CONGENITAL ANOMALIES

KERALA.HEALTH

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GOVERNMENT OF KERALA

Pinarayi Vijayan

CHIEF MINISTER

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MESSAGE

Congenital anomalies pose a significant public-health challenge, contributing to neonatal and infant morbidity, mortality, and long-term disability. Addressing these concerns demands a comprehensive and continuum-based approach encompassing prevention, early detection, timely clinical management, rehabilitation, and sustained support for affected children and their families. Kerala has consistently demonstrated leadership in maternal and child health through a strong primary healthcare system, high institutional delivery rates, and progressive public-health interventions, further strengthened by advances in antenatal and newborn screening, diagnostic services, and specialised care.

Congenital Anomaly Document presents a systematic and evidence-based overview of congenital anomalies, covering epidemiology, risk factors, prevention strategies, screening and diagnostic pathways, clinical management, referral mechanisms, and follow-up care. Emphasising multi-sectoral collaboration across health, nutrition, social welfare, and education sectors, it reflects our collective commitment to ensuring that every child in Kerala has the opportunity to survive, thrive, and realise their full potential. I am confident that this document will serve as a valuable guide for policymakers, programme managers, clinicians, and public-health professionals, and I wish every success to the initiatives undertaken by the Health Department in this crucial area.

Pinarayi Vijayan

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Message

Congenital anomalies remain a significant public-health concern, contributing to neonatal and infant morbidity, mortality, and long-term disability. Addressing this challenge demands a comprehensive, integrated approach that spans prevention, early detection, timely clinical intervention, rehabilitation, and sustained support for affected children and their families.

Kerala has a strong maternal and child health care system because of the State's robust public-health initiatives, high antenatal screening and institutional delivery services, and the extensive network of diagnostic and specialty care facilities. But Congenital anomalies remain a significant public-health concern, contributing to neonatal and infant morbidity, mortality, and long-term disability. Addressing this challenge demands a comprehensive, integrated approach involving early detection, timely intervention, rehabilitation, and sustained support for affected children and their families. State is committed to further strengthening the prevention, surveillance, and management of congenital anomalies through evidence-based health interventions.

This document presents a comprehensive framework on congenital anomalies, detailing their epidemiology, risk factors, prevention strategies, screening and diagnostic pathways, referral systems, clinical management, and follow-up care. It underscores the importance of a continuum-of-care approach from preconception and antenatal periods to new born, childhood, and adolescent stages.

I am confident that this document will serve as a practical guide for programme managers, clinicians, public-health professionals, and partners involved in maternal, new born, child, and adolescent health. I appreciate all those who are committed to ensure that all children, including those born with congenital anomalies, are given the opportunity to survive, thrive, and achieve their fullest potential.

Veena George

Foreword

Kerala Health has been taking various initiatives in a life cycle approach right from " cradle to grave". It gives me immense satisfaction that the health system does analysis of morbidity and mortality and based on the analysis specific interventions are planned and taken for implementation. One such intervention is detection and corrections of congenital anomalies. This often referred as birth defects, constitute a significant public health concern across the world. They constitute a wide spectrum of structural and functional disorders that arise during intrauterine life, and their impact affects not only the affected individual but the families, communities, and healthcare system alike.

Inspite of the many advances in medical science, the challenges still remain in the prevention, early detection, and comprehensive care. By adopting the current knowledge and best practices, we can reduce the incidence of preventable anomalies, improve survival rates, and enhance the quality of life for affected children. The department has specific new born screening program Hridyam focusing at cardiac problems and Shalabham focusing at birth defects. The document extensively covers the details of all these interventions and achievements. We appreciate the works done by the teams at the field as well as at hospital level.

This document provides a clear overview of congenital anomalies, highlighting epidemiology, causes, diagnostic approaches, and management strategies. We sincerely hope that the document will serve as a practical guide for health care workers, researchers and policy makers for better and informed program implementation as well as it will be informative to people.

I appreciate the contributions done by Dr Sankar, Dr Deepa, Dr Soumya, Dr Rahul and the field teams. I appreciate the efforts taken by Dr Prathapa Chandran and Dr Sasikumar in supporting me and in coordinating with all for publishing the document.

Together we can strengthen the health system to ensure that every child has the opportunity to thrive and to reach their full potential.

Dr Rajan Khobragade IAS
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PREFACE

The birth of a child is a moment of profound joy and hope, symbolizing the continuation of life and the promise of the future. However, for many families, this joy is overshadowed by the challenges posed by congenital anomalies, which can significantly impact a child's quality of life. These structural and functional abnormalities present at birth can lead to long-term disabilities, placing considerable emotional, social, and economic burdens on affected families and societies.

This document aims to shed light on the global and regional burden of congenital anomalies, with a particular focus on the Indian and Kerala contexts. By examining the prevalence, causes, and risk factors associated with these conditions, we strive to underscore the importance of early detection, intervention, and prevention. The state of Kerala, with its advanced healthcare infrastructure, provides a unique case study of how integrated public health initiatives and specialized medical care can effectively address the challenges posed by congenital anomalies. The comprehensive analysis presented here covers the various dimensions of congenital anomalies, including environmental, socioeconomic, and maternal factors that contribute to their occurrence. Additionally, this document highlights existing and emerging programs in Kerala designed to mitigate the impact of these conditions, such as the "Hridyam" and "Shalabham" initiatives under the Ardram scheme. These programs represent significant strides in reducing infant and maternal mortality rates, improving early detection and intervention, and ensuring that all newborns receive the care they need to thrive.

In compiling this document, we aim to provide healthcare professionals, policymakers, and the public with a deeper understanding of the complexities surrounding congenital anomalies. By raising awareness and promoting informed action, we hope to contribute to the ongoing efforts to improve the quality of life for children affected by these conditions and to support their families in overcoming the challenges they face.

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Chapter 1
Introduction

Babies enter the world as a symbol of hope, reminding us that humanity, with all its diversity and complexity, continues to thrive. While the arrival of a new-born brings immense joy to those around, there are, unfortunately, some babies whose birth is overshadowed by sadness and concern due to birth defects. These congenital abnormalities, which may become apparent either immediately after birth or later, can affect a new-born's structure, function, or metabolism. Such conditions often result in physical and mental disabilities and, in some cases, can be life-threatening. Congenital disorders can be defined as structural or functional anomalies that occur during intrauterine life. Also called birth defects, congenital anomalies or congenital malformations, these conditions develop prenatally and may be identified before or at birth, or later in life. Congenital disorders can contribute to long-term disability, which takes a significant toll on individuals, families, health care systems and societies. Beyond their immediate medical impact, congenital anomalies carry profound psychosocial and economic consequences. Families often face long-term treatment costs, social stigma, and reduced quality of life for the child. For health systems, the challenge extends beyond acute care to life-long rehabilitation, inclusive education, and social protection.

1.1 Global Scenario

Congenital anomalies are one of the leading causes of global disease, responsible for a staggering 57.7 million disability-adjusted life years (DALYs) lost worldwide (WHO 2013). The burden of congenital anomalies is not limited to mortality. They contribute substantially to years lived with disability (YLDs), affecting schooling, employability, and family well-being. The economic cost is significant, with WHO estimating billions of dollars lost annually in health care expenditure and lost productivity.

In recognition of this, WHO and partners have emphasized the integration of birth defect surveillance, prevention, and care into maternal and child health programmes. The WHO Birth Defects Surveillance Toolkit (2020) and global commitments under the Sustainable Development Goals (SDG 3.2) – reducing neonatal mortality and under-5 deaths – explicitly underscore congenital anomalies as a key frontier.

The Global Burden of Disease study 2013 identified congenital anomalies among the top ten causes of mortality in children less than five years of age[12]. While congenital anomalies are the leading cause of death in children in this age group in the high-income countries, they are not considered to be significant public health problems in low- and

middle-income countries (LMICs). The key reasons for this public health under-prioritization in LMICs relate to the inherent characteristics of these conditions. They are low in prevalence, their proportionate contribution to mortality is significantly lower as compared to other perinatal causes, infections and malnutrition, and their management is resource intensive.

An estimated 6% of babies worldwide are born with a congenital disorder, resulting in hundreds of thousands of associated deaths[1]. Of the 5 million children under-5 years who died in 2020, approximately 400,000 died from congenital conditions, representing nearly 8% of total under-5 deaths globally. A large proportion of children with congenital conditions (over 90%) are born in low- and middle-income countries[3].Birth defects or congenital anomalies are an invisible tragedy around the world. Every year an estimated 295 000 new-borns die due to congenital anomalies during the first 28 days of life.

Congenital disorders cause a further 170 000 deaths of children between the ages of 1 month and 5 years[2]. The most common, severe congenital anomalies are heart defects, neural tube defects and Down's syndrome[4].

A small proportion of these conditions are genetic i.e. chromosomal abnormalities (e.g. Patau's Syndrome or trisomy 13). Environmental factors like maternal infections (rubella, Zika), exposure to radiation, certain pollutants, maternal nutritional deficiencies (e.g., iodine, folate), illness (maternal diabetes) or certain drugs (alcohol, phenytoin), also increase the risk. The cause of most birth defects is unknown. Complex genetic and environmental interactions are proposed but these have not yet been clearly elucidated.

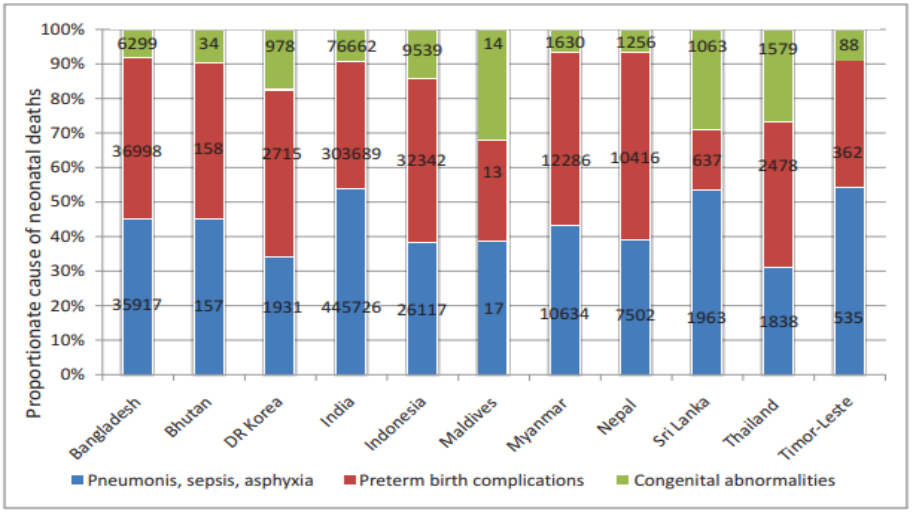


Figure 1 : Neonatal Mortality by Cause in Countries of the Southeast Asian Region[14]

Figure 1 presents the proportionate causes of neonatal deaths in several Southeast Asia region countries. Pneumonia, sepsis, and asphyxia account for a large share of neonatal deaths in all the countries. Variation between countries may reflect the differences in health system capacity, perinatal care quality, and reporting practices. Congenital anomalies represent a smaller but still a significant proportion in many countries including India.

1.2 Indian Scenario

According to WHO (2013), in India, birth defects cause the death of around 7% among under-5 children, 9.5 % of perinatal, and 9.9% of stillbirth defects. A nationwide analysis in 2014 estimated the prevalence of congenital anomalies at 70 per 10,000 births[13]. The perinatal period is the early postnatal period relating to birth which includes the period up to 7 days after birth and stillbirth are the late fetal deaths with a gestational age of 28 weeks or more. A quarter of global neonatal deaths occur in India. In 2013, the country reported a neonatal mortality rate of 29 per 1000 live births, responsible for 753,000 neonatal deaths (WHO, 2015) and the congenital anomalies constituted the fifth leading cause, being responsible for an estimated 9% of neonatal deaths in the year 2010 (Liu et al., 2012). The pattern and prevalence of congenital anomalies may vary over time with socioeconomic status, a complex interaction of known and unknown genetic and environmental factors. In India, the mortality and morbidity associated with low birth weight, birth asphyxia, and sepsis are of greater concern than congenital anomalies. [**Annexure:1**; List of different regions of India showing the occurrence of congenital anomalies][13]. The government is focusing significant efforts on addressing these leading causes of neonatal mortality. Although the Government of India has established the Rashtriya Bal Swasthya Karyakram (RBSK) at the district level for early intervention, aiming to identify and treat congenital anomalies promptly, the ground reality still lags. Various maternal factors contribute to congenital anomalies, many of which are preventable. Therefore, studying the patterns of congenital anomalies at the local level is crucial for implementing effective interventions. Contributing factors to the increased incidence of congenital defects in developing nations include genetic predisposition, socioeconomic challenges, environmental influences, and certain lifestyle-related diseases[13].

1.3 Kerala Scenario

In Kerala, congenital anomalies are a growing public health concern, contributing significantly to infant morbidity and mortality. Despite the state's advanced healthcare

infrastructure, the prevalence of congenital anomalies remains a challenge. However, Kerala has been proactive in addressing this issue through a combination of early detection, prenatal screening programs, and timely corrective interventions. The establishment of specialized paediatric centers and the Kerala Birth Defects Registry has enhanced the state's ability to monitor and manage these conditions effectively. Advances in neonatal surgery and postnatal care have significantly improved the survival and quality of life for children born with congenital anomalies. Kerala's integrated approach, involving public health initiatives and specialized medical care, continues to set a benchmark in managing and correcting congenital anomalies in India.

A new initiative by the Kerala state government will ensure that all new-borns born in public health facilities will be screened for various developmental diseases and birth defects within 48 hours of delivery. The universal Comprehensive Newborn Screening (CNS) programme will be the first of its kind to be launched in India. Kerala is focusing on the quality of survival of the new-borns. The low infant mortality rates do not reflect the quality of life of the surviving children. Our target is to reduce the Infant Mortality Rate (IMR) further from the current rate of 5 /1000 live births. This initiative will help to improve the quality of life of surviving children.

A study from Kerala[26] reported,

- Among 141,540 new-borns [140,558 deliveries: 139,589 singleton, 957 twins (6.81%), 11 triplets (0.078%), and one quadruplet] screened, 615 (0.44 %) were stillbirth and Major Congenital Anomalies were seen in 1,370 (0.97 %) new-borns.
- **Clubfoot** (404, 0.295%) was the most frequent major congenital anomalies followed by **hypospadias** (152, 0.21 % among male new-borns), **congenital heart disease** (168, 0.12%), cleft lip/palate (149, 0.11 %), **Down syndrome** (104, 0.07%), and **neural tube defects** (72, 0.05 %).[26]

Major Visible Birth Defects

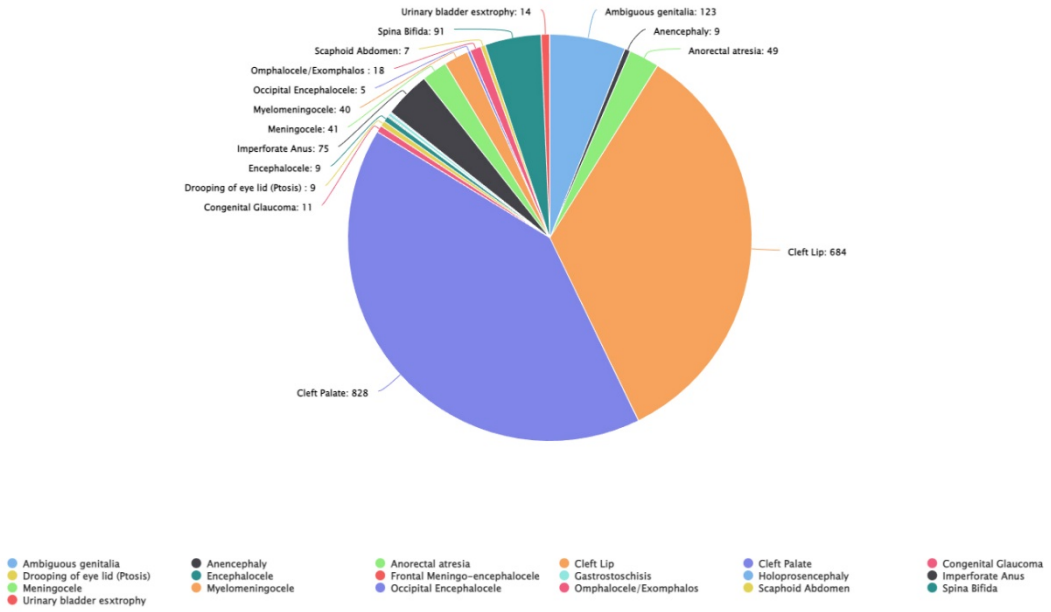


Figure 2 : Major visible congenital defects – Kerala study[26]

Chapter 2
Causes and Risk Factors of Congenital Anomalies

Congenital anomalies, also known as birth defects, are structural or functional abnormalities present at birth that can affect various parts of the body. These anomalies result from a complex interplay of genetic, environmental, nutritional and maternal factors. Understanding the causes and risk factors associated with congenital anomalies is crucial for prevention, early detection, and management. Congenital anomalies are highly associated with risk factors, and they have no definite patterns rather being vary in a pattern based on risk factors. However, several studies found that musculoskeletal systems are most associated with congenital defects mostly congenital talus equine.

