

Case Report

H SYNDROME IN A 14 YEAR OLD INDIAN GIRL

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ABSTRACT

H syndrome is an autosomal recessive multisystemic disorder characterized by cutaneous hyperpigmentation and hypertrichosis, hepatosplenomegaly, hearing loss, heart anomalies, hypogonadism, low height (short stature), hyperglycemia/insulin-dependent diabetes mellitus (IDDM), and hallux valgus/flexion contractures. A case of girl patient showing symptoms of H-syndrome has been reported here.

Keywords: H syndrome, Diabetes Mellitus Type 1, Growth retardation, Hypertrichosis

INTRODUCTION

A 14-year-old girl was presented to us in emergency department with recurrent episodes of fever, abdominal distention, hyperpigmentation and hypertrichosis of lower extremities and trunk. The patient also used hearing device because of her progressive hearing loss over the past 6 years. There was no similar history in any of her family members. Examination revealed hyperpigmented, thickened indurated plaques with marked hypertrichosis present over the extensor aspect of hands and forearm [Figure1], medial aspect of thighs, and both the feet [Figure 2].



Figure 1



Figure 2

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General physical examinations revealed moderate pallor, hepatomegaly and ascites. Sub mandibular and suboccipital lymphnodes were palpable. There was loss of sub cutaneous fat over forearms. She was short statured for her age 129cm. She also had mild scoliosis.

Routine laboratory investigations revealed haemoglobin to be 4.7 g/dl. Fasting blood sugar was 189mg% and HbA1C of 10%. Renal function test, Liver function test, ANA, Anti DS DNA, C3, C4, Anti SM Antibody, uric acid, ESR, CRP, sputum for acid fast bacilli, serum protein electrophoresis, urine protein electrophoresis were normal. Vitamin D3 was low, 49.12 (normal 81–250 nmol/L). TSH was high, 18 μ l(0.4-4.4 μ l). Serum cortisol, PTH were in the normal range. GH level was reduced to 0.76 ng/ml (normal range 1–14.4 ng/ml). Echocardiography was normal, no pericardial, or congenital anomalies were seen. Xry Thoracic spine showed scoliosis. Ultrasonography abdomen revealed hepatomegaly with moderate ascites. Mesenteric and para aortic lymphnodes were enlarged. Contrast enhanced CT scan of abdomen was done and its findings were consistent with those seen on ultrasound.

Skin biopsy taken from the affected areas of the skin showed increased melanin deposition in basal keratinocytes, irregular acanthosis. Thickened collagen bundles in the outer dermis were seen. Mild perivascular infiltrate of histiocytes was noted. Immunohistochemistry was positive for CD68 and CD45 in dermal perivascular histiocytic infiltrate and for CD34+ in vessel endothelium.

DISCUSSION

H syndrome is an autosomal recessive multisystemic disorder characterized by cutaneous hyperpigmentation and hypertrichosis, hepatosplenomegaly, hearing loss, heart anomalies, hypogonadism, low height (short stature), hyperglycemia/insulin-dependent diabetes mellitus (IDDM), and hallux valgus/flexion contractures. The disorder results from mutations in the SLC29A3 gene, encoding human equilibrative nucleoside transporter 3 (hENT3), which is a late endosomal/lysosomal nucleoside transporter [Molho-Pessach *et al.*, 2008]. The Syndrome was first described in 2008 by Molho-Pessach *et al.*, [Molho-Pessach *et al.*, 2008; Molho-Pessach *et al.*, 2008b]. Thereafter, about 100 patients of H syndrome have been described worldwide. Lymphadenopathy may be present, generalised or localised. Massive lymphadenopathy as noted in Rosai-Dorfman disease (RDD) can also be seen. Generalised lymphadenopathy was seen in this patient also. Differential diagnosis includes Torg-Winchester syndrome, hemochromatosis, POEMS syndrome and Rosai-Dorfman disease [Molho-Pessach *et al.*, 2014].

Other features seen include progressive hearing loss, short stature, Type 1 Diabetes causing hyperglycaemia, musculo-skeletal deformities, hepatosplenomegaly, and cardiac anomalies [Molho-Pessach *et al.*, 2014].

These patients present with recurrent febrile episodes, this patient had presented with fever as one of the complaints. From India approximately 10 cases have been reported so far [Mehta *et al.*, 2015].

H syndrome is a form of multi systemic histiocytosis. It has characteristic cutaneous manifestations with cutaneous hyperpigmentation, hypertrichosis, and sclerodermatous thickening [Tekin, 2015] They can have pancytopenia, reticulocytopenia, red cell aplasia, and myelofibrosis. Moderate anaemia was present in this patient. H Syndrome patients can have pancreatic exocrine deficiency and proptosis. Insulin-dependent diabetes mellitus may be the sole presentation in a few patients [Hussain *et al.*, 2009; Molho-Pessach *et al.*, 2015].

Learning Points

H syndrome is a rare disorder found in India.

Awareness about this disorder is important to avoid unnecessary repeated investigations and aggravating the agony of the patient.

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