics (stroke prevention, intensive care, surgery and education). Persistent gaps include limited access to diagnostics, medicines and robust data systems, alongside significant financial barriers that demand progress toward universal health coverage.

Clubfoot *(Ethiopia and United Republic of Tanzania)*: Effective clubfoot management in LMICs relies on strong government leadership, early detection and integration into routine maternal–child health services. The Ponseti method, supported by trained providers and taskshifting, underpins care. Early detection, referral and treatment are crucial for good outcomes, supported by specialized clinics in hospitals and health centres that improve accessibility. Coordination among child health, physiotherapy, orthopaedics and assistive technology sectors is important, as is long-term family education and support. Public–private partnerships, adequate supplies, robust information systems, and inclusion in national insurance schemes ensure accessibility, longterm followup and sustainability.

Asma was born with ano rectal malformations which were surgically repaired. Today she is a talented singer and painter, training to be an engineer at university ©Tahmina Banu

Anorectal malformations *(Bangladesh):* Effective management of anorectal malformations in Bangladesh requires strong Ministry of Health and Family Welfare leadership, trained surgical and neonatal teams, and clear referral pathways for urgent care. Workforce development is essential: staff must be trained to perform comprehensive head-to-toe newborn examinations, and skilled general and paediatric surgeons, anaesthetists, paediatricians, neonatologists and nurses are needed for both intraoperative and postoperative care. Community health workers also play a vital role in supporting families after discharge. Inadequate transport, high costs and stigma hinder access to care. Essential supplies, robust information systems, and universal health coverage are critical to ensure safe treatment and complete followup.

Congenital heart defect *(Punjab, Pakistan):* Punjab's CHD programme relies on strong state leadership and multidisciplinary teams (cardiologists, surgeons, obstetricians, nurses, community health workers and counsellors). The programme's success depends on teamwork, not individual specialists. Patientcentred services supported by robust infrastructure, including screening equipment at all levels of care, organized referral systems, tertiary care facilities with cardiac catheterization laboratories, operating rooms, cardiac ICUs, paediatric anaesthesia, and a reliable supply chain for consumables across all levels of care as needed. The continuum of care – from screening and diagnosis to prioritization, referral, treatment and follow-up – must be maintained. All health system levels are involved: tertiary hospitals provide advanced care, secondary hospitals support diagnostics and nutrition, and primary centres handle basic screening and referrals. Integrated public–private partnerships, sustainable financing (progress towards universal health coverage) and strong information systems are essential to maintain continuity and equity of care.

Post operative examination of a newborn after surgical repair of a congenital heart defect ©Department of Health, Government of Kerala, India

Strong nursing care post operatively for newborns with a congenital heart defect is key to recovery ©Department of Health, Government of Kerala, India

Health system requirements to ensure feasibility of screening, diagnosis and management of five birth defects in LMICs

The five tracer conditions discussed above reveal several common prerequisites including state leadership, human resource strengthening, contributions from multiple specialties (and thus multidisciplinary teams), and the involvement of all levels of the health system. Each clinical condition also has specific requirements. It is therefore helpful for countries to engage with their professional clinical bodies and relevant stakeholders to define the detailed requirements for managing a particular condition under consideration for prioritization.

Feasibility assessment: Kerala's state-led Hridyam programme (India)[2]

Kerala's efforts to reduce infant mortality, which has remained unchanged at 12 per 1000 live births for a decade from 2006, led to the strategic prioritization of CHDs. Analyses showed that congenital anomalies were the second leading cause of infant deaths, with CHD identified as the major contributor. A feasibility assessment in 2016 mapped existing resources for newborn and infant cardiac surgery, identifying 4000 annual CHD cases, of which 25% were critical; however, only half of these critical cases received timely surgery. Programme strengths included strong political commitment, physician support and existing surgical centres. Key gaps included limited echocardiography capacity, delayed diagnosis, inadequate financing, poor provider awareness, and fragmented referral and transport systems.

Kerala subsequently implemented wideranging system-strengthening measures. These included: (i) expansion of early detection through statewide pulse oximetry screening and provider training; (ii) capacity building, such as training radiologists, obstetricians, paediatricians in paediatric echocardiography, organizing interventional cardiology workshops, training intensive care unit nurses in paediatric cardiac care, and improving access to essential medicines; (iii) formalization of referral pathways and establishment of emergency transport systems through public–private partnerships, (iv) introduction of a centralized digital platform introduced for case registration, triage, surgical slot allocation, claims processing and community followup; (v) centralized pooling of surgical referrals and slots to create predictable waiting times; and (vi) financial protection measures that ensured cashless treatment in public and empanelled private hospitals through RBSK and state funding.

These combined actions made large scale, equitable management of CHDs feasible within Kerala's health system.

Feasibility challenges: Sri Lanka's Newborn Hearing Screening Programme

Sri Lanka's Newborn Hearing Screening Programme, launched in 2017, advocated for by the College of Otorhinolaryngologists of Sri Lanka, addresses a clear need for early detection of congenital hearing impairment. Led by the Family Health Bureau of the Ministry of Health with support from national and international partners, the programme faces significant feasibility constraints. Coverage remains low (8% of the birth cohort screened) due to limited equipment, malfunctioning otoacoustic emission devices and slow expansion. Confirmatory testing often entails long wait times, and access to cochlear implants remains restricted. Workforce shortages, high staff turnover and the absence of a dedicated budget further impede progress. Weak data systems and limited rehabilitation services hinder continuity of care, underscoring the need for adequate human resources, wellmaintained equipment and sustainable financing to ensure a functional hearing screening programme.

4.1.3 Affordability for sustainable implementation

This session opened with an overview of the rationale for NBS, discussing the difference in financial and economic costs, economic evaluations and how they support advocacy for NBS. This was followed by a short presentation on NBS, diagnosis and management falling within the ambit of universal health coverage, and on how regressive payment mechanisms hinder its implementation. Country presentations highlighted how national initiatives are funded, the financial sustainability of programmes and the challenges they face.

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[2] The statistics in the country presentations in this section are reported by the respective countries and do not represent official WHO statistics.

The aim of population-based NBS and economic evaluations

The primary aim of populationbased NBS is to improve health outcomes for children with birth defects by reducing mortality, preventing disability and enhancing longterm quality of life for affected children and their families. Governments invest in NBS because it advances equity – ensuring that rare but serious conditions receive the same attention as more common childhood illnesses – and because early detection prevents costly complications. Economic rationale is therefore central to decisionmaking.

Cost analysis for NBS distinguishes between direct financial costs (laboratory operations, programme infrastructure), and broader economic costs (lost productivity, family burden, longterm societal impact). While direct financial costs are relatively straightforward to estimate, economic costs are more complex and may not be immediately visible. Reliable cost estimates often draw on pilot programmes, timeuse data, or evidence from comparable national programmes. The involvement of health economists is particularly helpful.

Economic evaluations – including costeffectiveness and cost–benefit analyses – are sometimes required by governments to justify largescale implementation. Such analyses may compare outcomes for latediagnosed cohorts to highlight severe outcomes and lost productivity or compare health care utilization and outcomes for children with specific conditions. For example, prior to the passage of the Philippines NBS Act, an economic analysis estimated that achieving full coverage would cost US$ 12 million and showed that substantial savings were achieved by expanding to multiple conditions and leveraging existing systems rather than screening for single disorders.

Many countries (including high- and upper-middle-income nations) initiate NBS without initial economic evaluations, often due to limited data. Nonetheless, economic evidence remains a powerful tool for advocating programme adoption, guiding expansion and supporting sustainable health sector investment.

Universal health coverage

Universal health coverage means that all people have access to the full range of quality health services they need, when and where they need them, without financial hardship. It covers the full continuum of essential health services, from health promotion to prevention, treatment, rehabilitation and palliative care, across the life course *(14)*. Newborn screening, diagnosis and management of birth defects form an integral part of universal health coverage by enabling early detection, timely care and improved longterm outcomes for affected children.

State financing – through taxfunded national insurance (as in Kerala (India), the Philippines) or governmentcoordinated donor support (as in Uganda) – was seen to be the most equitable and sustainable approach, ensuring universal access and integration into routine maternal and child health services.

Countries sometimes used outofpocket financing during pilot phases, prior to full state-led implementation. Routine outofpocket payments position NBS as an “extra” service, accessible only to those who can afford it. This approach is regressive and risks excluding the most vulnerable newborns, especially those at highest risk due to factors like maternal undernutrition or infection. It was noted that outofpocket models transform newborn screening from a public health good into a market commodity, reinforcing inequities. Sustainable national financing remains essential for scaleup, quality and longterm programme viability.

Country experiences in financing

Financing Uganda’s sickle cell disease programme

Uganda carries a substantial burden of sickle cell anaemia, with 1% of newborns affected with the disease and 14% carrying the trait. An estimated 20 000 babies are born with SCD annually, yet up to 80% die before the age of five years due to limited access to early diagnosis and care *(15)*. The national NBS programme currently reaches only 3% of infants because of funding constraints; among the 500 000 newborns screened since the programme began, 35 000 tested positive.

An infant comfortably asleep with abduction braces for club foot correction ©Miracle Feet

Financing relies on modest government contributions, some outofpocket payments and extensive external support through donations, loans and research grants. Donor funding has strengthened capacity but remains unstable, with shifting priorities and risks associated with loanbased financing. Research funding provides valuable evidence but does not always translate into scalable services. To enhance sustainability, Uganda aims to integrate SCD services into routine care, expand national health insurance, build local pharmaceutical production and secure longterm financing. Sustained progress requires strong government leadership and reduced reliance on external funding.

Financing the Hridyam programme in Kerala, India

Kerala's Hridyam programme for CHDs began with a comprehensive needs assessment to identify gaps in surgical capacity, human resources, infrastructure, protocols and logistics. At launch, the state had limited public cardiac centres, uneven regional access, inadequate catheterization laboratories, shortage of trained specialists, and weak referral and transport systems. Data management was fragmented, with hospitals maintaining separate registries and families registering at multiple centres to secure timely surgery. This highlighted the need for a single, webbased registry to coordinate between patients and the different hospitals. Existing government programmes allowed partnerships for service purchase but lacked formal agreements and systems for monitoring service quality.

Financing for the initiative was drawn from multiple government sources, including the Kerala State Plan, National Health Mission (RBSK) and Finance Commission grants, supplemented by NGO support for capacity building. Activities were phased:

- Phase 1 – case management reforms (2017–present): Care pathways were defined by prioritizing cases based on urgency and allocating them to appropriate centres, with mutually agreed costs and pricing between the state and private hospitals, clearly outlining the responsibilities of both public and private partners.
- Phase 2 – system strengthening (*2017–2019*): Screening capacity was expanded, human resource capacity building, neonatal ICUs were upgraded, a referral transport system was initiated and paediatric cardiologists and surgeons were trained.
- Phase 3 – longterm investments (*2018–2021*): New hospitals were established, district echocardiography capacity was expanded, and intensive care unit facilities were upgraded.

The budget centred on treatment costs – including surgery, consumables, drugs and transportation – which were the largest component, followed by human resources (recruitment of additional staff and training of existing staff), diagnostic equipment (such as echocardiogram machines) and infrastructure (operating theatres and functional cardiac catheterization laboratories for paediatric cardiac interventions). Universal pulse oximetry screening was introduced across public birthing facilities, and district hospitals were equipped with echocardiography and surgical capacity. However, financial planning challenges emerged, including rising treatment costs, underestimated needs for repeated hospitalizations and investigations, funding delays, and static reimbursement rates for private partners. Longterm sustainability remains constrained by maintenance costs, adultcare needs and outofpocket burdens associated with continuing followup.

Hridyam's experience highlights the learning curve with financial planning and the importance of preparing budget space for and adapting to evolving needs and challenges, particularly in the context of public–private partnerships.

Hearing rehabilitation for a child fitted with hearing aids ©Hear the World Foundation

Financing the NBS, diagnosis and management programme in the Philippines

The Philippines NBS programme has expanded substantially since its launch in 1996, growing from 24 hospitals to more than 7000 by 2023, with approximately 4600 dried bloodspot samples collected daily. Its financial evolution reflects the country's efforts to shift from outofpocket payments to sustainable government and insurance funding. During the pilot phase, families paid US$ 7 per test, prior to full state ownership. Following the Newborn Screening Act of 2004, national insurance (PhilHealth) was mandated to cover screening and related services, although meaningful financial protection was only realized once government resources were secured. Coverage improved progressively with full insurance support for basic NBS (*2011*) and later for expanded NBS (*2019*), which now includes 29 conditions with the total programme cost reaching US$ 40 million. However, small outofpocket costs persist.

It is noteworthy that when outofpocket payments dominated, coverage was less than 1%. Even with full government coverage, uptake only reached 40%, showing that insurance alone does not guarantee high coverage. Education and acceptance among health professionals and the public were more influential in increasing uptake than financial coverage alone. Communication with parents about insurance coverage and periodic review of insurance fees have also been important for programme success.

Summary learnings from LMICs on paying for state-led initiatives

LMICs with large programmes have prioritized equity in access to population-based newborn and child screening programmes, aiming to ensure that no child is left behind. In most LMICs, national health insurance systems – such as those in Brazil, Cuba, Egypt, India and the Philippines – cover the costs of screening, diagnostic tests, management and follow-up. This approach minimizes or eliminates out-of-pocket expenses for families, promoting equitable access.

In settings where national insurance is not yet established, ministries of health have coordinated donor funding to launch and expand programmes. Uganda, for example, has relied on donor resources to initiate and scale up its SCD screening programme, with a modest contribution from the state. However, donor funding can be unpredictable, and long-term sustainability is often challenged by fluctuating resource flows.

Pilot or academic projects sometimes begin with out-of-pocket payments by families, but these are typically modest and short-lived, with national insurance or government funding eventually taking over (as in the Philippines). It is emphasized that out-of-pocket payments by families do not support equity and are not suitable for establishing or sustaining large-scale, population-based NBS programmes.

Other relevant learnings linked to financial sustainability include:

- *Sustainable Financing: Integration* into national health insurance or government-funded packages to promote equity and ensure long-term viability.
- *Donor support: While* useful for initial scale-up, these must transition to stable, government-led funding for sustainability.
- *Public–private partnerships and strategic purchasing,* requiring periodic review and renegotiation of agreements by the state to maintain population access at costs that are realistic and affordable for the health system.
- *Education and advocacy efforts*, which improve not just with funding, but also through public and professional awareness, community engagement and strong advocacy.
- *Data systems and monitoring*, with effective tracking, follow-up and quality assurance are critical for programme success and resource allocation.
- *Programme adaptability* to changing needs, resource constraints and evolving health system capacities is also crucial for sustainability.

Rapport building with the surgical and anesthesia team prior to surgery for an orofacial cleft ©Operation Smile

A medical specialist at the Continuity Clinic of the Philippine Children's Medical Center measures the height of a patient with congenital adrenal hyperplasia (CAH) during a routine follow-up visit ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

5. Preparing health systems: reviewing the feasibility of the continuum of care for a prioritized birth defect in the health system

Birth defects are highly heterogeneous and do not lend themselves easily to vertical programmes, making it challenging for ministries of health in LMICs to address them through vertical programmes. Countries typically begin by selecting one or more birth defects that are most relevant to their context. A prerequisite for this selection is a systematic feasibility assessment of health system capacity.

The feasibility framework developed during the consultation provides a structured method to identify strengths and gaps across the continuum of care. This enables countries to determine whether a condition can be effectively integrated into existing systems or whether significant barriers – such as absent specialist services, insufficient infrastructure, or limited human resources – preclude implementation at this time. For example, a lowincome setting may adopt screening for CHDs, but the absence of surgical capacity or regional referral arrangements may limit the ability to provide timely, lifesaving treatment. In such contexts, selecting conditions that are contextually relevant and more feasible to screen, diagnose and manage, may be more appropriate (e.g. congenital hypothyroidism, where laboratory-based screening and ongoing treatment can be readily established; or clubfoot, which is simpler to diagnose and treat).

