Newborn screening and testing: Picking up pace in India?

By Kalyani Sharma — On Sep 6, 2022

NEWS GENOMICS HEALTH POLICIES

Understanding the need and viability of newborn screening programmes in India is crucial to secure every newborn's health

As per WHO, In India, more than 1.7 million children are born with birth defects every year. The term 'birth defect' encompasses a diversity of health conditions and has been recognised globally as a major contributor to neonatal and infant mortality and disability. This is where the role of New Born Screening or NBS comes in.

NBS detects rare congenital metabolic disorders in children which are commonly called as Inborn Errors of Metabolism or IEM. If not treated, these disorders will lead to mental retardation, disability or death. NBS is the only way to detect these disorders early in the new-born period.

There are as many as 500 IEMs which lead to congenital disorders. Vast number of these disorders are genetic in nature due to defects in the gene that codes for enzymes in various metabolic pathways such as carbohydrate metabolism, amino acid metabolism, fatty acid metabolism etc. These defects will lead to accumulation of toxic substances which interfere normal functioning of cells in our body leading to diseases.

Talking about the need for newborn screening and testing in India, Neeraj Gupta, Founder & CEO, Genes2Me added, "Do you know that India tops the list when the context is about most births in the world? Moreover, the infant mortality rate in India is pretty high too. Keeping all these factors in mind, it is essential to conduct newborn screening testing. Catering to the basic medical requirements across the length and breadth of a country like India is tough."


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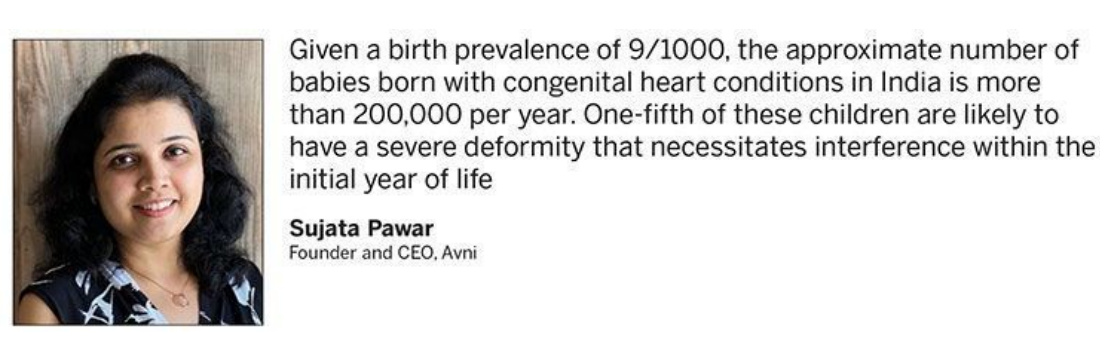
In India, infant mortality is higher compared to other developed countries. Many newborns are susceptible to various diseases that are not visible at the time of birth but affect babies later in their lives.

Explaining about the inborn errors, Dr Karthik Nagesh, Chairman & HOD-Neonatal ICUS & Manipal Advanced Children's Centre, Manipal Hospitals said, "For a newborn, the first month is considered the most crucial as it lays the foundation for the baby's healthy development. It is the period where any metabolic disorders/diseases if inherited genetically may present with severe symptoms. Many neonates however, are asymptomatic and clinically present with significant morbidity or mortality even later. These inherited IEMs, if not diagnosed early in life may unfortunately pose life-threatening problems and irreversible neurological and multi-organ damage to the child. Hence these disorders need to be detected early, soon after the baby's birth. The NBS also known as a baby's first test is a proactive initiative to detect early and prevent genetic metabolic disorders in a newborn. These tests are conducted within 48-72 hours of the birth of the infant."

