A CASE OF PAROXYSMAL NOCTURNAL HAEMOGLOBINURIA (PNH)

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SUMMARY

A 35 year old Chinese female with classical presentation of paroxysmal nocturnal haemoglobinuria is reported. The various aspects of these conditions are discussed.

INTRODUCTION

Paroxysmal nocturnal haemoglobinuria is a chronic haemolytic disorder due to an acquired red cell membrane defect. This disorder was described as early as 1882 by Paul Strubbing.[1] In 1928 it became recognised as a clinical entity following a case report by Marchiafava who called this condition "Chronic Haemolytic Anemia with perpetual haemosiderinuria." Michelli[1] published the condition in Marchiafava's second patient and proposed calling it 'Splenomegalic Haemolytic Anemia with haemoglobinuria – haemosiderinuria, Marchiafava type'. Hence it is now commonly called Marchiafava Michelli disease.

CASE REPORT

A 35 year old Chinese female was admitted in December 1984 with a history of passing 'sarsaparilla' coloured urine intermittently for about four months. It was noted that urine passed after awakening from sleep in the morning was darker but cleared up as the day progressed. In addition, she also noticed progressive pallor, yellowness of the eyes and easy fatiguability since the onset of her illness. Physical examination revealed a rather pale young woman with mild jaundice. There were no other significant findings noted. The haemoglobin on admission was 6.9 gm% with a reticulcyte count of 7.9%, platelet count of 218,000/ul and a white cell count of 3,100/ul with 84% neutrophils, 2% eosinophils and 13% lymphocytes. The urine sediments did not contain any red cells or white cells. Urobilinogen, haemosiderin and oxyhaemoglobin were easily detectable in the urine. Except for mild elevation of unconjugated bilirubin the liver function tests were unremarkable. Hydroxybutyrate dehydrogenase was markedly elevated at 2280 miu/ml. The acid haemolysis test performed according to the method of Ham was positive. Serum haptoglobulin was undetectable. Coomb's test was negative and bone marrow aspiration showed erythroid hyperplasia with iron deficiency and a low neutrophil alkaline phosphatase score of 10 (normal 14–100). Diagnosis was confirmed on the basis of the above findings. After transfusing her with two pints of saline washed red cells, she was discharged.

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DISCUSSION

This patient presented with a typical history and diagnosis was suspected when first seen. Investigations later confirmed the diagnosis Beal et. al.[2] listed four criteria for PNH: (i) positive Ham's test (ii) decreased level of erythrocyte acetylcholinesterase (iii) low or absent neutrophil alkaline phosphatase and, (iv) presence of haemosiderinuria. Whereas the other three are non-specific, a positive Ham's test is specific for PNH and should be routinely done in any patient with haemolytic anaemia or aplastic anaemia. In addition when both Ham and sucrose haemolysis tests are abnormal, the diagnosis of PNH is established beyond doubt (Conrad and Borton).[3]

The problem of management in patients with PNH is varied and there is no cure for the disorder. Transfusion should be given only when absolutely necessary using saline washed red cells to remove

the plasma containing complement from the red cells.

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