5.1 Core health system services for birth defects

To ensure effective care for prioritized conditions, the health system must be able to deliver five core functions:

1. **Screening** using laboratory, clinical or visual methods.
2. **Confirmatory diagnosis** through specialized testing or expert clinical assessment.
3. **Referral** to appropriate care pathways.
4. **Management and followup**, including medical, surgical, rehabilitative and long-term care.
5. **Communication, counselling and data recording** through systematic family engagement and documentation across all phases.

5.2 Operational framework to assess health system feasibility

The operational framework (Fig. 1, page 33), developed to review the feasibility and preparedness of the health system to provide the necessary services for a priority condition, was derived from country programmes and insights synthesized from country presentations and expert discussions held across the consultation rounds. These exchanges revealed common principles across countries, feasible entry points, and shared system challenges that shape how NBS, diagnosis, management and followup can be integrated within routine health services in LMICs. The framework consolidates these learnings into a practical, visual screening algorithm to support ministries of health in assessing feasibility, identifying system gaps, and planning the phased implementation of stateled initiatives for birth defects.

If a country selects a particular birth defect to focus on, then specific parts of the health system that provide the services necessary for the screening, diagnosis and management of that condition will need to be strengthened. The screening method – broadly, though not absolutely – can help inform the requirements for downstream pathways to diagnose and manage the condition within the health system (Fig. 1, page 33).

Screening pathways

The heel prick method collects blood for newborn screening by pricking the lateral side of the heel and placing drops onto a special filter card ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

Biochemical screening such as for congenital hypothyroidism or SCD (Fig. 1 – pathway on the left) requires a functional and reliable laboratory network, efficient sample transport systems, and adequately trained personnel to ensure timely sample collection, testing, reporting and initiation of treatment. Effective programmes must include mechanisms to track each infant from screening through confirmatory diagnosis and treatment initiation. Ongoing clinical followup, appropriate medication, specialized diets where needed, and early childhood development and rehabilitation services are essential components of care. Longterm monitoring, structured family counselling, and caregiver education strengthen treatment adherence and support optimal developmental outcomes. Ensuring opportunities for inclusive social interaction is also important, enabling children to participate fully in family and community life and supporting their overall wellbeing.

Screening for congenital heart disease using pulseoximetry in Kerala, India ©Department of Health, Government of Kerala, India

Clinical screening such as for CHDs or hearing impairment (Fig. 1 – middle pathway) depends on specialized equipment (e.g. pulse oximeters or auditory testing devices) and skilled specialists for confirmation (e.g. echocardiography specialists) and treatment. Robust referral pathways, surgical and intensive care capacity (for CHD), and provision of assistive technologies are often required.

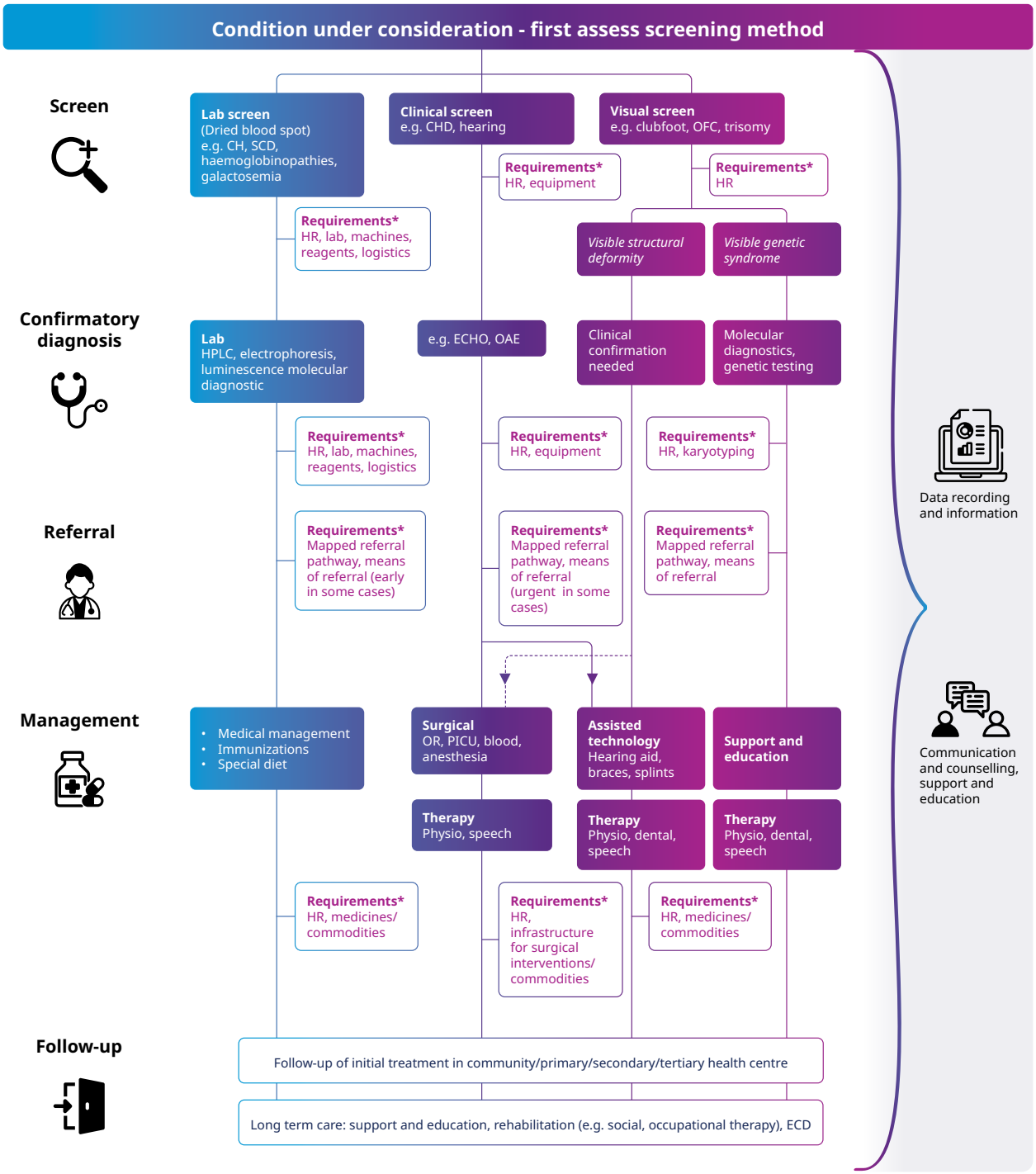
Clubfoot is a birth defect where one or both feet turn inward and downward; with early treatment in infancy, correction is usually achieved within 6–8 weeks (Philippines) ©MiracleFeet

Visual examination at birth such as for clubfoot, gastroschisis, orofacial clefts and Down syndrome (Fig. 1 – pathway on the right) requires trained frontline health workers capable of recognizing abnormalities and initiating timely referral. While certain conditions can be identified reliably through careful physical examination immediately after birth, others may require confirmatory genetic testing. Some conditions require urgent surgical intervention, whereas most necessitate coordinated, multidisciplinary management.

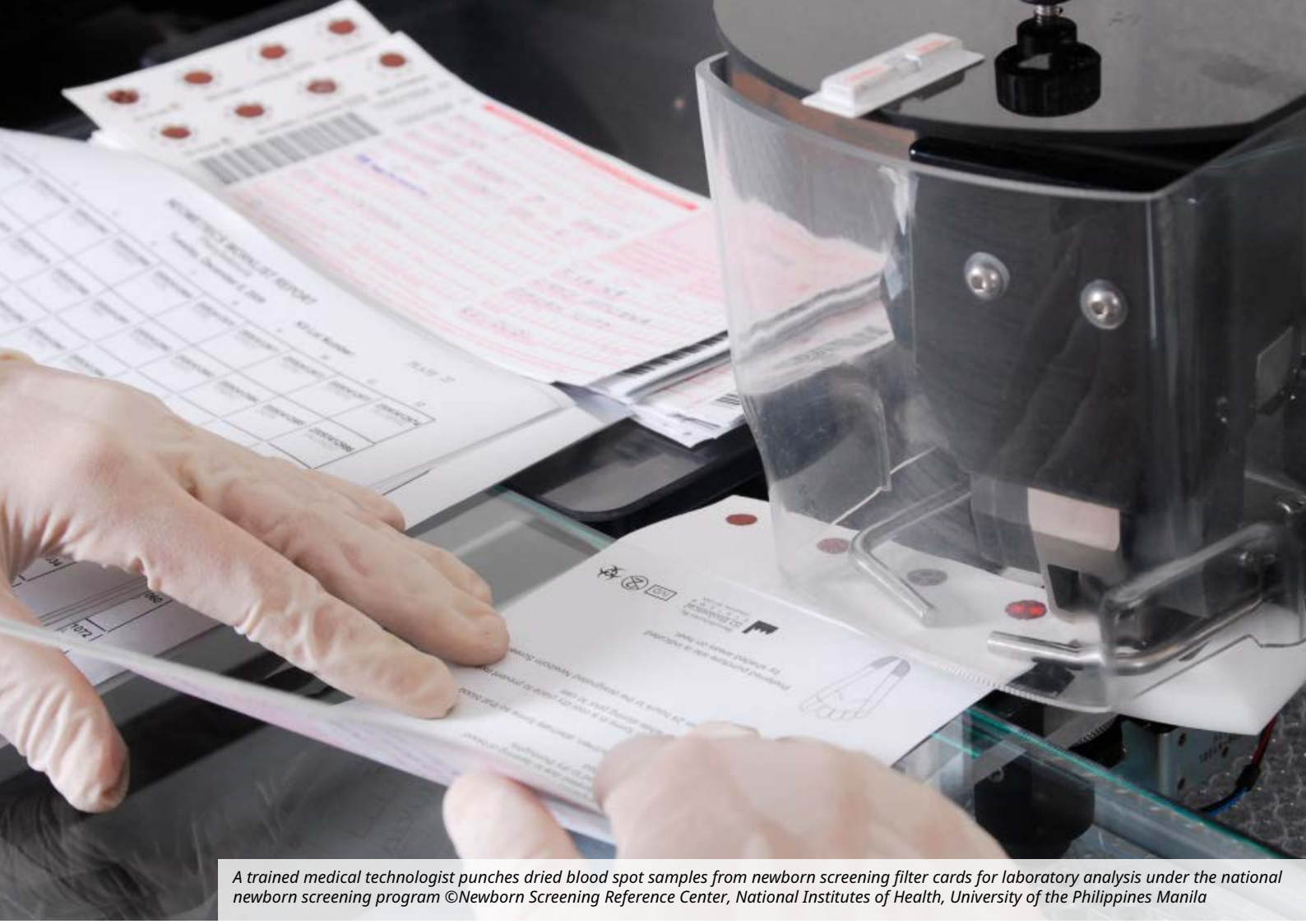
Because infants may present with coexisting anomalies, prompt referral pathways are essential to ensure comprehensive assessment and care. Counselling for families, provision of supportive therapies, and structured longterm followup are integral components of achieving optimal developmental and health outcomes.

In conclusion, across all screening approaches, effective communication with families, clear education materials and functional data systems are critical to ensure that newborns receive timely and appropriate care. The operational framework for the feasibility algorithm (Fig. 1) provides a practical tool to support countries in aligning condition selection with system capacity and planning necessary investments to ensure sustainability and quality of care. As shown in the figure, the importance of communication, education and robust data systems are key to a successful initiative for the screening, diagnosis and management of one or more birth defects.

Fig. 1. An operational framework to assess health system preparedness for newborn screening, diagnosis and management of a selected birth defect



CH: congenital hypothyroidism; ECD: early childhood development; ECHO: echocardiography; HPLC: highperformance liquid chromatography; HR: human resources; OAE: otoacoustic emissions; OFC: orofacial clefts; OR: operating room; PICU: paediatric intensive care unit; SCD: sickle cell disease. (requirements listed are not comprehensive but indicative)*



A trained medical technologist punches dried blood spot samples from newborn screening filter cards for laboratory analysis under the national newborn screening program ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

Janelle, who was screened and diagnosed with congenital hypothyroidism through newborn screening, is now 29 years old, works as an engineer ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

6. State-led partnerships to address birth defects

Partnerships led by ministries of health are central to the successful implementation of NBS, diagnosis, and management initiatives in LMICs. Countries highlighted several partnership types that strengthen programme delivery**.**

Public–private partnerships: In countries with large private health sectors, ministries of health often collaborate with private hospitals, clinics, and laboratories to expand service availability for the population, especially for specialized diagnostics and surgical care (e.g. Kerala, India). These partnerships help address gaps in public sector resources, reduce treatment delays and improve access for children needing complex interventions.

Nongovernmental organizations and civil society: NGOs provide technical expertise, supplementary funding, training, and community outreach. They are especially important for capacity building, advocacy, and family support).

Academic and research institutions: Universities and research centres generate pilot data, support monitoring and evaluation, and provide evidence that informs policy decisions and programme scaleup.

Community and parent groups: Engaging families and parent support groups strengthens advocacy, reduces stigma and ensures that programmes are responsive to the lived experiences and priorities of affected communities.

Other government departments: Collaboration with departments of education, social welfare, and local government is essential for integrated care, inclusive education and social support. Multisectoral coordination ensures children receive comprehensive services beyond health.

6.1 The Philippines: newborn screening programme – partnerships between the Department of Health, academia and national insurance

The Philippines NBS programme demonstrates the critical role of partnerships in scaling and sustaining national health initiatives. Initially launched as a research project, the programme expanded into a nationwide effort through collaboration between government agencies, academia and local governments. The Department of Health became the lead agency following the Newborn Screening Act of 2004, while the Philippine Health Insurance Corporation integrated NBS into its newborn care package, ensuring financial coverage. The University of the Philippines provided technical oversight, further strengthening the program's scientific foundation.

Regionalization of laboratories and the establishment of NBS continuity clinics – staffed by nurses and paediatricians – were achieved through coordinated efforts between the Department of Health and academic partners. Decentralization required close collaboration with municipal, provincial and city governments to maintain national standards and adapt policies to local contexts. These partnerships have been essential for ensuring programme continuity, quality and sustainability.

Lessons from the NBS programme highlight that shared leadership, legislative support, insurance coverage, and cross-sector partnerships are vital for success; ongoing engagement with local governments and stakeholders, as well as effective communication and coordination across sectors, are necessary for full implementation and long-term impact.

6.2 United Republic of Tanzania: Clubfoot Treatment Program – partnerships between the Ministry of Health and NGOs

The United Republic of Tanzania's Clubfoot Treatment Program exemplifies the importance of robust partnerships in scaling up care for birth defects. The Ministry of Health, NGOs and the President's Office, Regional Administration and Local Government collaborate to improve access to clubfoot treatment. Previously, treatment was limited, costly, and lacked coordination. Through partnership, the programme now uses the cost-effective Ponseti Method, with NGOs supplying materials and braces, while the Ministry of Health funds staff and facilities. The Ministry of Health leads policy development, stakeholder coordination and system strengthening, while NGOs provide training, supervision, data collection and resource mobilization. Service delivery is integrated into maternal and child health services, with 67 clinics functioning nationwide and community health workers trained for early detection and referral. The Steering Committee, led by the Ministry of Health and main partners, oversees governance, guideline development and joint supervision. Supplies are distributed by the Medical Stores Department, and efforts are underway to include clubfoot care in national insurance. These partnerships ensure standardized, sustainable and equitable care, though challenges remain in financing, integration into insurance and resource allocation. The collaborative model, which has now enrolled >50% of newborns with clubfoot into treatment, demonstrates how multisectoral engagement is essential for effective, scalable birth defect programmes.

Perspective
Jealous Chibare, Zimbabwe, father of Sympathy, born with clubfoot

My daughter Sympathy was born in 2014 at our home in Zimbabwe. At the time, we had no idea that she was one of 600 babies born with clubfoot that year in our country. Many children with clubfoot in LMICs don't receive the simple treatment that could prevent lifelong impairment, but fortunately, my daughter was not one of them.

