Case Report

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A 13 year old girl with alopecia and dysplastic nails. Clouston syndrome - brief revisit

Abstract

Clouston Syndrome is named after the person who first described this condition in 1929.Clouston Syndrome is also known as Hidrotic Ectodermal Dysplasia type 2 (HED2), is a rare autosomal dominant genetic disorder and is thought to be a mutation in the GJB6 gene located on chromosome 13q12.

Keywords: clouston syndrome, alopecia, hidrotic ectodermal dysplasia, hypoplastic nails

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Introduction

Clouston Syndrome is caused by mutations in a connexin gene, GJB6 or connexin-30. The features of this syndrome are alopecia which is characterized by hair which is brittle, wiry. They have sparse eyebrows and nails which are dystrophic. This is seen most commonly in people with French Canadian Ancestry. The disease is diagnosed by physical features in childhood and genetic testing for mutation analysis. Treatment is supportive.

Case presentation

13year old white female admitted for psychiatric reasons had had deformed nails, sparse eyebrows, and hair with thicker than normal palms and soles. She reports that her father, uncle and maternal grandmother have the same condition. Her father had the same nail problems but had shaved his hair because of its brittleness and being sparse. She had been diagnosed with Clouston syndrome.

Discussion

Font

13year old white female admitted for psychiatric reasons had had deformed nails, sparse eyebrows, and hair with thicker than normal palms and soles. She reports that her father, uncle and maternal grandmother have the same condition. Her father had the same nail problems but had shaved his hair because of its brittleness and being sparse.

She had been diagnosed with Clouston syndrome. Clouston Syndrome is named after the person who first described this condition in 1929 Figure 1. Clouston Syndrome is also known as Hidrotic Ectodermal Dysplasia type 2 (HED2), is a rare autosomal dominant genetic disorder and is thought to be a mutation in the GJB6 gene located on chromosome 13q12.[1,2]

Most of the people affected by this disease are of French Canadian descent but it has been described in people of other ancestries like German, Danish, Welsh, Chinese, and Russian.[3]

This syndrome is comprised of thick ridged hypoplastic nails which are very prone to paronychia, sparse brittle hair, hyperkeratosis (thick palms and soles), and dark skin over joints eg: knees and elbows, white patches inside the mouth especially the cheeks Figure 2. In addition to these features they can have thin or absent outer eyebrows, absent or reduced eyelashes. They have normal teeth and sweat and the intelligence is normal.[1,4] Diagnosis/testing. HED2 is suspected after infancy on the basis of physical features in most affected individuals. GJB6 is the only gene known to be associated with HED2 Figure 3 it is possible to perform prenatal testing.[2,3]

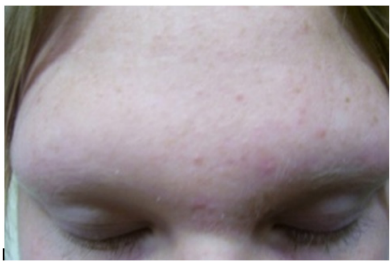

Figure 1 Eyebrows of patient.

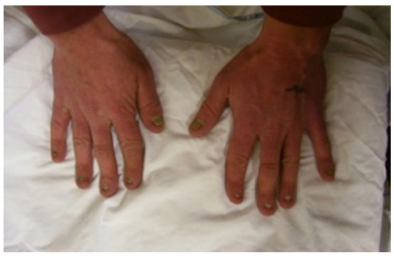

Figure 2 Fathers hands.

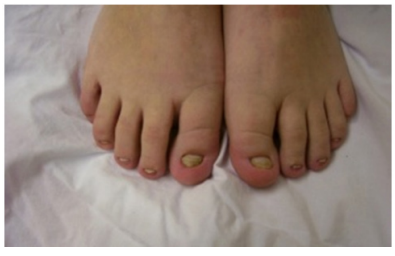

Figure 3 Feet of patient.

Management involves the use of special hair care products to help manage dry and sparse hair; wigs; artificial nails; emollients to relieve palmoplantar hyperkeratosis Figure 4.

The patients have a normal life span. Genetic counselling is offered if one parent is affected but is not common for HED2.

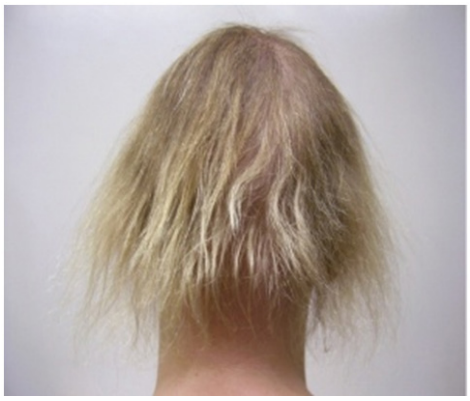

Figure 4 Hair of patient.

Acknowledgements

None.

Conflict of interest

The author declares no conflict of interest.

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