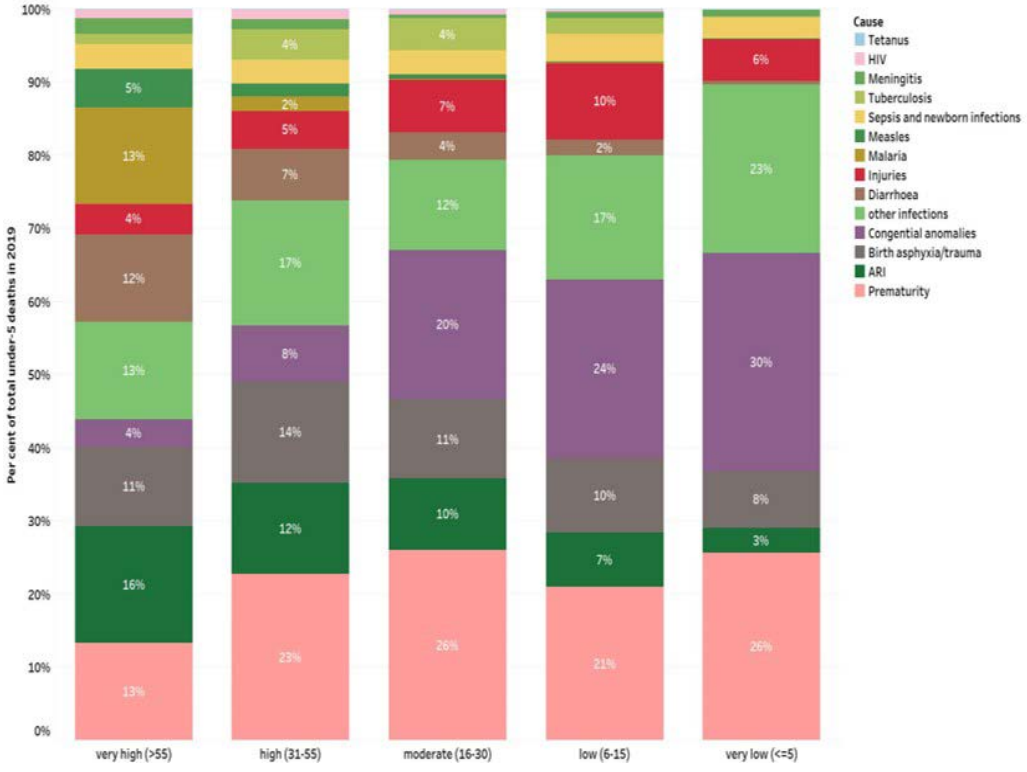


Figure 3 : Changes in causes of under 5 deaths as under 5 mortality rates decline

The proportion of under-5 deaths due to congenital disorders increases as other causes of under-5 deaths are controlled. Figure.3 shows the distribution of causes of under-5 deaths across countries grouped by their under-5 mortality rates. Countries with very high mortality show a substantial share of deaths from malaria, sepsis, diarrhoea, and injuries. In very low mortality settings, causes like congenital anomalies, prematurity, and birth complications dominate. (Ref: 2)

Conditions that may contribute higher incidence of birth defects in developing countries Inadequate periconceptional intake of folic acid, Iodine deficiency in mother's diet, Diabetes mellitus, Lack of vaccination against Rubella, Women giving birth after 35 years of age, Consanguineous marriage, Alcohol consumption during pregnancy, The use of teratogenic medications and oral contraceptives, Low birth weight and maternal malnutrition

The congenital anomalies originate during the intrauterine life of a fetus and show the different prevalence worldwide. The higher occurrence of hereditary disorders in a particular population could be the outcome of various social, cultural, and genetic factors. Though 40-60% congenital abnormalities are idiopathic in origin, 15-20% is contributed by multifactorial heredity, 10-20% by gene mutation, and less than 10 % by environmental factors.[13]

2.1 Genetic Causes

Genetic anomalies arise from alterations in DNA or chromosome structure, leading to developmental disruptions. Key causes include:

a. Chromosomal Abnormalities:

- Numerical Abnormalities: An extra chromosome can cause conditions like Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), and Patau syndrome (trisomy 13).

b. Single-Gene Disorders:

- Autosomal Dominant: One mutated gene copy causes the disorder.

- Autosomal Recessive: Both gene copies must be mutated.

- X-linked Disorders: Caused by mutations in X chromosome genes.

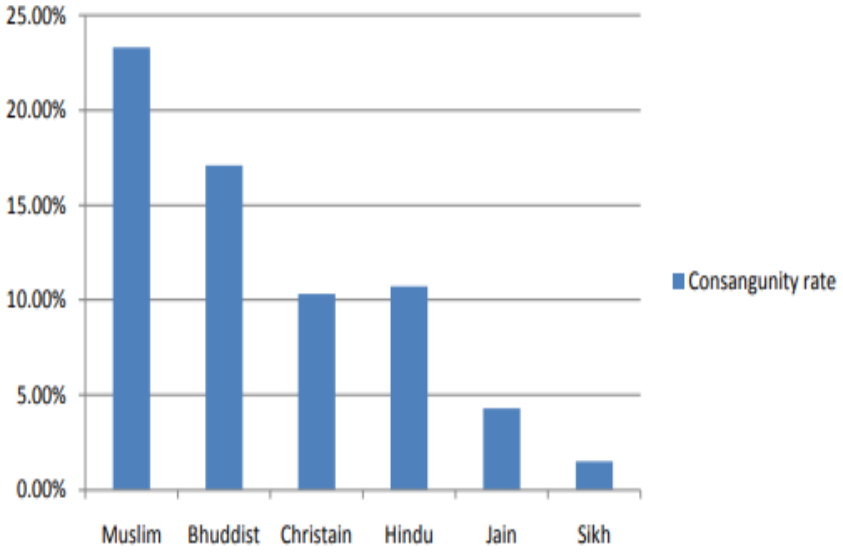


Figure 4 : Consanguinity rate in India[13]

Consanguinity (when parents are related by blood) also increases the prevalence of rare genetic congenital disorders and nearly doubles the risk for neonatal and childhood death, intellectual disability, and other anomalies.

2.2 Environmental Factors: Environmental influences significantly impact congenital anomalies. Key factors include:

- **Teratogens:** Substances like thalidomide and valproic acid can cause malformations when taken during pregnancy.
- **Infections:** Maternal infections like rubella, cytomegalovirus, and toxoplasmosis can lead to congenital anomalies.
- **Chemicals and Radiation:** Exposure to lead or high radiation levels can disrupt fetal development.
- **Nutritional Deficiencies:** Lack of folic acid, iodine, or vitamin A can result in defects like spina bifida or brain development issues.

2.3 Socioeconomic and Demographic Factors:

- Congenital disorders are more common in low- and middle-income countries, often due to limited access to nutrition, healthcare, and increased exposure to harmful factors like infections or alcohol

2.4 Maternal Factors: Certain maternal conditions increase the risk of congenital anomalies:

- **Maternal Age:** Women over 35 are at higher risk for chromosomal abnormalities like Down syndrome.
- **Chronic Health Conditions:** Poorly controlled diabetes and hypertension can lead to heart defects, neural tube defects, and other complications.
- **Maternal Stress and Poor Antenatal Care:** Limited vaccination (rubella, tetanus), inadequate supplements, and infrequent antenatal visits reduce opportunities for prevention.
- **Lifestyle Factors:** Smoking, alcohol consumption, and drug use during pregnancy are linked to various congenital anomalies.

2.5 Lifestyle Diseases:

- Maternal obesity, diabetes, and other lifestyle diseases are associated with an increased risk of congenital anomalies, including neural tube defects and cardiac malformations.

2.6 Unknown and Multifactorial Causes

- While many congenital disorders have unknown causes, some arise from complex genetic and environmental interactions. Conditions like cleft lip and congenital heart defects are often influenced by both genetic predispositions and external factors.

2.7 Impact and Challenges

Congenital anomalies may result in miscarriage, stillbirth, neonatal death, or lifelong physical and psychological challenges. In low-resource settings, limited availability of rehabilitation, counselling, and inclusive education worsens outcomes, leading to significant social and economic burdens.

Chapter 3
Focus on Rare Diseases or Unknown Causes

While many congenital anomalies are caused by well-known genetic or environmental factors, a significant proportion arise from rare diseases or unknown causes. Understanding and addressing these anomalies is crucial for several reasons.

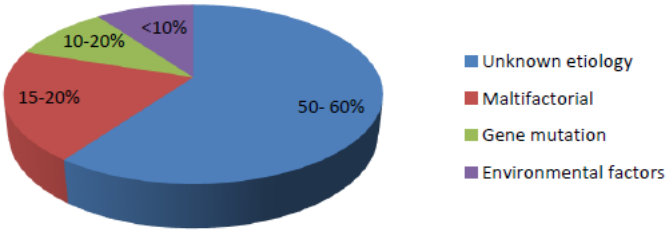


Figure 5 : Etiology of Congenital anomalies[13]

- The frequency of congenital anomalies is increased in deficient essential precautionary measures for public health. There is a tremendous need for organizing public health surveillance programs so that the data collected from patients will be used to improve public health. As of now, 50-60% of causative agents are unknown. Hence more emphasis should be given to prevention via regular antenatal care and avoidance of exposure to known teratogenic agents. Antenatal diagnosis, management facilities, genetic counselling and better diagnostic facilities should be provided to improve the outcome[13].
- Research into the unknown causes of congenital anomalies can uncover new genetic mutations, environmental factors, and biochemical pathways, leading to potential new treatments. If these conditions are not properly treated, they can result in serious complications and expensive medical care.
- Rare diseases, though individually uncommon, collectively affect a significant portion of the population and often result in congenital anomalies that go undiagnosed or misdiagnosed due to their rarity and limited awareness among healthcare providers. This gap in medical care underscores the importance of focusing on rare diseases, as it can lead to the development of targeted diagnostics and treatments.
- Prioritizing rare diseases and unknown causes in congenital anomalies is crucial, not only for advancing medical research but also for improving patient care, for reducing the out-of-pocket expenditure and for ensuring the necessary attention and support for the vulnerable population.

Chapter 4
Existing Programmes in Kerala

Kerala has several notable newborn and child health programs and initiatives which brought its health indices like IMR and NMR comparable to the developed countries in the world. Kerala continues to strengthen the system with innovative schemes like universal newborn screening (Shalabham), coordinated congenital-heart-disease care (Hridyam), and Clubfoot Free Kerala.

4.1 HRIDYAM For Little Hearts Programme

Hridyam for Little Hearts is a pioneering initiative under the Rashtriya Bal Swasthya Karyakram (RBSK) in Kerala, aiming to reduce infant and neonatal mortality and morbidity caused by Congenital Heart Diseases (CHDs). It ensures free identification, referral, surgical management, and follow-up care of children up to 18 years of age, irrespective of their socio-economic status. This web-based initiative serves as a comprehensive registry for CHD cases, allowing for the monitoring of program progress, identification of implementation bottlenecks, real-time tracking of case status, and evaluation of program outcomes[18].

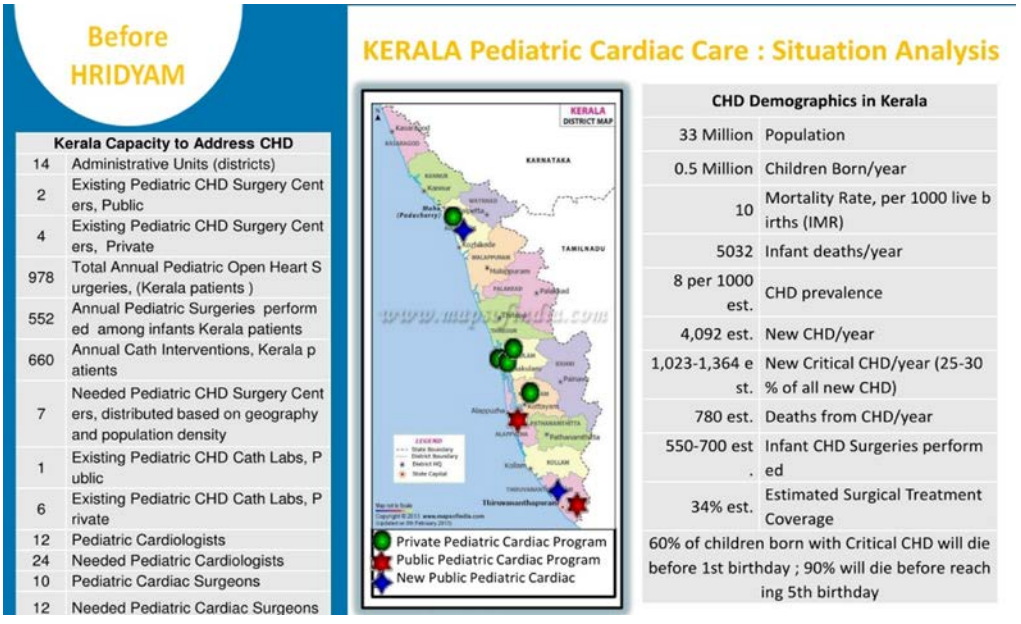


Figure 6 : Kerala paediatric cardiac care: situation analysis[17]

4.1.1 Hridyam - Methodology and Workflow

The Hridyam program follows a structured methodology and workflow to ensure systematic case management. Children suspected or confirmed with CHD are registered on the Hridyam online portal, where a unique case number is generated and linked to the respective District Early Intervention Centre (DEIC). Pediatric cardiologists then review the uploaded clinical data and categorize cases into specific priority groups—Category 1 (a–g) for emergency and semi-emergency cases, Category 2A and 2B for staged or elective surgeries, and Category 3 for cases requiring medical management and monitoring. These specialists, drawn from designated tertiary cardiac care centers, may re-categorize cases as clinical status changes. Surgical allocation is facilitated through empanelled government and private hospitals with system login access, where hospitals regularly update the availability of surgical slots. Based on case priority and parental consent, children are assigned to suitable centers for surgery and further care. At present 3 Govt. and 5 Pvt. Hospitals are empaneled for surgery. More hospitals are being added to the list. DEICs facilitate the entire process, including referrals to empanelled hospitals in consultation with the family. State level administration will grant preauthorization for empanelled hospitals for surgery and will monitor the entire process. Follow-up up to 1year post-surgery is also provided.

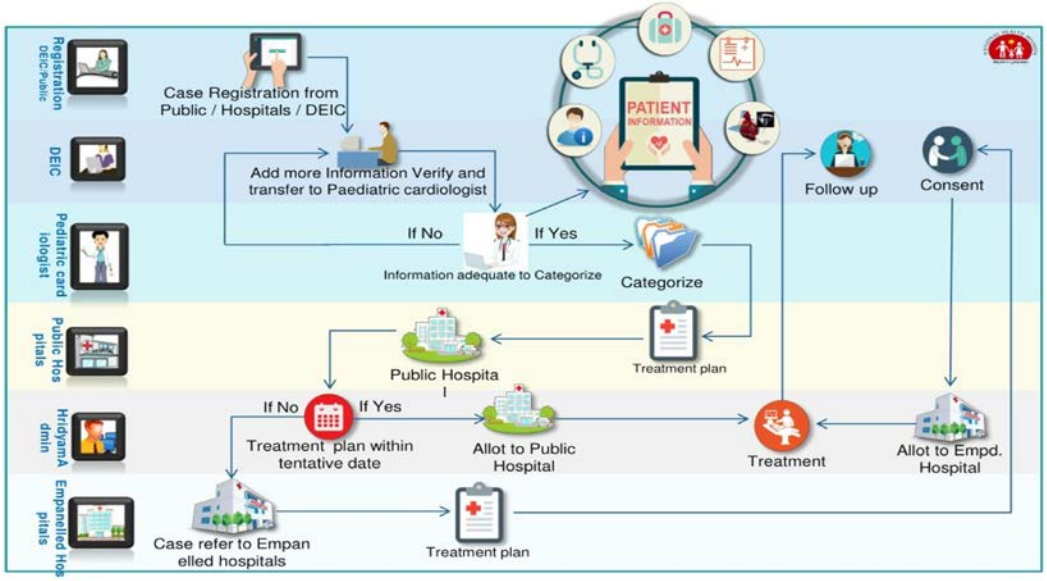


Figure 7 : Hridyam process map[25]

4.1.2 Hridyam - Surgery details:

Table 4.1: Number of Surgeries / Interventions under Hridyam

Year	Number of Surgeries / Interventions
2017–2018	295
2018–2019	748
2019–2020	926
2020–2021	1,102
2021–2022	1,428
2022–2023	1,694
2023–2024	1,512
2024–2025	886

Source: Hridyam Portal

4.1.3 Public Awareness and Communication: To increase public awareness and to facilitate informal communication, a Facebook page named “Hridyam,” a dedicated WhatsApp number, and a Twitter account have been established.

4.2 SHALABHAM programme

The Kerala government's new initiative to screen all newborns for developmental diseases and birth defects within 48 hours of delivery is a significant step forward in child health in the state. This universal **Comprehensive Newborn Screening (CNS) programme**, the first of its kind in India, reflects the state's commitment to improving early detection and intervention. By implementing this comprehensive screening package, Kerala aligns with the goals of the national Child Health Policy, aiming to enhance health outcomes for children through timely and effective diagnosis of birth defects[18].

Government of Kerala launched the Comprehensive New Born Screening program as part of the Ardram scheme to augment the existing Newborn screening programs. This includes Visible Birth Defect (VBD) screening, screening for Inborn Errors of metabolism, Hearing screening by OAE (Oto Acoustic Emission testing), Pulse Oximetry screening for Critical Congenital Heart Disease (CCHD) & Retinopathy of Prematurity (ROP) screening and aims to bring all under a single umbrella.