We first noticed her right foot didn't look normal at birth; she cried in pain when I tried to hold it. I was overwhelmed with fear and uncertainty about her future. The shock and hopelessness were immense. Our community's reaction made things harder, people believed my wife was bewitched, and we became socially isolated. Despite this, I resolved to be strong and support my family, determined to give Sympathy the best life possible. I told my wife that we would not let fear dictate our actions and that we would find a way to help our daughter thrive.

About a week after her birth, my wife and I visited the local clinic, 5 km away from our home to register Sympathy and receive postnatal care. Here a nurse diagnosed clubfoot and referred us to a hospital. There, we learned about the Ponseti method – plaster casts, a tenotomy, and a brace – which was provided free by a local NGO. Treatment began when she was 11 days old. It wasn't easy: we faced logistic challenges, negative comments, and long clinic waits. But after two months of casts, her feet were straight, and she moved to the braces stage, which she accepted after some initial protests. Her treatment was successful, and she started enjoying activities like running and playing.

Seeing other parents struggle, I became a volunteer at the clinic to support and encourage them. Many families face stigma, isolation and financial hardship. Mothers often endure abuse and abandonment. My greatest joy is seeing children successfully treated and encouraging fathers to be advocates for their families. I hope one day clubfoot treatment is routine and accessible for all. Today, Sympathy enjoys learning, playing and sports, and I am proud to have played a part in her journey and in supporting other families like ours.

Jealous Chibare with his daughter Sympathy
©Jealous Chibare

6.3 India (Kerala): Hridyam programme for congenital heart defects – public–private partnership with the private health sector for provision of surgical services and follow up

The public–private partnership model in Kerala's Hridyam programme for CHDs was established to expand access to timely diagnosis, surgery and followup care, and to promote equity across the state. Under this model, the government partnered with private hospitals with established capacity to diagnose and manage CHD and negotiated standardized reimbursement rates to ensure affordability and wide population coverage. Key strategies included expanding surgical access to address historical backlogs in CHD surgeries caused by limited public sector capacity, implementing standardized clinical protocols to ensure consistent care, strengthening surveillance and monitoring, and facilitating capacity building and knowledge sharing through national and international partners. Emphasis was also on improving emergency transport for critical cases, enhancing followup systems, and aligning practices with global standards.

Implementation relied on costceiling agreements with private hospitals, a centralized digital platform integrating public and private data, and coordinated training across sectors.

Challenges in programme implementation were addressed through targeted systemstrengthening measures. Limited provider awareness was mitigated through standardized protocols and focused training across public and private hospitals. To compensate for weak rural diagnostics and followup capacity, empanelled urban hospitals conducted periodic outreach clinics. Regionalization expanded the number of capable centres, reducing travel distances and improving timely access to surgery. Because complex neonatal cases required intensive resources, the government financed presurgical care, introduced incentives, and revised costing models for followup services. A continuous monitoring and evaluation system enabled realtime tracking of outcomes, supporting timely corrective action and sustaining programme quality.

The Kerala experience illustrates how structured public–private partnerships can effectively expand specialized newborn care and strengthen systemwide quality.

Confirming a positive screen for congenital heart disease by echocardiography in Kerala, India ©Department of Health, Government of Kerala, India

6.4 Role of champions in newborn screening

Champions – clinicians, parents, academics and advocates – have contributed significantly to advancing NBS for birth defects in some LMICs. Emphasis stems from sustained commitment, often catalysing programme initiation, scaleup and continuity, and helping maintain momentum and accountability once services are integrated into routine health systems.

In the Philippines, champions drove early legislative change and insurance coverage, though longterm sustainability was achieved only when government leadership and system integration were established. In other contexts, such as in Sri Lanka, India's RBSK and Kerala's Hridyam programme, public health authorities led programme development with limited reliance on champions, guided primarily by political commitment.

Experts noted that while champions are valuable, conditionspecific advocacy can risk creating vertical, poorly integrated systems. Effective approaches involve multidisciplinary collaboration, coalitionbuilding across conditions and alignment with stateled strategies to ensure equitable, scalable and resilient NBS programmes.

Munni (extreme right) was born with ectopia vesicae (i.e the inner surface of the bladder is visible through a defect in the abdominal wall). She underwent surgery as an infant to close the bladder and abdominal wall, and again as a teenager to correct incontinence. She completed high school, is married and delivered a healthy boy, who is now 2 years old ©Tahmina Banu

7. Data systems in initiatives to address birth defects – linking newborn screening to diagnosis, management and follow up

Newborn screening is not an end in itself; it must be linked to subsequent steps in the continuum of care. The discussions began with the importance of a data system in an NBS initiative, followed by a presentation on the data systems being used in LMIC programmes to address birth defects.

The NBS framework consists of screening, follow-up, diagnosis, management (including long-term care), and evaluation (of the programme), unified by education. Data systems, which may be simple, help ensure that the newborn moves through the continuum of care, optimizing outcomes for the child. They also support monitoring of programme quality, and provide data for economic evaluations, burden of disease studies and other analyses.

Simple data systems are essential and sufficient to register and track newborns and children from screening through diagnosis, referral and followup. Many LMIC programmes begin with basic data tools that ensure clinical continuity and enable monitoring of programme performance. However, data systems remain a common challenge in LMICs

Data collection process

Data collection for NBS begins at the birthing facility, where demographic information is recorded during registration and newborn details – such as record number; date of birth; weight; and sex – are added at birth. For bloodspot screening, laboratories document specimen information and return results to the facility. For clinical screening (e.g. hearing or pulse oximetry), results are captured in clinical notes or screening registers. Families of newborns with positive screens are promptly contacted for confirmatory diagnosis, and these results are incorporated into the database. Confirmed cases are linked to treatment pathways and longterm followup systems that track treatment adherence, service use and outcomes. The same data system can be expanded to include additional conditions over time, regardless of screening modality. Countries emphasized starting with simple systems, avoiding unnecessary data collection. Providing feedback to data providers is important to improve data quality, strengthen decisionmaking, and support better patient management.

Nurses play a key role in the clinical care of the child and support to family of a child with an orofacial cleft (Guatemala) ©Operation Smile

7.1 Data systems for programme success and organizational transformation – experience from the Hridyam programme in Kerala, India

The Hridyam programme for CHDs uses a comprehensive, stateled data system that tracks each child from initial registration through longterm followup, covering all cases up to 18 years of age. The system supports both individual case management and programme implementation, including hospital allocation for surgery and financial processing.

Data collection begins at registration, where demographic and clinical information is recorded. Multidisciplinary teams – including DEICs, cardiologists, surgeons and community nurses – enter details on assessments, procedures and ongoing management.

Data processing includes quality checks, tracking of bed availability and surgical slots, and prioritization of cases based on urgency. Hospital assignments are displayed transparently, and community nurses are allocated for followup. Payments and reimbursements are automated, with strong data security measures, including encrypted storage on government servers.

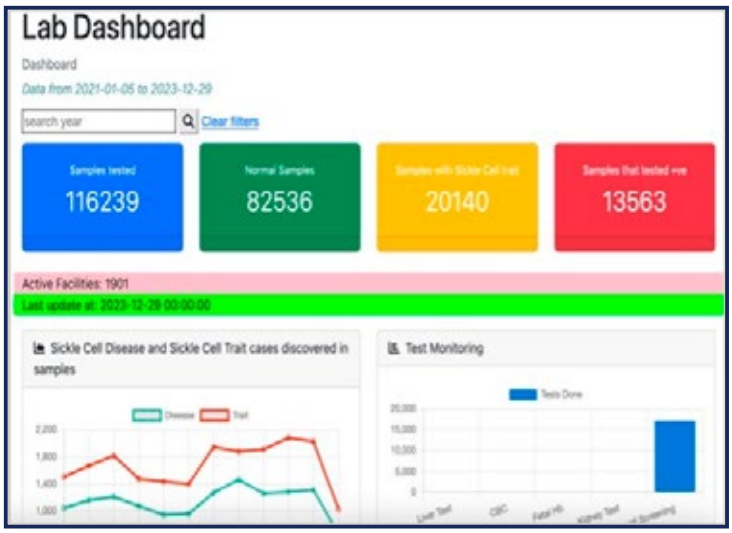
Data visualization is enabled through a realtime CHD registry and dashboard that monitor patient status, programme performance and key indicators. Spatial and demographic analyses inform planning, capacity building and policy refinement.

The system has strengthened operational efficiency, accelerated expert reviews, optimized referral pathways and facilitated responsive programme adjustments. Datadriven insights have improved diagnostic protocols, reduced patient travel and enhanced communication – supporting timely escalation of urgent cases and overall programme quality.

7.2 Data systems set-up – experience from the sickle cell disease programme in Uganda

The programme uses both paper and electronic forms to collect data for (i) NBS, treatment monitoring (including laboratory data), and (ii) clinical management. Paper forms are completed at the point of care and later entered into electronic systems.

Laboratory data and dashboard: Data from paper forms accompanying samples sent for screening and hydroxyurea treatment monitoring are entered into the Sickle Cell Laboratory Information Management System. This system provides real-time dashboards that display key metrics (samples tested, trait positivity, disease positivity) and hydroxyurea monitoring results when performed.



Laboratory dashboard: Sickle cell program, Uganda ©Sickle Cell program, Uganda

Clinical visit data and dashboard: The SCD clinical visit form records comprehensive information on each patient's treatment, including vital signs, history, examinations, investigations, diagnoses and management. Data from ten Centres of Excellence is uploaded to a central database, enabling real-time monitoring and tracking of key indicators (e.g. patients presenting with complications, patients started on hydroxyurea).

Data-driven decision-making: The integrated dashboards support datadriven decisionmaking. Programme data have been used to target newborn screening in highburden districts, resulting in increased case detection.

Future developments include establishing a longitudinal SCD register and a patient passport to support continuity of care and improve portability of clinical information. Indicators related to SCD are being incorporated into national reporting systems. NBS information is now included in the national newborn register and on child health cards to prompt screening before discharge and during immunization visits. These efforts aim to strengthen routine SCD care and improve data accessibility across the health system.

7.3 Data systems for linking screening, diagnosis and management – experience from the congenital heart defects programme in Argentina

Argentina's Federal Network for the Care of Congenital Heart Defects demonstrates how integrated national data systems can strengthen programme coordination, quality and equity. A central information platform links hospitals, government agencies and clinical teams, enabling realtime tracking of every child from diagnosis through postsurgical followup.

The system supports both clinical and administrative functions, including patient allocation, financial management and hospital accreditation. Integrated digital tools facilitate allocation of infants to appropriate hospitals based on urgency, complexity and proximity, while monitoring bed availability and programme capacity nationwide.

A National CHD Registry ensures mandatory reporting and standardized medical records, improving continuity of care and interoperability across facilities.

Automated billing and payment systems streamline financial processes, and compliance incentives encourage timely reporting, acceptance of referrals, and completion of clinical documentation.

The data system also underpins performance monitoring. Surgical hospitals are evaluated through a composite scoring framework that assesses their contribution to the network, quality of care, patient experience, and surgical outcomes benchmarked against international standards. These evaluations inform action plans led by the Ministry of Health. Key lessons highlight the importance of focused data collection with clear objectives, phased system expansion, standardized and interoperable tools, dedicated data management capacity, and alignment of financial and reporting mechanisms. Together, these elements enable effective digital transformation and improved national CHD outcomes.

Club foot is correctable with the Ponseti method- a series of weekly casts over 4-6 weeks, delivered by trained workers across the health system, including primary care (Cambodia above) and in community (Cote d'Ivoire below) ©MiracleFeet

8. Engaging multiple levels of the health system in newborn screening

Engaging every level of the health system is essential for equitable access to early detection, effective referral, management and comprehensive care for birth defects. An integrated approach improves outcomes, reduces preventable disability and mortality, and strengthens overall system performance.

This section presents examples from national programmes for three conditions, illustrating how different healthsystem levels contribute to birth defect initiatives.

8.1 National programme for congenital hypothyroidism (Brazil)

Primary care: Basic health units and family health programmes collect and transport dried bloodspot samples, educate families, communicate test results, refer newborns, provide levothyroxine, and support adherence and followup.

Secondary care: Specialized clinics and general hospitals run NBS laboratories, coordinate multidisciplinary teams, confirm diagnoses, guide treatment, and offer ongoing followup, education, and counselling.

Tertiary care: Highly specialized hospitals manage rare or complex cases, though most congenital hypothyroidism activities occur at primary and secondary levels.

Strengths and challenges
National guidelines and diagnostic algorithms are in place, and early sample collection (within five days) supports prompt treatment. However, variability in sample collection timing across states and delays in followup can hinder timely care. Strengthening primarylevel NBS services remains a priority for improving effectiveness.

8.2 National Sickle Cell Anaemia Elimination Mission (India)

This Mission is a comprehensive, multilevel initiative designed to address the high burden of SCD, particularly among tribal populations. The Mission is structured to involve all tiers of the health system, from community outreach to tertiary care – ensuring a continuum of prevention, screening, management and support.

Community and primary care: Screening begins at the grassroots, with door-to-door campaigns and health camps in communities, as well as institutional screenings at birthing points, primary health centres and schools. Community health workers play a vital role in raising awareness, conducting screenings, monitoring treatment adherence, educating families and facilitating disability certification. They are also instrumental in ensuring follow-up and connecting families to higher levels of care.

Secondary and district-level care: District hospitals provide separate wards for SCD patients and serve as referral centres for confirmatory diagnosis and management of screen-positive cases. They are equipped to handle complications and provide ongoing care, including prophylactic therapies (vaccinations, folic acid, penicillin) and essential medications such as hydroxyurea. Workforce training and the development of clinical guidelines ensure standardized care at this level.

Tertiary and specialized care: Centres of Competence at the tertiary level offer advanced diagnostics, surgery, and bone marrow transplantation. These centres also support advanced research,

including gene-editing technologies, and are integrated into the national health insurance system to provide cashless treatment.

System integration and partnerships: The Mission is supported by a dedicated SCD registry and portal for tracking patients, the introduction of telemedicine in endemic areas, and public–private partnerships to ensure affordable screening and uninterrupted supply of equipment and drugs. Interdepartmental collaboration – across health, tribal development, education, research, social welfare, and information technology – ensures a holistic approach.

By engaging every level of the health system, the mission strengthens prevention, early detection, comprehensive management and long-term support for persons with SCD.

8.3 Clubfoot programmes

Primary care in clubfoot treatment (multiple countries)

Primary care is foundational for clubfoot initiatives, even though treatment often occurs at secondary or tertiary levels. Primary care is crucial for raising awareness, early identification, referral and supporting long-term treatment adherence.

- Early identification and referral: Screening during newborn care, routine checkups, immunizations and wellchild visits enables early detection and prompt initiation of treatment.
- Supporting families: Primary health care providers guide families through the long treatment period, conduct community visits to reinforce brace use, encourage followup attendance, and monitor for complications.

Training and referral data: Across 15 countries, 75% of referrals originated from primary care sources, highlighting the central role of primary care in successful clubfoot programmes.

Country examples of involvement of primary health care and community-based approaches in clubfoot management:

- **Ethiopia:** Primary care units and community health workers are trained to integrate clubfoot screening into newborn and immunization visits. Support materials, clear referral pathways, and mobile apps for tracking and access are used to strengthen the system.
- **Nigeria:** Primary care workers are trained using visual aids and demonstrations, with a focus on addressing stigma. Their close community ties have increased early treatment rates and reduced dropouts, with additional support through home visits and peer groups.
- **Pakistan:** In regions with high home birth rates, lady health workers are trained to identify and refer clubfoot cases, supported by mobile apps for efficient tracking and follow-up.
- **Nepal**: The “Motorbike Mobile Ponseti Service” enables practitioners to reach children at home, reducing dropout rates and strengthening links with local providers. Telemedicine is also used for ongoing support.

