Extended Abstract

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Cornelia De Lange Syndrome with Hypertrichosis in 4-Month-Old Infant: A Case Report

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Hair may be a distinctive character of mammals and has various functions. There are approximately 100,000 to 150,000 hair follicles found within the human scalp. Skin produces hair everywhere the body with the exception of the only of the foot, palm of the hand, buccal surface of the lip, and parts of the external genitalia. We report the case of a 4-month-old Saudi girl delivered to our pediatric endocrinology service due to generalized hypertrichosis. She was noted to possess dysmorphic features and underwent laboratory investigations with resonance imaging (MRI) of the lumbosacral spine. On the idea of those findings, Cornelia de Lange syndrome was diagnosed and genetic counselling was provided to the family.

Introduction

Hair may be a distinctive character of mammals and has various functions. There are approximately 100,000 to 150,000 hair follicles found within the human scalp. In humans it's a special and defended feature, especially in females, but its main functions are in protection of the skin from mechanical injuries and to facilitate homeothermy [1,2]. Skin produces hair everywhere the body with the exception of the only of the foot, palm of the hand, buccal surface of the lip, and parts of the external genitalia; moreover, human hair growth is reduced with tiny and virtually colorless hair on most of the body surface, whereas hair is longer, thicker, and heavily pigmented in other areas, like the scalp, eyelashes, and eyebrows [3]. Here we report a case of a 4-month-old female infant presenting with generalized hypertrichosis over the entire body and located to possess multiple congenital anomalies. On further investigation she was diagnosed with Cornelia de lange syndrome.

Case Presentation

A 4-month-old Saudi girl with a known case of Tetralogy of Fallot and pulmonary atresia was delivered to our pediatric endocrinology service due to generalized hypertrichosis. She was a product of term , delivered by spontaneous vaginal delivery after a boring pregnancy, with no birth complications. She had a weight of two .5 kg at birth. case history was positive for consanguinity and her other siblings were normal. She has dysmorphic features, including bushy eyebrows, synophrys, long curled eyelashes, anteverted nostrils, a skinny upper lip, an extended philtrum, micrognathia, short neck, overlapping of second and third toes bilaterally, syndactyly, prominent antihelix, and hypertrichosis. The hair was short (2 cm), thin, and sometimes unpigmented. Additionally, she has sacral dimple. She underwent laboratory investigations as shown in Table 1. additionally , she underwent resonance imaging (MRI) of the lumbosacral spine, which showed closed posterior ectoderm defect at the extent of L5- S1

Discussion

Human body is roofed by sizable amount of hair follicles. The follicle type and size are often changed in response to several factors, specifically androgens [4]. Human hair are often categorised into three types: terminal, vellus, or intermediate. The body parts covered by hormonal dependent hair like scalp, beard, chest, axilla, and pubes consists of terminal hair which are characterised by long (2 cm), thick (60 mm in diameter), pigmented, and medullated [4,5]. Terminal hair usually extends quite 3 mm into the hypodermis. the remainder of the body in adults is roofed with vellus hair (androgen-independent hair, i.e., eyebrows and lashes), which is brief (2 cm), thin (30 mm in diameter), often unpigmented, and increasing just 1 mm into the dermis. Its primary role is to guard the skin and keep the body warm. it's related to syndromic cases like Cornelia de Lange Syndrome. Finally, some hair follicles are classified as intermediate hair, which may exist during a transitional phase between terminal and vellus forms.

One of the main classes of abnormal hair growth patterning is hypertrichosis, the term used for excessive hair growth during a nonsexual distribution which is an androgen excess.

Moreover, hypertrichosis could also be localized or generalized growth of vellus type hair and should be either acquired or congenital. this will present as an solitary finding or could also be accompanying with other abnormalities. The underlying mechanism isn't clear

Conclusion

We present a case of infant CdLS with generalized hypertrichosis that we consider of interest to extend pediatrician awareness of the differentiation between hypertrichosis and hirsutism, which are two differing types of excessive hair growth. Moreover, pediatricians should remember of the possible causes of generalized hypertrichosis.