β -Thalassaemia intermedia masquerading as β -thalassaemia major

Abhijai Singh,¹ Subhash Varma²

¹Department of Internal Medicine, St Vincent Hospital, Worcester, Massachusetts, USA
²Postgraduate Institute of Medical Education and Research, Chandigarh, India

Correspondence to
 Dr Abhijai Singh,
 abhijaisingh@gmail.com

Accepted 13 November 2014

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 I 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'

Figure 2 Sagittal view of the X-ray of the skull demonstrating a 'crew-cut appearance'. Vertical striations due to extramedullary haematopoiesis give rise to this appearance.

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,¹ although alloimmunization with positive Coombs test is common in Asian patients with thalassaemia who have received multiple transfusions.² Our patient also had the most common type of genetic mutation reported in north Indian patients.³

Figure 1 Classical 'Chipmunk facies' with frontal bossing, maxillary expansion, saddle nose and depressed cranial vault.

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.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Cividalli G, Sandler SG, Yatziv S, *et al.* Beta 0-Thalassemia complicated by autoimmune hemolytic anemia. Globin synthesis during immunosuppressive therapy. *Acta Haematol* 1980;63:37–43.
- 2 Singer ST, Wu V, Mignacca R, *et al.* Alloimmunization and erythrocyte autoimmunization in transfusion dependent thalassemia patients of predominantly Asian descent. *Blood* 2000;96:3369–73.
- 3 Panja A, Ghosh TK, Basu A. Genetics of thalassemia in Indian population. *J Community Nutr Health* 2012;1:39–46.

To cite: Singh A, Varma S. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2014-207637

Copyright 2014 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow