Hair-on-end appearance in a case of thalassemia intermedia

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DESCRIPTION

The hair-on-end sign is a finding seen in the diploic space on skull radiographs and has the appearance of long, thin vertical striations of calcified spicules perpendicular to bone surface that look like hair standing on end. It is classically seen in children/adolescents with haemolytic anaemias, in particular, thalassemia major.

A 24-year-old man, born out of consanguineous marriage with history of splenectomy done at the age of four and history of requiring frequent blood transfusions, that is, every 5–6 months, came with complaints of breathlessness on exertion, generalised fatigability and weakness. On examination the patient had frontal bossing, haemolytic facies and hepatomegaly. His haemoglobin (Hb) was 5.6 g/dl and peripheral smear revealed hypochromic, microcytic red blood cells with target cells. Serum iron studies revealed serum iron of 300 mg/dl (200–400 mg/dl) and serum ferritin of 1500 ng/ml (20–300 ng/ml). Haemoglobin electrophoresis showed fetal haemoglobin F of 7 g/dl and adult haemoglobin A of 8.8 g/dl. A skeletal survey was done which revealed evidence of extra medullary haematopoiesis with a classical hair-on-end appearance seen on x-ray skull (figures 1 and 2). The patient was given three packed cell transfusions. The patient was advised tablet folic acid daily and iron chelation therapy. The patient was discharged on a Hb of 9 g/dl.

Figure 1 Hair-on-end appearance—lateral view.

Figure 2 Hair-on-end appearance—frontal view.

Cooley and Lee described skeletal changes associated with haemolytic anaemias, more than 80 years ago.¹ Hair-on-end appearance refers to the skull abnormalities seen predominantly in patients with haemolytic anaemias that is, thalassemia, major, sickle cell anaemia, pyruvate kinase deficiency—hereditary elliptocytosis and spherocytosis.² It is due to periosteal reaction with neo-osteogenesis of the outer cranial table which results in marked calvarial thickening, external displacement and thinning of the inner table. The changes are due to marrow hyperplasia. Hair-on-end appearance is also seen in congenital syphilis—syphilitic periostitis of tibia, metastatic neuroblastoma, iron-deficiency anaemia, cyanotic—right-to-left shunt—congenital heart disease, osteomyelitis, polycythaemia vera, thyroid acropachy and haemangiomas.^{3 4}

It is debatable whether the hair-on-end sign may be reversed following treatment of anaemia. The classical presentation is rarely seen these days. Hair-on-end is uncommon in the milder thalassemia intermedia. Similar appearance in facial bones is rare and suggests an extreme degree of medullary erythropoiesis. Our patient had facial involvement as well.

Learning points

- ▶ Hair-on-end appearance refers to the skull abnormalities seen predominantly in patients with haemolytic anaemia and suggests an extreme degree of medullary erythropoiesis and marrow hyperplasia.
- ▶ This classical presentation is rare now-a-days.

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