

Newborn Screening Methods in India: A Review of Policies, Rare Diseases, and Emerging Technologies

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Abstract

Newborn screening (NBS) is a critical public health intervention enabling early detection of treatable congenital disorders before clinical symptoms manifest. India, with approximately 26 million annual births, has been slower than developed nations in implementing universal NBS. This review examines the current state of newborn screening in India, including the burden of rare genetic diseases, national policies (National Policy for Rare Diseases 2021, RBSK 2.0), state-led initiatives (Delhi's ANMOL scheme, Kerala's Shalabham program), and technological methodologies (dried blood spot collection, tandem mass spectrometry, ELISA, TRFIA, and genomic sequencing). Epidemiological data on congenital hypothyroidism, congenital adrenal hyperplasia, G6PD deficiency, and inborn errors of metabolism are synthesized. Challenges related to mandatory screening, cost, infrastructure, and awareness are discussed alongside future directions for comprehensive NBS implementation in India.

Keywords: Newborn screening, rare diseases, congenital hypothyroidism, tandem mass spectrometry, India, RBSK, ANMOL, UMMID

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I. Introduction

Newborn screening (NBS) involves checking apparently healthy newborns for serious but treatable conditions, enabling intervention before symptoms or permanent damage occur [1]. In the United States, NBS programs screen approximately 4 million infants annually, detecting treatable disorders in about 1 in 300 newborns [1]. However, in India—with the world's highest annual birth rate (approximately 26 million live births)—newborn screening remains largely non-mandatory and non-standardized across healthcare centers [2,3].

The burden of genetic and congenital disorders in India is substantial. Congenital malformations and genetic disorders rank as the third most common cause of neonatal mortality in India's urban areas [4]. Factors contributing to this high burden include a large population with high birth rates, consanguineous marriage practices in many communities, and limited access to prenatal and neonatal diagnostic services [4]. Rare diseases—80% of which are of genetic origin—affect an estimated 70 to 100 million Indians [4,5].

The Government of India has launched several initiatives over the past decade. The Rashtriya Bal Swasthya Karyakram (RBSK), established in 2013 and updated to RBSK 2.0 in 2026, screens children from birth to 18 years for the “4Ds”: Defects at Birth, Diseases, Deficiencies, and Developmental Delays [6,7]. The National Policy for Rare Diseases (NPRD) 2021 provides a framework for prevention, diagnosis, and treatment of rare diseases [4]. State-led programs—Kerala's Shalabham and Delhi's ANMOL (Advanced Newborn Monitoring and Optimal Life Care)—represent India's first large-scale government-funded expanded NBS initiatives [8,9].

This review aims to synthesize current knowledge on NBS methodologies, Indian policies and programs, epidemiological data on rare disorders, and the path forward for universal NBS implementation in India.

II. The Indian Rare Disease Landscape

2.1 Burden and Epidemiology

Rare diseases in India are defined as conditions affecting fewer than 1 in 5,000 individuals [5]. However, with India's population exceeding 1.4 billion, even “rare” conditions affect substantial absolute numbers. Approximately 80% of rare diseases have a genetic basis, and 70% present in early childhood [4,5].

Congenital hypothyroidism (CH) is one of the most common preventable causes of intellectual disability in India. Indian studies report CH incidence ranging from 1 in 2,000 to 1 in 2,500 live births—higher than the global average of 1 in 3,000—likely due to iodine deficiency in certain regions [2,10,11]. With 26 million annual births, an estimated 10,000 to 13,000 babies with CH are undiagnosed or diagnosed late each year in India [11].

A three-year study (2019–2021) screening 8,007 neonates in Pune reported the following birth prevalence: congenital hypothyroidism (1:2,002), congenital adrenal hyperplasia (1:2,002), G6PD deficiency (1:900), galactosemia (1:8,000), and biotinidase deficiency (1:8,000) [12]. Another study from Uttar Pradesh screening 396 neonates found G6PD deficiency prevalence of 2.27%, with CH and congenital adrenal hyperplasia each at 0.25% [3].

A comprehensive neonatal screening study in tribal populations of central India screened 382 newborns, demonstrating alarmingly high prevalence rates: sickle cell disease (7.3%), thalassemia (9.1%), and G6PD deficiency (8.9%), highlighting the need for targeted screening in underserved regions [17].

2.2 Consanguinity and Genetic Disorders

Consanguineous marriage is practiced in several Indian communities, particularly in South India and among certain Muslim populations. Consanguinity increases the risk of autosomal recessive disorders, as both parents are more likely to carry the same pathogenic gene variant [8]. This underscores the importance of population-based screening and genetic counseling in India.

III. Indian Newborn Screening Policies and Programs

3.1 National Policy for Rare Diseases (NPRD) 2021

The Ministry of Health and Family Welfare released NPRD 2021, establishing a framework for prevention and early detection of rare diseases through screening programs, creation of Nidan Kendras (diagnostic centers), financial support up to ₹20 lakhs for treatment of certain rare diseases, and development of registries for epidemiological data collection [4,5]. The policy explicitly recognizes NBS as a key strategy for early identification of treatable rare diseases [5].