Primary care underpins successful clubfoot initiatives by enabling early detection, continuous family support and effective referral. Strong integration with secondary care – supported by training, community platforms and digital tools – is essential for sustained programme performance in diverse LMIC settings.

9. Timing of newborn screening

Hospital admission for childbirth, prior to discharge of the mother and newborn, offers an opportune time for NBS. In some situations, certain LMICs have postponed the screen to a later point – for example SCD screening during immunization visits. In other countries, NBS programmes may face challenges due to early discharge of mothers and babies from hospital. These consultation sessions discussed the timing of screening in different LMICs, and the strategies used to address challenges related to early discharge.

9.1 Newborn screening in the context of early hospital discharge after birth (<24h)

In many countries, mothers and newborns are discharged within 24 hours after birth, which can make it difficult to complete NBS before they leave the hospital. This situation has been encountered in both high- and low-income settings. In high-income countries, home visits for screening after discharge are not uncommon. Below are some approaches adopted by a few front-runner LMICs.

Newborn screening for congenital hearing impairment ©Otto Mejia

The Philippines

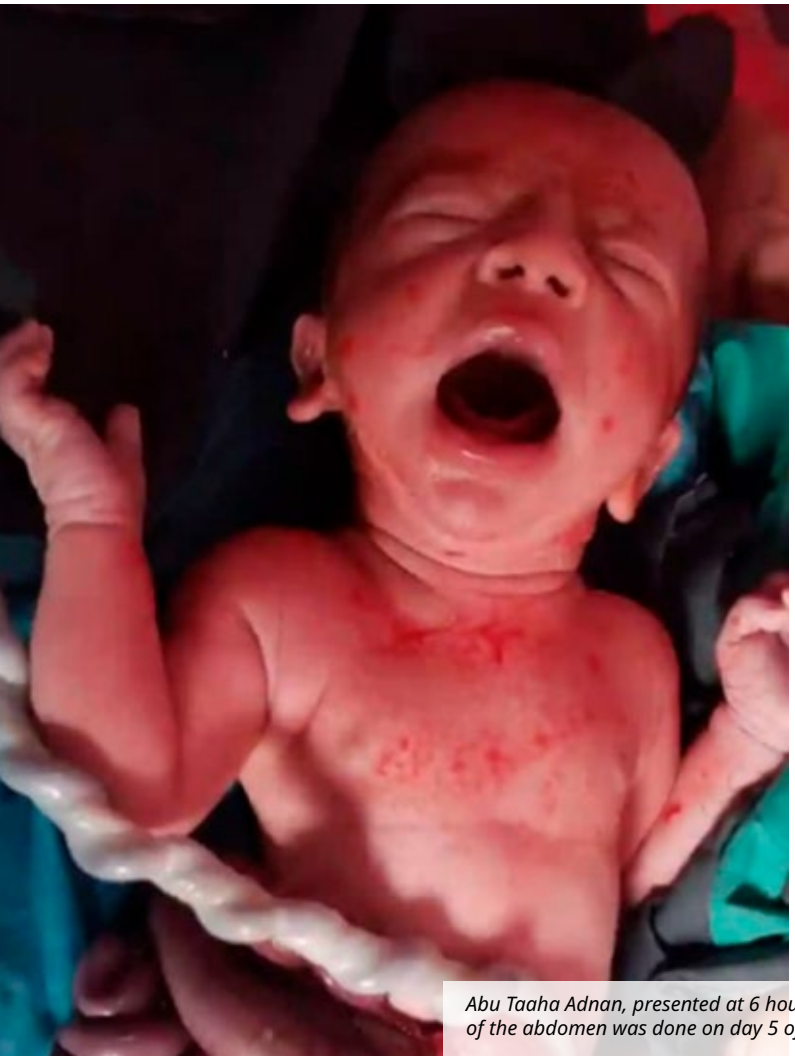
Hospitals in the Philippines are encouraged to keep mothers for at least 24 hours so dried bloodspot testing can be performed at that time. Testing before 24 hours is avoided to reduce the need for retests. Despite this, some infants still miss screening due to early discharge or because of the small number of home births. National communication campaigns encourage families to return promptly if screening was missed. Each year, approximately 1.4 million newborns (97%) are screened, leaving about 40 000 unscreened. Recent outreach has reached an additional 20 000 infants in remote southern islands, with ongoing efforts to reach the remaining unscreened newborns.

Strategies used: High coverage is achieved through strong communication efforts encouraging families to return at 24 hours for NBS; standardized protocols supported by the Ministry of Health; and communitylevel tracking and recall systems that help ensure that missed newborns return for screening.

India

Nine states, including Kerala and Tamil Nadu, have policies mandating a minimum 48hour postpartum stay. This ensures adequate time to complete NBS prior to discharge.

In India, NBS focuses primarily on clinical methods. For CHD, screening is recommended after 24 hours of life to reduce falsepositive results. Similarly, early hearing screening may yield higher falsepositive rates; timing is adjusted to optimize test accuracy.



Abu Taaha Adnan, presented at 6 hours of age. After initial stabilization and placement of a silo, bowel reduction and closure of the abdomen was done on day 5 of life. He went home 5 days later. Now he is a happy, playful 3 year old ©Nusrat Mona

9.2 Newborn screening for sickle cell disease in Ghana, in the absence of a population based newborn bloodspot screening programme

Sickle cell disease becomes clinically apparent between four and six months of age, making early detection – preferably before four months – critical. Early identification enables timely counselling, initiation of penicillin prophylaxis, ageappropriate pneumococcal vaccination, malaria prevention, introduction of hydroxyurea after nine months, and prompt management of acute complications.

In highincome countries and several LMICs (e.g. India, the Philippines, Uganda), SCD screening is integrated into universal NBS programmes using centralized dried bloodspot testing. These systems are supported by strong laboratories, reliable logistics, and qualityassured highvolume processing, allowing expansion to multiple disorders.

However, in many LMICs – particularly in subSaharan Africa – implementation is constrained by limited laboratory capacity, early postnatal discharge (often within six hours), and high rates of home births.

Ghana's experience: A pilot initiated in 1995 led to the launch of a national NBS programme in 2010, followed by a scaleup plan adopted in 2011. However, as of 2024, coverage remains incomplete. Data from a major teaching hospital show a 74% retrieval rate for positive cases, with 14% lost to followup. Delays in result turnaround and limited tracking systems impede timely counselling and enrolment in care – challenges that are common across the region.

Pointofcare testing as a transitional approach: These tests offer lower costs, easier integration into primary care, and higher retrieval rates, enabling counselling and digital data capture. However, they are less suited for populationlevel programmes due to manual data entry and limited scalability. Evidence from a district in Ghana shows that integrating point-of-care testing into immunization clinics (0–5 years) resulted in good enrolment, timely initiation of prophylaxis and hydroxyurea, and improved outcomes.

In settings with high SCD burden and limited NBS infrastructure, point-of-care testing can serve as an interim solution to facilitate early identification and timely intervention until populationbased NBS is established.

Newborn screening by blood spot in the Philippines – blood spot cards on a rack awaiting further processing ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

Newborn with a congenital heart defect being transported to a higher level facility ©Department of Health, Government of Kerala, India

Ambulance for neonates with a congenital heart defect who require urgent referral. This ambulance is designed for neonates and can be used for emergency referral regardless of condition ©Department of Health, Government of Kerala, India

Shifting a newborn with a congenital heart defect to a higher level facility ©Department of Health, Government of Kerala, India

10. Emergency referral for newborns with structural birth defects

Referral transport is a critical component of NBS systems, ensuring that infants identified with potential disorders receive timely diagnostic confirmation and life-saving treatment. Without functional transport, the early detection provided by screening cannot be translated into improved health outcomes.

This has been a challenge in many LMICs. Several life-threatening birth defects require infants to be transferred urgently to centres where they can receive lifesaving treatment. Participants from Brazil and Kerala, India presented their efforts to facilitate emergency referral transport.

10.1 Emergency referral transport in the Hridyam programme, Kerala, India

The Hridyam programme commonly encounters four types of referral scenarios in newborns with CHDs: (i) a stable newborn with a failed pulse oximetry screen, (ii) a baby who desaturates before screening and is admitted to the neonatal ICU, (iii) a newborn in the neonatal intensive care unit with a known CHD diagnosis, and (iv) an infant scheduled for surgery or experiencing a postsurgical emergency. The first scenario is not urgent, as it is relatively low risk, while the remaining three require urgent, wellcoordinated emergency transport.

Before Hridyam, neonatal transport systems were inadequate. Few ambulances were equipped for newborns, no specialized neonatal transport teams or ventilators were available, and transfers were often conducted in adult ambulances without prior communication to receiving hospitals, leading to delays and safety risks.

Hridyam, a public–private partnership programme, strengthened emergency referral by deploying six strategically located ambulances identified through GIS mapping. Staff received specialized neonatal transport training, and standardized protocols were developed with international partners and NGOs. A mobile application now links referring facilities, receiving hospitals, and transport teams, enabling realtime sharing of patient information and continuous monitoring during transit.

This strengthened system has safely transported more than 200 critically ill children, with all costs covered by the government, eliminating outofpocket expenses for families.

10.2 Referral pathways for critical congenital health defects in Brazil

Since 2017, Brazil has implemented a national plan for paediatric CHD covering prenatal and newborn diagnosis, pulse oximetry screening, safe transport, surgery and multidisciplinary care. Critical CHD cases follow two referral pathways: *prenatal detection*, allowing delivery at specialized hospitals for immediate management; and *postnatal diagnosis*, where infants identified through examination, pulse oximetry, or echocardiography are referred to surgical centres with neonatal intensive care capacity. Free ground or air transport is provided by the government.

However, major regional disparities persist. Compared to the south, the north of the country has fewer hospitals, longer distances, limited intensive care capacity, with referrals often managed manually and sometimes requiring interstate air transport.

Key challenges include administrative delays for interstate transfers, intensive care unit overcrowding, and long waiting lists, all of which increase the mortality risk for affected newborns.

Dana, diagnosed with phenylketonuria through the newborn screening program, at a follow up clinic, where she is monitored and managed with special formula and diet ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

Dana, diagnosed with phenylketonuria through the newborn screening program, is well today and enjoys her music ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

11. Short- and long-term follow-up in initiatives for newborn screening, diagnosis and management of birth defects

Consultations emphasized that screening alone was not sufficient. Responsibilities and processes for patient follow-up after NBS ensure that infants with a positive screen are recalled, a confirmatory diagnosis is made, and families are referred along care pathways for management to optimize outcomes. Programmes in LMICs have approached these processes differently, particularly regarding: (i) who tracks patients with a positive screen to ensure a confirmatory diagnosis, (ii) who refers patients to specialists for further treatment, and (iii) who monitors adherence to the advised management. This segment of the consultation began by describing short- and long-term follow-up in the context of NBS programmes to ensure continuity of care, followed by country examples of follow-up care.

Effective followup in NBS requires a reliable database to track screened conditions. Countries may use a single system for all conditions or separate databases for each one. The purpose of followup is to ensure that infants with positive or unclear results are quickly located, receive confirmatory testing, and are connected to appropriate treatment and support. Followup data also help evaluate and improve programme performance.

Short-term follow-up involved rapid communication of positive or invalid results to families – usually through the birth facility – and facilitating access to specialist services for diagnostic confirmation. It concludes once a diagnosis is confirmed and initial management begins.

Long-term follow-up provided ongoing, coordinated care for children with confirmed conditions. It ensures that interventions continue over time and supports assessments of whether the NBS programme is improving health outcomes.

Insight-sharing sessions elaborated that for both types of follow-ups, programmes must clearly define what constitutes a positive case, establish protocols for confirmatory testing, and maintain thorough documentation to support effective treatment and monitoring.

11.1 Stakeholder roles in newborn screening follow-up

The roles outlined below were discussed during the consultations and reflect shared perspectives on how families, programmes and providers can contribute to effective NBS follow-up.

- **Families** play a central role by engaging with health care providers, sharing accurate information, and attending examinations and tests. Their involvement ensures timely care and completion of all steps in screening and management.
- **Screening programmes** follow established protocols, support families, facilitate confirmatory testing after a positive screen, and document each case to maintain quality and accountability.
- **Primary health care providers** conduct initial screening, communicate results, link families to the NBS programme, and assist with followup. They are essential for early detection and for guiding families through the next steps.
- **Paediatric specialty providers** perform confirmatory testing, advise on clinical management, and work with primary care providers to ensure accurate diagnosis and appropriate treatment.
- **Specialized clinicians** (e.g. endocrinologists, surgeons, physiotherapists) initiate treatment for diagnosed conditions and provide longterm followup, collaborating with primary care providers to monitor progress and adjust care as needed.

11.2 Planning resources for short- and long-term follow-up

Reflections on the Philippines newborn screening programme

The Philippines NBS programme initially focused on testing and shortterm followup, but it soon became clear that planning for the full care pathway – from screening and diagnosis to treatment and longterm management – was essential. As the programme expanded, the budget was adjusted to include broader followup needs. The insurancefunded NBS fee now supports the continuum of care, including some aspects of longterm followup.

Sustainability has become a priority. The programme clearly distinguishes shortterm needs (confirmatory testing, specialist fees, recalls and transport for indigent families) from longterm needs (laboratory tests, ongoing treatment, specialized foods, consultations, followup staff and compliancerelated transport). Each added service requires careful budgeting. While integration into public health funding is ideal, limited government resources mean that regional offices, donors and some outofpocket payments still contribute – though reducing outofpocket costs remains a key goal.

Reflections on the Hridyam programme in Kerala, India

The Hridyam programme for CHDs relies on strong human resources, with trained staff handling screening, diagnosis and communitylevel followup. A dedicated information technology system supports coordinated data management integrating inputs from nurses and physicians to maintain a seamless continuity of care. Parent education materials are adapted to local languages with support from partners.

Infrastructure has been strengthened at district and tertiary levels, including facilities for followup, echocardiograms and neurodevelopmental assessments. Districts are equipped with echocardiography machines, and RBSK nurses are provided with pulse oximeters.

To improve efficiency, paediatricians and neonatologists are trained to perform echocardiograms, and RBSK nurses manage followup care. The programme also leverages existing DEICs for neurodevelopmental screening and therapy. Capacity building is supported through partnerships with private hospitals and NGOs.

