Neonatal screening for congenital hypothyroidism in India

I
dine was characterized as an essential element of thyroid tissue by Baumann in 1895. The efficacy of iodine to prevent goiter was demonstrated by Marine in Northern USA in 1916–1920. Severe endemic goiter and cretinism had been almost entirely eliminated from continental Western Europe and Northern America before the 1930’s.

Congenital hypothyroidism (CH) due to the absence of thyroid gland in babies was found to be the cause of cretinism in most of the infants in early 1930s, and this was considered a separate entity from iodine deficiency. Screening for CH started in many countries almost 50 years ago, after the screening for Phenylketonuria was invented by Dr. Robert Guthrie. Screening for CH is the most cost-effective public health program.

In India, there are several studies demonstrating the incidence of CH to be higher than the rest of the world. The current issue of Indian Journal of Child Health has published a study by Prasad et al.[1] which is a similar attempt to screen the newborns with CH in the state of Jharkhand. However, the limitation of this recent study is that it has too small a sample size to talk about incidence of CH in India. Furthermore, though many studies are quoted for reference by the authors regarding the incidence of CH, lot of studies have not done nuclear scans for confirming the absence of thyroid and indeed. Indeed, many of these studies either have done ultrasound of the neck or treated based on thyroid stimulating hormone (TSH) values, which is notable for false negative results. This may lead to “over treatment of CH” as many cases either have transient hypothyroidism or are due to uptake problem which resolves within 1 or 2 years. Kumar et al.[2] showed that the screening for CH is overdue in India, with all their babies having undergone nuclear medicine scans to confirm the absence of thyroid gland and also, they have noticed many of their babies had uptake problem too, which were not considered as CH.

It is well-known fact that newborn babies have a TSH surge in the first 36–48 h - so the value of TSH depends on the “timing” of the sample and also “accuracy of the sample” in the filter paper. Prasad et al. have taken the samples at different times accounting for the variable results. Furthermore, the results vary depending on the prematurity. Kumar et al. study have shown that the TSH values in CH are generally in 100s and if $<80$, it is suggestive of transient hypothyroidism. The present era is of mass spectrometry and next-generation sequencing methods expanding of the newborn screen panel. These might address the technical issues such as turnaround time, and decrease the false positive and false negative rates [4].

It is high time, India with its vast resources, should start the screening program for CH. However, to do that we need to have guidelines from academic bodies like Indian Academy of Pediatrics and National Neonatology Forum about the timing of screening, TSH values for each day till day 7, educate the common pediatrician and public about the importance of the accuracy of sample to prevent false negative cases and transport to the regional lab promptly. We can emulate the guidelines by the European Society for Paediatric Endocrinology [3].

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