4.2.1 Program Overview

In a country where 99% of births occur in hospitals, the Comprehensive Newborn Screening Program is designed to examine all new-borns born in government hospitals. The program includes the following critical screening tests:

1. **Screening for Visible Birth Defects -** within 24 hours post-delivery
2. **Pulse Oximetry screening for CHD -** 24-48 hours post-delivery
3. **Oto Acoustic Emission (OAE) screening for hearing -** 24-48 hours and before discharge
4. **Invasive blood spot examination for IEM -** around 48 hours and before discharge.

This needs to be performed by the Delivery point Staff Nurse (Post Natal Ward or anyone designated) /RBSK nurse, under the guidance of the Pediatrician or Medical officer

4.2.2 Approach to Newborn Screening

- Immediately after birth, each newborn undergoes an examination by a pediatrician, where APGAR, respiratory rate, and heart rate are recorded. The child's weight, length, head circumference, and chest circumference are also measured. All findings are meticulously documented in the newborn's case sheet. The four primary screening tests are completed before the mother and child are discharged from the hospital, with a strong recommendation for a thorough examination within 48 hours of birth.
- High-risk cases, such as preterm infants, low birth weight babies, and those admitted to SNCU, undergo additional tests, including checks for neurodevelopmental delays. These tests can be scheduled on any day or after SNCU discharge. Guidelines and checklists are available under the RBSK: VBD scheme for conducting these examinations.

4.2.3 Detailed Screening Procedures

4.2.3.1 Visible Birth Defect Screening

This screening aims to detect visible or hidden birth defects early. An Android application, Jatak Seva, has been developed to facilitate the screening process. Staff nurses enter new-born details into the application, including basic information like birth date, weight, length, head circumference, and parents' information (figure.8). If any defects are found during the physical examination, images of the affected area can be captured and uploaded. Each child is then assigned a VBD-UID, which is recorded on the case sheet.

The collected data is reviewed at DEICs (District Early Intervention Centers), where necessary treatments are ensured in consultation with the paediatricians. The RBSK

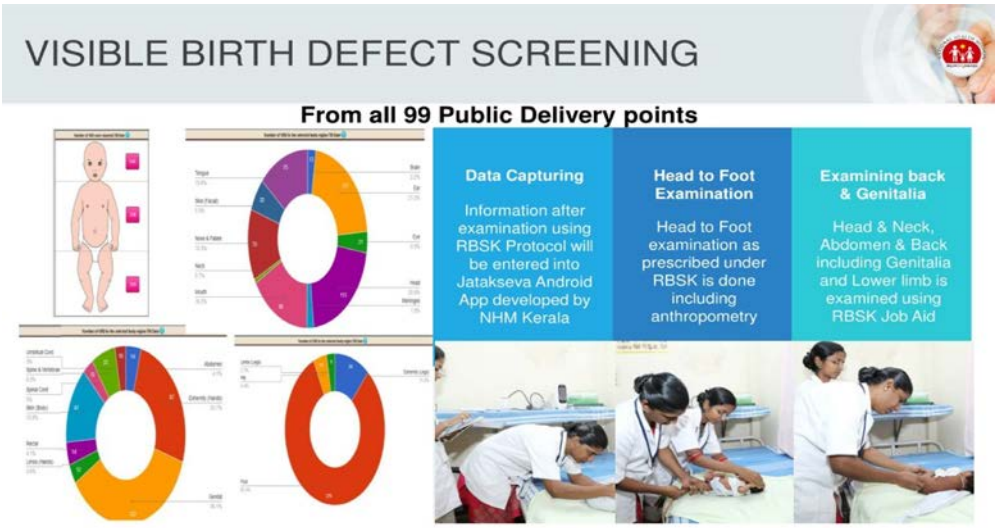


Figure 8 : Visual Birth Defect Screening

Nursing staff receives SMS notifications of these cases, enabling effective follow-up at the field level.

4.2.3.2 Functional Birth Defect Screening

4.2.3.2.1 Oto Acoustic Emission (OAE) Screening: This screening is conducted at all major delivery points in Kerala, supported by the Kerala Social Security Mission (KSSM), capture data from individual units for each child in VBD software. The data coming to this portal will be shared to NHM portal for Birth Defect in a separate module and the results will be shared to DEICs and District officials along with concerned area RBSK Nurse and Field staff. This will ensure follow up of children identified with hearing impairments for consultation with ENT surgeons, to do a BERA for confirmation, hearing aid support and speech therapy from DEICs/ other centers.

4.2.3.2.2 Pulse Oximetric Testing: This test is the first step in detecting congenital heart disease in new-borns. Hospitals with more than 50 deliveries per month and 14 DEICs are equipped with state-of-the-art pulse oximeters. These devices accurately measure oxygen saturation in the arms and legs of the new-borns, with results linked to the VBD-

UID. The data is collected through the device's software and is available on the Hridyam portal. Cases with abnormal oxygen saturation levels are subjected to further

examination, including echocardiograms, with treatment facilitated through the Hridyam scheme.

4.2.3.2.3 Retinopathy of Prematurity (ROP) Screening: Premature, low birth weight, and SNCU/NICU-admitted infants are screened for ROP, which affects vision. Early detection and treatment can prevent blindness. Screening is conducted by trained staff nurses using Midriatic camera and the pictures uploaded to central console where this will be read by experts /trained ophthalmologists. Those children screened positives need to be referred to experts available in districts and will be further screened and treatment made available through medical colleges. The data will also be captured in the Birth Defect system portal and will be followed up through this system using RBSK Nurses.

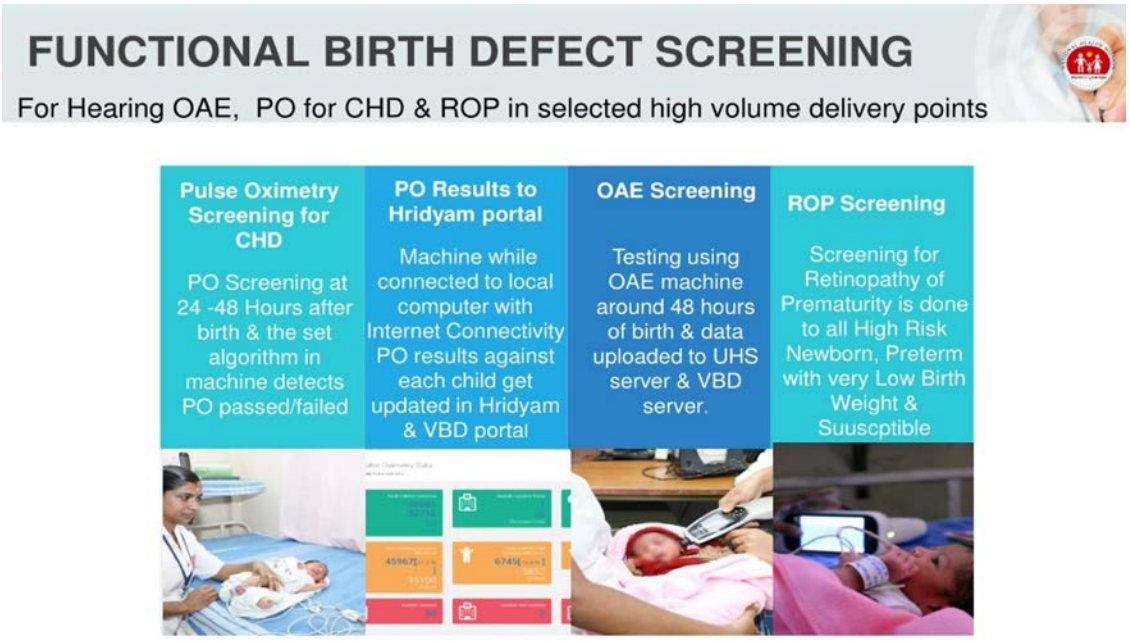


Figure 9 : Functional Birth defect screening

4.2.3.3 Inborn Errors of Metabolism (IEM) Screening: IEM blood tests are conducted at major delivery points across the state to detect four disorders (CAH- 17 OHP, Congenital Hypothyroidism, G6PD Deficiency & Galactosemia). The VBD-ID is recorded on the blood sampling card, and the samples are tested in regional labs. The results are integrated into the VBD Portal, allowing for coordinated treatment through DEICs.

4.2.3.4 Neurodevelopmental Defect Screening

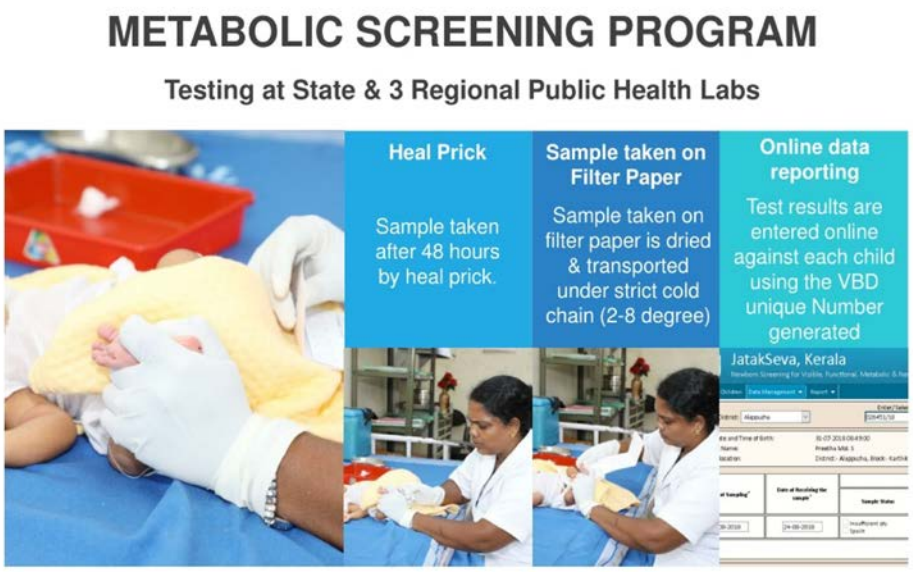


Figure 10 : Metabolic Screening Programme

High-risk new-borns in SNCU/NICU are regularly screened for neurodevelopmental defects as part of the RBSK scheme. The examination focuses on growth, development, and any defects or deficiencies. Babies diagnosed with issues receive expert treatment through DEICs or Anuyatra mobile units.

4.2.4 Technological Support and Implementation: To ensure the effective implementation of the Visible Birth Defect Screening, a dedicated mobile application was developed. Additionally, 1306 tablet computers were distributed to all delivery points for enhanced program management. As part of the Anaemia Mukt Bharat project, 1070 Digital Haemoglobin meters were provided in 2019-20 for anaemia screening by RBSK Nurses across all districts.

4.3 CLUB FOOT PROGRAMME

Club Foot is one of the most common congenital deformity affecting new-borns, causing physical limitations and impacting quality of life. Effective treatment through the gold standard Ponseti technique requires early detection and intervention. The Kerala government, in partnership with CURE India, launched the "Club Foot Free Kerala Programme" to address this issue. In this programme emphasis was on early detection through screening at birth and prompt referral to club foot clinics for treatment. Treatment

is provided free of cost at 43 public health facilities like Medical Colleges, District & General Hospitals and Taluk Level Hospitals spread across 14 districts of the state. Training programs have been conducted for doctors, nurses, and other health workers to enhance treatment capacity. Awareness campaigns have been taken up to educate communities, improve understanding of clubfoot and increase program enrolment. Since 2021, over 4000 children have received successful treatment, significantly reducing the physical, emotional, and financial burden on families.

Figure 11 : Clubfoot clinic

Club foot treatment involves raising awareness, followed by identifying and referring the child to a clubfoot clinic equipped with trained professionals and the necessary resources. Treatment is not a one-time intervention; it demands ongoing dedication and effort over several years. A team of trained professionals consisting of doctors, nurses and other support stuff is required for provision of comprehensive care. The entire team, including the child's family, plays a crucial role, as the family is a vital partner in the treatment process.

Table 4.2: Clubfoot clinics in Kerala

CLUBFOOT CLINICS IN KERALA

Sl No	District	Institution
1	Thiruvananthapuram	MCH Trivandrum
2	ALAPPUZHA	MCH Wandanam
3	Kottayam	MCH Kottayam
4		GH THRISSUR
5	Thrissur	MCH THRISSUR
6	Kozhikode	MCH Kozhikode
7	Kannur	DH-KANNUR
8	Kasargod	TH THRIKARIPUR
Sl No	**District**	**Institution**
9		DH Nedumangaud
10	Thiruvananthapuram	GH Neyyattinkara
11		THQH Karunagappally
12		THQH Kottarakkara
13		MCH Paripally
14	Kollam	DEIC Kollam
15		THQH Ranni
16		GH Adoor
17	Pathanamthitta	THQH Thiruvalla
18		DH MAVELIKARA
19	Alappuzha	THQH CHERTHALA
20		THQH Vaikom
21	Kottayam	GH Pala
22		DH IDUKKI
23	Idukki	DH THODUPUZHA
24		DH ALUVA
25		GH MUVATTUPUZHA
26	Ernakulam	MCH KALAMASERRY
27		THQH KUNNAMKULAM
28	Thrissur	GH IRINJALAKUDA
29		THQH,MANNARKKAD
30		DH Palakkad
31		THQH,OTTAPALAM
32		GTSH Kottathara
33		THQH Alathur
34	Palakkad	THQH,CHITTUR
35		GMCH MANJERI
36		THQH THIRURANGADI
37	Malappuram	DH TIRUR
38		DH Vatakara
39	Kozhikode	TH Thamarassery
40	Wayanad	DEIC WAYANAD
41		GH THALASSERY
42	Kannur	TH PERAVOOR
43	Kasargod	GH KASARAGOD

Every new-born child delivered at Government Health Facilities is screened by the Delivery Point Staff or Nurses under the Rashtriya Bal Swasthya Karyakram (RBSK) for Visual Birth Defects. Club-foot detection in the State generally occurs during this time and the case registered in Shalabham portal. As the treatment using Ponseti technique needs to be initiated in the first week of birth, the child is immediately referred to the nearest clubfoot clinic convenient to the parents after due counselling by the paediatrician.

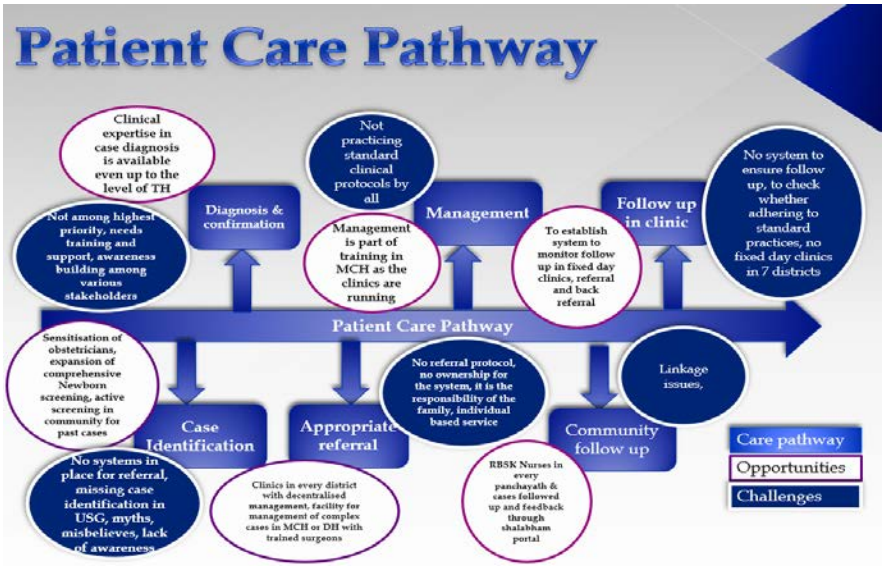


Figure 12 : Patient Care pathway for Clubfoot

The course of treatment including the date of casting should be captured to ensure adherence to the treatment protocol. Appropriate counselling to the family is an integral part.

There should be an established mechanism to screen for all those babies brought to the immunisation clinics and born in private sector institutions. All identified cases from the community by ASHA workers and HI/JPHNs along with those identified in the immunisation sessions should be registered in Shalabham portal and followed up to ensure complete cure and any disability due to clubfoot.

Chapter 5
Comprehensive Screening and Early Diagnosis

Health care before and near conception (preconception and peri-conception) includes basic reproductive health practices, as well as medical genetic screening and counselling. Screening can be conducted during the 3 periods listed[2] - Preconception, Periconception and Neonatal screening.

5·1 Pre-conception Screening

This can be useful to identify those at risk of specific disorders or of passing a disorder onto their children. Screening includes obtaining family histories and carrier screening and is particularly valuable in countries where consanguineous marriage is common.

A preconception examination, also known as a preconception visit, is one of the best ways to ensure a healthy pregnancy. The goal is to assess the overall health and to identify any risk factors that can complicate a pregnancy. A preconception examination can include any of the following[6]:

- **Family medical history**: A doctor will assess the medical history of couple's biological parents to see if any family member has had medical problems such as high blood pressure, diabetes, or intellectual disability.
- **Genetic testing:** A doctor will assess any possible genetic disorders that can be passed down to the baby. Some of the genetic disorders can be detected by blood tests before pregnancy.
- **Personal Medical History**: to determine if the mother has any medical conditions that may require special care during pregnancy (anemia, epilepsy, diabetes, high blood pressure); to gather information about previous surgeries; and to obtain information about past pregnancies such as complications, losses, and length of gestation.
- **Vaccination status:** To assess the immunity to diseases such as tetanus, rubella (German measles) that can cause miscarriage or birth defects. Vaccine can be given at least three months prior to conception to provide immunity.
- **Infection screening:** Infection screening determines if a woman has a sexually transmitted infection, a urinary tract infection, or any other infection that can be harmful to her or to the fetus.