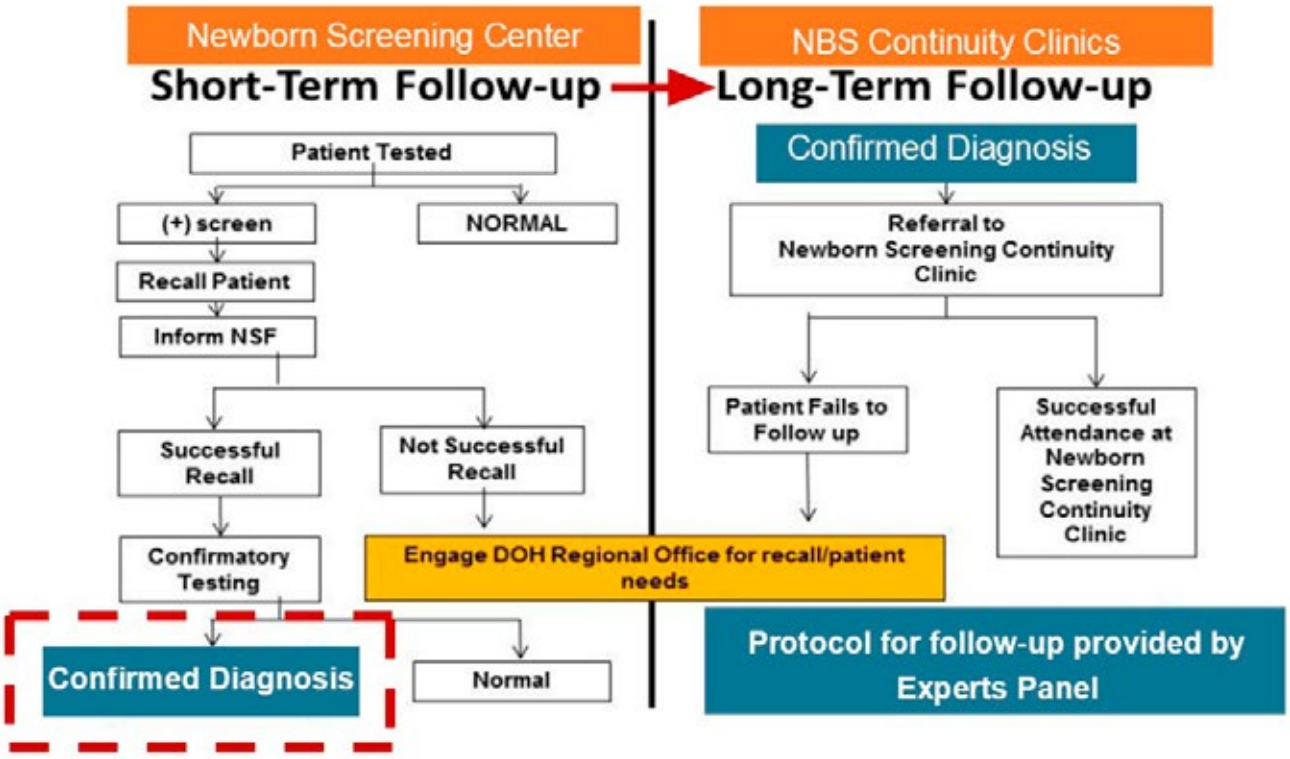
11.3 Mechanisms for short-term and long-term follow-up

11.3.1 The Philippines newborn screening programme

Dried bloodspot samples from hospitals and birthing centres are sent to seven newborn screening centres across the Philippines. When a result is positive, the newborn screening centre's shortterm followup team works with the facility's NBS coordinator to recall the newborn, assess the infant and arrange confirmatory testing. They ensure timely communication of abnormal results, confirm diagnoses and guide families into appropriate care. Confirmed cases are referred to newborn screening continuity clinics for longterm management.

If families cannot be contacted, newborn screening centres coordinate with regional health offices and local health workers for home visits; persistent nonresponse results in "lost to followup." For inborn errors of metabolism, urgent confirmation, specialist consultation, and hospital admission for sick infants are prioritized. Challenges include reaching families in remote areas, coordinating promptly with subspecialists, and ensuring collaboration with local health units.

Longterm followup is provided by 33 continuity clinics, mostly in tertiary hospitals, established in compliance with the Newborn Screening Act of 2004, as part of the network for referring and managing all positive cases. They are staffed by nurses and parttime physicians with access to specialists such as nutritionists, endocrinologists, haematologists and geneticists. These clinics offer ongoing consultations, treatment guidance, diet plans, monitoring of treatment adherence, appointment management, counselling, and coordination of additional support services. Regional health offices support activities, help track patents, and facilitate specialist consultations, home visits and delivery of medication or diets, if necessary.



Pathways for short- and long- term follow up in the newborn screening program of the Philippines ©Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila

11.3.2 Hridyam programme in Kerala, India

The Hridyam programme for CHDs provides a structured system for detecting and managing CHDs in newborns. Because nearly all births occur in hospitals, screening is widely implemented. Newborns with positive screens receive confirmatory echocardiograms at tertiary hospitals or DEICs.

The programme maintains strong monitoring systems, tracking screening, failed screens, followup echocardiograms, confirmed cases, registrations, surgeries and cases lost to followup. Once CHD is diagnosed, cases are registered electronically, and RBSK nurses conduct followup according to conditionspecific protocols. All interactions are recorded in a centralized portal to ensure continuity between primary care teams and specialists.

Screening is ideally done within 24–48 hours, but infants missed at birth are screened later during routine immunization visits. Noncritical cases are followed up according to the immunization schedule, while critical cases receive neonatal surgery and protocol-driven intensive followup. The protocol allows cases to shift quickly from non-critical to critical, if complications arise.

Key challenges include high falsepositive rates from pulse oximetry, reliance on equipment and trained staff, significant staff turnover, the risk of missing milder cases, and community stigma that affects followup.

Continuity of care into adulthood for individuals with congenital conditions

Based on inputs gathered during the consultation, countries had not yet formally planned for continuity of care into adulthood for individuals with congenital conditions, resulting in gaps once they aged out of childhood services. Country-specific experiences outlined below demonstrate both the challenges and emerging solutions.

India (RBSK): The RBSK provides care only up to 18 years. After this, the responsibility shifts to the Department of Empowerment of Persons with Disabilities, which manages disability identification, education, skills training and rightsrelated support. Coordination between ministries occurs through DEICs.

The Philippines: Followup care for congenital conditions is mandated by law. Transition clinics are being developed in government hospitals to support the shift from paediatric to adult care, with plans for national scaleup.

Kerala, India (Hridyam programme): By the third year of implementation, the need for adolescent and adult CHD services became clear. Efforts are underway to develop adult CHD care and provide counselling, particularly for women with complex CHD who are planning pregnancy. These services currently exist in only a few hospitals and require further advocacy, clinician training and formal guidelines for broader implementation.

11.4 Communicating with families after a positive screen

11.4.1 Experience from the Philippines newborn screening programme

The Philippines NBS programme prioritizes early, effective communication with parents, beginning in the prenatal period. Health workers are trained to explain the purpose and importance of screening and to prepare families for possible results. When a newborn has a positive screen, the Newborn Screening Centre alerts the attending physician or the facility NBS coordinator, who must promptly inform the parents and arrange for their return for confirmatory testing.

Facility coordinators receive specific training on communicating results, releasing them appropriately, and initiating confirmatory tests. These communication skills are reinforced through workshops and integrated into NBS team training.

Family participation is strongly encouraged. Counselling often includes multiple family members, reflecting the view that supporting the child is a shared responsibility. In some communities, religious leaders may also be involved to help families understand results and strengthen trust between families and health workers.

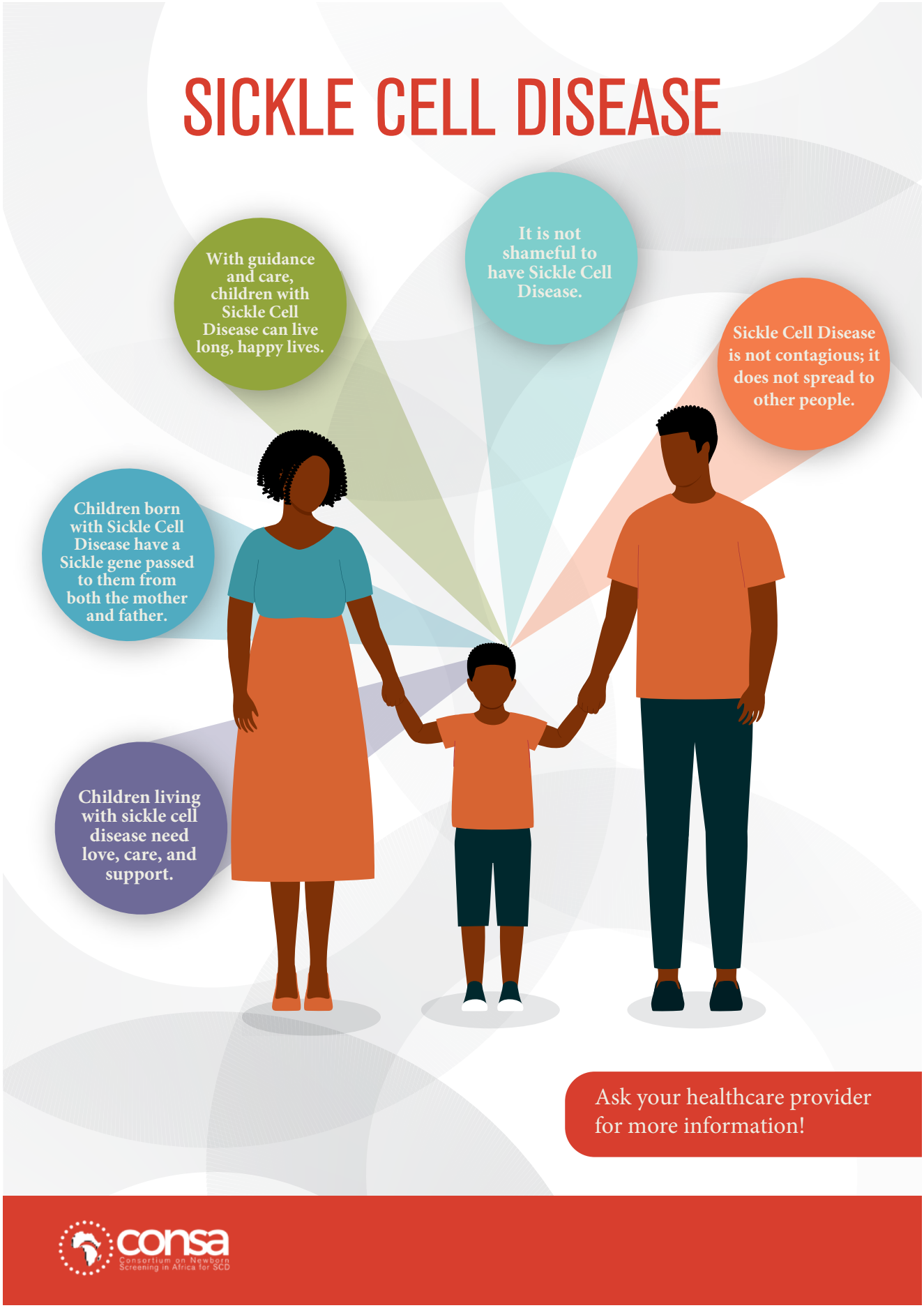
11.4.2 Experience from Uganda's sickle cell disease programme

Uganda's SCD programme focuses on proactive, supportive communication with families from the time a newborn's sample is collected. Families are informed about the purpose of testing, expected timelines, and are contacted immediately if the result is positive. Health workers use simple job aids to explain the diagnosis, its implications, inheritance patterns and key aspects of care. Both parents are encouraged to participate, and siblings are offered testing.

Health workers are trained in counselling skills – listening, clear communication, negotiation, flexibility and empathy – though limited staffing can hinder necessary support.

Community outreach needs strengthening, and there is no dedicated communication cadre. Instead, trained health workers – and even trained patients – volunteer to counsel others, enhancing peer support.

Family involvement can be challenging, particularly when some men distance themselves from the diagnosis. Encouraging both parents to be tested helps shift responsibility away from mothers alone and improves shared understanding. Broader family participation strengthens support and care continuity.



Information for families with a child diagnosed to have sickle cell disease ©Consortium on Newborn Screening in Africa

11.5 Patient support groups

Reflections from the Hridyam programme in India, NBS in the Philippines and the SCD programme in Uganda

In Kerala, India, early awareness campaigns and patient support groups helped strengthen the NBS programme, though sustaining volunteerbased efforts is often challenging due to time and financial constraints faced by potential volunteers.

In the Philippines, patient support groups are a core part of the NBS system, providing peer learning and emotional support for newly diagnosed families. The Newborn Screening Act of 2004 reinforces this by requiring health workers to inform families about screening, embedding communication into routine practice.

In Uganda, civil society groups formed by affected individuals offer advocacy and support, often linked to clinics. Sustaining these groups is challenging, but small stipends for specific tasks help maintain involvement and create job opportunities. This approach mirrors successful strategies from HIV programmes.

In conclusion, longterm sustainability in the context of NBS programmes in ensuring continuity of follow-up care, depends on consistent resources, integration into health systems, and recognition of stakeholder roles and the value these groups contribute.

Clubfoot treatment begins with a series of weekly casts that gently manipulate and stretch the feet (Liberia) ©Miracle Feet

12. Rehabilitation services and inclusive health systems for children with birth defects

This session opened with an explanation of key concepts related to rehabilitation and its importance for children with birth defects. This was followed by presentations on Uganda's strategic plan to include rehabilitation at primary care and community levels, as well as presentations from Cuba, India and Mexico on rehabilitation services for children with birth defects in their respective countries. An example of rehabilitation services delivered by non-specialists was provided from Brazil in the context of the congenital Zika syndrome.

Rehabilitation is defined as a set of interventions aimed at optimizing functioning and reducing disability in individuals with health conditions, in interaction with their environment *(16).* For children with birth defects, rehabilitation is crucial – not only for orthopaedic issues but also for physical, cognitive and communication functions essential to early childhood development, such as playing, eating and communicating. Globally, about 2.6 billion people could benefit from rehabilitation, including those with CHD, orofacial cleft, neural tube defects and clubfoot. Rehabilitation should be integrated at all levels of care – from community and primary care to tertiary hospitals – and included in universal health coverage. Early intervention maximizes developmental potential, prevents complications and supports family adaptation, while delays can lead to missed opportunities and increased complexity.

12.1 Country experiences

12.1.1 Uganda: integrating rehabilitation into primary health care

The Uganda National Rehabilitation and Assistive Technology Strategic Plan (*2024–2028*) aims to expand access to rehabilitation, especially at primary and community levels. It includes interventions for children with birth defects and applies the WHO basic package of rehabilitation interventions *(17)* to train primary health care workers in basic rehabilitation and referrals. Key steps include building consensus for tasksharing due to specialist shortages, assessing the capacity of community and primarylevel workers, and selecting priority conditions such as cerebral palsy, neurodevelopmental disorders and clubfoot. Service delivery models are co-designed for local integration, with NGO partners and rehabilitation professionals serving as mentors. Referral pathways and a hub-and-spoke model link primary care with regional and national centres. Ongoing training, supportive supervision and community engagement are emphasized.

12.1.2 India RBSK: district early-intervention centres providing rehabilitation services

District early-intervention centres are a cornerstone of India's RBSK, designed to deliver comprehensive management and rehabilitation for children with birth defects, developmental delays, diseases or deficiencies. Their primary aim is to optimize children's functional abilities and minimize impairments through early intervention. DEICs employ an interdisciplinary approach, consolidating multiple specialties under one roof to streamline access for families and reduce delays in care. They are equipped with age-appropriate tools and staffed by trained specialists across domains such as medical, physical therapy, audiology, speech, psychology, vision, cognition, special education, laboratory, sensory, and service coordination. This ensures that developmental delays are addressed with targeted, repetitive interventions.

Hearing aid fitting for a child with hearing impairment ©Hear the World Foundation

Besides rehabilitation services, DEIC's services include screening, assessment, diagnostics, condition-specific intervention and management, evaluation, review, follow-up, referral to tertiary hospitals for surgery, and coordination for aids such as spectacles and hearing aids through collaboration with other health programmes and ministries.

There are presently 437 DEICs across 36 Indian states and union territories, with the goal of establishing one in every district. Children are identified for referral to the DEIC through mobile health teams in schools and crèches, special newborn care units, primary and community health centres, and community-level workers. Once referred to the DEIC, diagnostic confirmation and condition-specific interventions are implemented followed by further referral to tertiary hospitals or NGOs as needed.

The National Health Mission supports the establishment of DEICs by providing infrastructure, equipment, human resources and capacity building, with guidelines ensuring uniformity in manpower and equipment. Critical staff include physiotherapists, occupational therapists, clinical psychologists, paediatric optometrists, paediatric audiologists and speech pathologists. Other staff such as paediatricians, nurses, special educators and social workers also contribute.

Since their launch in 2014–2015, the number and utilization of DEICs have expanded across the country, though challenges remain, including infrastructure delays, fund-flow bottlenecks, skilled manpower shortages and travel costs for families. Funding is shared between central and state governments, though states can partner with private hospitals or NGOs for specialized care. To address follow-up challenges, digital solutions are being developed for therapy management while local innovations – such as garden-based sensory therapy and mobile therapy camps – help reach underserved areas.

