Newborn screening for hemoglobinopathies

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India has enormous burden of birth defects. 800,000 babies are born annually with birth defects, of this 14,000 are born with thalassemia, as per 2013 report on birth defect surveillance of Southeast Asian region [1]. Carrier frequencies rate of various hemoglobinopathies ranges from 17% to 30% for sickle cell disease, 0.3% to 15% for β-thalassemia. HbE is found in eastern half of Indian subcontinent with carrier rate of as high as 60%, and for milder forms of α-thalassemia, it varies from 15% to 80% (tribal population) in north eastern parts of India [2]. The current aim is to decrease thalassemia births by 50% with main focus on primary prevention in the form of prenatal screening in the first trimester of pregnancy [3].

In this issue of Indian journal of Child Health, Kumar et al have published a research where they carried out carried out secondary prevention study of hemoglobinopathies by doing neonatal screening within the 1st week of postnatal life [4]. Population screened was from catchment area of West Bengal, Jharkhand, Bihar, and Orissa. This belt has high carrier rate of β-thalassemia and hemoglobin E. 4000 neonates were screened over a period of 1½ year. Isoelectric focusing was used to detect hemoglobin variants. This method has an advantage over high-performance liquid chromatography (HPLC) method due to its high resolution; hence, bands are not overlapped, but at the same time, it needs expertise and training of handling the machine. Isoelectric focusing (IEC) has not only high sensitivity but also has high specificity, thus preventing false negativity in double heterozygous cases. Whereas, more conventional method, HPLC is more user friendly and easy to interpret. It was found to have good kappa agreement with IEF as first line screening test for hemoglobinopathies in this study. The current study did not mention about total eligible neonates for the screening test and how many neonates were missed on first screening. This could have been the reason behind not reporting the true prevalence of hemoglobinopathies in the study population. Previous studies from West Bengal have reported 14–29% rate of hemoglobinopathies [2].

Hemoglobin variants have a significant prevalence in India and population-based prevention programs are needed. Antenatal screening can bring reduction in disease load of birth defects attributed by hemoglobinopathies, and in case, both parents are carrier, they should be screened in each pregnancy irrespective of previous conception result [5]. In case, antenatal screening was missed then neonatal screening for hemoglobinopathies using HPLC or IFC method may be carried out so that anemia and related morbidities are decreased [6].

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