Sujata Pawar, Founder and CEO, Avni stresses, "Most developed nations include a hearing test in newborn screening, as well as measure the level of oxygen in a baby's blood to identify newborn babies who need to see a cardiologist right away; neither testing involves a bloodspot. Given a birth prevalence of 9/1000, the approximate number of babies born with congenital heart conditions in India is more than 200,000 per year. One-fifth of these children are likely to have a severe deformity that necessitates interference within the initial year of life. Many Neonatal Intensive Care Unit (NICU) admissions are also attributed largely to IEM. Many infants develop mental disabilities, intellectual difficulties, autism, dyslexia, behavioural abnormalities, and scholastic backwardness later in their lives if they go undiagnosed and left untreated."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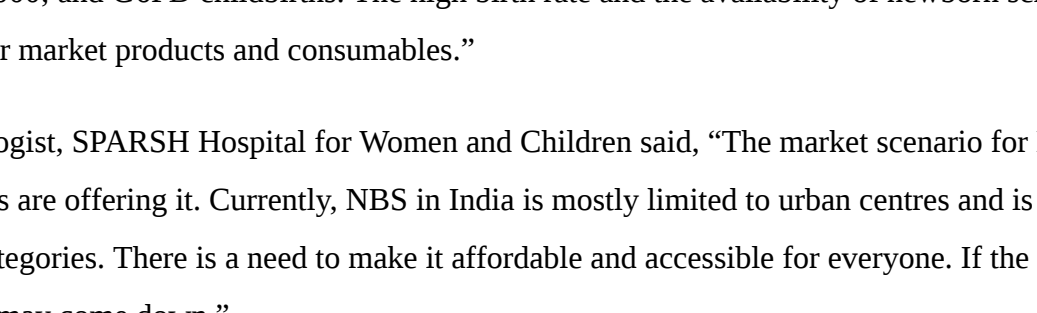
Market scenario

According to data provided by Indian Paediatrics, in India, the prevalence of IEMs is 1 in 2,497 newborns; the incidence of congenital hypothyroidism is 2.1 per 1,000; and G6PD childbirths. The high birth rate and the availability of newborn screening in public hospitals indicates a strong demand for market products and consumables."

Dr Paras Kumar J, Neonatologist, SPARSH Hospital for Women and Children said, "The market scenario for NBS has definitely improved and many hospitals are offering it. Currently, NBS in India is mostly limited to urban centres and is not very affordable for people from all economic categories. There is a need to make it affordable and accessible for everyone. If the screening is done on a mass scale, the costs of screening may come down."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Dr Lathiesh Kumar Lead Neonatology, Consultant-Paediatrics and Neonatology, Aster Women and Children Hospital said, "The size of the global newborn screening market is anticipated to increase by 8.3 per cent CAGR from 2021 to 2026, from USD 0.9 billion to USD 1.3 billion."

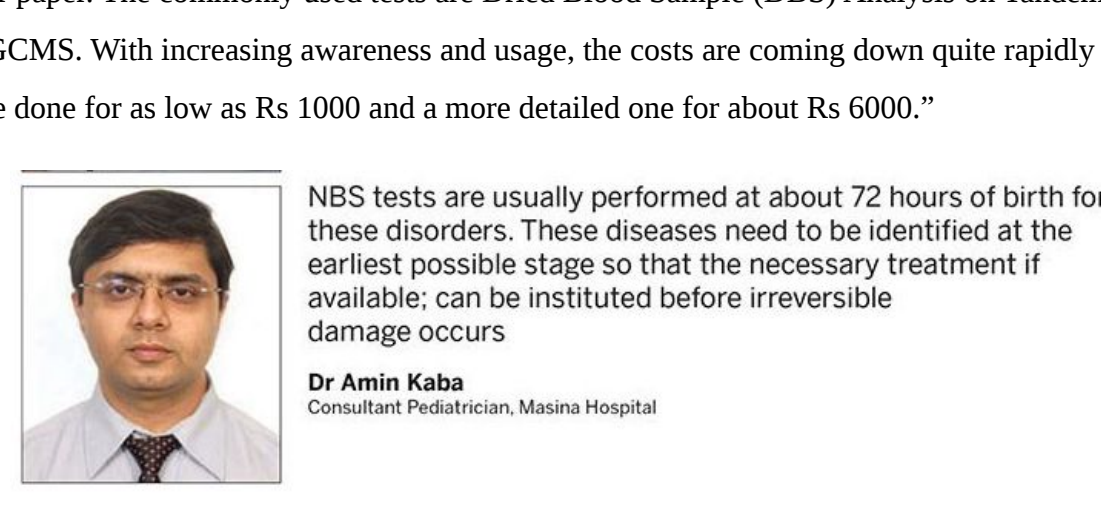
Current available test for NBS in India

Early diagnosis of IEM is a challenge for pediatricians since they will not present any symptoms in the new-born period. Most of the IEM cases are diagnosed after a few months or even after a few years of birth and by this time life threatening irreversible damage to the brain or other disability has already happened. NBS is the only way to pick up these disorders early so that the child can get an early intervention and treatment. Experts explain some of the current available tests for newborn screening in India.