3.2 Department of Biotechnology UMMID Initiative

Launched in 2019, the UMMID (Unique Methods of Management and treatment of Inherited Disorders) initiative operates on the principle that “prevention is better than cure” [4]. Key components include NIDAN Kendras (National Inherited Diseases Administration centers) established in government hospitals, training of clinicians in biochemical genetics and molecular genetics, population screening in aspirational districts, and building evidence for future research [4].

A three-year UMMID program (2019-2022) conducted in Ranchi, Jharkhand, screened newborns for five metabolic disorders and antenatal mothers for thalassemia and other hemoglobinopathies [16]. Results showed G6PD deficiency as the most prevalent metabolic disorder in Ranchi, with 13.6% of screen-positive cases confirmed positive for hemoglobinopathies. The mutation c.20 A>T was identified as the most common mutation among carriers [16]. As of November 2025, over 60,000 beneficiaries have received antenatal screening and more than 33,000 newborns have been screened under UMMID [4].

3.3 RBSK 2.0: Updated National Child Screening Program

In May 2026, the Ministry of Health and Family Welfare released RBSK 2.0 Guidelines [6,7]. Key features include expanded scope covering developmental disorders, mental health issues, and NCD risk factors; a lifecycle approach from birth to 18 years; digital integration with digital health cards and real-time data systems; strengthened referral pathways; and multi-sectoral convergence between health, education, and women & child development systems [6,7]. Screening services continue through Mobile Health Teams at Anganwadi Centers and schools [6].

3.4 State-Level Initiatives

Kerala’s Shalabham program is the only state to have implemented a free NBS program before 2026, screening for congenital, metabolic, and genetic disorders within 48 hours of birth [8,9].

Delhi’s ANMOL scheme, launched in the 2026–27 Budget with an allocation of ₹25 crore, provides 56 free diagnostic tests to newborns in all Delhi hospitals [8,9]. Led by Dr. Seema Kapoor at Maulana Azad Medical College, key features include 56 tests from a single dried blood spot sample, screening ideally within 24 hours up to 7–10 days after birth, integration of point-of-care hearing and heart screening, and use of Tandem Mass Spectrometry (MS/MS) technology [8,9,13]. Dr. Kapoor notes: “There are roughly 800 births per minute in the country. Screening can save about one in 300 lives” [8].

3.5 The Case for Mandatory Screening

Despite evidence supporting NBS, screening is not mandatory across India. A 2024 publication noted: “Newborn screening is done in India but is not mandatory. As of now, the number of diseases in the list of screening is not standardized among hospitals and laboratories in India” [3]. The authors concluded that screening for treatable metabolic diseases should be made compulsory in all healthcare centers [3].

Dr. Bradford Therrell, writing in *Indian Pediatrics* in 2019, stated: “Unfortunately, a national NBS policy aimed at early screening, detection and treatment...has been slow to evolve. The net result is thousands of individuals and families who must unnecessarily endure the adverse consequences of an easily and cheaply treated disease” [11].

IV. Newborn Screening Methodologies

4.1 Dried Blood Spot (DBS) Method

The DBS method is the foundation of biochemical NBS worldwide, involving collection of capillary blood from an infant’s heel onto specialized filter paper cards (Guthrie cards) [14]. The procedure involves warming the heel, cleaning with alcohol, pricking the side of the heel, applying blood drops to pre-printed circles, air-drying for 3–4 hours, and transporting in gas-impermeable bags with desiccant [14]. Advantages include minimal invasiveness (<100 µL blood), stability of analytes at room temperature, reduced biohazard risk, and cost-effectiveness for large-scale programs [14].

4.2 Tandem Mass Spectrometry (MS/MS)

MS/MS is the primary technology for expanded NBS, simultaneously detecting over 30–60 metabolic disorders from a single blood spot by measuring amino acids and acylcarnitines [13,15]. Under Delhi’s ANMOL scheme, MS/MS screens for over 50 conditions [9]. Detectable disorders include amino acid disorders (PKU, MSUD), organic acid disorders (methylmalonic acidemia, propionic acidemia), and fatty acid oxidation disorders (MCAD deficiency) [15].

4.3 Enzyme-Linked Immunosorbent Assay (ELISA)

ELISA is a plate-based immunoassay used to quantify hormones and proteins. In India, ELISA is commonly employed for screening congenital hypothyroidism (TSH), congenital adrenal hyperplasia (17-OHP), and biotinidase deficiency [12,14]. The Pune study utilized ELISA along with colorimetric and DELFIA techniques for screening 8,007 neonates [12].

4.4 Time-Resolved Fluorescence Immunoassay (TRFIA)

TRFIA uses lanthanide chelates (e.g., europium) as labels, offering superior sensitivity through time-resolved measurement eliminating background fluorescence. The dissociation-enhanced lanthanide fluorescence immunoassay (DELFLIA) was used in the Pune screening study [12]. TRFIA is particularly valuable for detecting low-concentration analytes in dried blood spots.