12.1.3 Cuba: rehabilitation services for children with birth defects

In Cuba, children with birth defects are identified at birth and immediately referred to rehabilitation services available in each polyclinic. During the first three months, these children receive further assessment at the community level by multidisciplinary teams, including genetic counsellors, rehabilitation specialists, nutritionists, psychologists, family physicians and other key experts. Rehabilitation specialists are trained within the health system, and the country's health care system annually reviews and adjusts the composition and capacity of rehabilitation services to ensure they meet the population's needs.

12.1.4 Mexico: speech and language rehabilitation for children with orofacial clefts

In Mexico, the health system is divided into public and private sectors, with care delivered at primary, secondary and tertiary levels. However, speech therapists for children with orofacial clefts are mainly available at secondary and tertiary levels in the public system, and their numbers are limited. In rural areas, online therapy – supported by national and international organizations – helps bridge access gaps, but speech therapy is not available at the primary care level, leading many families to seek private consultations. Appointments are infrequent and treatment for velopharyngeal insufficiency often starts late, especially when primary surgeries are delayed.

Support systems include free language therapy in schools through an Education Support Services Unit and low- or no-cost therapy through family support systems via national and international organizations and foundations, supplement public services, though most clinics focus on surgery rather than rehabilitation. Ongoing efforts aim to expand speech and language rehabilitation services, improve protocols and guidelines, and recruit specialists to increase access for all children who need this service.

Speech therapy as part of rehabilitation in Morocco ©Operation Smile

12.2 Inclusive health systems for children with disabilities from birth defects

Disability results from the interaction between individuals with a health condition (e.g. cerebral palsy, Down syndrome or orofacial clefts), and personal and environmental factors including negative attitudes, inaccessible transportation and public buildings, and limited social support. A person's environment has a huge effect on the experience and extent of disability. Inaccessible environments create barriers that often hinder the full and effective participation of people with disabilities in society on an equal basis with others *(18)*.

Globally, 240 million children have disabilities *(19)*, and many face significant health inequities, including limited access to both specialized rehabilitation and routine health services such as immunization and nutrition. These inequities lead to higher mortality, delayed development and poorer lifelong health outcomes for children with disabilities *(20,21)*. Stigma, negative attitudes and discrimination further exacerbate these challenges.

The WHO global report on health equity for persons with disabilities *(21)* calls for prioritizing disability inclusion in all health services, including the development of rehabilitation. Contrary to common misconceptions, disability-inclusive rehabilitation is not a choice only high-income countries can afford to make; a disability-inclusive health sector is likely to bring dividends for all individuals and communities. Ministries beyond health – such as social protection, planning and finance – are also key stakeholders in advancing inclusivity.

An inclusive health system for children with disabilities ensures equitable access to both routine and specialized health services. It addresses the unique needs of this population through integrated, systemwide approaches.

Perspective
Villaney Remegesau, Republic of Palau, born with arthrogryposis provides a perspective on an inclusive health system for a person with limited mobility

I am Villaney, born with arthrogryposis – a condition that limits my mobility and has shaped both my personal life and my advocacy for disability rights and inclusion in Palau and beyond. Living with this disability has made me realize how essential quality health services are for the well-being, independence and dignity of people with impairments. Timely and appropriate health care is not just about treatment, but also about prevention and rehabilitation, supporting our right to live a full life.

I have experienced both positive and negative encounters in the health system. When I was rushed to the emergency room, the doctors were respectful and attentive, speaking directly to me and accommodating my needs, which made me feel valued. However, I have also faced situations where I was ignored or spoken to indirectly, as if I wasn't present. Physical barriers, like high examination beds and narrow doorways, made hospital visits frustrating, highlighting that the system was not designed with people like me in mind.

To improve inclusion, we need accessible infrastructure, disability-inclusive training for health workers, and strong, accountable policies that prioritize accessibility and equity, ensuring no one is left behind.

Health inequities and barriers: Children with disabilities face substantial health inequities due to negative attitudes, stigma, discrimination and systemic barriers. These inequities manifest as higher rates of serious illness, increased mortality and limited access to essential services such as immunization and nutrition programmes *(21)*. They are often marginalized from routine health services and de-prioritized for critical care compared to their peers.

The twin-track approach: This approach is recommended as it makes all routine health services accessible and inclusive for children with disabilities, while also providing specialized care for specific impairments for those who need them. Effective implementation requires careful planning, targeted actions to remove access barriers, and systemwide integration rather than isolated, specialist-only interventions.

Health systems barriers and training: Health systems face major barriers due to limited knowledge and discriminatory attitudes among health care workers. Training on disability inclusion – integrated into core curricula for all staff – is important, along with dialogue between providers, decision-makers and people with disabilities to improve awareness. Social and behaviour change communication helps reduce stigma that deters families from seeking care.

Integrated models of care: This can address both clinical and stigmarelated barriers by strengthening referral pathways, increasing community awareness, and ensuring that policies include children with disabilities in routine services such as immunization and nutrition programmes.

Improving rehabilitation: National plans focused on inclusion, accessibility audits and active engagement of families and support groups can help tailor services to children's needs and strengthen rehabilitation.

Shine from Ghana, with her mother ©Operation Smile

13. Stigma

This session opened with a discussion on the stigma surrounding birth defects, the burden of which often falls disproportionately on mothers, and the impact this has on families. It emphasized that providing information and increasing awareness is the most effective way to combat stigma. This session was introduced by a social scientist and the parent of an affected child.

13.1 Stigma in the context of birth defects

Stigma surrounding birth defects is a multifaceted issue, affecting individuals and families both externally and internally. Externally, stigma manifests as exclusion, discrimination and disenfranchisement of those with birth defects and their families. Internally, individuals may absorb negative societal attitudes, leading to self-deprecation, reduced access to health and social opportunities, and increased mental health distress. Anticipated stigma can result in self-exclusion, where individuals or families withdraw from opportunities due to fear of negative judgment.

13.2 Sources of stigma

Stigma arises from limited understanding of specific conditions, misconceptions about their causes or perceived "infectiousness," and misleading portrayals in media or social environments. Cultural norms, religious beliefs and superstitions further reinforce negative attitudes. Structural contributors – including inequitable policies, discriminatory practices and the absence of protective legal frameworks – also perpetuate stigma.

Perspective
Ruth Nankanja, 50 years old, born with SCD in Uganda – mother, teacher, entrepreneur, and advocate sharing her experience of stigma

I was first diagnosed with SCD when I was six months old. The doctor told my parents not to spend much money on me, as I was expected to die young. My parents initially blamed each other, but ultimately chose to stay together, love me and support me so I could be a happy baby.

Growing up with SCD was tough. Whenever I fell sick, people in the community would say, "this time it's terrible, she's not coming back." Being taken to the hospital felt like a one-way trip. At school, stigma was a constant companion. I was excluded from physical education, which was especially painful because it marked me as different. No child wanted to sit near me or share food with me, and I began to isolate myself.

At university, I hid my condition, even during painful crises, pretending it was something else. After graduation, my father advised me not to mention SCD on my curriculum vitae, fearing I wouldn't get a job. I followed his advice and got a teaching job, but left after one term, worried that night shifts would trigger a crisis.

Ruth Nankanja, birn with sickle cell disease in Uganda ©Ruth Nankanja

13.3 Impact of stigma

The impact of stigma is seen in several areas with extensive consequences:

- Health care access: Individuals may avoid essential health services due to anticipated discrimination.
- Diagnosis: Families may delay confirmatory testing, fearing longterm social or financial repercussions, including insurance challenges or denial of unrelated care.
- Education: Children with visible conditions may face exclusion, with schools refusing admission or lacking adequate support systems.
- Employment and social integration: Stigma reduces employment opportunities, limits social participation, and increases isolation.
- Gender disparities: Girls are disproportionately affected, encountering greater obstacles to health care, education and broader opportunities.

Overall, stigma restricts autonomy and undermines personal dignity.

Perspective
Harikrishnan, 27 years old, born with CHD in Kerala, India on the impact of stigma around the condition

The challenges have been many: my parents faced social challenges from stigma and practical challenges from the absence of a clear roadmap for my treatment and care. The schooling system was unsupportive in dealing with my absences due to ill health. I had to complete my education as a private candidate, studying alone at home. There is a gap in my curriculum vitae because of my surgeries. Despite performing well academically, this gap has been construed by potential employers as a reason to reject my candidature for engineering positions. I have tried but failed to successfully set up support groups for CHD in Kerala. This is due to reluctance of families to publicly acknowledge that they have a child with CHD because of stigma and the knock-on effects on social and marital prospects.

The other challenge has been health insurance which I cannot avail because of my CHD. CHD is a disqualifying criterion for private health insurance in India, and the national insurance programme caters to patients below the poverty line.

My family's support has been my biggest blessing. They have not made any demands on what I should achieve and allowed me to follow my interests and choose my career.

Manifestations of stigma on families and caregivers

Mothers often carry a disproportionate share of caregiving responsibilities, which may require leaving paid employment. They may also experience blame for the child's condition, social exclusion or pressure to abandon the child or have additional children.

Families as a whole face significant social and economic impacts. Siblings may withdraw from or change schools due to stigma, while fathers may encounter community pressure to separate or remarry. These combined pressures contribute to social isolation, heightened psychological stress, reduced opportunities and increased financial hardship for the household.

13.4 Addressing stigma

Combating stigma requires targeted information, education and awareness. Providing condition-specific education, information on disease progression and quality-of-life expectations helps families prepare. Community and family education can alleviate social and psychological burdens, especially for women. Educating decision-makers, health care workers, social service providers, religious leaders, teachers and staff in orphanages is vital. Anti-discrimination frameworks are important for systemic change.

14. Support systems for families in LMICs

Peer to peer family support, play and speech therapy for children with orofacial clefts (Ghana) ©Operation Smile

Perspective
Bhagyalekshmy, 46 years old, Thrissur, India – mother to Nivedith Prakash, 17 years old, born with ataxic cerebral palsy. Accountant and freelance online service provider

My husband, also an accountant like me, and I are carers for our 17-year-old son, Nivedith, who was born with ataxic cerebral palsy. He is non-verbal but understands conversation, has no behavioural issues, and can sit with support. He moves by rolling on the floor and needs help with daily activities like going to the toilet, but he enjoys company and social interaction. My father used to live with us and helped care for our son until he passed away. Now, my husband and I share all caregiving responsibilities, and we love our son deeply. When Nivedith was three months old and unable to hold his neck, we visited many doctors and tried various treatments. It took about a year to get a diagnosis and begin therapies – occupational, physiotherapy, speech and music. Many doctors reassured us he would walk or improve, but in hindsight, these were sympathetic gestures rather than honest assessments. Knowing the correct diagnosis and its implications earlier would have helped us prepare psychologically and professionally and adjust our lives to the reality of raising a differently abled child. As a young mother, I had never heard of cerebral palsy and had to educate myself. Connecting with other parents of differently abled children was invaluable. Initially, therapy was difficult for Nivedith, and I often worried whether to continue, but learning from others helped us persist.

We spend a lot of time with our son, and he shares a close bond with his father. We talk to him daily, and he enjoys these interactions, though most of his social contact is limited to us. We try to take him out on our scooter, which he loves, but wish he had more opportunities to interact with others. There is no organized social support system – extended family is kind but not always available. It would be good if there were other students in college or high school who could come and interact with him in a friendly way, take him out sometimes, so he has some social contact and exposure – it would broaden his world. It would also be good if there were special education facilities to be able to tap the talents that differently abled children have and to educate them to the extent possible, to open up new interests and possibilities for them.

We have an extended family, they are polite, but they keep a distance because they know that I usually need help. As parents, we have arranged our professional life to ensure we are available at home whenever necessary. Both my husband and I work from home, which is a privilege, but financial stress remains. State support is minimal; the small grant we receive is spent on essentials like diapers. We would like to renovate our home for better accessibility for our son, but costs are prohibitive, and we are not yet able to afford that.

We have to pay for his treatments, wheelchair, diapers, etc., which we are happy to do, but it means we do have to make sacrifices – sometimes I have to think multiple times before I buy a new dress. Some health insurance cover for differently abled persons would be very helpful for the family.

Between my husband and me, we must always be available, which is sometimes tiring. Sometimes as the mother, I am sick or unwell I usually take a pain killer and continue my caregiving duties, to my son whom I love very much. I will forgo going to the doctor so that I am available to give the care needed to my son at home. Though my husband is very supportive and has a very good relationship with my son, I wish there were some ways for mothers to find some time off, to rest and do something outside to refresh oneself. I think men are able to find such opportunities more than women.

Often, there is little consideration given to the parents of differently abled children. Though we love our children, the physical and psychological strain is undeniable. We also need to be able to focus on our own health needs and take care of ourselves. If we cannot take care of ourselves as parents – particularly as we age while carrying caregiving duties – we will be unable to care for our differently abled child. But this is often overlooked, the health and well-being of parents of differently abled children is important for us as parents and for the well-being of the differently abled child. We contribute a lot to society through our caregiving responsibilities. It would be very helpful for us as parents to have some sort of insurance to cover our own health needs as the parents of differently abled children. The needs of parents, often primary caregivers for a child with severe impairments, are not often considered by planners, governments or society.

What could have helped or can help? Early diagnosis, honest communication, peer support, ways of social inclusion for us and our child, educational opportunities to help differently abled children express their potential, financial assistance to cover costs of health care, make adaptations to our home to make it more comfortable for our son. An often overlooked but very important aspect here – is the health and well-being of parents. Parents are often simply expected to be caregivers with no consideration of our own health needs. We often deny ourselves the health care we need despite the immense physical and mental strain that full-time caregiving involves. This is particularly important as we age. State provision of insurance to support the health needs of parents of differently abled children would be invaluable. Such support enables us to fulfil our caregiving duties which benefits not just our differently abled child and family but also society at large. While we carry out this care with love for our child, support for our own health is an important consideration for governments to provide. The challenges aside, we love our son deeply, and caring for him brings us happiness and meaning.

Bhagyalekshmy with her son, at home
©Bhagyalekshmy

14.1 State-led health and social support systems: learnings from Kerala, India

An effective social support system would provide access to early detection and diagnosis, timely intervention, family counselling and psychological support, financial assistance, inclusive education and robust community-based rehabilitation.

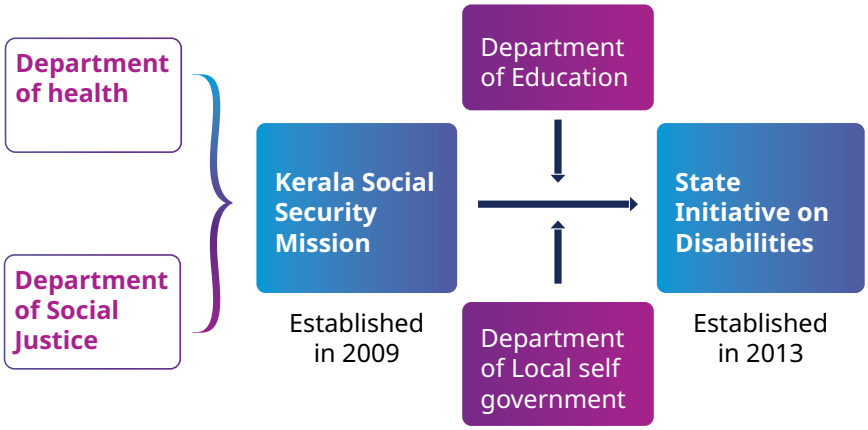
Key challenges and solutions

Kerala's journey highlights several systemic challenges common in low- and middle-income settings, along with targeted solutions:

1. Fragmented systems across departments. Services for children with birth defects were previously scattered across health, education and social sectors, causing duplication and service gaps.
   Solution: Kerala created an integrated model through the Kerala Social Security Mission, which later expanded to include education and local government, ensuring coordinated, interdepartmental action.
2. Limited access to services. Specialized care was concentrated in urban centres, leaving rural children with poor access.
   Solution: Kerala decentralized care by establishing DEICs, regional and apex centres, mobile intervention units, and communitybased preschools to extend early intervention and homebased support.
3. Financial burden on families. High costs of surgeries, therapies and assistive devices, combined with limited insurance, placed families under significant strain.
   Solution: Financial support schemes – Aswasakiranam, Samashwasam and special loans – were introduced to reduce outofpocket expenses.
4. Social stigma and discrimination. Misconceptions about birth defects led to isolation and reduced opportunities for affected children.
   Solution: Awareness campaigns and community engagement initiatives were launched to reduce stigma and promote inclusion.