In India, the 'National Neonatology Forum' of India has published 'Clinical Practice Guidelines' for the early diagnosis of patients with metabolic disorders to enable early intervention and prevent morbidity and mortality.

Dr Nagesh while explaining the guidelines said, "According to the guidelines, there are three levels of newborn screening tests that are to be carried out at the hospitals. Level 1 screening test is strongly recommended universally for all newborns in the country to enable detection of the two commonest diseases that is Congenital Hypothyroidism and Congenital Adrenal Hyperplasia (CAH). Level 2 screening should be conducted in a high-risk population (with family history of intellectual/physical deformity, symptoms of IEM and in critically ill neonates). A number of disorders can be detected in this test including phenylketonuria, galactosemia, tyrosinemia, Glucose-6-phosphate dehydrogenase deficiency, biotinidase deficiency, cystic fibrosis, maple syrup urine disease (MSUD) etc. Level 3 screening is offered to families of a newborn in tertiary care hospitals in high resource centres where the screening is offered for nearly 50 more neonatal disorders."

Dr Amin Kaba, Consultant Pediatrician, Masina Hospital explains that, "NBS tests are usually performed at about 72 hours of birth for these disorders. These diseases need to be identified at the earliest possible stage so that the necessary treatment if available; can be instituted before irreversible damage occurs. The screening tests are usually done either on a urine sample or a heel pin prick single drop of blood taken on a filter paper. The commonly used tests are Dried Blood Sample (DBS) Analysis on Tandem Mass, Spectrometry (TMS) and Urine Analysis by GCMS. With increasing awareness and usage, the costs are coming down quite rapidly and a basic screen for 5 common diseases can be done for as low as Rs 1000 and a more detailed one for about Rs 6000."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Dr Yogesh Kumar Gupta, Consultant-Paediatrics, Fortis Hospital said, "Metabolic disorders make a big chunk of problems in children which go undetected or in diagnosed till they present which can be late and sometimes with irreversible damage. Screening of their rare but serious disorders is the only way forward to prevent the morbidity and mortality. We were earlier carrying out screening for congenital hypothyroidism which still remains the most common cause of mental retardation. With advent of advanced technology, we have expanded the testing to many rarer metabolic conditions."

Dr Sujit Chatterjee, CEO, Dr L H Hiranandani Hospital highlights, "NBS test as the name suggest is a group of tests done within three days of birth to screen for diseases that may be sub-clinical and have long term implications on quality of life. Some diseases, though subclinical initially, can have lasting and irreversible damage if detected later in life, hence the importance of NBS. It is a cost-effective intervention to decrease the burden of disease in the community and to intervene early so as to prevent morbidity and mortality arising from the otherwise un-detected disease. Commonly the NBS includes: 3 drops of blood on a filter paper collected by needle prick on sole of newborn, urine soaked on a filter paper by keeping the paper near urethra, hearing screening by otoacoustic emission study and if refer then confirmed later by ABR before 3 months and eye and fundus examination by dilating the pupils of both eyes with eye drops to detect disorders like IEMs, congenital hypothyroidism, congenital adrenal hyperplasia, G6PD deficiency, cystic fibrosis, hearing deficits and visual impairments."

Upcoming trends

These tests are mandatory in developed countries but in a developing country like India, their availability and accessibility were an issue. However, with the trend that NBS is slowly picking up, these tests are now available, accessible and even prescribed by specialists and not limited to a few labs.

Dr Kumar highlights, "One of the key trends is that earlier NBS was available mostly in high-tech laboratories. This scenario has changed quite a bit. NBS is now easily accessible in urban centres and in fact, in many hospitals they do it as a routine screening for all babies. The basic five panel tests are offered to all the newborns."

Dr Madhavi V, Consultant Geneticist, Fernandez Foundation said, "Phenylketonuria was the first disease screened by NBS from dried blood spots by bacterial inhibition assay. With improvements in technology and the introduction of newer techniques like Enzyme-Linked Immunoassay (ELISA), High-Performance Liquid Chromatography (HPLC), and Liquid Chromatography-Mass Spectrometry (LCMS), more disorders are being incorporated into the NBS panel. Currently, more than 100 metabolic and genetic disorders are being screened in several laboratories. It is estimated that about 10000 babies affected with beta thalassemia are born every year. As it has a high carrier frequency, newborn screening for hemoglobinopathies may be considered as an additional strategy. Hearing loss has a high incidence, and if not corrected before six months of age, may lead to permanent hearing and speech impairment. Hence, screening for hearing loss is important."