4.5 Genomic and Molecular Methods

Genomic newborn screening (gNBS) leverages genomic sequencing for early detection of genetic diseases, a departure from traditional biochemical approaches [15]. Over the past decade, advances in sequencing technology have expanded the scope of detectable conditions. While traditional biochemical methods fail to detect conditions without early biochemical markers, gNBS enables early diagnosis and identification of actionable genetic risks [15]. Key molecular techniques include Next-Generation Sequencing (NGS) for targeted gene panels and whole exome/genome sequencing; High-Performance Liquid Chromatography (HPLC) for sickle cell disease and thalassemia screening, particularly valuable in tribal populations [17]; Quantitative PCR (qPCR) for SCID and SMA screening; and Sanger Sequencing for confirmatory testing [4]. India has unique factors including large birth cohorts, established health delivery systems, increasing rare-disease initiatives, and developing national genomic databases—creating an opportunity to develop a gNBS program focusing on actionable results [15].

V. Screening vs. Diagnostic Testing

A fundamental principle of NBS is that a positive screening test does not diagnose a disorder. Screening determines whether a baby is more likely to have a condition; diagnostic testing (quantitative plasma assays, enzyme studies, or genetic sequencing) tells whether the baby actually has the condition [1]. In India’s ANMOL scheme, based on predefined cut-offs, results are categorized as positive, ambiguous, or negative, with follow-up diagnostic testing where needed [9]. Parents of infants with positive screens receive counseling and referral for confirmatory testing [12].

VI. Challenges to NBS Implementation in India

6.1 Lack of Mandatory Universal Screening

Unlike the United States where NBS is mandated by state law [1], India has no national mandate. A national NBS policy has been slow to evolve, resulting in thousands of children with treatable conditions suffering preventable disability [11].

6.2 Non-Standardized Screening Panels

The number of diseases included in NBS panels is not standardized across Indian hospitals and laboratories [3]. Private hospitals typically screen for only 5–6 common conditions, while expanded panels are limited to specialized centers [9].

6.3 Cost and Infrastructure

Advanced tests such as MS/MS can cost between ₹15,000 and ₹1 lakh in private setups, and treatment for rare disorders can range from ₹50,000 to over ₹10 lakh per year [13]. Government programs like ANMOL and RBSK 2.0 address this by providing free screening in public facilities, but MS/MS equipment remains expensive, requiring specialized infrastructure and trained personnel [9].

6.4 Awareness Gaps

Awareness regarding genetic disorders remains limited, particularly beyond urban centers. “Upper middle- and high-income groups usually opt for genetic screening due to affordability and access,” notes Dr. Sanjeev Gupta [13]. Some doctors remain unaware of available treatments for rare disorders [8].

6.5 Follow-Up and Confirmatory Testing

A robust follow-up system is essential to ensure that infants who screen positive receive timely confirmatory testing and treatment. RBSK 2.0 addresses this through strengthened referral linkages and tracking systems [6].

VII. Future Directions

7.1 Toward Universal Newborn Screening

The successful implementation of state programs like ANMOL and Shalabham provides models for national expansion. Dr. Seema Kapoor expressed hope that the Delhi model will be replicated nationwide [8]. Key steps include enacting national legislation mandating NBS for a core panel of conditions, standardizing screening panels across states, and integrating NBS into existing RBSK 2.0 infrastructure.

7.2 Integration of Biochemical and Genomic Screening

Experts emphasize that genomic screening is unlikely to replace biochemical tests entirely. An integrated approach—combining MS/MS, immunoassays, and targeted NGS—provides the highest accuracy and broadest detection capability [4,15]. The UMMID initiative’s focus on both biochemical and molecular genetic diagnostics reflects this integrated vision [4].

7.3 Digital Health Innovations

RBSK 2.0 introduces digital health cards, real-time data systems, and integrated tracking platforms [6], which can improve program efficiency, reduce loss to follow-up, and enable evidence-based decision-making.

7.4 Building Evidence for Cost-Effectiveness

While international studies have demonstrated the cost-effectiveness of NBS, India-specific data are needed. The economic burden of untreated conditions—lifelong institutional care for intellectual disability due to missed CH—far exceeds the cost of screening [11].

VIII. Conclusion

Newborn screening in India is at a transformative juncture. The convergence of national policies (NPRD 2021, RBSK 2.0), targeted initiatives (UMMID), and pioneering state programs (ANMOL, Shalabham) is creating momentum toward universal NBS. With 26 million annual births, early detection of congenital hypothyroidism alone could prevent intellectual disability in over 10,000 children each year [11].

Technological methodologies—from dried blood spot collection to tandem mass spectrometry, ELISA, TRFIA, and genomic sequencing—are available and validated. The remaining challenges are primarily systemic: mandating screening, standardizing panels, building infrastructure, training personnel, and ensuring follow-up. As Dr. Therrell concluded: “The time to begin NBS is now. Let’s Just Do It!” [11]

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Conflict of Interest

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