5·2 Periconception Screening

Maternal factors may increase the risk, and screening results should be used to offer appropriate intervention to mitigate the risk. Maternal factors include maternal age, use of alcohol, tobacco or other drugs, uncontrolled diabetes, hypertension, and hypothyroidism. Ultrasound can be used to screen for Down syndrome and major structural abnormalities during the first trimester, and for severe fetal anomalies during the second trimester. Maternal blood can be screened for placental markers to predict the risk of chromosomal abnormalities, neural tube defects, and for free fetal DNA screen for many chromosomal abnormalities. Diagnostic tests such as chorionic villus sampling and amniocentesis can be used to diagnose chromosomal abnormalities and infections in women at high risk.

5·3 Neonatal Screening

Screening the newborn is an important step towards detection of congenital anomalies which facilitates in early referral and the initiation of medial or surgical treatment which in turn reduces the mortality and morbidity. Early screening for hearing loss provides an opportunity for early correction which helps the child to acquire better language, speech and communication skills. Early screening of newborns for congenital cataract or ROP allows early referral and surgical correction to improve the vision.

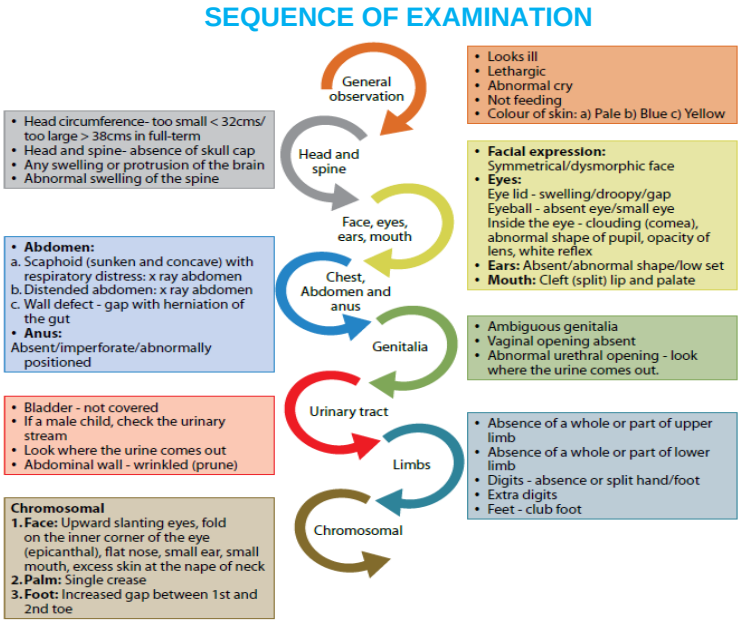


Figure 13 : Sequence of examination [16]

Newborns may be screened for certain metabolic, hematologic and endocrine disorders, many of which may not have immediately visible effects at birth. The conditions screened for may vary depending on the prevalence and cost.

5·3·1 Comprehensive New-born Screening (CNS) Programme in Kerala

A new initiative by the Kerala state government, Comprehensive Newborn Screening (CNS) programme, will ensure that all newborns delivered in public health facilities will be screened for various developmental, functional and birth defects within 48 hours of delivery. This universal Comprehensive Newborn Screening programme is the first of its kind to be launched in India[16].

Kerala has low infant mortality rates. As per the Sample Registration System (SRS) 2023, Kerala has achieved an Infant Mortality Rate (IMR) of 5, surpassing the earlier target of reducing IMR to 8 by 2020. Hence the state is focusing on the quality of survival of the new-borns. The low infant mortality rates do not reflect the quality of life of the surviving children. The focus is on enhancing the quality of survival and developmental outcomes among newborns.

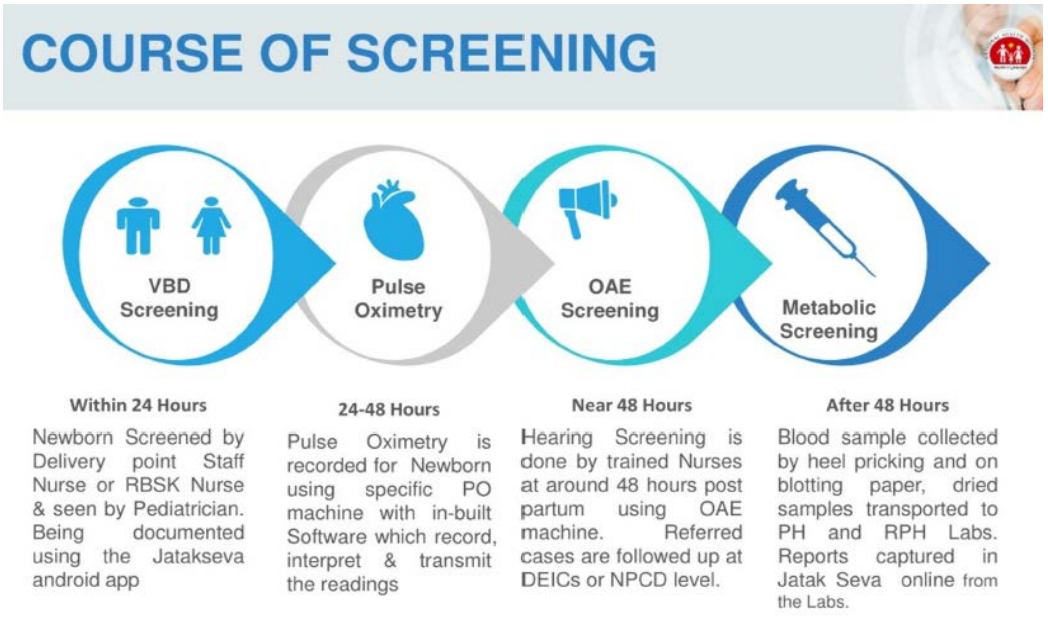


Figure 14 : Course of screening in Shalabham programme[17]

Visible birth defects are those that can be seen in infants, such as clubfoot, cleft lip and cleft palate. Under functional birth defect screening, babies will be tested for visual, auditory and heart defects as well.

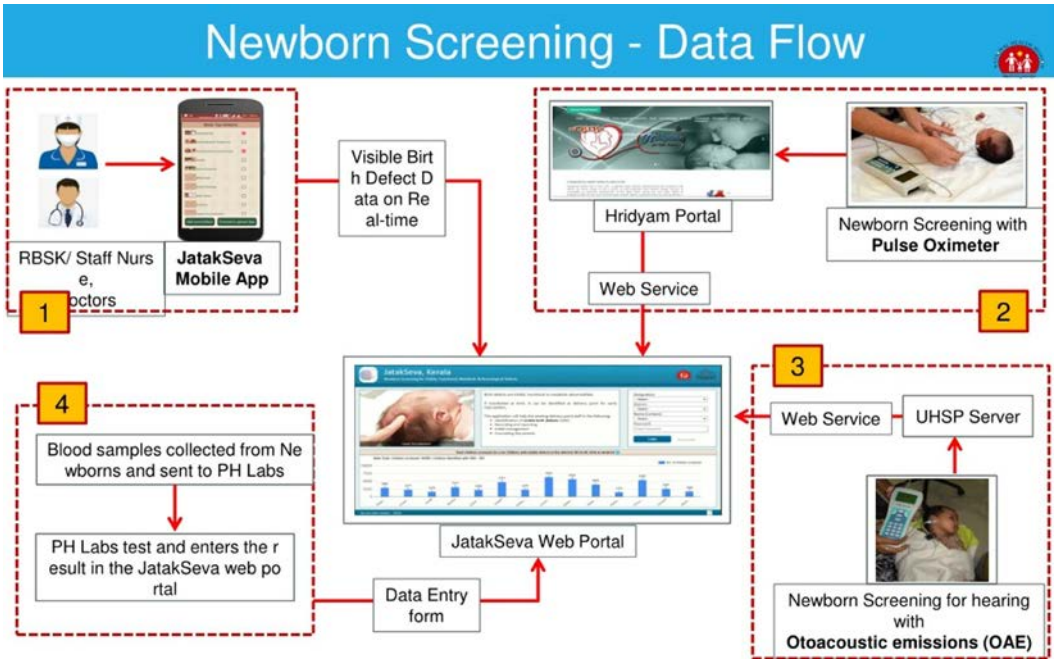


Figure 15 : Comprehensive Newborn Screening; data flow

Newborns will be screened for hearing problems using **Oto-Acoustic Emissions test** (OAE) and those identified with defects will be followed up to take corrective measures before 6 months of age.

Babies will also be screened for Congenital Heart Diseases (CHD). An innovative initiative called **Hridyam** was launched by the state with the goal of decreasing the infant mortality rates and morbidity due to congenital heart diseases.

Retinopathy of prematurity (ROP), which can lead to blindness, is another functional birth defect which will be screened for. The prevalence of low birth weight and preterm new-borns has seen an increase in the state. Often, premature babies in the intensive care units are given a high flow of oxygen. This high concentration oxygen flow may lead to retinopathy which can possibly result in blindness. Screening of such high-risk babies for ROP will help in early diagnosis, and treatment which will prevent blindness.

Though screening for **metabolic disorders** such as congenital hypothyroidism was in place for last 4 years, it had not been made universal. Now under CNS programme, every child born in a public health facility will be screened for Congenital Hypothyroidism, Congenital Adrenal Hyperplasia(17OHP), Galactosemia and G6PD deficiency.

Though **Neurodevelopmental defects** cannot be identified at birth, screening helps to detect whether any babies will require a follow-up in the future.

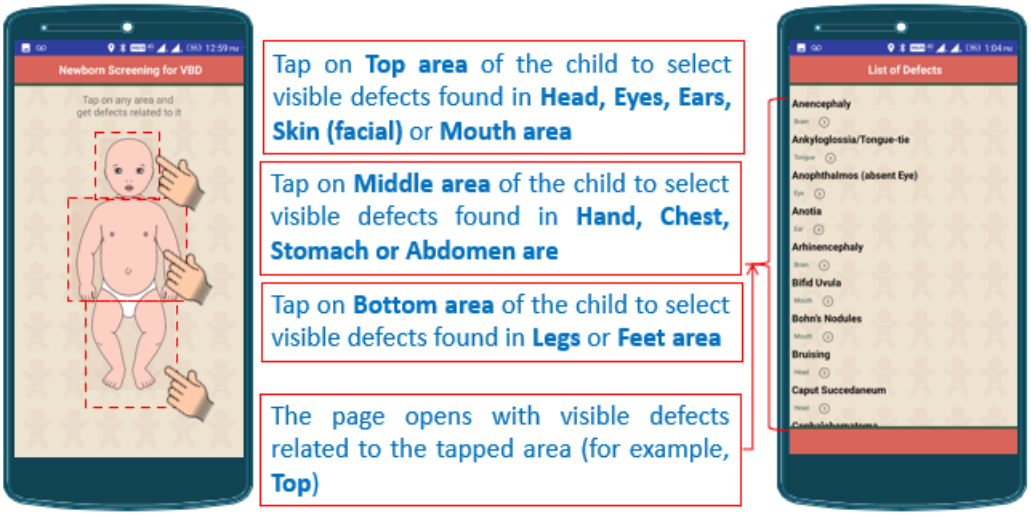


Figure16 : Jatak Seva Screening App

We need to identify all these cases at the earliest and act on them so that the disabilities due to them can be minimized. An Android application, Jatak Seva App, for documentation has been developed which will be used at the delivery point wherein data on 21 parameters will be collected. The information will come to a central server where the database is maintained. All the screening results against a unique number generated

by the Android application will be available for every child. If any baby is found to have an anomaly, the information will go to the concerned area field staff under the health department which will help in the follow up and treatment by the medical team. The state government will also offer treatment support using Rashtriya Bal Swasthya Karyakram (RBSK), and Arogyakiranam funds.

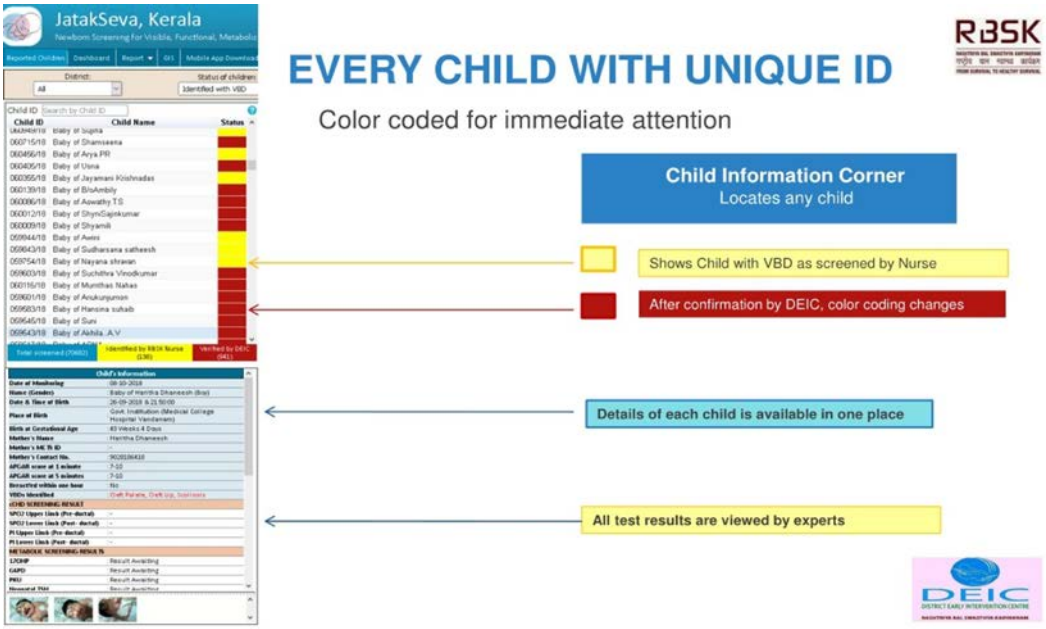


Figure 17 : Android application for maintaining database[17]

5·4 Child Development Centre (CDC), Medical College, Thiruvananthapuram was started on 1st of August 1987, as a project and later elevated to an autonomous centre by the Government of Kerala. During the year 1995 CDC was registered as a society (under Travancore-Cochin-Literary, Scientific and Charitable Societies Registration Act-XII of 1955, Reg. No.363/95) by the Government of Kerala with the intention of bringing up the centre as an autonomous centre of excellence in Early Child Care & Education, Adolescent Care & Education, Pre-marital Counselling, Women's Welfare and other related fields.

CDC's research and development involve mainly in the field of disability and its management with reference to preschool children, adolescent care and women and youth welfare. The Centre also conducts community extension programs such as school health programs, medical camps and undertake various training programs for health and ICDS functionaries in the State.

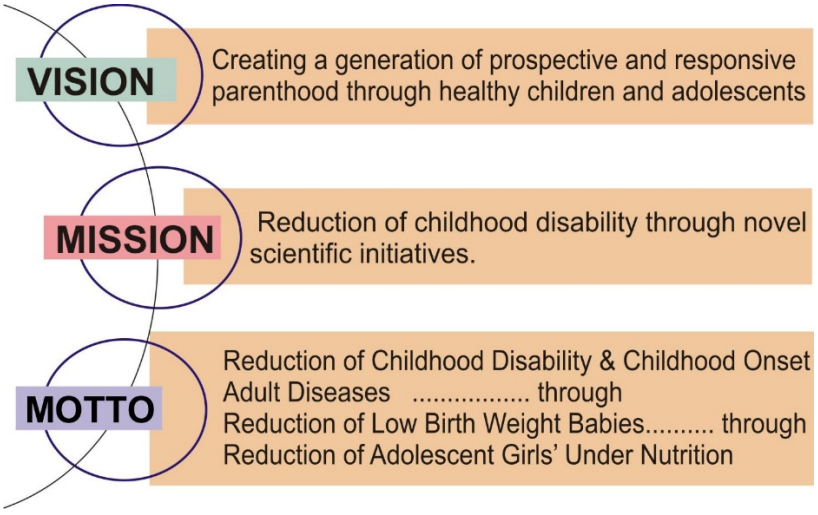


Figure 18: CDC – Vision, Mission and Motto

Table 5.1: Services offered by CDC

CDC SERVICES

- **Early stimulation and Early intervention clinic**
- **Developmental Evaluation Clinic**
- **Preschool Intervention Clinic**
- **Neurodevelopmental Disorders Evaluation and Intervention Clinics**
- **Adolescent Care Counselling Clinic**
- **Genetic clinic & Laboratory**
- **Sradha Project (Fetal Medicine Unit)**

Developmental assessment is being done at development evaluation clinics I, II, III according to the stipulated age using simple but effective screening tools designed and validated at CDC and gold standard Developmental tests.

5·4·1 Developmental Evaluation Clinic I

Children with Developmental Delay of the age group 0-3 years are referred to the evaluation clinic for providing assessment and intervention facilities.

Objectives

1. Identifying children with developmental delay using various screening and diagnostic tools
2. Growth monitoring using growth charts
3. Following up the children at regular intervals
4. Parental Counselling

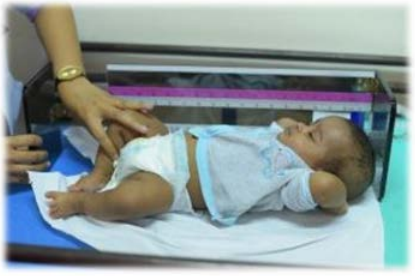


Figure 19: Development evaluation clinic I

5·4·2 Developmental Evaluation Clinic II

Intervention is aimed at enhancing language and communication, social play, pre-academic and independent living skills of young children so that they may take better advantage of opportunities in their communities and may require less professional attention as they grow older. Intervention is mainly given for children who are diagnosed as having Attention Deficit Hyperactive Disorder (ADHD), Learning Disorders (LD),

Intellectually disabled and ASD (Autism Spectrum Disorder). Intervention packages are available for children with different problems.

