# Hereditary hypotrichosis simplex of the scalp: A case report with 10 affected members in a family

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## ABSTRACT

Hereditary hypotrichosis simplex of the scalp is an autosomal dominant disorder, characterized by sparse or absent scalp hairs without structural defects, in the absence of other ectodermal or systemic abnormalities. Hairs are usually normal at birth but thin progressively during childhood to become very sparse or may be absent by the third decade. Hair loss is confined only to the scalp with normal growth of facial and body hairs.

**Key words:** *Autosomal dominant, Hereditary alopecia, Hypotrichosis simplex of the scalp*

Hereditary hypotrichosis simplex (HHS) of the scalp is an autosomal dominant type of genotrichosis [1]. Patients with HSS of the scalp present with normal hairs at birth and in the 1st year of life. Gradually, the patient starts experiencing progressive hair loss, which is limited to the scalp only with the normal growth of the facial and body hairs. Normally, it begins at the latter half of the first decade, and by the third decade, there will be an almost complete loss of scalp hairs. This condition is not associated with any ectodermal defects or systemic abnormalities [2].

This condition has been largely regarded as autosomal dominant, with variable severity described within families. It affects both sexes equally. Scalp looks normal except for a reduction in follicular density. Histopathology will show vellus-like hair follicles devoid of any associated inflammation. This condition has no definite treatment. A detailed family history should be taken, and patients should be effectively counseled regarding the progression of hair loss. Herein, we report the case of an HHS in a 10-year-old girl, with nine of her family members affected with the same disorder.

## CASE REPORT

A 10-year-old girl born of a non-consanguineous marriage presented to our dermatology outpatient department with a chief complaint of sparse and short scalp hairs. She apparently had normal scalp hairs at the birth but noticed gradual hair loss and inability to grow lengthy hairs for past 1 year. She did not have any significant medical history.

On examination, she had short hairs with diffuse thinning on scalp mainly in the frontal area (Fig. 1). Follicular ostia showed no erythema or scales but were notably fewer in number. There was no evidence of scarring on the scalp. She had normal facial and body hairs. A hair pull test was negative. Oral cavity, nails, teeth, and perspiration were normal.

Complete blood count, erythrocyte sedimentation rate, liver function tests, thyroid function tests, renal function tests, serum electrolytes, urinalysis, blood glucose, serum copper, zinc and ferritin, quantitative immunoglobulins, and amino acids were normal. Trichogram revealed normal hairs with no structural defects. As the patient was not willing, a scalp biopsy could not be done.

Family history revealed gradual hair loss in her father, two paternal aunts, paternal grandmother, and paternal great-grandfather (Fig. 2). One of her paternal great aunts, her son, daughter, and one granddaughter were also affected with the same problem. All of them had normal hairs on the scalp in early childhood but started losing hair by the age of 10 years and had very sparse hair on the scalp by the third decade of their life. None of the affected family members had a history of any significant systemic illness. Her father had an almost complete absence of scalp hair by the age of 30 years (Fig. 3). Her 40-year-old paternal aunt had sparse and thin scalp hair (Fig. 4). Clinical history, family history, and examination findings pointed toward a diagnosis of HHS of the scalp. Since there is no effective treatment of HHS of the scalp, the patients were counseled regarding the disease and its consequences. They were also guided about the use of wigs or additional over-the-counter cosmetic options for the same.

## DISCUSSION

HHS is an autosomal dominant type of genotrichosis (Online Mendelian Inheritance in Man [OMIM] 146520) and was first described by Toribio and Quinones, in 1974, in a large Spanish

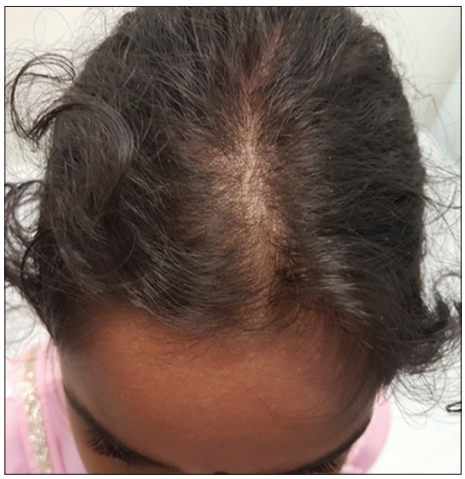

**Figure 1: Sparse and thin hair mainly on frontal area of scalp in the patient**

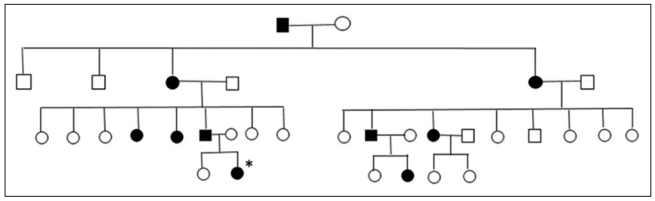

**Figure 2: Family pedigree chart. Squares represent males, circles represent females, shaded squares and circles represent affected members (*represent patient)**

family presenting with normal scalp hairs at birth followed by gradual diffuse hair loss [1]. Hair loss that usually began in school-aged children with subsequent few fine hairs remaining on the scalp by the third decade of life was identified in these individuals. Eyelashes, eyebrows, pubic, axillary, and other truncal hairs were normal [1]. HHS of the scalp is inherited in an autosomal dominant fashion, with the exception of one reported sporadic case [2]. It was an autosomal recessive variant and rare form of HHS, characterized by the sparse hairs on the scalp, and affected individuals are nearly devoid of eyebrows, eyelashes, axillary hairs, and body hairs. This disorder is associated with mutations in the CDSN gene located on 6p21.3. CDSN encodes a protein called corneodesmosin that is exclusively expressed in certified squamous epithelia [3].

