CASE REPORT

G 6PD DEFICIENCY AND BETA-THALASSAEMIA IN THE SAME PATIENT

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Abstract: A report of 8 months old female child with a rare combination is presented who had inherited G6PD deficiency from father and Bela-Thalassaemia from mother.

INTRODUCTION

Haemoglobin is a conjugated protein and its heredity disorders may be classified into two broad groups, the haemoglobinpathies and the thalassaemia. At birth, Hb- Foetal accounts for from 70-90% of the total haemoglobin. It then falls rapidly to 25% at one month and 1% at six months, remaining at this level throughout life. It is elevated in some haemoglobinpathies and thalassaemia syndromes. Study done in Karachi suggest an incidence of 1.5% for Beta-Thalassaemia trait.[1] Study done at Abbottabad showed that 1.3% subjects had elevation of Hb-Foctal.[2]

Those who have a deficiency in G6PD often have haemolysis. Haemolysis in G6PD deficient subjects may occur in Favabean, infection and due to antimalarial, antibacterial and antituberculosis drugs. A study by Robert et al reported that 2.5% males and 1.3% females were found deficient in G6PD activity.[3] Another study showed that G6PD deficiency is one of the cause of neonatal Jaundice and such cases had increased neonatal deaths.[4] A study entitled "Hb-Foetal level and G6PD deficiency in suspected anaemia and their relationship" is being conducted at PMRC-Research Centre, Ayub Medical College, Abbottabad. We have so far conducted 78 cases and an unusual case was detected which is being presented here.

CASE REPORT

An 8 months' female child was presented with weakness and pallor. On physical examination she was markedly anaemic. She was referred to PMRC Research Centre, Ayub Medical College, Abbottabad for determination of haemoglobin (Hb), Hb-Foetal and G6PD. Hb was estimated by cyanmethaemoglobin method,[5] Hb-F by alkali denaturation technique[6] and sigma test kit 400 was used to determine the G6PD deficiency.[7] The result of these tests showed Hb 2.7 gm/dl, Hb-F 67% and G6PD deficiency. Her parents gave history that earlier two children were born (a female and a male), they died at the age of 3 years and 1 year respectively. Since, the child was found to have higher Hb-F and low G6PD levels, her parents were also investigated. The Hb of her 30 years old mother was 7.8 gm/dl, Hb-F 38% and had normal G6PD level. Her 33 years old father had Hb 8.3 gm/dl, Hb-F 0.9% (normal upto 1%) and G6PD deficiency. Her parents were 1st degree cousins, the mother being the daughter of her father's maternal uncle.

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DISCUSSION

The child showed an unusual combination of G6PD deficiency and high level Hb- Foetal (β_2 r^2) consistent with Beta-Thalassemia. Her father was G6PD deficient and mother was Beta-Thalassemic. These two traits are carried by genes on two different chromosomes, that for G6PD on X-chromosome and the one for β/r chains of Hb on Chromosome No. 11. Each parent had transferred its trait to the child who, therefore, inherited these disorders from the parents. The chances of survival of the child are poor because she has two inherited disorders. Her sister and brother died at a very young age, most probably due to the same disorders.

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