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Editorial

Newborn screening in India: A pressing priority

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September is newborn screening awareness month

Newborn screening (NBS) is a preventive healthcare process by which newborns are screened just after birth for disorders that can cause severe illness or death unless detected and treated early. NBS entails using blood collected from the baby's heel which is placed onto spec. Which is placed onto special absorbent paper, air dried and transported to a screening laboratory for analysis. It aims to identify at the earliest possible moment, hereditary disorders to prevent serious and life-threatening consequences by timely intervention.

Due to the high birth rate in our country, a large number of infants are born with congenital malformations, genetic conditions like inborn error of metabolic (IEM) disorders. Many of these IEMs are treatable if diagnosed at an early age. Interestingly, there are few conditions such as congenital hypothyroidism (CH), congenital adrenal hyperplasia (CAH) and Glucose 6 phosphate dehydrogenase deficiency that are treatable with a minimum of cost if identified early, which is the primary norm for NBS. Hence, the presence of a screening program that screens newborn babies for common treatable conditions is essential to identify affected babies early. This shall aid in significant reduction of infant morbidity, mortality in the country with a significant reduction in National Health Burden. India, a country with the second largest population in the world, does not have a national NBS programme as part of its health policy. Dried blood spot test introduced by Robert Guthrie in 1961 to screen phenylketonuria initiated the concept of NBS which became a national programme of the US in 1962. The benefits of NBS have formed a topic of discussion since then. NBS helps in reducing preventable developmental delay, disability, morbidity and mortality during infancy and childhood. The outcome is a better quality of life for the affected child, with an increased cognitive, intellectual and monetary contribution to society. The decision to implement the NBS programme for a country largely follows the Wilson and Jungner criteria (World Health Organization, 1968). The most important criterion is that the disorders detected by a screening test should be treatable.

NBS is a simple and cost-effective strategy for the early identification of IEM disorders in the neonates immediately after birth. The basic idea of carrying out NBS is to identify newborn with common treatable conditions before the manifestation of symptoms. Early treatment in these infants can prevent or minimise serious irreversible complications such as mental retardation, metabolic complications and disabilities. This shall have a direct consequence on a significant reduction of infant morbidity and mortality. In addition, in the presence of a successful NBS, the financial burden incurred on the families and the society will be far less as compared to that without NBS because these tests are cheap and the available treatment options have a good prognosis.

The key components for an effective NBS include: (a) educating the participating families about the benefits of the screening program, (b) timely sample collection, (c) recruitment of trained professionals for performing appropriate tests, (d) generation of reports and conveying the same to the patients, (e) providing counselling when required and (f) monitoring the long-term outcome and cost-effectiveness of the screening program. NBS is a mandatory procedure in many developed countries such as the USA , UK, Australia, Sweden, Finland and Japan for screening as many as 54 different inherited conditions. However, despite India having a high birth rate of ~60,000 children born every day, NBS is not a routine practice. A high rate of consanguinity and endogamy in many communities in India, as well as a lack of awareness and lack of adequate diagnostic facilities in many parts of the country, are likely to be the major contributors to the high incidence of IEM. Nonetheless, this emphasises the need for implementing a national-level centralised NBS program. The major hurdles of implementation of an effective NBS in India are: (a) unavailability of accurate epidemiology data of common treatable conditions for the Indian population in different regions of the country, (b) lack of awareness of treatable genetic conditions in the medical community, (c) dearth of necessary infrastructure and trained manpower for the program and (d) absence of a national policy for NBS.

Dr. Meena Desai was the first one to initiate NBS for CH in the country in the early eighties in Mumbai followed by Dr. Appaji Rao *et al*. who carried out the newborn metabolic screening program for amino acid disorders in Bengaluru in 1988. Indian Council of Medical Research (ICMR), through a Task Force on Inborn Metabolic Disorders, had set up a screening program that lasted from 2007 to 2012. In this study, a total of 207,561 newborns were screened for CH and CAH from 5 centres in India, Delhi, Mumbai, Chennai, Hyderabad and Kolkata. They observed that the incidence of CH varied from 1 in 727 in Chennai to 1 in 1528 in Mumbai. Likewise, for CAH the incidence varied from 1 in 2036 in Chennai to 1 in 9983 in Mumbai.

NBS in India has gained impetus in the last decade. There are several public and private NBS programs being carried out in different parts of the country. The key challenge is setting up a centralised policy to supervise the program to achieve maximum outcome from the same. Likewise, it is the need of the hour to create awareness among the medical community regarding this group of treatable conditions. Finally, setting up the required infrastructure and employing trained personnel is critical for successful implementation of such a gross newborn screening program in every district of the country. NBS plays a crucial role in saving and improving babies' lives. India has one of the highest infant mortality rates in the world. To improve infant and child mortality rates, a national NBS policy should be adopted.

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