

Pregnancy Outcome in Homozygous Haemoglobin C

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Abstract

Haemoglobin C homozygous is a benign haemoglobinopathy, and the individual belong to the high risk group in pregnancy, because of the associated moderate anaemia. However, with multidisciplinary approach to management, the maternal and foetal outcome is good.

INTRODUCTION

Haemoglobin C disease is an autosomal recessive disorder, and it is considered one of the benign haemoglobinopathy. The disease is caused by a mutation in the beta globin gene, resulting in the substitution of glutamic acid for lysine at position 6 of the globin chain, the same site valine replacement occurs in haemoglobin S₁. Haemoglobin C primarily occurs in persons of African ancestry, but it has also been reported in persons of Hispanic and Sicilian ancestry. The prevalence of haemoglobin C reaches 40-50 % in West Africa (Burkina Faso, Ivory Coast, and Ghana). The disease is also found in Togo and Benin (20%), in individuals of African descent in the Caribbean (3.5 %) and in the USA (3 %), in North Africa (1 to 10 % in Morocco and Algeria) and in Southern Europe (Italy, Turkey)₂. Haemoglobin C disease has been characterized as a mild haemolytic anaemia with splenomegaly; the anaemia is the result of a shortened survival of the erythrocyte as well as a relative lack of erythrocyte production₃. Ferrokinetic studies in haemoglobin C disease have demonstrated an inadequate compensatory increase in erythrocyte production in the presence of anaemia₄. The mild anaemia usually is not associated with symptoms or crises as seen in other haemolytic anaemias₅. Although the heterozygous state, haemoglobin AC has not been associated with clinical abnormalities, the combination of haemoglobins S and C results in anaemia and a particularly severe clinical picture during pregnancy₅. Haemoglobin C in combinations with the normal haemoglobin A, and the abnormal haemoglobin S has been frequently encountered in obstetric practice, however, the homozygous haemoglobin C (haemoglobin C disease) is rare in pregnancy. We therefore report a case that was managed in our unit.

CASE REPORT

Mrs. JO was a 27 year old Para 0⁺² student who presented for antenatal care booking in our hospital on 2nd October, 2006.

The booking investigations were as follows: urinalysis was within the normal limits, the haemoglobin concentration was 7.7g/dl, the blood group was B Rhesus positive, and the haemoglobin genotype was CC; the retroviral screening was non-reactive.

Obstetric ultrasonography done at 20th week revealed no gross foetal anomaly, and was compatible with date.

Patient was not aware of a previously low haemoglobin concentration, and there were no symptoms or signs of anaemia, except for the buccal mucosa pallor at presentation. Mrs. JO was also not aware of her haemoglobin genotype because she never had cause to check it.

Abdominal examination revealed a uterus that was compatible with 18th week gestational age; the spleen was 8cm enlarged below the left costal margin, the liver was not palpably enlarged, and the kidneys were not ballotable.

Additional investigations to unravel the cause of the anaemia revealed no abnormality, except for the normocytic hypochromia, marked target cells on peripheral blood film, and a mildly lowered mean corpuscular volume (MCV) of 75fl.

She kept the antenatal clinic appointments, and she was on routine oral haematinics, and antimalaria prophylaxis. The haemoglobin concentration ranged between 6.7-8.0g/dl throughout pregnancy, however, she was asymptomatic. The

blood pressure and the pulse rate were within the normal limits. The regular measurement of the symphysio-fundal height was compatible with the gestational age.

She went into spontaneous labour at 40 weeks gestational age, and had spontaneous vertex delivery of a live male infant with birth weight of 4.0kg, and Apgar scores 7 and 10 at one and five minutes, respectively.

The puerperium was uneventful. Mrs. JO haemoglobin concentration at the postnatal clinic visit was 8.7g/dl. The baby was gaining weight adequately, and the immunization schedule had been commenced.

DISCUSSION

This reported case of haemoglobin C disease in pregnancy demonstrated the clinical and laboratory features of haemoglobin C disease that had been documented in the literatures.^{3,5} Haemoglobin C disease being a benign haemoglobinopathy is rarely associated with acute and severe crises that characterized sickle cell anaemia, although mild to moderate musculoskeletal pain may be present.⁴

The absence of serious acute crises may be the reason for the patient not being diagnosed until adulthood, as in the case presented that was only diagnosed in the course of routine antenatal investigations. Haemoglobin C was diagnosed using electrophoresis at an alkaline PH (8.4-8.6) using a cellulose acetate membrane as a substrate; this method had been shown to be simple, rapid, sensitive and generally satisfactory in detecting most common haemoglobin variants.⁶

Our patient experienced moderate anaemia throughout pregnancy. For a patient who had been mildly chronically anaemic pre-pregnancy, it would have been expected that the anaemia would be worsened in pregnancy as a result of the volume change that occur normally in pregnancy, and the malaria attack in the endemic zone, which is worse in primigravidae as in the case presented.⁴ Mrs. JO had sulphadoxin/pyrimethamine (as Intermittent Preventive Treatment, IPT) for malaria prophylaxis in pregnancy which reduced episodes of malaria attack; but more importantly,

the amino acid change in the haemoglobin C molecule had been shown to impair malaria growth and development.^{7,8}

In spite of the moderate anaemia, the foetal outcome in the case presented was good, and this supports the previous findings that mild to moderate anaemia may not adversely affect foetal outcome.¹⁰

Although homozygous haemoglobin C pregnancy is an high risk pregnancy, multidisciplinary approach to management ensures a good outcome, both for the mother, and the baby.

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