

Case Report

Hair: more than just an appendage

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ABSTRACT

Hypotrichosis is a relatively common feature of a number of complex hereditary syndromes. However, the isolated variant, called hereditary hypotrichosis simplex (HHS), is uncommon. We present a 4 years old child and his father who presented to us with features suggestive of HHS. No other associated ectodermal and systemic abnormalities were noted.

Keywords: Hypotrichosis, Hereditary hypotrichosis simplex, Ectodermal defects, Trichoscopy

INTRODUCTION

Hair defects become significant when they produce an unsightly cosmetic appearance or when they lead to the diagnosis of an underlying metabolic disorder or genetic syndrome.¹ Hereditary hypotrichosis simplex (HHS) [Online Mendelian inheritance in man (OMIM) 146520] of the scalp is a rare autosomal dominantly inherited disorder where there is sparsity of scalp hair with no abnormalities in the other ectodermal derivatives. Autosomal dominant hypotrichosis is of two types, generalized hereditary hypotrichosis simplex (hypotrichosis type 1) and hypotrichosis simplex of the scalp (hypotrichosis type 2). The former type can affect hair growth all over the body whereas the latter affects only the scalp.²

CASE REPORT

A 4 years old male child, second born of non-consanguineous marriage presented with complaints of gradual thinning of scalp hair since 6 months of age. Birth history and development history was uneventful. Child had normal hair density at birth. History of similar sparse hair in patient's father. His sibling had normal hair growth.

Haematological investigations like complete hemogram, liver function test, renal function test, serum electrolytes, urine analysis, blood glucose (random) thyroid profile and serum zinc levels were done and found to be normal. On light microscopic examination showed varying diameters of the hair shaft but was otherwise normal (Figure 3). Trichogram also showed hair of varying diameters, sparse hair with minimal atrophy of the scalp (Figure 4). Biopsy was not done as the parents were not willing for the same.

Congenital atrichia and hypotrichosis disorders, loose anagen syndrome, short anagen syndrome and hair shaft disorders were the broad differential diagnosis we considered. In view of the following features: sparse thin hair since 6 months of age, no other ectodermal or systemic defects, normal hair shaft, no significant trichoscopic finding and blood investigations being normal, a diagnosis of hereditary hypotrichosis simplex was made.

In a similar case reported by Pasricha et al, topical applications of 0.1% fluocinolone acetonide acetate ointment showed significant improvement in hair growth so the same was started for our patient.³ But the outcome could not be assessed as the patient was lost to follow-up.

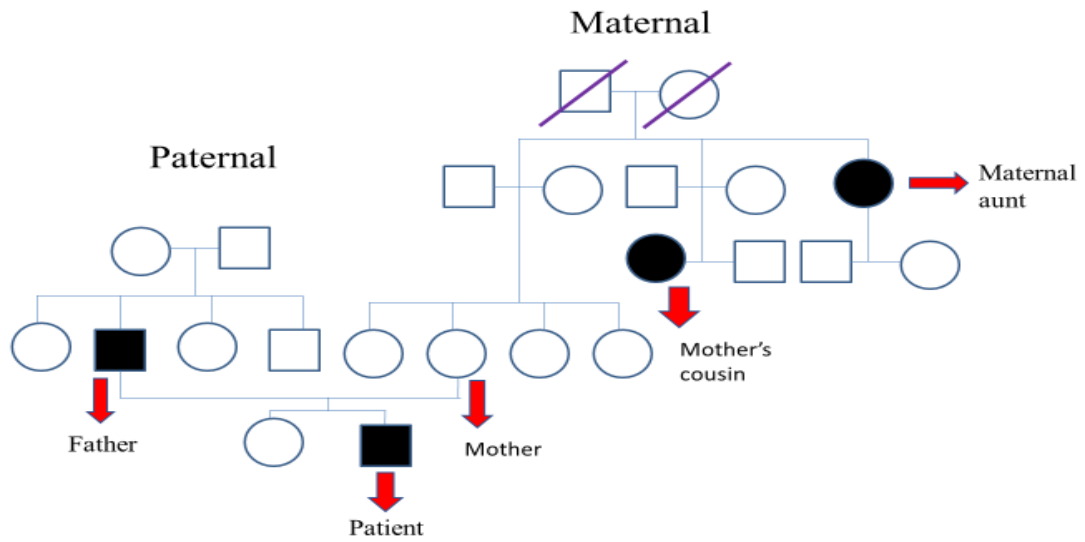


Figure 1.



Figure 2: Scalp of patient showing sparsity of hair.



Figure 3: Scalp of father showing sparsity of hair.

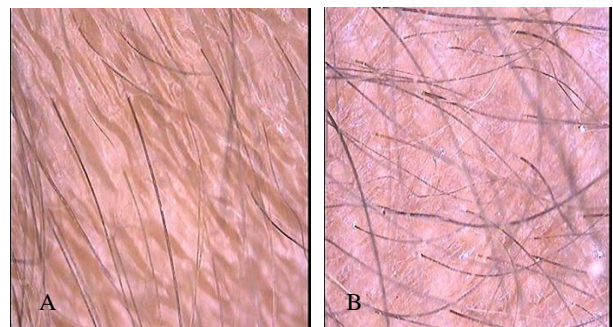


Figure 5 (A-C): Trichoscopy.

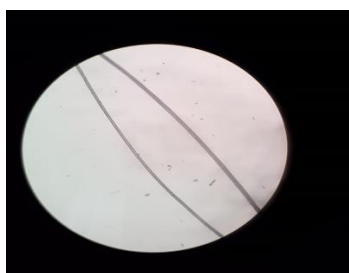


Figure 4: Light microscopy.

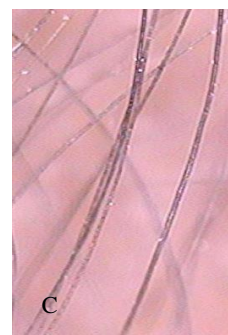


Table 1: Examination findings of father and child.

Clinical features	Father	Child
Scalp hair	Sparse	Sparse
Eyebrow hair	Normal	Sparse
Eyelashes hair	Normal	-
Skin, nails, teeth	Normal	Normal
Intelligence	Normal	Normal
Development	Normal	Normal
Hair pull test	Negative	Negative
Systemic abnormalities	None	None

DISCUSSION

Hypotrichosis represents a very heterogenous and vast group of disorders characterized by a reduced density of hair follicles. Phenotypic variability is the rule and can be striking even among affected members of the same family, which often complicates the diagnosis.⁴ Patients with HHS present with normal hair at birth and in the first year of life, progressive, gradual thinning and loss of hair begin in the middle of first decade.⁵ Mutation in APC downregulated gene 1- APCDD1 located on chromosome 18p11 or nonsense mutation CDSN gene encoding for corneodesmosin has been suggested to be responsible.⁶ Development and intelligence is normal. Scalp biopsy shows decreased number of follicles, with no specific pattern and no structural changes. Based on our literature search, only 5 families with this type of hypotrichosis have been reported.⁷

CONCLUSION

Human hair is an important phenotypic marker that can aid in the diagnosis of underlying metabolic or genetic syndromes. We report this case because of its uncommon occurrence. Although evidence-based treatment options

are mostly non-existent at this stage for most genotrichosis, a prompt diagnosis may help ruling out any underlying systemic abnormalities.

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Conflict of interest: None declared

Ethical approval: Not required

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