Objectives

1. Identification of developmental and learning problems of pre-schoolers.
2. Assessing the behavioural pattern of children
3. Early detection and intervention services for children between 3 or 6 years

Service Offered

1) Growth monitoring of children – Weight, Height & Head Circumference
2) Various Developmental Screening Tests.
3) Psychological evaluation
4) Educational counselling for scholastic backwardness
5) Evaluation, diagnosis, and treatment plan for medical problems by Developmental Paediatrician

Figure 20 : Development evaluation clinic II

5·4·3 Developmental Evaluation Clinic III

As part of developmental evaluation, various intervention programmes are being conducted for this group (6-12years), which include pre-adolescent children also. The Behavioural Pediatrics and Scholastic Backwardness unit take care of the cases attending this clinic.

Service Offered

1) Growth monitoring of children – Weight, Height & Head Circumference
2) Educational counselling for scholastic backwardness
3) Various Developmental Screening Tests
4) Psychological evaluation
5) Evaluation, diagnosis and treatment plan for medical problems by Developmental Pediatrician

5·4·4 At Risk Baby Clinic

At-risk babies are more likely to develop developmental delay and hence regular developmental follow-up at fixed interval is planned for these children. All children will be categorized on risk basis as mild, moderate, and severe. All mild risk babies will be followed up at 4th month, then at 1 year, moderate-risk babies will have the follow up regime at 2,4,8,12 months. All babies in severe risk categories will be followed up monthly till one year.

Objective

1. Reducing childhood disability through early detection and intervention program
2. Assessing the developmental status of new-borns who are at risk for developing developmental delay
3. Assessing and monitoring the growth status

Table 5.2 : Services offered by At Risk Baby Clinic - CDC

Services Offered

1. Health Education – Breast-feeding, weaning, supplementary foods, immunization, parenting etc.
2. Growth monitoring– weight, length & Head Circumference measurement
3. Developmental assessment using Trivandrum Developmental Screening Chart (TDSC) & CDC Grading for motor milestones
4. Developmental Screening using Denver Developmental Screening Test (DDST)
5. Neurological evaluation using Ameil-Tison method of neurological evaluation
6. Assessment of social age using Vineland Social Maturity Rating Scale
7. Early Intervention services (general stimulation techniques) for all children
8. Specific mother-oriented therapy for those children showing mild developmental delay
9. Screening for hearing problems using Oto Acoustic Emission test
10. Screening for visual abnormalities by the ophthalmologist
11. Screening for medical problems by the Developmental Paediatrician.
12. Appropriate referrals to various specialty, if found necessary.
13. Follow up in pre-school – 3 ½ years and further need based follow up

5·4·5 Imageology Division

5·4·5·1 SRADHA Project – Foetal Medicine Clinic

The "Sradha project" aims at reduction of childhood disability through various antenatal intervention including anomaly scanning using the 4D Ultrasonography machine and advanced equipment available at Genetic & Metabolic unit of CDC. The major services offered are First trimester scan (11-14 weeks), Early Anomaly Scan – 16 weeks), Target Anomaly Scan (18 to 27 weeks), Target Anomaly scan with fetal ECHO, Growth scan with fetal Doppler objectives,

- Early detection of foeto- maternal problems and timely intervention
- To train Post Graduate Doctors and Consultants
- To establish a full- fledged state-of-the-art dedicated Maternal Foetal Medicine unit

5·4·5·2 Neurosonogram

Objective: To do neurosonogram as a routine to all children attending Newborn follow up clinic I.

Clinic set up: The clinic is held on all Tuesdays from 9.00 am to 1.00 pm except on public holidays.

Services offered

- Neurosonogram for babies attending newborn follow up clinic I
- Follow up scans in new borns with positive findings like PVE to detect progression to periventricular leucencies or cystic PVL.
- Neurosonogram in newborn with head circumference more as per Dyne's formula*
- Neurosonogram in 0–1-year-old infants with developmental delay with adequate A7 size

Figure 21 : Sradha clinic

*Category of patients – all new borns in Newborn follow up I and referred cases from DEC I, who are less than 1 year with wide A7 and increased head circumference

Table 5.3 : Sradha clinic cases 2020-23

Cases registered under Sradha Clinic				
Year	**2020**	**2021**	**2022**	**2023**
Total Cases registered	**562**	**940**	**1157**	**1358**

source: CDC data

6

Chapter 6
Treatment and Care

The Kerala government resolved to develop a comprehensive plan to address the problem of Congenital Heart Disease (CHD) in neonates and infants. Consultations were held with various stakeholders representing the public and private health sectors like UNICEF Kerala and Children's HeartLink (a US-based NGO). With input from the stakeholders, Children's Heart Link conducted an assessment and developed a continuum of care model[25] describing the lifetime path for such children, rather than viewing their care as a one-time surgical event. Some congenital disorders can be treated with medical or surgical interventions. Access to this care may vary by country and by different levels of a health system, though complex care is increasingly available in low- and middle-income settings.

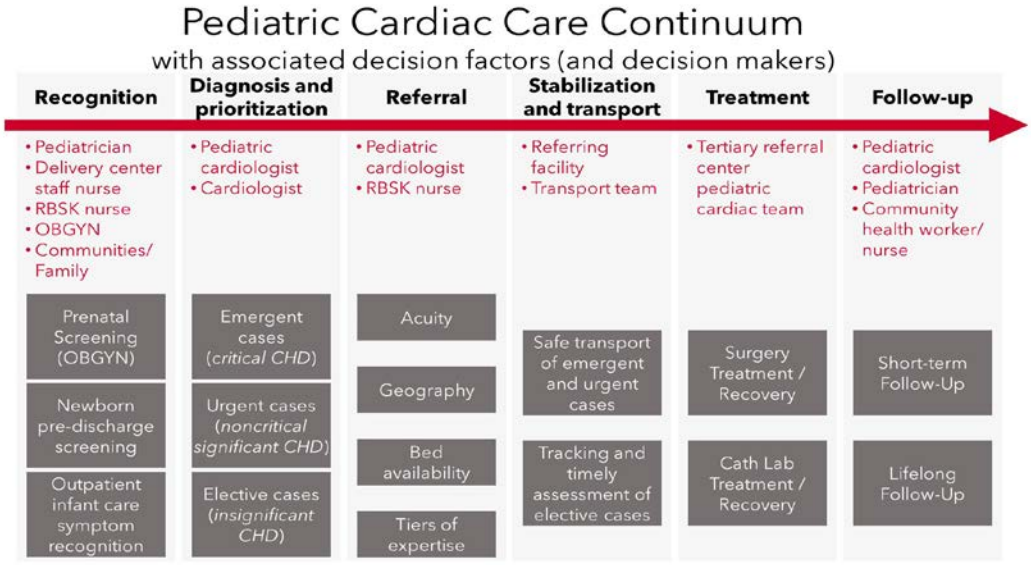


Figure 22 : CHD Patient Care Continuum

Surgery with good follow up care can often mitigate the potential lethality (as in the case of congenital heart defects) or the morbidity (as in clubfoot, cleft lip/palate) associated with structural congenital disorders. The contribution to reducing mortality and morbidity of this aspect of the treatment is often underestimated. Outcomes are improved with early detection at lower levels of the system through screening, referral and management (at specialist centres in case of some issues like cardiac defects)[2].

Medical treatment for certain metabolic, endocrine, and hematological conditions can improve quality of life. A clear example is congenital hypothyroidism, where early detection and treatment allow full physical and mental development to healthy adulthood, whereas a missed diagnosis or unavailability of a simple treatment carries a risk of serious intellectual disability[2].

Children with some types of congenital disorders may require long term support including physical therapy, speech therapy, occupational therapy and support from families and community.

6·1 Current Treatment Methods under Hridyam and Shalabham

Under the Hridyam and Shalabham care pathways, which were developed after a thorough assessment of the strengths and weaknesses within the health system, targeted implementation strategies were put in place. These strategies included engaging the private sector, as exemplified by Hridyam, with a primary focus on strengthening public sector systems. As these systems were expanded to address various health conditions, significant gaps in the existing health infrastructure were identified and addressed. For example, in the area of clubfoot management, additional centres were established to ensure effective service delivery. Similarly, new BERA (Brainstem Evoked Response Audiometry) units were introduced, with at least one unit in every district to facilitate hearing testing. The capacity for cochlear implants was expanded by equipping more centers, while Retinopathy of Prematurity (ROP) screening facilities were set up in all medical colleges along with treatment capabilities. Comprehensive Vision Impairment (CVI) clinics were established in all District Early Intervention Centers (DEICs).

A three-tier system for disability prevention was also implemented which includes:

1. DEICs (District Early Intervention Centers) at the base level, located in every district, which handle referrals from delivery points and screening centers for infants and toddlers. These are supported by mobile intervention units from the Kerala State Social Security Mission (KSSM).

2. REICs (Regional Early Intervention Centers) at the second level, jointly established by KSSM and the National Health Mission (NHM) in all medical colleges and centers of excellence such as CDC and ICCONS.

3. Centers of Excellence at the third level, which provide advanced specialized services and support.

These measures collectively aim to enhance the overall effectiveness of health service delivery and address critical gaps within the system.

6·2 Club Foot Management

As per the **Ponseti method** there are two phases of case management, Corrective phase and Maintenance phase. The tentative schedule of treatment is as follows. In the first phase, **the corrective phase**, is stretched across 4-6 weeks of serial casting (usually on a weekly basis) and then a tenotomy after 6 weeks and further three weeks of casting. Then it goes to the **Maintenance phase** where there will be bracing (first brace for three weeks and next two bracing for one month each) and advised to wear it for 23 hours a day. Then the brace can be used only during night and Nap time for five years with a quarterly visit to the clinic for follow up. Every activity in this scheme of treatment will be captured in the Shalabham portal through RBSK Nurses in the community and any defaulters will be prompted to follow the advised protocol by the concerned field level functionaries in Health Department.

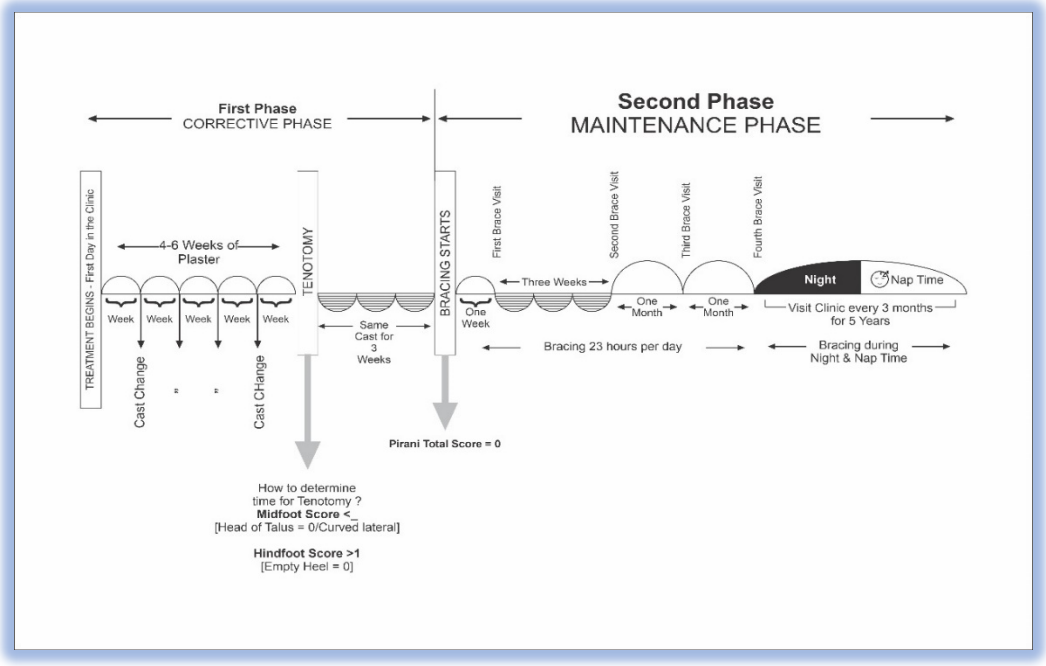


Figure 23 : Two phase Club foot case management

6·3 Significance of counselling the parents

Counselling plays a pivotal role in the care of children with congenital anomalies. Parents often experience shock, grief, guilt, and anxiety upon learning of their child's condition. Providing the right information helps to make an informed decision, find realistic ways to solve their problems and remove myths and misconceptions. Birth Defect is not due to any fault of the parents including their lifestyle, but due to natural, complex circumstances that were beyond the control of either of the parents. Reassure the parents and the family that the cause of their child's Birth Defect was not due to any fault on their part.

- Importance of early and timely intervention: Birth Defects, if not addressed early in life, can lead to developmental delays and disabilities causing permanent damage to the brain. 90% of the brain development happens during the first two years of life through the right kind of inputs from the sensory system such as vision, hearing, smell, touch, taste, movements of limbs etc. Damage to any of these sensory systems will affect the brain growth.
- Focus on parent's acceptance: along with the immediate priority of bonding, especially between mother and her child, feeding and warmth through proper clothing.
- Inform about existing intervention and available services. The possible interventions are:
    - Medical treatment
    - Surgical treatment
    - Neurodevelopmental therapy (NDT) and rehabilitation services
    - Genetic counselling and psychosocial support
- Importance of feeding and maintaining warmth during transport of infants.
- Prevention of Birth Defects in subsequent pregnancies.

Interventions to prevent complications

One of the major interventions in this area is Neurodevelopmental follow-up for children with CHD established through DEICs and is a model for LMICs

Chapter 7
Surveillance System and Follow-up Mechanism

A robust surveillance and follow-up system is essential to ensure that children identified with congenital anomalies through newborn screening programmes receive timely referral, treatment, rehabilitation, and ongoing support. The follow-up mechanism for the Hridyam and Shalabham programs was designed to enhance the comprehensive care provided to newborns.

Shalabham program is aimed to document Comprehensive Newborn Screening. The State Health Department, through the National Health Mission (NHM), sought to integrate this initiative with existing verticals. This included the establishment of management and follow-up protocols for various conditions, such as Congenital Heart Disease (CHD) (under the Hridyam initiative), Clubfoot, Cleft lip and palate, Inborn Errors of Metabolism, Hearing Impairments, Retinopathy of Prematurity (ROP), and Cerebral Visual Impairments (CVI).

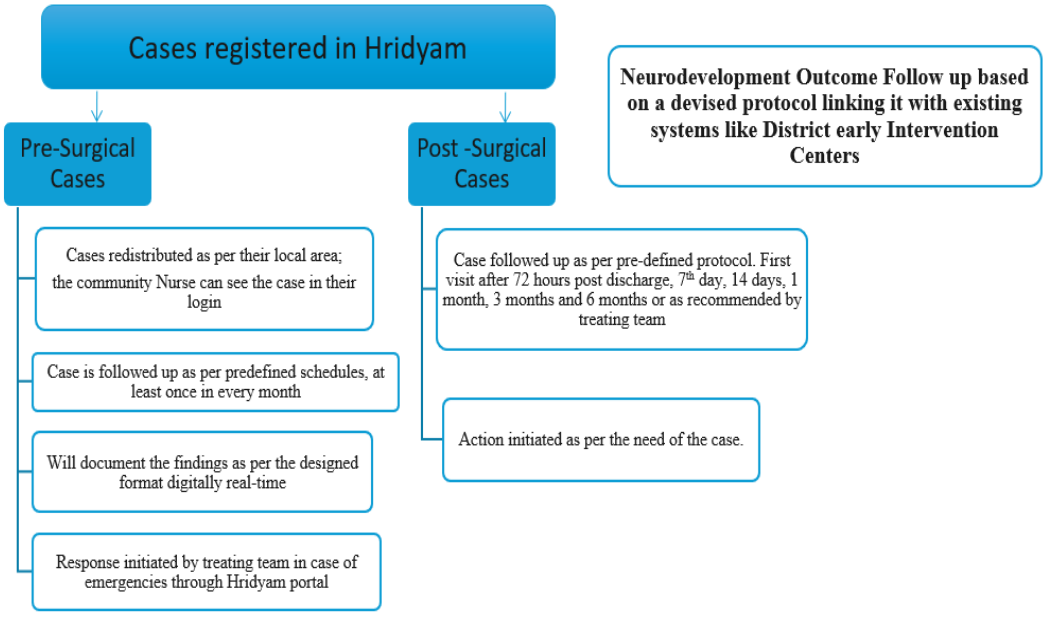


Figure 24 : Follow-up of Hridyam cases in community

Additionally, a general referral protocol was established for other conditions, directing the cases to the District Early Intervention Centers (DEIC) via the Shalabham portal and RBSK Nurses to facilitate and ensure effective follow-up care.