Integrated system for care and coordination

The State Initiative on Disabilities – "Anuyatra" (meaning "walking together") – reflects a comprehensive life-course approach. The institutional mechanism is structured around key life stages: pre-conception, the first 1000 days, ages 3–5 years, 5–18 years, and adulthood. Each department (Health, Social Justice, Local Self Government, Education) is assigned specific responsibilities, with action plans tailored to each developmental stage. This ensures continuity of care, from early detection to inclusive education and, where possible, employment and independent living. For those unable to live independently, the system provides access to assisted living arrangements.



State-specific social support programmes

Kerala's support programmes for children with birth defects and disabilities are organized into four key categories:

1. **Health care programmes:** Hridyam (for CHDs), Kathoram (for hearing impairment) and Kerala United Against Rare Diseases. The rare diseases programme uses crowdfunding to help finance costly treatments for rare conditions.
2. **Financial assistance programmes:** Schemes including Aswasakiranam, Samashwasam and special loan programmes that reduce the financial burden on families and caregivers.
3. **Education support programmes:** Inclusive education initiatives like Samagra Shiksha Abhiyan Kerala, BUDS schools for children with intellectual disabilities, and special Anganwadis to ensure educational access.
4. **Communitybased social safety nets:** Local Self Government Department implements community rehabilitation services – the Kudumbasree Care programme (engaging women entrepreneurs in caregiving) and Assisted Villages for individuals needing longterm support.

Ongoing challenges and lessons learned

Despite robust systems, Kerala faces ongoing debates about coverage – balancing the number of patients, types of products (medicines, devices), and available resources. Kerala's experience demonstrates that a coordinated, multisectoral approach, with strong institutional mechanisms and community engagement, can significantly improve outcomes for children with birth defects and their families.

Perspective
Rinsha, visually impaired from birth, pursuing her university studies in sociology on challenges faced during education

I am Rinsha, a national award winner for best creative child, honoured by the former President of India. Currently, I am pursuing a bachelor's degree in sociology, now in my second year at university. Despite these achievements, my blindness from birth has made my educational path challenging in ways that it need not have been. I share my story to advocate for other students like me.

Blindness is often misunderstood – many assume that visually impaired people cannot study, be independent, or excel in science, mathematics or technology. Society tends to see us as inspirational simply for existing, rather than recognizing us as equals. In my early years, even basic activities like walking on the streets were unsafe due to the absence of tactile footpaths and auditory traffic signals. Attempts to explore and play were often discouraged, and before I even entered a classroom, society had already limited my future.

In primary school, I attended a special school for blind children, where we had Braille textbooks but lacked reference books in regional languages and adequate library resources. In high school, the absence of advanced Braille textbooks and digital materials compatible with screen readers forced me to rely on YouTube lessons and volunteers, which were insufficient for deep learning. Mathematics and science were particularly inaccessible –there were no Braille books for complex equations, graphs were unreadable and science laboratories were not designed for us. These barriers restricted my study choices as I had to drop these subjects although I enjoyed them. Some teachers lacked awareness or held prejudices, resulting in unfair grades, though others were supportive and proved inclusion is possible with the right mindset.

Today, assistive technologies like Braille displays and talking calculators exist, but many families cannot afford them. The availability of tactile pedagogy and inclusive tools is expanding, but access is limited. For me at university, simple things, such as finding qualified writers for exams, has been challenging, and the inadvertent social exclusion by sighted peers during group work has been hurtful.

What will help us: expanding assistive technologies and tactile pedagogy across more educational institutions, training teachers, ensuring state support for special materials, and fostering social inclusion.

Every student, sighted or visually impaired, deserves the right to dream, learn and achieve their full potential.

Rinsha, visually challenged since birth, is pursuing her university studies ©Rinsha

14.2 Family support from local organizations

Several organizations in LMICs focused on specific birth defects to provide support to families with affected children. These organizations strengthen family resilience by organizing peer support groups, awareness seminars, and organizing advocacy.

For this consultation, the Down Syndrome Association of the Philippines, presented their work to support parents of children with Down syndrome. Examples of activities undertaken by the Association and other similar family support organizations include:

- ***Collaboration*** with local self-governments and corporate social responsibility programmes.
- Building and maintaining a strong ***volunteer network*** and dedicated staff to support families.
- Working with ***stigma reduction and advocacy***, and information for families of children with Down syndrome.
- Delivering ***early intervention support***; evolving from parenttoparent assistance to involvement of medical professionals and therapists. This includes periodic online seminars to expand access and encourage shared learning.
- Organizing ***outreach seminars*** for parents, caregivers, teachers, and health professionals to increase awareness and practical skills.
- ***Holistic family support***, including sibling support groups to address family-wide needs.
- Operating ***free clinics*** that provide comprehensive specialist consultations.
- Expanding ***employment opportunities*** through partnerships with private companies.

Employment opportunities for young persons with Down Syndrome in a pharmacy
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Annex 2. Details of country programme presentations

Details of state-led national/regional initiatives for newborn screening, diagnosis and management of one or more birth defects in health systems

Details of presentations made by ministries of health from front-runner countries are described here.[4] *Where relevant, references to support further reading are provided.*

The Philippines: newborn screening programme

***Initial pilot project**:* The Philippines' journey in NBS began in 1996 [IMR = 30/1000] *(22)* with a pilot project in 24 hospitals in Metro Manila, targeting five key disorders using dried bloodspot screening: congenital hypothyroidism (CH), congenital adrenal hyperplasia, galactosaemia, phenylketonuria (PKU), and homocystinuria. The pilot aimed to establish the prevalence of these conditions and develop recommendations for a nationwide programme. Seven core strategies were implemented: shared leadership with a core group and hospital-based NBS coordinators; robust communication efforts to educate both health practitioners and families; continuous capacity building for hospital staff; provision of a comprehensive NBS package for each hospital (including educational materials and sample collection tools); standardized flow of information (from health provider to parent, sample from hospital to laboratory, result from laboratory back to hospital, hospital provider to family); adoption and later adaptation of confirmation algorithms for the laboratory; and the development of treatment protocols, particularly for congenital hypothyroidism. The cost to families was kept low (US$ 7), and positive cases were recalled for confirmatory testing, diagnosis, and referral for management.

***National programme*:** After the pilot, the programme was scaled up and evolved into the Comprehensive National Newborn Screening and Management Programme, now covering 29 endocrine and metabolic disorders across 7000 hospitals and birthing centres, with approximately 4600 samples screened daily. *Key principle: only disorders with treatment supported in the country were included.*

The programme's infrastructure currently includes seven accredited screening laboratories and 33 continuity clinics for long-term care and follow-up, supported by teleconsultation with specialists. Expert panels oversee protocols for each condition. The cost of screening, US$ 30, is now covered by national health insurance. Coverage has reached 98% of births.

***Legislation** has played a pivotal role:* the Newborn Screening Act of 2004 established the Department of Health as the lead agency, mandated insurance coverage, and required all health institutions to provide NBS as a condition for licensure. Accreditation and regular evaluation ensure quality, and all health workers are required provide parents with information regarding NBS.

Strengths: These include legislative backing, insurance coverage, accreditation and expert-developed protocols. **Challenges:** include early discharge of newborns, patient recall, access to information and specialists, treatment adherence, and programme resilience in the face of disasters or pandemics.

[4] The statistics in the country presentations in this annex are reported by the respective countries and do not represent official WHO statistics.

Expansion: The programme, which has thus far focused on conditions screened using dried bloodspots, is expanding to include four visible anomalies (e.g. clubfoot, neural tube defects, orofacial clefts, abdominal wall defects). In a second phase, the plan includes congenital cardiac and hearing defects (separate pilots are currently underway). This expansion leverages existing infrastructure with new management algorithms and referral pathways being developed by multidisciplinary teams. The Department of Health will continue to lead, with public hospitals providing treatment and partnerships with the private sector for broader service delivery.

India: national Rashtriya Bal Swasthya Karyakram for birth defects

Launched in 2013 [IMR = 40/1000] *(23)* by India's Ministry of Health, RBSK is a nationwide programme aimed at early identification and intervention for children with birth defects, diseases, deficiencies, and developmental delays – the "4Ds." The focus on birth defects was driven by an epidemiological transition: as under-five mortality from infectious causes declined, birth defects became a more prominent cause of child morbidity and mortality. Other triggers included a recognition of the preventable or treatable nature of many conditions reducing the risk of lifelong impairments, and a high out-of-pocket expenditure for affected families.

***Programme planning and scope*:** RBSK was designed after a review of available datasets from academia, hospitals in the country, and international databases. The initial focus was on nine major birth defects selected based on prevalence, ease of diagnosis, treatment availability and affordability within the system. These included neural tube defects, Down syndrome, orofacial clefts, clubfoot, developmental dysplasia of the hip, congenital cataracts, congenital deafness, CHDs and retinopathy of prematurity. Screening for congenital hypothyroidism, sickle cell disease, and beta-thalassemia was made optional for states where these defects were priorities.

***Programme expansion and funding**:* The programme started with screening for nine birth defects (visible or detected on clinical screen) and expanded to 109 by 2016, including functional and metabolic defects in provinces where resources and system readiness allowed. The approach was to start small with visible birth defects, as these were relatively easy to detect at birth with low infrastructure needs, and then scale-up in a phased manner to include functional defects like CHD, followed by metabolic defects in some states, depending on the availability of funds and the readiness of the health care system in the different states. Screening for chromosomal anomalies/genetic disorders is presently limited to tertiary centres. Management relies on robust referral networks, public–private partnerships, and financial support through national health assurance programmes.

National insurance schemes cover treatment, especially for children below the poverty line. The Pradhan Mantri Jan Arogya Yojana insurance programme offers health coverage of approximately US$ 6000/family/year for secondary and tertiary care, including the management of birth defects.

Target population and programme components

The programme targets India's annual birth cohort (27 million, with 80% in the public sector) and 80 million preschool children in state-run creches (anganwadi). The key components include:

- Comprehensive NBS: Conducted at over 22 000 birthing points, with capacity building for staff and community health workers screening babies during home visits.
- Mobile health teams: Over 11 800 teams (each team comprising two doctors and two nurses) screen children in schools and creches (anganwadis), coordinate referrals, transport children and manage post-surgery follow-up.
- DEICs: 430 centres provide diagnostic (echocardiography, radiology, confirmatory tests for hearing impairment) and management services, staffed by multidisciplinary teams (paediatrician, audiologist, physiotherapist, speech therapist, psychologist, optometrist).
- Tertiary care: Genetic tests and care for complex cases.

The aim of the programme is to facilitate the management of interventions with the goal of zero out-of-pocket expenditure for families.

Achievements and challenges In the past three years, 28.2 million children have been screened out of 61.2 million births, with 0.9 million identified with a birth defect, of whom 0.3 million received services at higher-level hospitals.

Challenges include a shortage of skilled providers, infrastructure gaps and the absence of a national database. Screening data capture occurs in most but not all states in the union presently. Challenges exist in some states to track and follow-up children over extended periods necessary with most birth defects. A countrywide online data portal is under development, and long-term follow-up for parents is facilitated at DEICs. Families still incur out-of-pocket costs particularly linked to transportation.

Monitoring and evaluation: The programme emphasizes regular monitoring and evaluation through monthly and quarterly reporting, field visits, internal reviews, third-party assessments and periodic meetings with state nodal officers. Key performance indicators have been defined which are reviewed periodically.

The RBSK programme is a large-scale, phased initiative focused on early detection and management of birth defects, with a strong emphasis on system strengthening, stakeholder engagement and continuous monitoring. While the programme has expanded rapidly and integrated funding and insurance mechanisms, it faces challenges in human resources, financing, logistics and data management. The challenges are recognized and efforts are being made to generate solutions which are important for the long-term sustainability and effectiveness of the programme.

Sri Lanka: approach to newborn screening and birth defect management

Sri Lanka, despite being an LMIC, has achieved a relatively low infant mortality rate (7/1000). However, birth defects account for 40% of infant deaths, making their detection and management a national priority, especially in the context of Sustainable Development Goals. The country is currently drafting a National Strategic Plan (*2024–2030*) focused on birth defect surveillance, prevention and care.

The National Strategic Plan integrates screening for visible birth defects, CH and CHDs into routine newborn examinations before discharge. Results are recorded in the standard child health development record. Hearing defect screening is also conducted, although not nationwide. While the Ministry of Health funds screening, families may sometimes pay out-of-pocket for confirmatory diagnoses.

For CH, a pilot programme launched in 2007 led to a government scale-up, now covering 80% of the national birth cohort. Screening is performed close to discharge by nurses, with dried bloodspot specimens sent to centralized laboratories. A web-based database enables rapid communication of positive results to parents and community medical officers for follow-up. Confirmed CH cases are managed by specialists, and public health midwives ensure ongoing compliance and follow-up. Comprehensive resources have been developed for health care staff, including guidebooks, standards (from quality of dried bloodspot to ensuring treatment is started within two weeks), and public education materials. Challenges include securing financial resources for reagents and supplies and some out-of-pocket costs for families.

For hearing impairment, screening is available in select teaching hospitals covering up to 30% of births. Initiated by a national professional body, the programme uses otoacoustic emission screening, with positives referred for further testing. Challenges include equipment costs, maintenance costs, long wait times for cochlear implants, and underdeveloped care pathways.

For critical CHDs, pulse oximetry screening began in 2017, in specialty hospitals, prior to discharge. Positive screens are referred for echocardiograms, with management plans required before discharge. However, data on coverage is lacking, waiting lists for surgery are long, and more cardiac surgeons are needed.

For eye abnormalities, external examination and red reflex screening are part of the routine newborn examination with referral to an ophthalmologist if screened positive. Challenges include lack of coverage and care pathways.

Although Sri Lanka has a state-led programme, there are constraints related to financing resulting in limited coverage, infrastructural (equipment) constraints, and families must bear some out-of-pocket expenses. Data and monitoring gaps remain, making it difficult to monitor programme performance and plan for improvements. Care pathways for some conditions are still being developed. Despite these obstacles, Sri Lanka's commitment to integrating NBS into routine health services is notable. With ongoing efforts to strengthen infrastructure and improve data systems, important strides toward better outcomes for children with birth defects are being made.

Uganda: national sickle cell disease programme

Uganda's national SCD programme focuses on screening newborns and children up to two years of age in high-burden areas and providing care through specialized SCD clinics. The programme's foundation was influenced by donor-funded vertical programmes for human immunodeficiency virus, tuberculosis and malaria, which led to the creation of centrally located centres of excellence for diagnostics. However, these were difficult for patients to access, so a hub-and-spoke model was adopted, allowing samples to be collected near patients and transported to central laboratories – this system became the backbone for SCD screening.

In 2013, advocacy groups prompted the Ministry of Health to generate a national database on SCD. Partnerships with Uganda's Central Public Health Laboratory, Makerere University, and Cincinnati Children's Hospital enabled a national survey, testing 100 000 babies and revealing a 13.3% prevalence of the sickle cell trait and 0.7% for SCD. 14 high-burden districts, accounting for half the national SCD burden, became the focus of the national SCD programme.

