

Congenital Hypotrichosis Simplex

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We present a case of a seven year old female child, second in birth order. She was brought by her parents with complain of deficient scalp hair since her birth. There were no significant details in antenatal, past medical, past surgical, or family history. The parents were non-consanguineous. On general physical examination, the scalp hair was sparse (Fig 1 A, B, C) and could be shed with slight pulling. Eyebrows, eyelashes, nails, teeth, eyes and sweat glands were normal. Mental development and intelligence were normal for the age of the patient. Examination of abdomen, chest, and musculoskeletal and nervous systems did not reveal any abnormality. The parents were counselled and a diagnosis of congenital hypotrichosis simplex (CHS) was made after extensive work up and investigations, ruling out metabolic and systemic disorders. Isolated variant of CHS is a rare disorder though hereditary hypotrichosis is uncommonly reported in literature [1]. The defect affects only scalp hair and rest of hair-bearing areas are spared. The scalp hair are absent or sparse (decreased density of hair follicles per/cm²), short vellus type from birth or early childhood and thin out progressively with age. Morphologic studies of the hair usually show poor imbrications of cuticles and dysplastic bulbar structure of the anagen follicles [2]. In hereditary variants, the disease transmits as an autosomal trait with vari-

able penetration [2]. Using genetic linkage analysis, Shimomura Y et al mapped a new locus for the disease to chromosome 18p11.22, and identified a mutation (Leu9Arg) in the adenomatosis polyposis down-regulated 1 (APCDD1) gene [3]. In presence of a strong family history, diagnosis may be straight forward but in isolated cases like the one presented, the diagnosis of sporadic hypotrichosis simplex of the scalp should be made only after ruling out all other possible causes of congenital and hereditary hypotrichosis [4]. Several congenital disorders have hypotrichosis as one of the features but are generally associated with other physical, metabolic and mental disorders. These include conditions such as Graham-Little syndrome, cartilage-hair hypoplasia, Ofuji syndrome, hidrotic ectodermal dysplasia [5], Jeanselme and Rime hypotrichosis, trichorrhexis nodosa Marie Unna hypotrichosis, and metaphyseal chondrodysplasia. Congenital hypotrichosis simplex also needs to be differentiated from aplasia cutis congenita, alopecia triangularis and congenital atrichia. In aplasia cutis congenita, there is an embryonic developmental error and a baby is born with an absent patch of skin [6] resembling an open wound or an ulcer or simply with absence of hair follicles. Alopecia triangularis is similar in presentation to aplasia cutis but tends to affect a well-defined triangular patch of scalp above the temples. In congenital atrichia, a baby is born with a normal appearing scalp and hair but in early childhood, the hair are lost, never to regrow again [7].

Figure 1 : Congenital hypotrichosis simplex (A) Fronto-parietal view (B) Lateral view (C) Occipital view



ACKNOWLEDGEMENTS

The authors express gratitude to the parents of the patient who allowed the usage of the details and the images for academic purposes/publication.

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Conflict of Interest: None declared

This article has been peer reviewed.

Article Submitted on: 12th July 2014

Article Accepted on: 18th March 2015

Funding Sources: None declared

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Cite this Article: Naqash MM, Salati SA. Congenital hypotrichosis simplex. *J Pioneer Med Sci* 2015; 5(3):90-91

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