Generalized forms of hypotrichosis (OMIM 605389) have long been reported and described as a loss of scalp hairs with the involvement of eyebrows, eyelashes, and other body hairs. Genetic studies have allowed for genome-wide linkage analysis, linking three families with this more generalized HHS phenotype to chromosome 18 (18p11.32-p11.23); specifically, an Italian family with sparse scalp and body hairs but normal eyelashes and eyebrows [4]. A mutation in the APC downregulated 1 gene, APCDD1, also has been identified in these families [5]. These genetic findings indicate that the generalized form of HHS is a distinct syndrome.

HHS of the scalp typically presents in school-aged children in the form of hair loss, which is localized only to the scalp [6]. Most patients are unaffected at birth and otherwise healthy without abnormalities of the nails, teeth,

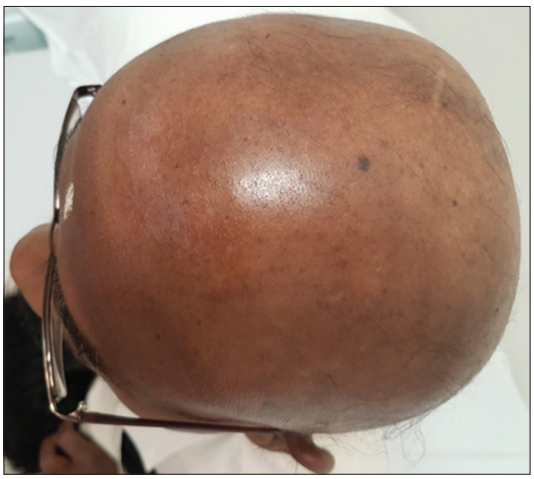

**Figure 3: Near total alopecia of scalp in patient's father**

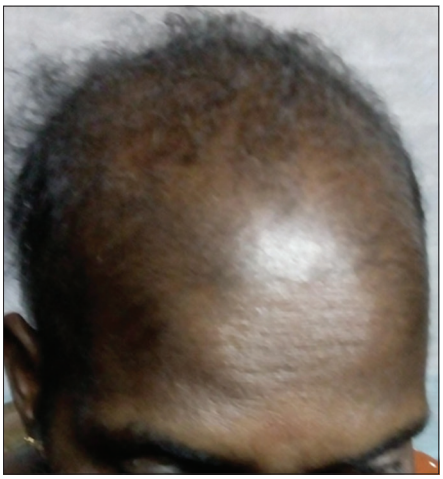

**Figure 4: Sparse and thin scalp hair in patient's paternal aunt**

or perspiration. They experience a progressive, gradual loss of hair that is limited to the scalp, beginning at the middle of the first decade and leading to almost complete loss of scalp hair by the third decade. A few sparse, fine, and short scalp hairs remain in some individuals. The body hair, beard, eyebrows, and axillary hair are normally developed. Mental development and intelligence are normal. This non-syndromic alopecia affects males and females equally. Examination of the scalp reveals normal follicular ostia and absence of scale and erythema; however, decreased follicular density may be noted [6]. The histopathologic findings of HHS are vellus-like hair follicles without associated fibrosis or inflammation [7]. Scalp skin biopsy would reveal a decreased number of follicles, especially in the telogen phase, with no specific pattern and no structural changes [8]. Examination of hair follicles with light microscopy is unremarkable [2,4].

The differential diagnosis of HHS includes Marie-Unna hereditary hypotrichosis, loose anagen hair syndrome, trichothiodystrophy, and androgenetic alopecia. Marie-Unna hereditary hypotrichosis presents as a near-complete absence of the scalp hairs at birth, development of wiry twisted hair in childhood, and progressive alopecia [2]. Loose anagen hair syndrome demonstrates a ruffled cuticle on hair pull test and remits

in late childhood. The polarization of the hair shaft can identify patients with trichothiodystrophy. Follicular miniaturization will point toward early-onset androgenetic alopecia in some patients.

HHS of the scalp in otherwise healthy individuals can occur as a separate entity or can be associated with other hair shaft defects. Different phenotypic variations have to be considered. Structural hair defects in patients with HHS of the scalp may be presented with a genetic disorder affecting hair growth or part of a congenital syndrome or may indicate underlying metabolic disorders, or may be associated with other diseases. Mutations in lipase H (LIPH) were responsible for autosomal recessive hypotrichosis simplex with woolly hair in three families [9]. Moravvej-Farshi *et al*. reported a case of HHS of the scalp associated with trichorrhexis nodosa with a history of the same disease in six members of a family [10].

There is no effective treatment of HHS of the scalp. Patients should be counseled that they may experience progressive or possible total loss of scalp hair by the third decade of life [1,2]. Wigs or additional over-the-counter cosmetic options may be considered [2]. The psychological impact of alopecia should not be overlooked, and psychiatric referral should be preferably provided. A thorough family history, as well as clinical monitoring, is recommended. Genetic counseling also may be offered [2].

## CONCLUSION

HHS of the scalp is a rare form of hypotrichosis that typically presents in school-aged children as worsening hair loss localized to the scalp. HHS of the scalp has been largely regarded as autosomal dominant, with variable severity also described within families. As there is no treatment for HHS, the affected individuals should be counseled that they will develop progressive total loss of scalp hairs.

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