Key programme components include:

- Screening at maternity centres, immunization clinics and health care centres for children 0–24 months of age.
- Training health workers in screening and care pathways. The programme utilizes existing staff, supported by national and regional trainers, with ongoing training and provision of guidelines.
- Efficient sample transport network and electronic transmission of results: A robust hub-and-spoke model for sample management, with electronic tracking and efficient result delivery.
- Laboratory capacity: dedicated SCD laboratories equipped through donor support, with trained staff and participation from about 200 facilities.
- Establishing 100 SCD clinics (including 10 Centres of Excellence) for prophylactic treatment and crisis management.
- Provision of hydroxyurea (limited by cost constraints).
- Data management systems for laboratory and clinic data.
- Follow-up: positive cases are contacted by phone for care enrollment; presently home visits are restricted due to funding constraints.
- Creating national guidelines, a strategic plan and a steering committee for SCD.
- Engaging stakeholders, including cultural, religious and political leaders, and increasing public awareness.

System challenges: Sustainable financing remains the biggest barrier, with funding being ad hoc and fragmented. There is no robust national SCD registry yet, but a clinical database is being developed at the centres of excellence. Since 2014, approximately 50 000 newborns have been tested annually (3% of annual birth cohort, 67% of testing done below six months of age). Testing is limited to high-burden areas due to funding constraints. The programme leverages existing public health infrastructure and donor resources for sustainability, but faces challenges such as limited coverage, sustainable funding, lack of health insurance, and the need for a long-term programme approach. Opportunities include new diagnostic technologies, increased hydroxyurea availability, global focus on SCD, ongoing research and strong civil society advocacy.

India (Kerala): Hridyam programme for congenital heart defects

Hridyam is a state-led programme in India (Kerala), designed to improve the screening, diagnosis and management of CHDs in newborns. The programme was launched in response to a stagnant infant mortality rate of 12/1000 for a decade since 2006, with CHD identified as a major contributor. Leveraging strong political leadership focused on bringing IMR down to 6/1000 by 2030 and government funding, Hridyam engaged both public and private hospitals in a state-led partnership, adopting a systemic approach that integrates actors (people, providers, state), functions (financing, service delivery, governance), and outcomes (health status, financial protection, trust in the system).

Early identification and referral for management: Before Hridyam, there was no system for identifying newborns with CHD or assessing care needs; providers operated independently with limited specialized staff. Since Hridyam began, early identification and registration of newborns with CHD is done through statewide screening and diagnosis, all cases are entered onto a common portal and allocated to accredited clinical centres for management. Protocols to determine and prioritize urgency are in place.

Strengthening inputs: Previously, CHD care was fragmented, with families seeking treatment on their own and limited facilities. Hridyam shifted to a population health approach, expanding NBS (pulse oximetry) and diagnostic capacity (echocardiography in all districts), improving intensive care and transport systems, and establishing centralized follow-up. Surgical slots are pooled and allocated by protocol, and a web registry tracks each case from registration to follow-up.

- *Expanding access*: The state leads a public–private partnership model, increasing pool of specialists and care units. Hridyam has added new centres and staff, launched e-learning for paramedics and nurses, improved intensive care capacity, and supported the availability of equipment and medicines.
- *Capacity building:* Training radiologists and obstetricians/paediatricians in paediatric echocardiography, organizing interventional cardiology workshops, and ongoing support from the Indian Academy of Paediatrics.
- *Electronic systems for centralized coordination:* Introduction of electronic data management software for case registration, verification and expert review. The electronic system matches cases with available surgical slots, allows families to choose hospitals, and manages claims and follow-up care, including community health worker visits. All surgical slots in Kerala were pooled, providing finite waiting periods and tentative surgery dates for registered children.

Financing: Cashless treatment under Hridyam has been made available in both government and empanelled private hospitals and is funded through the national RBSK and the Kerala state government. All newborns and children registered in the portal receive care regardless of family income. Payments to private partners are set out in agreements taking into consideration case complexity and volume.

Challenges: Despite government funding, adequate financing consistently has been difficult due to rising needs and competing priorities. Private sector stakeholder engagement has been challenging particularly around payments from the state. Staff turnover and bureaucratic delays have also been a challenge.

Strengths: The Hridyam programme in Kerala exhibits several key strengths:

- *Strategic prioritization:* The programme was launched after careful analysis showed that CHDs were a major contributor to infant mortality in Kerala. By targeting CHDs, the state addressed a leading cause of preventable deaths among infants.
- *Government leadership and clear targets:* The Kerala government set ambitious IMR reduction targets, which drove strategic planning and focused interventions. This high-level commitment assured attention and resource allocation to the programme.
- *Comprehensive system strengthening:* Hridyam integrated screening, diagnosis, treatment, and follow-up into a single coordinated system, improving access and outcomes for children with CHD.

- *Public–private partnerships:* The programme leveraged both public and private sector resources, expanding capacity for paediatric cardiac care and reducing treatment delays.
- *Monitoring and accountability:* Regular tracking of outcomes and transparent reporting have helped maintain programme quality and drive continuous improvement.

Achievements: The Hridyam programme has reduced mortality and morbidity, improved equity and transparency, and guaranteed services for registered patients. IMR dropped from 12 to 6 per 1000 *(24)*, with over 9000 children supported and 12 000 surgeries performed since 2017.

Egypt: national neonatal screening and birth defect initiatives

Egypt's initiative for early detection of hereditary disorders in neonates began in 1999 with dried bloodspot screening for CH and PKU at primary centres nationwide. In 2021, the programme expanded to include 19 genetic diseases, including CH, congenital adrenal hyperplasia, and cystic fibrosis, among others, with conditions reviewed and prioritized by a scientific expert committee. The Ministry of Health leads the programme, supported by the national health insurance authority. The Egyptian Centre for Disease Control and Prevention handles genetic analysis and confirmation, while university hospitals provide treatment and follow-up, including the provision of therapeutic milks. The private sector also participates. Equitable access is ensured through 317 screening points across all 27 governorates, and the programme is free to the user. Treatment is available for three to five of the 19 disorders. Since 2021, 500 000 newborns have been screened, identifying 3785 positive cases, with glucose-6-phosphate dehydrogenase deficiency being the most common.

For hearing impairment, a national programme launched in 2020 uses otoacoustic emission and auditory brain stem tests, screening seven million children since 2020 and covering 83% of the 2023 birth cohort. Cleft lip and palate screening and treatment are provided at a specialized hospital in Alexandria, with annual follow-up and new centres being established for early detection and care.

India: National Sickle Cell Anaemia Elimination Mission

India's Sickle Cell Anaemia Elimination Mission, launched in July 2023, is a comprehensive, multisectoral initiative targeting the high burden of SCD among tribal populations, with about 50 000 of the world's 500 000 annual SCD births occurring in India. The programme is implemented in 17 of 36 states, focusing on prevention, early detection and holistic management of SCD through coordinated action across all levels of the health system and multiple government sectors.

The programme is built on three pillars: health promotion through awareness (including pre-marital, pre-conception and antenatal counselling), primary and secondary prevention via universal screening and early detection, and holistic management that integrates all levels of care with community support.

Programme structure and health system involvement:

- Community and primary care: Screening began with door-to-door campaigns, health camps and institutional screenings at birthing points, primary health centres, and schools. Community health workers were pivotal, raising awareness, conducting screenings, monitoring treatment adherence, educating families, and facilitating disability certification. Their close ties to the community ensured high coverage and effective follow-up.
- Secondary- and district-level care: District hospitals serve as referral centres for confirmatory diagnosis and management of positive cases. They provide ongoing care, including prophylactic therapies (vaccinations, folic acid, penicillin prophylaxis up to five years of age) and essential medications like hydroxyurea (for children with repeated episodes of acute chest syndrome or >3 crises/year requiring hospitalization). Workforce training and clinical guidelines ensure standardized care at this level.
- Tertiary and specialized care: Centres of Competence at the tertiary level offer advanced diagnostics, prenatal testing, surgery and bone marrow transplantation. These centres also support advanced research, including gene-editing technologies, and are integrated into the national health insurance system to provide cashless treatment.

Key strategies

- Financial support is provided to states for SCD prevention and management, with separate hospital wards for SCD patients at the district level.
- Standard operating procedures, clinical guidelines and workforce training to ensure quality care.
- Availability of hydroxyurea (including paediatric syrup) and other essential drugs is ensured.
- Provision of prophylactic therapy and immunization.
- A dedicated SCD registry and portal enable tracking and monitoring, while telemedicine extends specialist support to remote areas.
- Public–private partnerships are leveraged for affordable screening, and local products are used to ensure uninterrupted equipment supply.
- SCD care is integrated into the national health insurance package, ensuring cashless treatment for patients.

Multisectoral collaboration: The programme's success relies on collaboration between the departments of health, tribal development, education, medical research, women and child development, social welfare, information technology and rural development. This multisectoral approach is deemed important to meet the stated goal of eliminating sickle cell disease as a public health issue by 2047. The programme has screened over 60 million individuals, identified 215 000 cases of SCD and 1.6 million carriers as of end 2025.

Argentina: national programme for newborn screening

Argentina has made significant strides in NBS of endocrine/metabolic/genetic conditions and CHD through comprehensive national programmes supported by legislation, governmental collaboration across federal and provincial levels, and a focus on equity.

The national NBS programme was launched in 2006 (IMR = 14/1000) following a survey that revealed less than 40% screening coverage in some provinces. In 2007, a new law mandated screening for six key conditions: PKU, CH, cystic fibrosis, galactosaemia, congenital adrenal hyperplasia, and biotinidase deficiency. The programme is characterized by a clear agreement between national and provincial authorities: the national budget covers equipment, reagents, blood collection cards and PKU formula for two years, while provinces manage human resources, logistics, diagnosis and treatment.

Coverage has dramatically improved, rising from 52% in 2006 to 98% in 2022. The NBS law allows for the addition of new conditions based on scientific evidence, with a special expert commission established to assess feasibility. The programme's components include dried bloodspot specimen collection at maternity hospitals (recommended between two to five days of life), timely transport and processing in one of 20 national laboratories, and rapid diagnostic confirmation, treatment and follow-up by medical specialists. The national programme supports provincial coordination by facilitating access to specialized centres, offering training, and covering initial treatment costs for certain conditions.

Strengths of the NBS programme include mandatory legislation, strong support from both national and provincial governments, and the legal requirement for NBS data reporting in the official health information system since 2022. However, challenges persist, such as unequal availability of treatment options, disparities in resources and specialist availability across provinces, and the absence of standardized process monitoring indicators.

Congenital heart defects: Recognizing that CHDs accounted for 23% of birth defect-related mortality, Argentina initiated a national CHD programme in stages. Early efforts (*2004–2008*) focused on creating a patient registry, categorizing waitlisted patients by urgency, listing surgical hospitals, and establishing a dedicated budget for surgeries. The next phase (*2009–2010*) integrated CHD care into Plan Nacer, a results-based financing programme for vulnerable populations, and expanded infrastructure, including equipment for hospitals and ambulances, a 24/7 referral coordination centre, and registries for patients and surgical centres.

The CHD Federal Network has led to a decrease in infant deaths, a 66% increase in CHD diagnoses, a 63.6% rise in surgeries, and an 86.3% reduction in waiting lists. The programme's strengths include a collaborative governance model, an advisory board for quality assurance, universal coverage institutionalized by law, and strategic purchasing and training. Challenges remain, such as disparities among provinces, the need for more specialists, transportation quality and the development of referral networks for pregnant women and adults.

Cuba: national programme for birth defects and genetic disorders

Cuba launched its national programme for the diagnosis, management, and prevention of birth defects and genetic disorders in 1981 [IMR = 18/1000] *(25)*. The Ministry of Health established a National Network of Medical Genetics and Services, integrating clinical services and 38 NBS laboratories across all three levels of the health system.

A strong emphasis was placed on capacity building. Clinical genetics specialists were (are) trained in six of Cuba's seventeen medical universities; two-year master's degree programmes in medical genetics and genetic counselling were introduced. Since 1998, over 900 genetic counsellors have been trained, 60% of whom are family doctors and 40% are higher-level nurses. In 2004, medical genetics was incorporated into the medical school curriculum, further strengthening the country's expertise in this field.

The National Center for Medical Genetics provides methodological oversight to the network, coordinating 116 clinical geneticists and 456 genetic counsellors. In 1985, Cuba introduced a National Registry for Birth Defects, creating a centralized repository for birth defects and genetic disorders in all 46 hospitals. This was later expanded to include community-based surveillance.

NBS is a key component, with dried bloodspot screening for six conditions: PKU, CH, congenital adrenal hyperplasia, galactosaemia, biotinidase deficiency and cystic fibrosis. Locally developed enzyme-linked immunosorbent assay-based technology is used for metabolic disorder screening. The testing system was decentralized 15 years ago, allowing all testing to be carried out at the municipality level. Cardiac surgery for CHDs is performed at two institutes, and pulse oximetry is planned as part of the NBS programme.

Brazil: National Neonatal Screening Program

Brazil's Ministry of Health launched the national NBS programme in 2001 [IMR = 27/1000] *(26)* building on a NBS programme for PKU initiated years earlier by a parent support group. The programme includes screening, tracking positive cases, confirmatory diagnosis, treatment and follow-up.

NBS began in phases: Phase 1 included screening for PKU and CH; Phase 2 added SCD and haemoglobinopathies; Phase 3 added cystic fibrosis; Phase 4 (*2012*) added congenital adrenal hyperplasia and biotinidase deficiency. In 2022, congenital toxoplasmosis was added. Between 2012 and 2023, NBS coverage ranged from 80–84% of births, with samples collected at over 23 000 points. The heel prick test is performed between two to five days of life for most newborns.

NBS for hearing loss became mandatory in 2010 for all newborns in public and private hospitals. The "Little Ear Test" is conducted within 24–48 hours of birth, using otoacoustic emission or auditory brain stem response tests. Confirmatory tests are done before three months of age, followed by interventions as needed (hearing aid, speech therapy or in some cases cochlear implants). In 2023, 43% of newborns were screened. The test is free in public hospitals, but disparities in access persist, especially in remote areas, making improved follow-up a key focus.

Brazil also conducts active surveillance for visible birth defects, focusing on those detectable at birth and manageable within the health system. Nationwide pulse oximetry screening ("Little Heart Test") for CHD began in 2017. However, surgical capacity remains insufficient, with a waiting list for children needing cardiac surgery. In 2022, a programme was initiated to enhance surgical care, and there are now 69 reference hospitals for cardiac surgery, though this is still inadequate

Armenia: universal newborn screening programme

Armenia, a UMIC with a population of over 3 million, has made significant progress in NBS since gaining independence in 1991. The Ministry of Health is the central regulator, funding outpatient, emergencies, and maternal and child health services, which are largely free at the point of care. Both public and private institutions participate in the state-covered universal NBS programme, provided they comply with Ministry of Health regulations. The health system includes a wide network of hospitals, polyclinics, and rural health centres. In 2023, Armenia had an IMR of 9/1000 *(27)*, of which 19% was attributed to birth defects. NBS in Armenia began with Swiss collaboration over a five-year period. Switzerland initially provided full funding, following which the Armenian state funded 20%, and increasing by 20% each year, so that at the end of five years, the Ministry of Health fully took over. Today, screening is universal and state-funded, covering conditions such as CH (started 2005), hearing loss (*2008*), PKU (*2008*), hip dysplasia (*2009*), CHD (*2015*) and congenital adrenal hyperplasia (*2024*).

The National Screening and Referral Laboratory at Arabkir Medical Centre, supported by international partners, assures quality control, with external proficiency testing from the United States Centers for Disease Control and Prevention. Training for laboratory and maternity staff began abroad and transitioned to local programmes, with ongoing awareness campaigns for health care staff and parents. Screening, referral, and follow-up roles are clearly assigned, with approximately 80% follow-up rates. Data is managed electronically, with efforts underway to further digitalize the system.

Sustainability is ensured through Ministry of Health regulations, funding and supervision. Key success factors include a robust health system, technical support from champions, and international partnerships. Challenges remain, particularly resource constraints for expanding screening to new conditions.

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