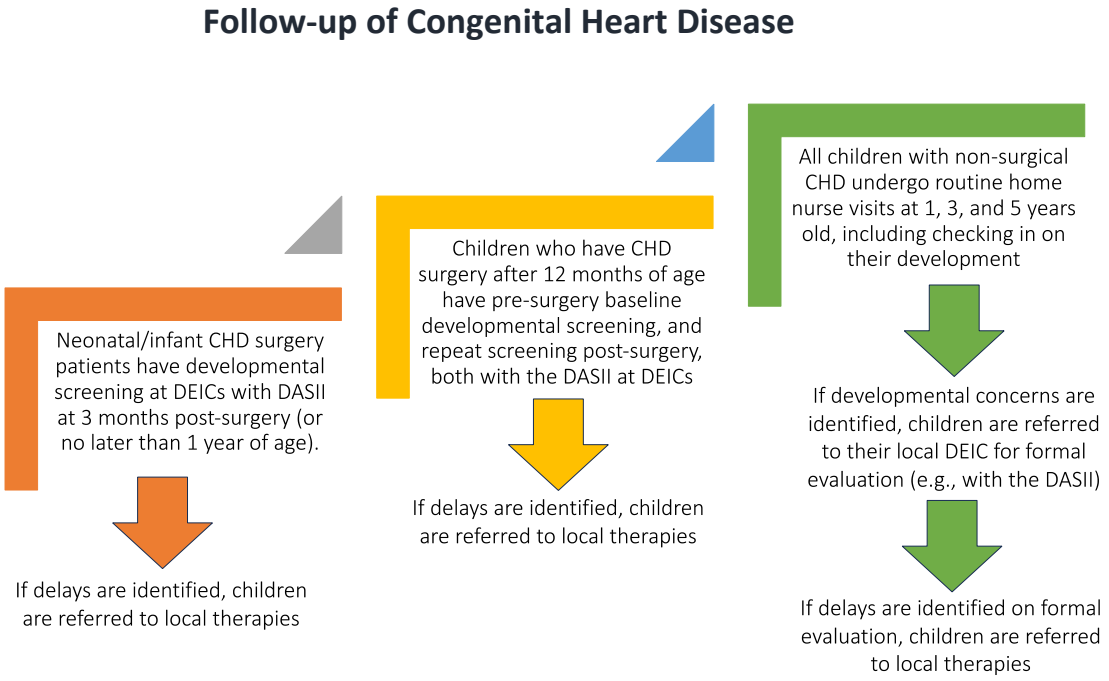


Figure 25 : Follow - up of CHD

8

Chapter 8
Prevention Strategies

8·1 Significance of Preconception and Antenatal Care in Reducing the Risk of Birth Defects

Preconception care refers to healthcare interventions and services provided to individuals or couples before pregnancy to optimize their health and promote favourable outcomes for both the mother and the baby**.** Extensive research has demonstrated a strong association between preconception care and improved neonatal and birth outcomes. By addressing modifiable risk factors and promoting optimal health before conception, preconception care can significantly reduce the incidence of various neonatal and birth disorders[20].

Key Components of Preconception Care[20]

- Health promotion and risk assessment
- Family planning and contraception
- Screening and managing pre-existing conditions (e.g., diabetes, hypertension, thyroid disorders)
- Genetic counseling and screening
- Immunizations and infectious disease prevention
- Psychosocial support and mental health considerations

8·2 The Importance of Preconception Care for Maternal and Fetal Health

- Improved Pregnancy Outcomes: Engaging in preconception care significantly reduces the risk of complications like preterm birth, low birth weight, and congenital disabilities. By addressing potential health risks before conception, individuals can enhance their health, leading to healthier pregnancies and more favorable birth outcomes.
- Enhanced Maternal Health: Preconception care helps identify and manage pre-existing conditions, such as diabetes, hypertension, and thyroid disorders, which can pose risks during pregnancy. Addressing these conditions before conception optimizes maternal health and minimizes complications during pregnancy.
- Management of Modifiable Risk Factors: Preconception care assesses lifestyle factors like smoking, excessive alcohol consumption, and poor nutrition, which can impact pregnancy outcomes. With proper guidance and support, individuals can make positive lifestyle changes, reducing the risks associated with these modifiable factors.

- Early Detection and Intervention: Preconception care includes screening and counselling to identify potential risks such as genetic disorders, infectious diseases, or mental health conditions. Early detection and intervention before pregnancy allow for timely management, helping to ensure healthier pregnancy outcomes.

Importance of preconception examination

A preconception examination, also known as a preconception visit, is one of the best ways to ensure a healthy pregnancy. The goals are to assess your overall health and identify any risk factors that can complicate a pregnancy. A preconception examination can include any of the following[6]:

Family medical history

A doctor will assess the medical history of couple's biological parents to see if any family member has had medical problems such as high blood pressure, diabetes, or mental retardation.

Genetic testing

A doctor will assess any possible genetic disorders that can be passed down to the baby. Some genetic disorders can be detected by blood tests before pregnancy.

Personal Medical History: to determine if mother has any medical conditions that may require special care during pregnancy (anaemia, epilepsy, diabetes, high blood pressure); to gather information about previous surgeries; and to obtain information about past pregnancies such as complications, losses, and length of gestation.

Vaccination status

To assess immunity to diseases such as rubella (German measles) that can cause miscarriage or birth defects. Vaccine can be given at least three months prior to conception to provide immunity.

Infection screening

Infection screening determines if a woman has any sexually transmitted infection, urinary tract infection, or any other infection that can be harmful to her or to the foetus. Immunizations and contagious disease prevention are vital in preconception care. Ensuring that individuals are current on recommended vaccinations, such as Rubella, Tetanus, and Hepatitis B, protects against infections that can pose risks during pregnancy. Prevention and management of infections, including sexually transmitted infections, is crucial for protecting maternal and fetal health[22]. Women who get certain infections during pregnancy are at a higher risk for having a child with birth defects. For example, Zika virus infection during pregnancy is linked with the congenital anomaly

called microcephaly, in which the brain and skull are atypically small. Zika infection in pregnancy is linked to other structural problems with the brain as well[7].

8·3 Significance of Micronutrient Deficiency among Mothers and Women of Reproductive Age

Micronutrient intake recommendations are determined by assessing the normal physiological requirements to support a healthy pregnancy. These requirements might be determined, in part, by a woman's diet and nutritional status before pregnancy, as well as the environmental stressors that can lead to inflammation of body tissues which might deplete, or divert the use of micronutrients. Congenital anomalies are largely preventable through improved nutrition in women of reproductive age, prenatal counselling, micronutrient supplementation like folic acid, iodine, zinc etc, and adequate antenatal care[23]**.** Micronutrients support maternal health and fetal development throughout gestation through processes that are integrated across maternal, placental and fetal compartments*(fig.16).*

Women in low-income countries often enter pregnancy malnourished, and the demands of gestation can exacerbate micronutrient deficiencies with health consequences to the fetus. Examples of efficacious single micronutrient interventions include folic acid to prevent neural tube defects, iodine to prevent cretinism, zinc to reduce preterm birth, and iron to reduce the risk of low birth weight. Folic acid and vitamin D might also increase birth weight. While extensive mechanism and association research links antenatal multiple micronutrients to plausible maternal-foetal health advantages, hypothesized benefits have often been absent, minimal, or unexpected in trials.

These findings suggest a role for population context in determining health responses and extensive gaps in knowledge. Multiple micronutrient supplements reduce risks of being born low birth weight, small for gestational age or stillborn in undernourished settings, and justify micronutrient interventions with antenatal care. Measurable health effects of gestational micronutrient exposure may persist into childhood, but few data exist on potential long-term benefits.

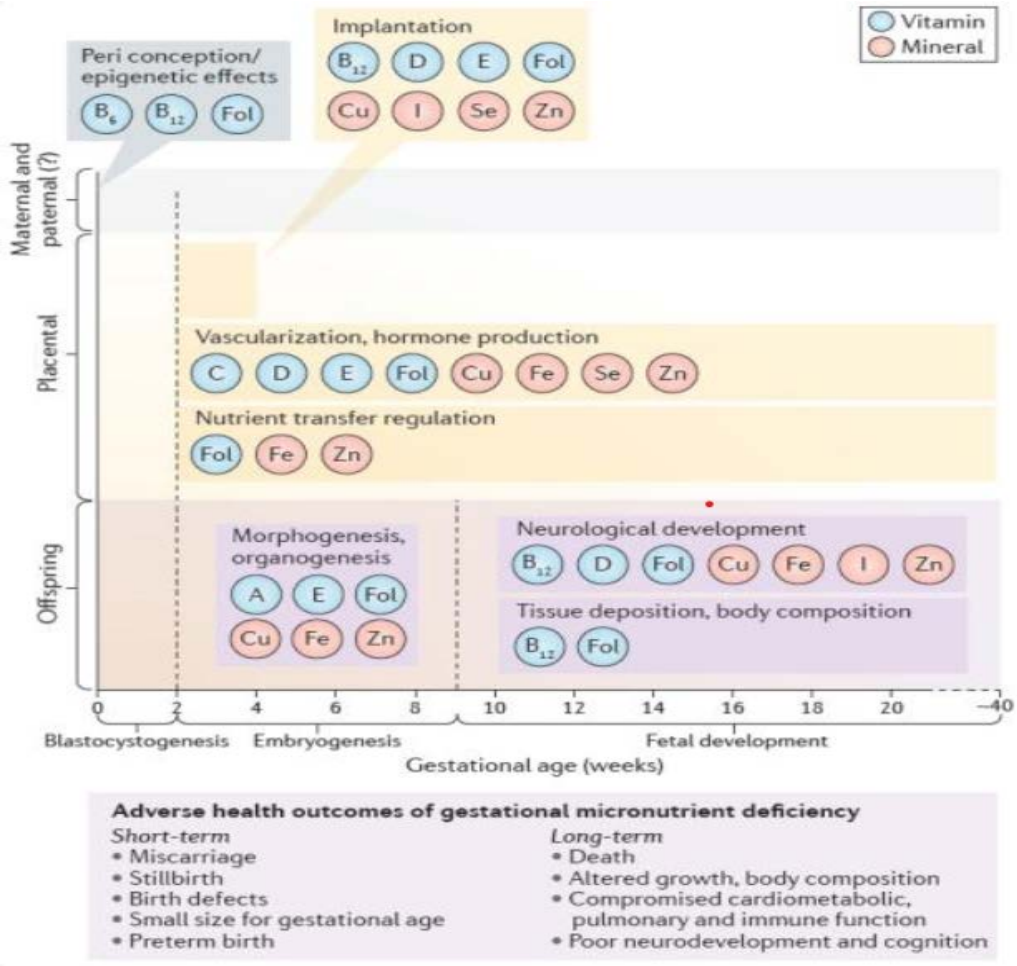


Figure 26 : Micronutrients support maternal health and fetal development throughout gestation

8·4 Importance of Awareness on Congenital Anomalies among Public

Congenital anomalies are under-prioritized in maternal and child health services of low- and middle-income countries (LMIC) (Darmstadt et al. 2016) where healthcare is not universal and assured to all. The focus of global agendas like the Sustainable Development Goal 3 is to prioritize activities to reduce preventable child mortality. Childhood disability remains under-recognized, and services are fragmented[24].

A study identifies that there was limited knowledge among public on congenital anomalies, their causation, and prevention. Common conditions that led to an anomaly were recalled only after probing, after adverse pregnancy outcomes like miscarriage, stillbirths, premature birth, or the birth of a child with a disability. Lay beliefs and misconceptions such as the association between maternal practices during an eclipse and congenital anomalies is well-documented in ethnocultural studies on congenital anomalies (Cohen et al. 1998)[24].

Childhood disability was viewed as a burden, imposed additional stress on mothers in child's care, maternal blaming, and financial burden on families. Stigmatizing attitudes towards children with disabilities were evident. Knowledge about medical care, rehabilitation, and early intervention services was minimal.

Despite availability of services, maternal experiences of guilt and blame, stigmatizing attitudes, and difficulty in negotiating access to welfare support were concerns.

Raising awareness about congenital anomalies is crucial for public health as these conditions, often present at birth, can lead to lifelong disabilities or even death if not detected and managed early. Educating the public about the causes, risk factors, and prevention strategies - such as proper prenatal care, nutrition, and avoiding harmful substances - can significantly reduce the incidence and severity of congenital anomalies. Awareness campaigns can also help to reduce the stigma, encourage early screening, and promote access to timely medical interventions, which will ultimately improve the quality of life of the affected individuals and their families.

Chapter 9
Current Challenges to Provide Care for Congenital Anomalies

9·1 Lack of Awareness and Knowledge among Healthcare Providers and Individuals

- A significant challenge in implementing effective care is the lack of awareness and knowledge among healthcare providers and individuals. The absence of accurate information about the benefits and services available further discourages individuals from seeking preconception care.
- Individuals often lack awareness of the importance of diagnosing congenital anomalies which can significantly impact their health and the health of future children
- Socioeconomic and Cultural Barriers
- Missed opportunities for timely interventions.

9·2 Challenges in Data Collection

- Inadequate Data Collection and Monitoring: Current data collection and monitoring systems for preconception care are often inadequate, making it difficult to assess the effectiveness of interventions and improved practices.
- Current estimates of congenital anomalies range from 4 to 12 cases per 1,000 births, but these figures likely underrepresent the true scope of the issue due to factors such as stigma and social exclusion (Bickler et al. 2010; Goksan et al. 2006; Wu and Poenaru 2013). Under reporting may be a problem even in Kerala.
- Incidence and prevalence data may be skewed by the survivability of certain anomalies. Children with non-life-threatening conditions are more likely to reach medical centers, which can lead to an underrepresentation of immediately life-threatening conditions in hospital-based data[5]

9·3 Mortality Rate Estimation Challenges

- The burden of disease associated with congenital anomalies is most often calculated as the mortality rate, not including the morbidity nor the cost of ongoing illness
- Autopsies are not performed in in all neonatal deaths. This might've provided additional information on the cause of death.
- Furthermore, proportion of children who do not reach a health facility and die at home might lead to hidden mortality.

9·4 Treatment Delays

- Mortality & morbidity from Congenital anomalies will go up from delay by health professionals to identify and treat anomalies and by pejorative cultural beliefs surrounding anomalies.
- Home deliveries and delay in reaching the medical facilities will delay the management.
- The misdiagnosis of anomalies as better-known infectious diseases, and added delays for invisible anomalies, may further hinder the provision of timely, appropriate services.

9·5 Recurrent Abortion and Infertility Evaluation

Intrauterine Fetal Death/ Still birth needs detailed evaluation for congenital causes to prevent recurrence in future pregnancies. The study detailed below show the importance of this observation:

- Frequency of stillbirth observed in the study was 4.35 ‰ (615 newborns) and MCA among stillbirths were about 20-fold higher at 190.24 ‰ (117 newborns) compared to 8.89 ‰ (1,253 newborns) among 140,925 live births ($P < .001$).[26] This stillbirth (6.72 ‰) and overall MCA (13.81 ‰) were highest among mothers in the age group of 30 years or more. Newborns with MCA among stillbirths were about 20-fold higher at 190.24 ‰ (117/615) compared to 8.89 ‰ (1,253/140,925) among live births ($P < .001$).[26]

10

Chapter 10
Data Analysis and Interpretation

This chapter would analyze quantitative and qualitative findings from the preceding sections - particularly Kerala's newborn screening, Hridyam, Shalabham, and Clubfoot programs - and connect them to trends, outcomes, and implications.

10·1 Overview

The available data on congenital anomalies from Kerala provides valuable insight into the magnitude, types, and outcomes of these conditions, as well as the effectiveness of the state's intervention programs. Analysis of the findings from newborn screening and programmatic data indicates measurable progress in early detection, intervention, and reduction of morbidity due to congenital disorders.

10·2 Prevalence and Pattern of Anomalies

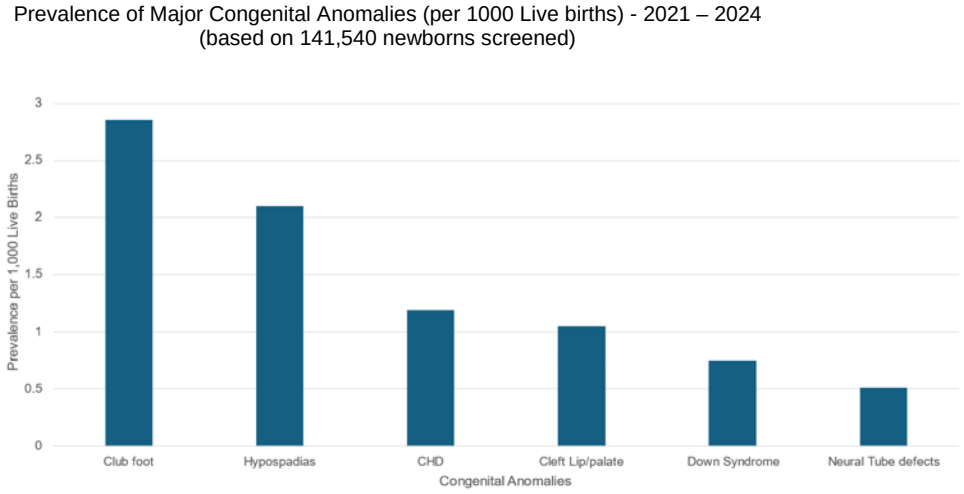


Figure 27 : Prevalence of Major Congenital Anomalies (per 1000 Live births) - 2021 - 2024

From the study conducted across 141,540 newborns in Kerala, the prevalence of major congenital anomalies was 9.68 per 1,000 live births, which is slightly higher than the national average but consistent with improved detection through universal screening.

- Clubfoot (0.29%) emerged as the most common anomaly, followed by hypospadias (0.21%), congenital heart disease (0.12%), and cleft lip/palate (0.11%).

- Genetic anomalies such as Down syndrome (0.07%) and neural tube defects (0.05%) accounted for a smaller but significant proportion.

