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Editorial

# Inherited Metabolic Disorders - Relevance of Newborn Screening in India

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Congenital metabolic disorders/Inherited metabolic disorders caused by deficiency of enzymes which result in substrate accumulation and cause mild or sever neurologic/psychological manifestations. This leads to death or permanent disability of the child. Some forms IEMs cause isolated stable mental retardation (MR) and global developmental delay [1]. About 25 million babies were born per year and infant mortality rate of 35 /1000 live births were reported in India [2]. Around 500 inborn errors of metabolism (IEM) were present in Indian population and it is stated that 1:20 Indian have some form of disorder or birth defects [3]. Pooled birth prevalence of IEM in the Eastern Mediterranean region is about 75.7/100 000 live births. Total fatality rate in developing countries is 33% accounting for 0.4% of child deaths all over the world [4].

IEM is classified into a) energy metabolism disorders like respiratory chain disorders b) intoxication syndromes like organic acidurias and amino acidurias c) lipidoses (lipid storage diseases), c) metal (iron, copper) storage diseases and d) neurotransmitter diseases (serotonin, dopamine). Some IEM have one time curative treatment while some requiring lifelong treatment and some others have definitive treatment but high cost and requiring lifelong therapy [5].

Tandem mass (TM) spectrometry can be used for the diagnosis of more than 20 inherited disorders while advanced high resolution mass spectrometry detects more than 400 metabolites. Uniform screening panel recommended by American College of Medical Genetics (ACMG) consists of 35 primary and 26 secondary disorders, out of these 45 can be screened using LCMS/MS. Several studies have been conducted in different parts of India on congenital hypothyroidism, congenital adrenal hyperplasia and glucose 6 phosphate dehydrogenase deficiency but the data varies between studies; congenital hypothyroidism 1:22 to 1:3400, congenital adrenal hyperplasia 1:200 to 1:6813, glucose 6 phosphate dehydrogenase deficiency 0.8% to 17.5%, and aminoaciduria 1:847. ICMR multi-centric study screened 143,344 babies and found prevalence of 1:1130 for congenital hypothyroidism and 1:5762 for congenital adrenal hyperplasia [6]. All these studies reveal the high prevalence of CA, CAH and G6PD in India with regional differences. Limited data is available on aminoacidemia (frequencies 1.7- 6.1 %,) and organic acidemia (1.3-10.7%) from India [7].

Newborn screening (NBS) consists of methodologies for education, screening, confirmation and treatment of inborn errors of metabolism to prevent death or irreversible brain damage. In India about 800,000 babies were born with congenital disorders, 25,000 with metabolic disorders 350,000 cases of G6PD and 15,000 cases of CH were detected per year [8,9]. Region wise NBS was started in different states like Goa, Chandigarh and Kerala which were mainly focused on CH, CAH and G6PD deficiency. Nationwide NBS in India should be started with proper planning with focusing on diagnostic techniques, management and logistics. Available data emphasize the need of routine NBS in India. Nationwide newborn screening program should be started for CA, CAH, G6PD, and high-risk population should be screened for amino acid disorders and organic acid disorders (phenylketonuria, tyrosinemia, homocystinuria, alkaptonuria, galactosemia, maple syrup urine disease, medium chain acyl CoA dehydrogenase deficiency, fatty acid oxidation defects, biotinidase deficiency, cystic fibrosis, sickle cell anemia and other hemoglobinopathies). Awareness is an important part of NBS. After the successful implementation of NBS program for above disorders, it can be extended according to prevalence of different disorders in different regions. Screening center and special clinics for follow up and treatment should be constituted in each state as it is highly relevant from socio-economic aspects to reduce the disease

burden. As the rate of screening and diagnostic facilities improves the number of positive cases detected increases. For them timely counseling, therapy and rehabilitation will be necessary.

The nationwide newborn screening may be a challenge in India but increasing awareness and different regional screening programs conducted for the last decade make it possible that more rare inborn errors were detected per year in India. This necessitates government's policy and support for expanded screening.

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