Mandatory Testing for Hemoglobinopathies: Need of the Hour!

Thalassemia is the commonest monogenic disorder in the Indian subcontinent. Average carrier rate of beta thalassemia trait is 3.3% against the global frequency of 1.5%. There are 1-2 per 1000 couples at risk of having an affected offspring each year [1]. Several Mediterranean countries have successfully controlled the birth of children with thalassemia with the implementation of robust prevention programs [2]. Prevention of the disease is more important in this part of the world, where resources are limited. National Health Mission has implemented thalassemia and sickle cell disease control programs; however, these are yet to reach the grassroot levels.

An 18-month-old child presented to us with complaints of progressively increasing pallor for 6 months with history of one blood transfusion. He was born through non-consanguineous marriage, following natural conception after 18 years of primary infertility. Mother had received regular antenatal care. Parents were residents of Varanasi and belonged to Sindhi community. On examination, child was pale with frontal bossing and hepatosplenomegaly. Investigations revealed microcytic hypochromic anemia with features of hemolysis in peripheral smear. High performance liquid chromatography (HPLC) showed HbA 2.0%, HbF 90.0% and HbA2 3.7%. Possibility of homozygous beta thalassemia was considered. Parents had microcytic hypochromic anemia. HPLC of father showed: HbA 4.4%, HbF 1.5%, HbA2 0.7% and HbD 90.7%. HPLC of mother was: HbA 82.9%, HbF 1.2%, HbA2 4.6% Mutation analysis revealed that the index case was compound heterozygous with cd 8/9 (+G) and 619 bp deletion, father was compound heterozygous HbD Punjab and 619 bp deletion and mother was heterozygous cd 8/9 (+G). Genetic counseling was done and child was started on regular blood transfusions.

In this case, father was found to be a compound heterozygous for HbD and beta thalassemia (HbDβ thalassemia). He was asymptomatic and had never received a blood transfusion. It has been reported that HbDβ thalassemia can have variable presentation with some of the patients requiring blood transfusions whereas others may remain asymptomatic [3,4]. Father was diagnosed only when the offspring was diagnosed with transfusion dependent thalassemia.

This case also highlights the importance of screening for hemoglobin variants in the community or at least during the antenatal period. Birth of a child with transfusion dependent thalassemia places a huge social and financial burden not only on the family but also on the health system as a whole [5]. Here the parents were educated, and belonged to a community with high prevalence of hemoglobinopathies, yet were never screened. Although a screening program for hemoglobinopathies is in place, people tend to ignore it as it is optional. Antenatal mothers present late in their pregnancy and many times the spouse is not available for screening. Many of our colleagues in obstetrics may also not be sensitized enough to carry out the screening routinely.

We propose that India should have mandatory prenatal screening as has been done in many Middle Eastern countries to control the birth of children with thalassemia [6], otherwise we will continue to see the birth of affected babies.

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