Case Report

Hemoglobinopathy E in 4 siblings of a North Indian family: A hidden malaise of social problem

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Abstract

The preference for male child and neglect of girl child for long has resulted in a hazardously poor sex ratio, but bride buying seems to be no solution for it as it has its own inherent consequences on society and public health. This article tries to look at a relationship between a state's poorly kept secret like bride buying and its unseen complications. Here, we report a family of six members, where all 4 children have hemoglobin E disease a relatively rare entity seen in this part of the country. Hemoglobin E (β26 Glu → Lys) is usually seen in northeastern parts of India. This case report has tried to highlight a relatively rare disease afflicting all children of the family due to gene mutations which are rarely seen in Haryana/Punjab.

Keywords: Chromosomal translocation, hemoglobin E, induced abortion, sex ratio

Introduction

Hemoglobinopathy is an inherited autosomal recessive blood disorder with a wide range of clinical signs and symptoms.[1] Hemoglobin E disease patients require lifelong blood transfusions and only definitive treatment available is bone marrow transplantation.[2]

In Indian state Haryana, there had been a continuous dip in sex ratio due to relatively low status and neglect of girl child, albeit have shown an increase in recent times.[3] Because of the social milieu of Haryana and earlier practices of female feticide, eligible males remain unmarried for a long time. This gap of supply and demand of eligible females for marriage had introduced an often ignored, abominable practice of buying brides from other northeastern states. Due to a lack of awareness where only *kundli milan* was done, there was no attempt to look at the probable diseases being prevalent in those regions resulting in the inheritance of hemoglobinopathies.

Case History

A 9 years old girl came to pediatric OPD with complaints of increased pallor for 6 months and breathlessness. The consent of patient and parents was obtained and on Examination, the nasal bridge was depressed and per abdominal examination showed splenomegaly (5 cm below costal margin). Work up for anemia showed a hemolytic picture. Laboratory investigations revealed abnormal hemoglobin F and hemoglobin A2 which was 31.8% and 56.0% respectively. Hemoglobin E in the double heterozygous state was illustrated by high-performance liquid chromatography (HPLC). A similar hematological workup was conducted for the rest of the family members as making the diagnosis of hemoglobin E-β thalassemia in Haryana was a dilemma because very few cases have been reported in the literature from this region. Her sister, eleven years old also had abnormal HbF and HbA2, 18.5% and 65.5%, respectively. HPLC showed the same HbE- β double heterozygous state.

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Her brothers, 6 years old and 1-year-old also showed similar findings. All siblings have been diagnosed with HbE and they had no previous blood transfusion history. All were sharing signs of failure to thrive. On HPLC, it was later found that father and mother were carriers for β thalassemia and HbE heterozygous trait, respectively. This prompted us to make an unusual connection that because of mother's lineage all children were having HbE thalassemia. The permission was obtained from the institutional ethics committee for publishing this case report.

The usual treatment regimen was followed. Treatment was started for HbE- β thalassemia without delay for proper management and control of the disease. Hydroxyurea therapy, folic acid, and calcium supplementations for 3 months were prescribed.[4] The patient is under follow-up. Since HbE is rare in north India, lack of appropriate genetic tests makes it harder to diagnose. The difficulty encountered was in definitive subtyping of hemoglobinopathies.

The family study revealed that the mother (28 years old) is an Assamese girl. She was married to a local Haryanvi male due to the prevalent social practice of bride buying. Hemoglobin E carrier rates of up to 10.2% are found in the northeastern states of India.[5]

Discussion

The chain reaction started with a poor sex ratio is ending dangerously on social, demographic, and genetic levels now. As in the above case, all the children were HbE- β thalassemic and would require a cumbersome and costly lifelong treatment for the same. This case highlighted that the issue if left undiagnosed will become the root of more such health problems for the society in the near future. It might be one of the numerous rising hidden consequences of the bride buying and low sex ratio. This also shows the propensity of geographically absent traits to express in new environments, leading to an increase in the burden on growing state's economy and health care facilities and the need for cautious clinical and social practices.

As healthy parents are the key to a healthy family, screening the genetic makeup of parents at an early stage should be done for detection, better control, and treatment. Also, HbE disease should be taken into consideration while assessing the differential diagnosis of children with symptoms, whose parents have not been screened for the mutation and it should be made a part of primary care practice. Premarital and prenatal screening for thalassemia instead of "*kundli milan*" could be more worthwhile. Awareness against such genetic anomaly should be increased on the public level.

Despite state government incentives, additional efforts of public health individuals and policymakers are required. Proper rules against such evil malaise should be framed so that such neglected, abominable practices could come to an end.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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