This pattern indicates that structural deformities amenable to correction through early medical or surgical intervention dominate the congenital anomaly profile in Kerala.

10·3 Programmatic Data Trends

10·3·1 Hridyam: The Hridyam web-based registry enabled real-time tracking of children with congenital heart disease (CHD).

- More than 10,000 CHD cases have been registered since inception, with a notable increase in survival and follow-up compliance due to systematic referral and surgery allocation protocols.
- Integration with DEIC and empaneled hospitals improved case management timelines and reduced dropouts.

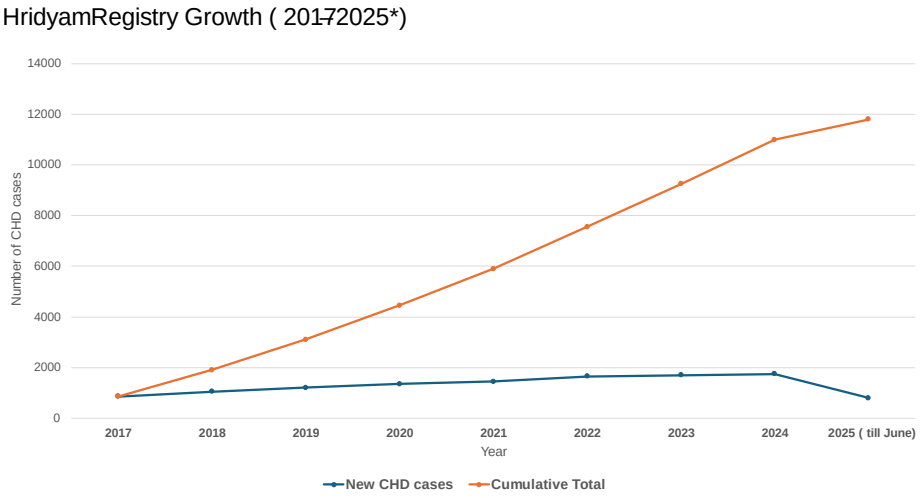


Figure 28 : Hridyam Registry (2017 – 2025*)

10·3·2 Shalabham (Comprehensive Newborn Screening):

- Introduced across all government delivery points, the program achieved screening coverage above 95% for visible and functional birth defects within 48 hours of delivery.
- Functional screening for CHD (Pulse Oximetry), hearing (OAE), and vision (ROP) resulted in early referral to specialized units, improving long-term outcomes.

- Digital innovations such as the Jatak Seva mobile app and integration with VBD-UIDs have enhanced traceability and follow-up.

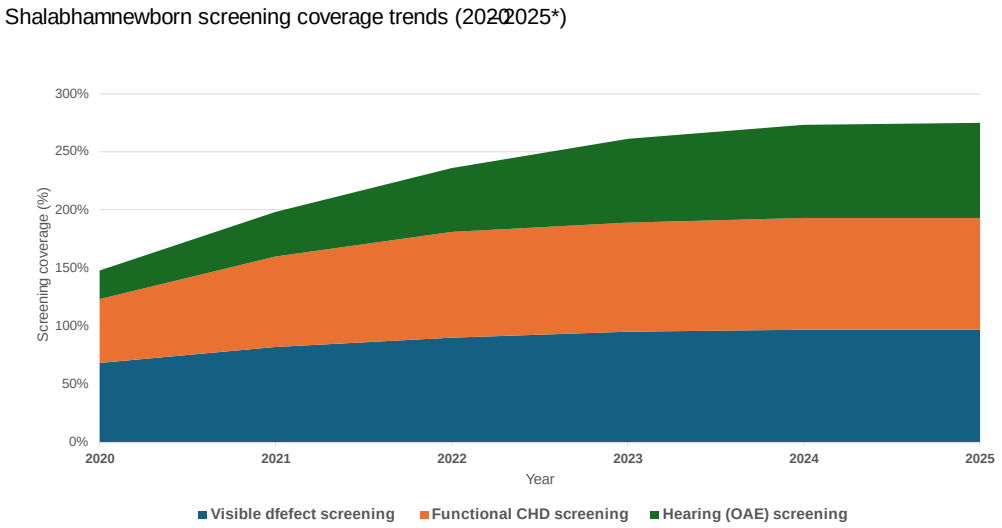


Figure 29 : Shalabham newborn screening coverage (2020 – 2025*)

10·3·3 Clubfoot Clinics:

- By 2024 Thirty six dedicated clinics were established across districts, ensuring decentralized access to Ponseti-based correction.
- Early initiation of casting within the neonatal period correlated with a 90% success rate in complete correction without surgical intervention.
- Continuous monitoring via Shalabham data confirms improved adherence and parental satisfaction.

Clubfoot clinics performance (2019 – 2024*)

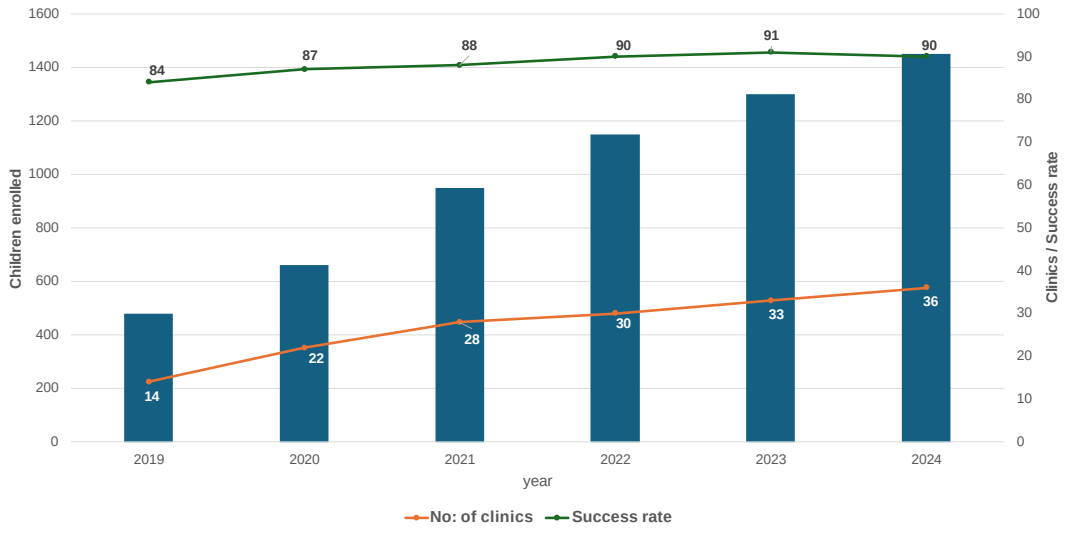


Figure 30 : Clubfoot clinics performance (2019 – 2025*)

10·4 Analysis of Surveillance and Follow-up Systems

Data from DEICs and RBSK field-level reports show that:

- Follow-up compliance rates exceeded 80% in Hridyam and Clubfoot programs when RBSK nurses were actively engaged.
- Regular DEIC-level case reviews helped identify high-risk infants and ensure timely intervention.
- Challenges remain in integrating data across vertical programs (e.g., Hridyam, Shalabham, and RBSK) for unified state-level analytics.

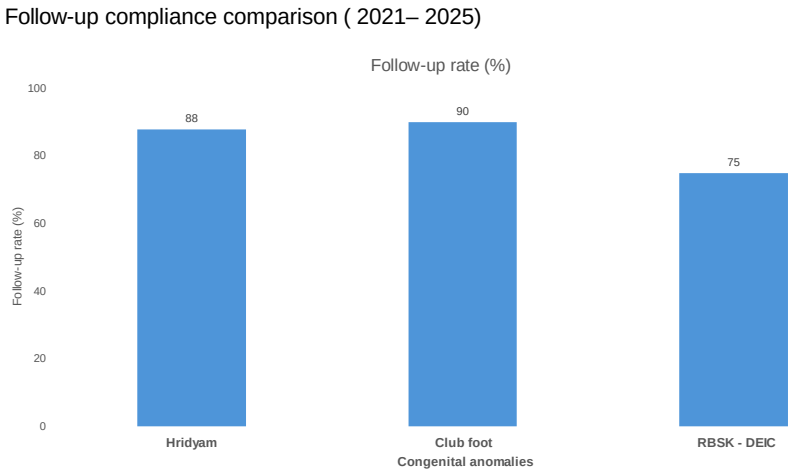


Figure 31: Follow-up compliance comparison (2021 – 2025)

10·5 Outcomes and Impact

- Kerala's Infant Mortality Rate (IMR) dropped from 12 (2015) to 5 (2024), for which the improved management of congenital anomalies also played its role.

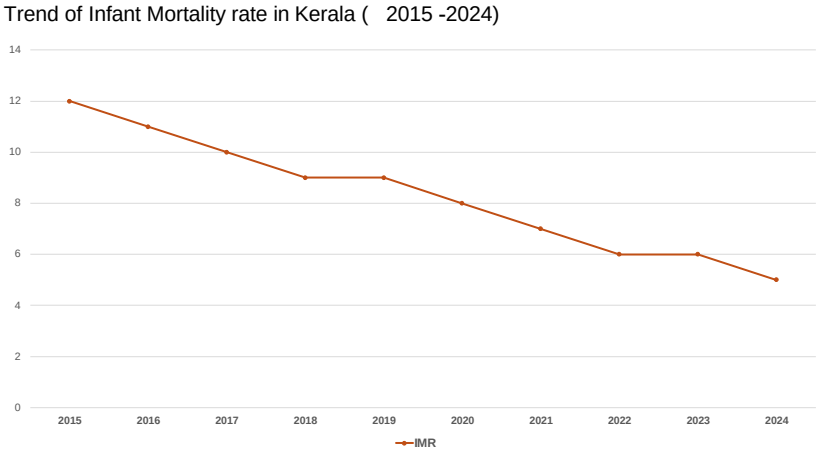


Figure 32: Trend of Infant Mortality rate in Kerala (2015 – 2024)

- The establishment of antenatal (Sradha), neonatal (Shalabham), and postnatal (Hridyam/DEIC) care continuum represents a replicable model for other states.
- Increased awareness and accessibility have reduced stigma and improved community-level acceptance of corrective procedures.

10·6 Key Insights from Data

Table 10.1: Trend of Infant Mortality rate in Kerala (2015 – 2024)

Indicator	Observed Trend	Interpretation
Detection rate of anomalies	Increasing	Reflects success of universal screening and improved reporting
Surgical correction success rate (CHD, clubfoot)	85–90%	Demonstrates effective referral and timely intervention
Parental counselling and awareness	Improving but variable	Indicates need for standardized IEC and psychosocial support
Follow-up adherence	>80%	Shows strength of RBSK and DEIC-based mechanisms
Research and data linkage	Limited	Highlights need for integrated birth defects registry

10·7 Limitations

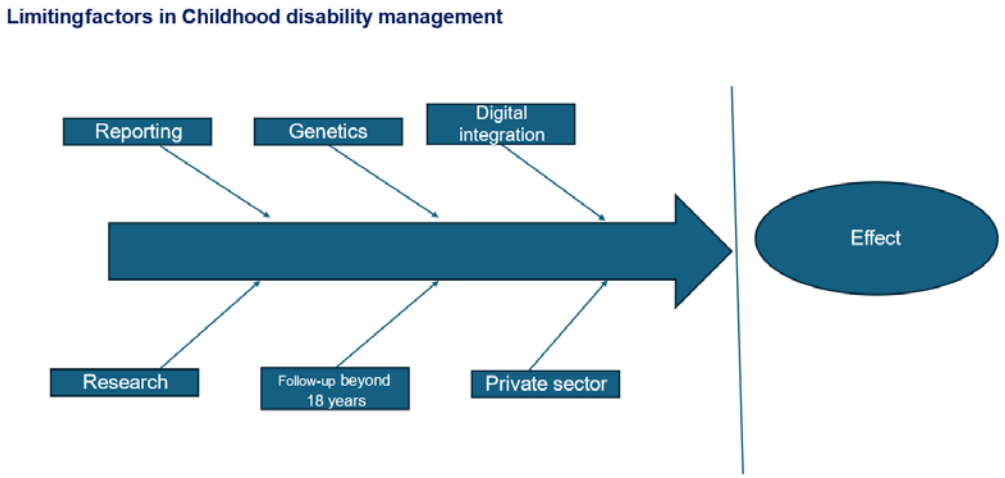


Figure 33: Limiting factors in Childhood disability management

- Incomplete private sector reporting,
- Limited genetic testing capacity. Need enhanced genetic testing capacity to identify idiopathic or multifactorial cases.
- Inadequate long-term follow-up for children transitioning into adulthood restricts evaluation of neurodevelopmental outcomes.
- Weak linkages across vertical programmes and lack of digital integration of all child-health platforms

11

Chapter 11
Road Ahead:
Congenital Anomaly Care in Kerala

Kerala has made exceptional progress in maternal and child health indicators, attaining an Infant Mortality Rate (IMR) of 5 (SRS 2023), one of the lowest in the country. However, congenital anomalies continue to pose a significant public health challenge—contributing to neonatal and infant mortality, lifelong disability, and emotional and financial strain for families. As the state transitions from ensuring survival to improving the quality of survival, a comprehensive, evidence-based, and integrated approach to the prevention and management of congenital anomalies becomes imperative.

Building on Kerala's success in maternal and child health, the next decade's focus must shift from survival to lifelong wellness. The vision is to establish Kerala as the first Indian state with a fully integrated congenital anomaly prevention and management system—from preconception to rehabilitation—through digital health, community participation, and interdepartmental convergence.

11.1 Strengthening Prevention and Preconception Care

- Preconception and Antenatal Services: Scale up folic acid and iron supplementation, rubella immunization, and routine screening for maternal infections such as TORCH and nutritional deficiencies. Ensure targeted interventions among high-risk populations such as tribal, migrant, and urban slum communities.
- Genetic Counselling and Risk Reduction: Establish prenatal diagnostic and genetic counselling units in all medical colleges and selected district hospitals to identify high-risk pregnancies early.
- IEC and Behaviour Change Communication: Intensify awareness on maternal nutrition, consanguinity risks, prevention of diabetes and hypertension, and avoidance of alcohol, tobacco, and teratogenic substances. Promote early antenatal registration and healthy lifestyle practices among women of reproductive age.

11.2 Early Detection and Newborn Screening

- Standardized Antenatal Screening: Ensure availability of high-quality antenatal ultrasonography and biochemical screening for foetal anomalies at all secondary and tertiary care facilities in all the districts.

- Strengthening the Screening Continuum: Enhance the functioning of ‘Shalabham’, ‘Hridyam’, RBSK, screening teams for early identification of congenital anomalies and link them with District Early Intervention Centres (DEICs). Expand the number of ‘Clubfoot’ clinics across the state.
- Newborn Screening Expansion: Expand *Shalabham* newborn screening to include metabolic and endocrine disorders, leveraging medical colleges, district hospitals, and specialized laboratories.
- Long term Follow-up: Develop standardized long term follow up protocols linking primary care facilities, DEICs, and tertiary care institutions for comprehensive follow up, for ongoing rehabilitation, outcome monitoring, and family counselling.
- Digital Integration: Develop unified dashboards linking *Shalabham*, *Hridyam*, DEIC, and Civil Registration data for end-to-end tracking from birth to outcome.
- Private-Sector Inclusion: Mandate reporting from private hospitals conducting anomaly scans and newborn screening to improve registry completeness.

11.3 Comprehensive Care and Rehabilitation

- Tertiary care Centres: Strengthen existing tertiary care hospital network and empanel private sector hospitals, to provide diagnosis, surgical correction, and rehabilitation of congenital anomalies.
- Integrated Referral Networks: Establish a hub-and-spoke model connecting private hospitals, Govt. hospitals, DEICs, SNCUs, and Labs with telemedicine-enabled specialist consultations.
- Mental-health integration: Integrate psychological support and caregiver mental-health counselling through district mental-health programs.

11.4 Capacity Building and Workforce Development

- Training and Skill enhancement of Health Workforce: Conduct periodic capacity-building programs for obstetricians, paediatricians, medical officers, ASHAs, RBSK teams, DEIC staffs on prevention, screening, and management of congenital anomalies.

- Integration into Existing Programs: Incorporate congenital anomaly prevention and management modules into the NHM and KSSM e-learning systems and CMEs.

11.5 Surveillance, Research, and Data Systems

- Kerala Birth Defects Registry: Expand the population-based state registry under the Directorate of Health Services, integrating data from DME and private sectors.
- Data Integration: Strengthen data reporting and triangulation through DEIC, RBSK, ‘Shalabham’, ‘Hridyam’ portals to capture anomaly-specific indicators.
- Research and Evidence Generation: Promote research through medical colleges on genetic, environmental, and nutritional determinants of congenital anomalies in Kerala.

11.6 Community Engagement and Family Support

- Family Counselling: Institutionalize pre- and post-natal counselling services for families, especially for high-risk pregnancies and identified cases.
- Parent Support Networks: Encourage formation of parental support groups for congenital disorders such as cleft lip/palate, CHDs, and Down syndrome.
- Inclusion and Social Support: Strengthen linkages with social welfare and disability schemes to ensure inclusion and financial protection for affected children and families.

11.7 Governance, Financing, and Partnerships

- Dedicated Resource Allocation: Earmark more funds under the State PIP for prevention, screening, and management initiatives.
- Public–Private Partnerships: Leverage collaborations with private diagnostic centres, NGOs, and professional bodies for specialized care, outreach, and awareness.
- Interdepartmental Coordination: Strengthen convergence among Health, Social Justice, Women and Child Development, and Education Departments for holistic child development.