Incorporation of NBS in National Health programme: Need of the hour

Although, NBS in India is slowly picking up pace, it is still not as par with the number of births and the current rate of infant mortality in the country. Cost, awareness and ignorance are among the factors for this reality.

The good news is that with increase in the uptake of these tests, the costs will also come down further making these tests more accessible to the general population as well. Incorporation of NBS in National Health Programmes is the utmost need of the hour. All the more crucial is its viability and proper implementation.

Explaining the current policy scenario, Dr Nagesh said, "The NBS initiative has been recommended for last two decades in many states and at the national level but is not yet implemented in full due to lack of funding and low prioritisation. Currently, there are a good number of NBS laboratories either public or private that offer NBS tests in India, and the number is growing. There is a lack of awareness among parents about NBS in the country, which hence keeps it from reaching its full potential."


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"The fact that 76 per cent of the Indian population is classified as low socioeconomic with another 20 per cent falling under the poor category, accessing basic healthcare facilities is certainly difficult particularly for the impoverished community in the rural parts of India. Even in the urban middle-class, there may be major difficulties in conducting basic NBS programs due to the limited availability of resources. Owing to its huge population, implementing the NBS program in India necessitates a careful prioritisation and structured approach. Segregating it into a three structured approach of Level-1 to 3 based on the state's budgetary allocation and the incidence of IEM in the community is the way forward. Prevention of avoidable infant deaths and mental retardation should be given a consideration and the benefits of implementing state wide universal NBS will lead to significant drop in neonatal mortality rates over time", he added.

Experts believe that this will give a better idea of the kind of disease load that's prevalent in our country.

Dr Gupta says that, "New-born screening has been made mandatory in most of the private institutes but still needs to reach the government setups in rural areas where we may be missing many cases. There are various labs which run tests for metabolic screening. But we need to be very specific and precise in looking for these metabolic disorders. I would always recommend a certified and skilled lab or institution whenever we go for newborn screening. These screening tests have to be very sensitive, so they pick the defect much before they present and that's the whole purpose of screening. Definitely by expanding the screening programme to various institutes and hospitals we can significantly diagnose and pick these disorders. By making it available on a mass scale, we can cut down on the cost of the tests too which is the major obstacle in implementing these tests routinely."

Commenting on the same, Dr Chatterjee said, "Incorporating NBS into national health programme would increase the coverage and make the NBS available to much needed economically challenged population. One of the best things government could do for the health of its people would be to budget for including NBS in the health programme."


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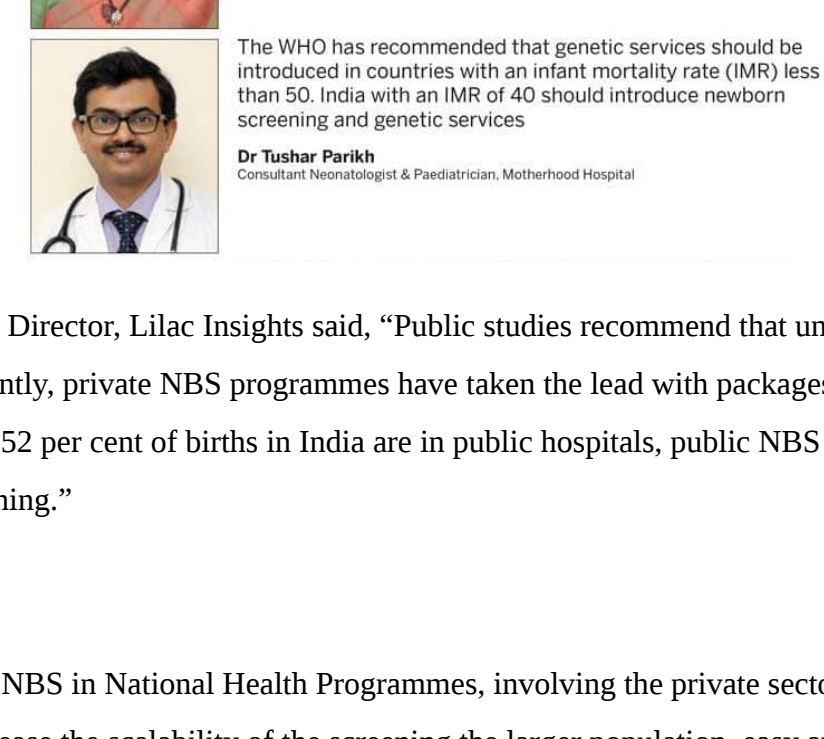
Stressing on a successful pilot, Dr Tushar Parikh, Consultant Neonatologist & Paediatrician, Motherhood Hospital said, "In the state of Goa 'Heel to heal' programme was carried out few years ago to screen all babies for newborn metabolic disorders. Surprisingly the incidence of metabolic disorder was to the tune of 1 out of 650 babies. This was a pilot project which gave high incidence of these disorders in our population. Total of 48,000 babies underwent screening. With so much burden of disorders in our country it is definitely important to screen the babies for NBST. India is going through a progressive transitional phase of control over infant mortality and morbidity due to infections, and emergence of genetic conditions. The WHO has recommended that genetic services should be introduced in countries with an infant mortality rate (IMR) less than 50. India with an IMR of 40 should introduce newborn screening and genetic services. The Indian Academy of Pediatrics strongly advocates inclusion of newborn screening in our public health policy. The current infant mortality rate for India in 2022 is 27.695 deaths per 1000 live births."

