

## Case Reports

### Congenital Hypotrichosis Simplex: A Case Report

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#### Abstract:

*Congenital hypotrichosis simplex of scalp is a rare disorder which only affect the scalp hair. Rest of the hair bearing areas are spared. In this disorder the scalp hair are absent or sparse. We describe a 5 month old boy presented with no growth of scalp hair since birth in this case report. After taking detailed history, a thorough physical examination was done. Other causes of alopecia were ruled out and finally the child was diagnosed as a case of Congenital Hypotrichosis Simplex (CHS).*

#### Introduction:

Congenital Hypotrichosis Simplex of scalp is a genetic disorder characterized by sparse or absent scalp hair without structural defect of the hair and absence of other systemic abnormalities<sup>1</sup>. Prevalence of the disease is <1/100000 population and male and female are equally affected<sup>2</sup>. Both autosomal dominant and recessive modes of transmission have been reported in this disorder<sup>2</sup>. In some patients the diagnosis of sporadic hypotrichosis simplex should be considered after rulling out all other possible causes of congenital hypotrichosis<sup>3</sup>. There is no abnormalities in other ectodermal derrivatives, so skin, nail and teeth remain normal in these patients<sup>4</sup>. There is no specific treatment of this disease . Due to variable expression of the disease proper counselling is of great importance<sup>5</sup>.

#### Case report:

A 5 month old boy, 2nd issue of non consanguineous parents was admitted at the Department of Paediatrics, Bangabandhu Sheikh Mujib Medical university (BSMMU) with the complaints of no hair growth since birth along with some itchy erythematous skin rashes over the scalp. His perspiration was normal and his eyebrow and nail growth were also normal. Other family members were healthy with no evidence of such type of illness.



**Fig.-1:** No scalp hair

On examination there was no scalp hair (Figure 1) but his eyebrows, eyelashes and nails were normal (Figure 2 and 3). There were some maculopaular rashes present over the scalp. The child was developmentally age appropriate. Examination of other system revealed normal findings. Provisionally he was diagnosed as a case of Congenital Hypotrichosis Symplex (CHS).

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**Fig.-2:** Normal nails in feet



**Fig.-3:** Normal nails in hands

Work out and investigations were done to rule out congenital hypothyroidism and diagnose the case which included normal T4 and TSH level. Neurometabolic diseases were also excluded as S.ammonia ,S.lactate, urinary ketone body and urinary PH were normal. Scalp biopsy was done which revealed perifollicular fibrosis and mild perifollicular infiltration of chronic inflammatory cells which were consistent with Congenital hypotrichosis simplex (CHS). Finally the baby was diagnosed as Congenital hypotrichosis simplex(CHS).

#### **Discussion:**

Congenital hypotrichosis simplex of scalp is a rare autosomal dominant genotrichosis characterized by a hair defect confined to scalp in the absence of other ectodermal and systemic abnormalities<sup>4</sup>. In this disorder other ectodermal derivatives such as eyebrows, eyelashes, nails and sweat glands remain normal<sup>4</sup>. Rafique et al. in their study showed that the genetic defect was located to chromosome 18q21.1<sup>6</sup>. Shimomura Y et al. identified a mutation in the adenomatosis polyposis down-regulated 1(APCDD1) gene in CHS<sup>7</sup>. If there is presence of strong family history the diagnosis is straight forward but in isolated cases like our one the diagnosis of sporadic hypotrichosis simplex of scalp can be done only by ruling out other possible causes of congenital and hereditary hypotrichosis.

Ectodermal dysplasia which is one of the important cause of hypotrichosis was excluded in our patient

as the eyelashes, nail and sweat glands were unaffected.

We also excluded congenital hypothyroidism and metabolic disorders where there may be hypotrichosis. Another rare congenital disorder is Marie Unna hypotrichosis where there is absence or scarcity of hair, eyebrows and eyelashes at birth, coarse and wiry hair during childhood and progressive hair loss begins around puberty<sup>8</sup>. But in our patient the eye brows and eyelashes were normal. CHS also need to be differentiated from aplasia cutis congenita, alopecia triangularis and congenital atrichia<sup>9</sup>.

In aplasia cutis congenita baby is born with absent patch of skin resembling to open wound<sup>10</sup>. In our patient the skin was normal. In patient with alopecia triangularis a well defined triangular patch of scalp is affected<sup>11</sup>. In our patient the whole scalp was affected. The congenital atrichia baby usually born with normal scalp hair but in early childhood hair is lost and never regrow again<sup>9</sup> which was also not found in our patient as he had no hair growth since birth.

We also did scalp biopsy in our patient which showed perifollicular fibrosis with perifollicular infiltration of chronic inflammatory cells consistent with CHS. In a previous case report of hereditary hypotrichosis, the scalp biopsy showed marked decrease number of hair follicles accompanied by mild chronic predominantly lymphocyte perifollicular infiltration<sup>1</sup>.

There is no effective treatment of CHS. Due to phenotypic variation patient need counselling<sup>5</sup>. We also counselled the parents about the disease during the course of admission and discharge. Besides that we prescribed an emollient for his rough skin over the scalp along with antihistamin syrup for itchy skin lesion.

#### Conclusion:

Congenital hypotrichosis simplex , a rare form of hypotrichosis has been largely regarded as a genetic disorder usually confined to the scalp. The disease can be diagnosed by excluding other possible causes of hypotrichosis. There is no specific treatment , counselling is the mainstay of management.As because it is a rare disease a good knowledge is required for early identification of the cases.

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