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**Neonatal Screening for Hemoglobinopathies**

A pilot study was undertaken to develop a feasible neonatal screening strategy for hemoglobinopathies. Isoelectric focusing using dried blood spots samples as a primary screening technique was standardized for the first time in India. The screened positives were confirmed by high performance liquid chromatography followed by parental screening, confirmation, and education.

**Key words:** Hemoglobinopathy, India, Isoelectric focusing, Neonatal screening, Prevention.

Hemoglobinopathies cause high degree of morbidity and mortality in India [1], there is an urgent need to detect the disorders as soon as possible after birth. We conducted a pilot study aiming to develop a feasible neonatal screening strategy. Following informed consent from parents, dried blood spot (DBS) samples were collected from 207 inborn babies within day 3-7 of life, over a period of two months. Primary screening by isoelectric focusing (IEF) (Perkin Elmer, Finland) [2] was done within 7 days of sample collection. Results were interpreted using ISOSCAN software (Perkin Elmer, Finland). The screened positive babies were recalled for confirmation by high-performance liquid chromatography (HPLC) (Biorad Laboratories)
using anticoagulated blood at a reference laboratory. Parents of the positive babies were also screened and confirmed. Complete hemogram of the recalled babies and parents was performed. Repeat screening by IEF as well as HPLC of 20 screen negative babies were performed to check whether the technique of IEF gives false negatives or not.

Among four babies positive for hemoglobinopathies, three had Hb E trait and one had HbE disease. All were term babies and clinically asymptomatic, with average hemoglobin concentration 9.6 g/dL. All the mothers of Hb E trait babies were carriers of Hb E. The father of the baby with Hb E disease was a carrier while the mother was affected with Hb E disease. After counselling the parents, the babies were referred to our outpatient department for further management and follow up.

We tried to develop a feasible screening program in our institute, which could subsequently be adapted in all parts of the country. The use of DBS samples ensures that samples might be easily transported without any special facilities with low probability of transmitting blood borne pathogens [4,5]. The results of repeat IEF and HPLC using anticoagulated blood matched with that of IEF results performed by DBS samples, thereby proving the stability of hemoglobin in DBS. Moreover, the results of the repeat testing of the screen negative babies matched with the first screening result of IEF and that of HPLC. IEF along with the ISOSCAN software was standardized for the first time in India, since it is a reliable [6] and cost effective screening tool. In the study, IEF results matched with the HPLC results, the gold standard method widely used in India for detecting hemoglobinopathies. IEF was able to differentiate between the heterozygote and homozygote cases. One limitation of the technique in common with HPLC is the inability to identify beta thalassemia traits in neonates. For this, babies need to be screened at the age of six months or more when the switching of Hb F to Hb A is usually complete.

We conclude that implementation of a neonatal screening program for hemoglobinopathies is feasible in India.

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