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Case Report

Pediatric Patient with Hereditary Pyropoikilocytosis in India

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Abstract

Hereditary pyropoikilocytosis (HPP) is an inherited red cell membrane disorder, which is characterized by an abnormal sensitivity of the red blood cell membrane. Hereditary pyropoikilocytosis is a severe, relatively rare congenital (autosomal recessive) hemolytic anemia with extensive features like severe microcytic hypochromic anemia with acanthocytes, dacrocytes, schistocytes, microcytes, normocytes, elliptocytes. Considerable evidence indicates that assembly of the red cell skeleton is abnormal in this patient and confirmed as hereditary pyropoikilocytosis based on the laboratory findings.

Keywords: *Hereditary Pyropoikilocytosis; Acanthocytes; Dacrocytes; Schistocytes; Elliptocytes; Microcytes*

Abbreviations

HPP: Hereditary Pyropoikilocytosis; CBC: Complete Blood Count; RBC: Red Blood Cell; NICU: Neonatal Intensive Care Unit; DNA: Deoxyribose Nucleic Acid; Hb: Hemoglobin

Introduction

Hereditary pyropoikilocytosis is a rare inherited red blood cell membrane disorder in which it is characterized by strikingly abnormal red cell morphology with micro spherocytosis, aniso-poikilocytosis, schistocytosis, elliptocytosis and an unusual sensitivity of the red blood cells [1].

However the recent studies indicates that HPP is a subset of the hereditary elliptocytosis due to either homozygous or compound heterozygous mutations in spectrin leading to severe disruption of spectrin self-association [2].

The membrane skeleton that maintains erythrocyte shape and consists mainly of actin, spectrin and protein 4.1 is particularly susceptible to mechanical stress in HPP patients. In HPP patients, membrane fragility is due to molecular defect in spectrin. Spectrin, an alpha-beta heterodimer, is the most abundant component of the RBC membrane skeleton [3].

Knowledge of DNA sequence and exon/intron organization of the cloned genomic DNA encoding the alpha 1 domain of spectrin has allowed demonstration of the specific nucleotide changes, by the replacement of amino acid codons [4].

Case Report

A 2 years old male child brought to the tertiary care teaching hospital with chief complaints of increasing pallor for 1 weak and the child had history of pallor and tiredness from 3 months of life. He was given with vitamin supplements and packed blood transfusions.

At 10 months of age, diagnosed as probable hereditary pyropoikilocytosis based on the blood pictures at Christian Medical College, Vellore and advised to have regular blood transfusions and folic acid supplementation.

Child birth history was full term normal vaginal delivery at home. No history of NICU admissions and child was exclusively breast feeded for 6 months. Immunization and milestone development history are appropriate. On examination he had frontal bossing and pallor, heart rate was between 112 and 124 beats per minute (112 - 124 beats/min) and respiratory rate was between 24 and 32 beats per minute (24 - 32 beats/min). Systemic examination reveals that he had hepatomegaly, splenomegaly, right Para cardiac infiltrates was present. Complete blood test reveals that RBCs shows anisocytosis, microcytes, macrocytes, normocytes, elliptocytes, dacrocytes and schistocytes. Whenever he seems to have signs of pallor, tiredness or weakness there will be depleted levels of hemoglobin. So that he was brought to the hospital for the packed cell blood transfusion every time to regain his Hb levels. He was given the blood transfusions for every 3 months to treat his condition. Every time he was given around 2 to 3 packed cell blood transfusions to level up his hemoglobin levels there by his condition will be improved and the therapy was given like deferasirox, iron folic acid, B complex whenever he is discharging the hospital for the rest of the period. Meanwhile the blood transfusions, the serum ferritin levels are monitored to notify the range around normal values so that the blood transfusions are given accordingly. Finally he was given blood transfusion for every 3 months along with the drugs to retrieve his condition.

Discussion

However in 1973 and 1975 Zarkowsky described three patients with moderately severe congenital hemolytic anemia with unusual and bizarre RBCs with anisocytosis, poikilocytosis and micro spherocytosis. The red blood cells of these patients showed marked fragmentation, building of the cell membrane and micro spherocyte formation when exposed to a constant temperature of 45°C or above for 15 minutes. this is in contrast to the red cells of the normal people which showed these changes only at 49°C or above. He termed the condition "congenital pyropoikilocytosis" and it is related to this child based on the complete blood counts.

Since then other cases have been reported, all in black American children [Liu., *et al.* 1981; Palek., *et al*. 1979, 1981; Prchal., *et al*. 1982; Walter., *et al*. 1977; Wiley and Gill, 1974; Zarkowsky., *et al*. 1973, 1975; Zarkowsky, 1979], and more extensive studies were done to clarify the basic defect in these RBCs. Abnormal calcium leak [Wiley and Gill, 1974], abnormal ratio of membrane phospholipid to protein [Walter., *et al*, 1977], altered spectrin assembly [Liu., *et al*. 1981; Palek., *et al*. 1981] and endovesicle formation [Prchal., *et al*, 1982] have been demonstrated [5].

According to the Dipti Sidam., *et al*. in a case report on hereditary elliptocytosis, they described in detail about the HPP, this variant presents in infancy or early childhood with moderate to severe hemolytic anemia, requiring intermittent transfusion, splenomegaly, retardation of growth are present [4]. These clinical manifestations are also seen in our patient along with elliptocytosis, anisocytosis, dacrocytes, microcytes, schistocytes.

HPP is most often seen in Blacks, Whites, Arabs and also seen in under developed countries and according to the Peterson LC., *et al.* 1987 the *in vitro* studies suggests that it may confer some resistance to infection with the malarial parasite *plasmodium falciparum* [2].

Hence based on the clinical presentation and laboratory data the patient confirmed that he had HPP showing that this inherited condition is not limited to one racial group. However the child CBC indicates many changes like anisocytosis, microcytes, macrocytes, normocytes, elliptocytes, dacrocytes and schistocytes.

Conclusion

Hereditary pyropoikilocytosis is mostly asymptomatic with rare presentation as haemolytic anemia. However our patient manifested as haemolytic anemia requiring frequent blood transfusion and was diagnosed at very young age. Our patient shows many abnormalities in complete blood count, splenomegaly, hepatomegaly and based on known history the child is confirmed as HPP. The patient was discharged with strict advice to monitor and regular health checkups are to be done frequently so that he may be promptly treated and managed. The patient of these conditions need to be followed for a possible need for blood transfusions in the first few years of life.

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