PRESIDENT'S PAGE

Newborn Screening in India

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Newborn screening for common metabolic and genetic disorders should be an integral part of neonatal care as early detection and treatment can help prevent intellectual and physical defects and life threatening illnesses [1]. The list of conditions for which screening is carried out differs from country to country, based on the prevalence of the condition and available resources. Universal screening for about 40 to 50 metabolic disorders is mandatory in US, Europe and many other countries across the world. Though universal screening is a cost-intensive exercise, the benefits far exceed the cost as it helps in reducing the mortality and morbidity of these diseases. In 1968, Wilson and Jungner [2] proposed the following criteria for inclusion of a condition in screening: (*i*) condition should have an important health problem/frequency; (*ii*) test should be acceptable to the population (reliable/ simple); (*iii*) disease does not manifest at birth/ routine examination; (*iv*) treatment will prevent mortality and morbidity; (*v*) delay in diagnosis will cause irreversible damage; and (*vi*) screening is cost-effective.

The conditions for which neonatal screening has been proposed in Indian scenario include hearing loss, congenital hypothyroidism, congenital adrenal hyperplasia (CAH) and glucose-6-phosphate dehydrogenase (G6PD) deficiency [3-10]. Hearing loss has a high incidence, and if not corrected before 6 months of age, may lead to permanent hearing and speech impairment. Congenital hypothyroidism also has a high incidence and is the most important preventable cause of intellectual disability. Congenital adrenal hyperplasia. If undetected at birth, can result in mortality, morbidity or genital abnormalities. G6PD deficiency has a relatively high incidence in Northern parts of the country, and cost of testing is affordable. Considering the prevalence of these conditions and huge financial implications for universal screening for a developing country like India, a practical approach will be to categorise the conditions as follows:

Category A (all newborns): Screening for congenital hypothyroidism and hearing should be a must in Indian scenario. Screening for CAH and G6PD deficiency may be added in a phased manner. G6PD screening should be done in Northern states of the country. Screening for Sickle cell disease and other hemoglobinopathies should be undertaken in pockets of high incidence.

Category B (High risk screening): Screening for the following disorders should be conducted in the high risk population (consanguinity, previous children with unexplained intellectual disability, seizure disorder, previous unexplained sibling deaths, critically ill neonates, newborns/children with symptoms/ signs/investigations suggestive of inborn errors of metabolism). These conditions include phenylketonuria, homocystinuria, alkaptonuria, galactosemia, sickle cell anemia and other hemoglobinopathies, cystic fibrosis, biotinidase deficiency, maple syrup urine disease, medium-chain acyl-CoA dehydrogenase deficiency, tyrosinemia and fatty acid oxidation defects.

Category C: Screening (in resource-rich setting/ expanded screening) for 30-40 inherited metabolic disorders may be offered to 'well-to-do' families, especially in urban settings where facilities for sending sample to laboratory are available.

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, and will offer its technical and logistic inputs to the Government of India for initiating this program.

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