Adding her views Dr Madhavi said, "The central government along with the state government has to initiate the universal implementation of NBS. The programme has to be initiated nationwide and the result has to be analysed by experts. With the already available facilities at public hospitals, dried blood spot collection can be done. Definitive diagnosis of high-risk neonates can be offered at selected centers where diagnosis, treatment, and prenatal test in subsequent pregnancies are available. The key to the success of the programme is continuous followup with the affected babies. Hence, follow-up of these babies can be done by Accredited Social Health Activist (ASHA) workers who are trained professionals, primarily women, with a focus on women and child healthcare, and come from the rural communities they serve. The ASHA workers can help in monitoring cases to ensure compliance and bring patients to the notice of the physicians if required."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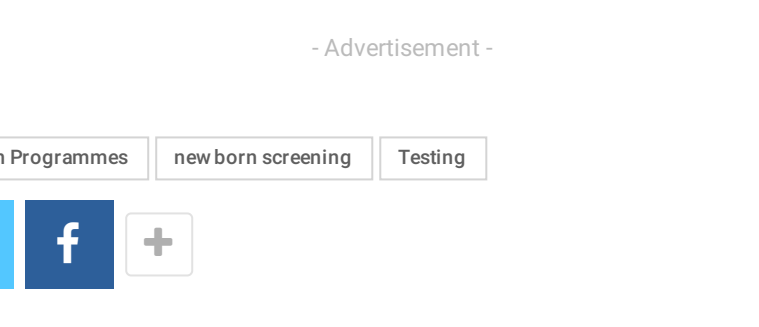
Subhamoy Dastidar, Co-Founder & Director, Lilac Insights said, "Public studies recommend that universal screening programmes should include two to four disorders. Presently, private NBS programmes have taken the lead with packages to detect three disorders to a comprehensive set (for 50+). Since 52 per cent of births in India are in public hospitals, public NBS screening programmes have the potential to achieve universal screening."

Way forward

For a successful implementation of NBS in National Health Programmes, involving the private sector and encouraging the PPP model can play an important role as it can increase the scalability of the screening the larger population, easy availability and accessibility of tests and can also help in making the parents aware about overall need and advantages of NBS.

Stressing on the need for PPP, Dr Srinivasa Murthy C L- Lead Paediatrics, Consultant-Pediatrics and Neonatology, Aster Women and Children Hospital, Bangalore said, "To lower infant mortality rates, Rashtriya Bal Swasthya Karyakram (RBSK) is being introduced throughout India as part of the National Rural Health Mission (NHM).

Public-private alliances might be taken into consideration in order to execute the NBS program on a nationwide scale, ensuring that every newborn will be able to benefit from it. Major government hospitals in significant cities ought to serve as the process' initial catalyst. Later, proceed to tier 2 and tier 3 levels while putting in place the new infrastructure."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Talking about the way forward, Pawar said, "Despite the numerous advantages and the absolute necessity of a screening programme in India, putting it in place presents its own set of barriers. NBS is a programme, not a test. It requires much more than tools and supplies. It necessitates meticulous planning, commissioning, follow-up and confirmatory testing, adequate dietary and healthcare assistance, and expert genetic pre-and post-test counselling. Therefore, the central and state governments, as well as private healthcare providers, must collaborate to demonstrate commitment to incorporating newborn screening testing into the national health programme."

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