Kerala's strong public health infrastructure and human resource base provide an ideal foundation to advance the agenda of congenital anomaly prevention and care. However, progress is contingent upon addressing persistent challenges. Key limitations include incomplete private sector reporting, weak linkages across vertical programmes, limited genetic testing capacity, and inadequate long-term follow-up for children transitioning into adulthood. Establishing a comprehensive Birth Defects Registry, enhancing genetic services, and integrating digital platforms will be vital to overcoming these gaps. Expanding research on environmental, nutritional, and genetic determinants will further refine the state's approach.

The way forward demands a strong and coordinated call to action. By strengthening the interdepartmental convergence, full digital integration of all child-health platforms, systematic inclusion of the private hospitals, sustained capacity building, and investment in family counselling and community support systems, the state can further reduce neonatal mortality and ensure that every child not only survives—but thrives.

12

Chapter 12
Conclusion

The journey through the intricate landscape of congenital anomalies has highlighted the profound medical, social, and policy challenges these conditions pose, not only for the affected individuals and their families but also for the broader healthcare systems and society. Congenital anomalies remain a silent but significant contributor to neonatal and under-five mortality, as well as to long-term disability and socioeconomic burden. In understanding the global, regional, and particularly the Kerala-specific contexts, it becomes evident that a multifaceted approach is essential to addressing the burden of congenital anomalies effectively.

Kerala, with its robust healthcare infrastructure and pioneering public health initiatives such as the "Hridyam" and "Shalabham" programs, has demonstrated that early detection, timely intervention, comprehensive care and structured follow up can significantly mitigate the impact of congenital anomalies. The state's efforts in integrating new-born screening into its broader public health strategies underscore the importance of early and accurate diagnosis, which is crucial for improving outcomes and enhancing the quality of life for affected children. However, the battle against congenital anomalies is far from over. Continued investment in research, healthcare infrastructure, and public health education is imperative. Moreover, fostering collaboration between healthcare professionals, policymakers, and communities is vital to sustaining and expanding the progress made thus far.

This document has sought to provide a comprehensive understanding of congenital anomalies, their causes, risk factors and the preventive measures that can be implemented at both the community and healthcare system levels. As we move forward, it is essential to maintain the momentum gained through these efforts, ensuring that every child has the opportunity to live a healthy and fulfilling life, free from the preventable burdens of congenital anomalies. The insights gathered here not only inform current practices but also serve as a foundation for future innovations in public health. By continuing to prioritize the health and well-being of new-borns, we can contribute to a healthier, more equitable future for all.

ANNEXURE: 1

Table 1: List of different regions of India showing the occurrence of congenital anomalies[13]

S no	Study area	No of birth during study time	Number of anomaly affected birth	Prevalence	Reference
1	Aurangabad, Maharashtra	13414	114	0.84%	Agarwal et al., 2019
2	North India	14530	76	0.52	Tiwari et al., 2020
3	Pondicherry	6134	140	2.28%	Rathod and Samal 2020
4	Haryana, India	920	12	1.2	Malik et al., 2019
5	Kozhikode, Kerala	1,08,024	911	0.84%	Jayasree & D'Couth (2018).
6	Hyderabad, Telangana	4120	123	2.93%	Tirumani and Khatija 2017
7	Bangalore, Karnataka	2,137	86	4%	Doddabasappa *et al.*, 2017
8	Trivandrum, Kerala	15227	379	2.48%	Vinodh & Balakrishna 2017
9	western Maharashtra	892	24	2.69%	Jain *et al.*, 2016
10	Vellore, Tamil Nadu	36,074	449	1.24%	Cherian *et al.*, 2016
11	Dharpur, Gujarat	2760	34	1.23%	Thaddanee *et al.*, 2016
12	Haryana	6143	109	1.7%	Marwah *et al.*,2016
13	Surat, Gujarat	5518	68	1.23%	Gandhi *et al.*,2016
14	Aligarh, UttarPradesh	5315	139	2.5%	Dewangan*et al.*, 2016
15	Ghanpur, Hyderabad	4628	189	4.08%	Pabbati *et al.*,2016
16	Pune, Maharashtra	4450	59	1.32%	Kumbhar and Leela 2016
17	Punjab	1554	75	4.82%	Marwah *et al.*, 2014
18	North India	20432	799	3.91%	Bhat *et al.*, 2016
19	Kashmir	1146	17	1.48%	Qurieshi *et al.*,2016

20	Bankura, West Bengal	14079	328	2.3%	Pal *et al.*, 2015
21	Bhopal, MP	3616	45	1.24%	Kanhere *et al.*, 2015
22	Pune, Maharashtra	2324	56	2.40%	Deshpande *et al.*,2015
23	Bankura, west Bengal	14079	328	2.3%	Murmu *et al.*, 2015
24	Ludhiana, Punjab	10,674	217	2.03	Ghorpade *et al.*,2015
25	Ahmedabad, Gujarat	4456	106	2.37%	Shah and Pensi 2013
26	Kolkata	12896	286	2.21%	Sarkar, *et al.*, 2013
27	Central Maharashtra	9386	179	1.91%	Taksande *et al.*,2010
28	Kolar, Karnataka	------	60	2.17%.	Bilodi & Gangadhar 2008
29	Nagpur	6076	84	1.38%	Bhalerao and Garg 2006
30	Maharashtra	2968	48	1.61%	Datta and Chaturvedi 2000

ANNEXURE:2

Table 2: List of some birth defects related studies conducted in India[8]

Study location	No. Of Malformed Babies	Risk Factors	Most Predominant Anomalies
Congenitalmalformations at birth in Central India: A rural medical college hospital based data. (Maharashtra January 2005 and 31 July 2007)	179	• Prematurity • Increased maternal age • Increasing birth order • Low birth weight	• Cardiovascular malformations • Musculoskeletal • Genitourinary anomalies.
A community-based survey [14] of visible congenital anomalies in rural Tamil Nadu (Rural Areas of Tamil Nadu (2004-2005)	166,833	• Family history • Consanguinity • Medication(anti-convulsants)	• Cleft palate
Birth defects surveillance[15] study. (Genetic Research Centre, National Institute for Research in Reproductive Health, Parel, Mumbai, India)	1694	• Low birth weight • Mothers having anaemia • Genetic ,Consanguinity • Maternal age	•Central nervous system anomalies •Polygenic malformation •Genetic disorders • Chromosomal abnormalities
Chromosomal abnormalities: genetic disease burden in India[16] (Guru Nanak Dev University, Amritsar, India, March 1991 - March 2005)	1950	• Consanguineous marriage • Maternal age greater than 35 years	• Down syndrome • Microcephaly • Mental retardation • Ambiguous genitalia/
Congenital Malformations at Birth - A Prospective Study From South[17] India. (Department of Pediatrics, Jawaharlal l~fftute of Post-Graduate Medical Education and Research, Pondicherry (September 1989 to December 1992)	469	• Consanguineous marriage • Low birth weight	• Musculoskeletal malformations • Cutaneous malformations • Genitourinary malformations • Central nervoussystem defects • Gastrointestinal defects
Pattern of distribution of congenital anomalies in stillborn[18]: a hospital based prospective study. (Gandhi Medical College, Hyderabad(July 2007 to December 2009)	28	• Consanguity • Previous history of abortion • Previous child with congenital anomalies • Low socio economic Group ,Maternal age	• Anencephaly • Astomia • Agnathia • Polycystic Kidney • Dextrocardia.
The incidence of major[19] congenital malformations in mysore(1967 through 1969)	46	• Maternal age	• Cleft palate • Anencephaly • Spinabifida
Congenital Malformations at Birth (Department of Obstetrics and Gynecology, Banaras Hindu University, Varanasi ,January 1988 to December 1989)[20]	48	• Multi Gravida • Low birth weight	• Anencephaly • Hydrocephalus • Talipes • Hypospadias

SOURCE OF DATA /INFORMATION ON BIRTH DEFECTS IN INDIA[27]

ANNEXURE: 3 – Head to Toe examination of Newborn for Visible Health defects

ANNEXURE:4

RBSK: SCREENING CUM REPORTING FORM FOR ALL BIRTH DEFECTS

FRONT SIDE

1. Location of reporting	State	District	Block
2. Source of identification (please tick √) 01) Sub Centre 02) PHC 03) CHC/BPHC 04) Rural Hospital 05) Sub divisional Hospital 06) District Hospital 07) Medical College Hospital 08) Other Govt. Hospital 09) Tertiary centre 10) Any other Details of DP:			
3. Reporting Month: …/….		**4. Reporting Year:** …/…./…./….	
5. Date of Identification (DD/MM/YY)	**6. Time of Identification** O at birth O < 1 month O 1-12 months O 1-6 yrs. O above 6 years O Prenatal diagnosis O Spontaneous Abortion O Autopsy		
Delivery outcome details			
7. Date of Birth (DD/MM/YY)	8. Outcome of delivery: O Live Birth O IUD/Still birth, <20 wks.		
9. No. of Babies: O Single O Twins O Multiple	10. Birth weight in gms		
11. Sex O Male O Female O Ambiguous O Intersex	12. Gestational age (completed weeks):	13. Last menstrual period (DD/MM/YY)	
14. Birth asphyxia O Yes O No	15. Autopsy shows birth defect (if applicable): O Yes O No		
16. Status of induction/augmentation: O None O Oxytocin O Misoprostol			
17. Place of birth O Home O Institution	State / Dist	Block	Municipality
Identification details: MCTS/Unique ID/Aadhar (any one)			
18. a) MCTS: b) Unique ID* (computer generated): c) Mother's Aadhar no. (if available): d) Mobile no. of Mother:			
19. Child's name	20. Mother's name	21. Mother's age (in completed years)	
22. Father's age	23. Caste: O SC O ST O OBC O Others		
24. Permanent address: House No. Street name Area Post office District State			
Antenatal details (If available)			

25. Folic acid details (peri-conceptional)	O Yes O No	26. H/O serious maternal illness	O Yes O No	27. H/O radiation exposure (x-ray):	O Yes O No	28. H/O substance abuse	O Yes O No
29. Parental consanguinity Specify degree if known	O Yes O No	30. Assisted conception IVF/ART	O Yes O No	31. Immunisation history (rubella)	O Yes O No	32. Maternal drugs	O Yes O No
		If yes, describe				details, if available	
33. History of anomalies in previous pregnancies	O Yes O No	34. No. of previous abortion:		35. No. of previous still birth:			
If yes, describe							

Neonatal details			
36. Head circumference:	37. Birth defects: O Single O Multiple		
38. Visible Birth Defects (Nervous system, Eye, Ear, face & neck, CHD, Respiratory, cleft lip & palage, digestive, genital, urinary, musculo skeletal, chromosomal, syndromes, skin, limbs)			

S.N	Type & Site	Description of the anomaly	Age at diagnosis	Code- ICD10	Confirmed or suspected
a.					
b.					

SECTION B (to be filled in by Medical Officer/Paediatrician)
39. Birth defects identified through instruments (Functional) (please tick √)
O a. Congenital deafness O b. Congenital vision defects O c. ROP O d. CHD
40. Birth defects identified through blood test (Metabolic) (please tick √)
A. IEM: O 1. CH O 2. CAH O 3. G6PD O 4. Others
B. Haemoglobinopathy: O 1. Thalassemia O 2. Sickle Cell Disease 03. Others

41. Details of Congenital birth defects detected through instruments or blood test (functional and metabolic)

S.N	Name	Description of the anomaly	Age at diagnosis	Code- ICD10	Confirmed or suspected
a.	*Congenital deafness*				
b.	*Congenital vision defect*				
c.	*ROP*				
d.	*Congenital Heart Disease*				
e.	*IEM*				
f.	*Haemoglobinopathy*				

42. Photographs/reports attached: O Yes O No

43. Investigation details

S.N	Relevant test	Result	Findings (if abnormal)
a.	Karotype/infantogram/ECG/US abdomen/Brain MRI/Any other	O N O AbN O Pending	
b.	Blood test, for O CH O CAH O G6PD O SCD O Others	O N O AbN O Pending	
c.	BERA, Fundoscopy etc.	O N O AbN O Pending	

44. Diagnosis

Anomaly	Syndrome	Provisional diagnosis	Complete diagnosis
Notifying person	Designation and contact details:	Facility referred	Provisional diagnosis status O Confirmed O Suspected

ANNEXURE:5

ANNEXURE - V
COUNSELLING PARENTS ON BIRTH DEFECTS

Who is at Risk for Birth Defects?

All pregnant women have some risk of delivering a child who suffers from a birth defect. Risk increases under any of the following conditions:

- Family history of birth defects or other genetic disorders.
- Drug use, alcohol consumption, or smoking during pregnancy.
- Advanced maternal age of 35 years or older.
- Inadequate prenatal care.
- Untreated infections or viruses, including sexually transmitted infections.

Preventing Birth Defects

Ensure that the pregnant woman:

- Does regular Antenatal checkup at least three times during pregnancy.
- Stays happy and stress free and rests at regular intervals.
- Folic acid intake should start soon after the marriage and continue during pregnancy. Also eat folic acid rich food like green leafy vegetables, liver and pulses.
- Goes for at least 4 antenatal check-ups.
- Do not smoke or drink alcohol.
- Minimize unnecessary medicines except those prescribed by ANM or medical officer.
- Maintain good hygiene by adopting safe sex practices and personal hygiene to prevent infection during pregnancy.
- Marriage among close relatives must be avoided, especially in families with a history of Birth Defects.
- Family members to maintain positive environment at home by avoiding any maternal stress and domestic violence.
- Immunization against rubella at least one month prior to pregnancy or during adolescence
- Always use iodized salt.
- Avoid taking any medications without consulting the doctor.
- Screening for diabetes during antenatal period and keep diabetes under control.
- Prevent infections by washing hands; cooking meat until it is well done, staying away from people who have infections; avoid cats during pregnancy.

- Always have safe and protected sexual activities.
- Be active and maintain a healthy weight.
- Avoid obesity and treat obesity before getting pregnant.
- Avoid exposure to hazardous environmental substance, heavy metals, pesticides and radiation exposures including X-rays.
- Consulting doctor for any family history of Birth Defects or genetic disease.
- Test for blood group and Rh compatibility.
- Timely referral of high risk cases of pregnancies including screening for advanced maternal age.

Neural Tube Defects

Neural tube defect:

Key messages to the parents:

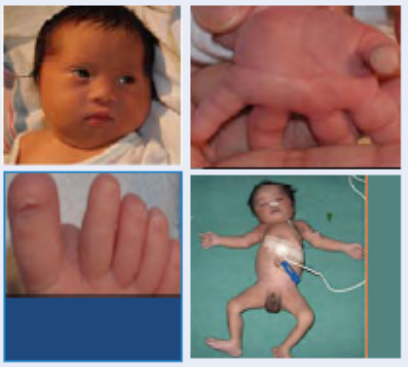
All pregnant women have some risk of delivering a child who suffers from a birth defect. Risk increases under any of the following conditions:

- Advise pregnant women to eat foods rich in folic acid like green leafy vegetables, pulses to prevent neural tube defects.
- Strictly avoid smoking/alcohol at any time and in any amount during pregnancy.

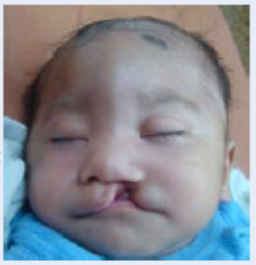
Down Syndrome

Key messages to the parents:

- With early intervention i.e., within the first three months, children with Down syndrome can lead a near independent life.
- Examination of eye, teeth, hearing and thyroid of the child should be done regularly.
- Special care of the child's neck and spine (avoid sudden movement to the neck)
- The child's speech can be improved if the parents are supported by a speech therapist.



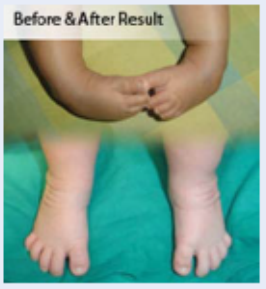
Cleft lift and palate



Key messages to the parents

- Cleft lift and palate can be corrected by performing timely surgeries.
- By 2-3 months of birth, surgery is performed to close the cleft lip; cleft lip could be corrected before the child speaks.
- Cleft palate surgery should be performed between 12 to 18 months of age.

Club Foot



Key messages to the parents

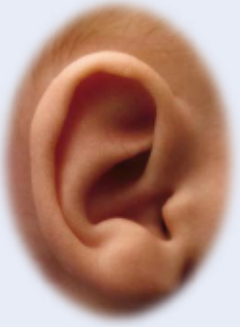
- Early detection and correction is important for child's development.
- If not treated at an early stage, this deformity can lead to lifetime disability.
- The mother should not try to correct the foot by pushing the sole towards the floor.
- Never pronate (rotate) and never put pressure on the heel
- Never use force while massage.
- Treatment to start within 7-10 days; requires multiple plaster 4-6 and changing weekly.

Congenital Cataract

Key messages to the parents

- This needs immediate referral, otherwise the child would become permanently blind
- If not corrected, the blindness would also affect the learning ability of the child

Congenital Deafness



Key messages to the parents

- Take the child for a hearing test at the District Hospital.
- Early management of hearing loss is important as it affects speech development.
- Reduce exposure to loud noise.
- Hearing devices, such as hearing aids and speech therapy can help in hearing.
- Every child has the ability to speak. Hence if a child's deafness could be confirmed by the age of 9 months and hearing aids could be provided before 2 years, the child could be prevented from becoming dumb.

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