β-Thalassaemia intermedia masquerading as β-thalassaemia major

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DESCRIPTION
A 19-year-old man presented with dysmorphic facies, progressive fatigability and exertional breathlessness from the age of 15 years. On examination the patient had classical ‘Chipmunk facies’ (figure 1), short stature, delayed puberty and mild splenomegaly. X-ray of the skull revealed a classical ‘crew-cut’ appearance (figure 2), a hallmark of extramedullary haematopoiesis.

Haemoglobin electrophoresis of this patient revealed fetal haemoglobin (HbF) of 97% and alpha2 haemoglobin of 2.1%. However, the presentation at adolescence with no transfusion since childhood established a diagnosis of β-thalassaemia intermedia. Genetic mutation analysis revealed the IVSI 6(T→C) mutation (thymine to cysteine substitution at the 6th nucleotide position of the intervening sequence 1 of the β-globin gene). Further testing revealed positive direct Coombs test. Other aetiologies of autoimmune haemolytic anaemia were ruled out. The patient was treated with hypertransfusion and prednisolone 0.5 mg/kg on which his haemoglobin stabilised at around 10 g/dL. He is awaiting a human leucocyte antigen-matched stem cell transplant from his younger sibling who is not a carrier of the disease.

This is the first case of β-thalassaemia intermedia presenting with advanced extramedullary haematopoiesis. Differential diagnoses of ‘Chipmunk facies’ and ‘crew-cut appearance’ include β-thalassaemia major, sickle cell anaemia, very severe iron deficiency anaemia and hereditary spherocytosis. Coexistence of autoimmune haemolytic anaemia and β-thalassaemia intermedia in non-transfusion dependent patients is also limited to case reports, although alloimmunization with positive Coombs test is common in Asian patients with thalassaemia who have received multiple transfusions. Our patient also had the most common type of genetic mutation reported in north Indian patients.

Learning points
▸ Awareness of differential diagnosis of ‘Chipmunk facies’.
▸ Importance of awareness of autoimmune haemolytic anaemia in β-thalassaemia and aggressive treatment of the same to attenuate the clinical severity and relieve some degree of extramedullary haematopoiesis.

Competing interests None.

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REFERENCES