Read Item - Congenital Hypertrichosis

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Publisher/Journal:
Date: 09/05/2000
Keywords: excess hair congenital hypertrichosis lanuginosa Coffin Siris osteochondrodysplasia X Linked Cornelia de Lange Leprechaunism Gorlin Syndrome Lissencephaly Rubinstein-Taybi Schinzel-Giedion Lawrence Seip Mucopolysaccharidoses Foetal alcohol POEMS

Abstract: Doctor's Resource on various forms of congenital general excess hair

Congenital Hypertrichoses

CONGENITAL HYPERTRICHOSIS LANUGINOSA

Introduction
This rare autosomal dominant disorder is characterised by the retention of and continued synchronised growth of foetal lanugo hair. Names such as apeman, dogman, manlion and wildman were used in older literature to describe this condition. The incidence is estimated at 1/1000 million live births.

Clinical Features and Investigation
The infants are either born with or develop within the first few months, a fur consisting of a thick coat of fine silky hair that can be up to 5 cm long and which covers the entire non-glabrous surface. The terminal scalp hairs are easily distinguished from the extensive silvery-grey body hair that only spares the palms, soles, lips, glans penis and the terminal phalanges.

During childhood the condition usually progresses, but occasionally it may regress or even stay the same. At puberty there is growth of sexual hair but without the usual conversion to terminal hair. Thus longer lanugo hairs grow in the pubic, axillary and beard areas.

There are generally no associated features, although neonatal teeth and pyloric stenosis have been described. This condition should be distinguished from hypertrichosis with gingival hyperplasia.

Management
The striking appearance of affected individuals has lead to their exploitation over the centuries as side-show freaks. Psychological support of the patient and family is an important part of the management. Shaving is often the only feasible means of controlling the hair growth at present, although the new, non q-swithched ruby lasers may prove to be effective.
Key points
A complete pelage of fine silvery hair makes this hereditary condition unmistakable.

Other Congenital Syndromes with Prominent Hypertrichosis

OTHER CONGENITAL SYNDROMES WITH PROMINENT HYPERTRICHOSIS

Hypertrichosis with gingival fibromatosis

Hypertrichosis in association with gingival hyperplasia and epilepsy was recognised as a rare autosomal dominant condition before the introduction of phenytoin . Eighty per cent of cases are familial and the condition has been associated with Cowden's disease, suggesting genetic linkage.

The hypertrichosis usually appears at birth and mainly affects the face, arms and back. During infancy it mimics congenital hypertrichosis lanuginosa, but the hair darkens with puberty. The gingival fibromatosis may not be noticed until the age of 10 years, but usually is detected earlier if the epilepsy is severe. The hypertrophy of the gums buries emerging teeth in a mass of redundant tissue. It has been suggested the phenytoin induced gingival hyperplasia relates to poor dental hygiene, however it is not known if the same holds true for familial cases.

Hypertrichosis with osteochondrodysplasia
This is an autosomal recessive condition combining generalised congenital hypertrichosis with a constellation of skeletal abnormalities that include a narrow thorax, generalised osteopaenia, hypoplastic pubic rami and premature growth arrest of the femur.

X Linked hypertrichosis
This X linked dominant condition is characterised by hypertrichosis predominantly over the face, back upper chest and pubic regions. The facial growth obscures the eyebrows and sometimes only the eyes and lips are visible beneath a dense growth of dark black hair.

Prepubertal hypertrichosis
This is a non-familial hypertrichosis, present at birth, that increases in severity during early childhood. As it occurs equally in Asians and Europeans, the term racial hypertrichosis is inappropriate. There is growth of terminal hairs on the temples spreading across the forehead, bushy eyebrows, and marked growth on the upper back and proximal limbs. In contrast to the synchronised growth of lanugo hairs in congenital hypertrichosis lanuginosa, the hair growth in this condition is unsynchronised. In adolescence this form of hypertrichosis is often confused with hirsutes. Misdiagnosis of this condition is why some so-called 'hirsute' cases fail to respond to anti-androgen therapy.

Coffin Siris Syndrome
This syndrome combines generalised congenital hypertrichosis, prominent eyebrows and eyelashes and sparse scalp hair with profound mental and growth retardation and congenital absence of the distal phalanges and nails of the fifth fingers and toes. Characteristic facies and abnormal dentition, have also been described. The inheritance of this syndrome has not yet been established.

Cornelia de Lange Syndrome
Hypertrichosis is a constant and distinctive feature of this rare syndrome. In addition to the mild generalised hypertrichosis there is abundant scalp hair with low frontal and nuchal hair lines. The eyebrows are bushy and meet in the centre and the eyelashes are long and curled. Other features are variable and include physical and mental retardation, characteristic facies, cutis marmorata, short digits, syndactyly of the second and third toes, a high arched palate, an unusual cry, increased susceptibility to infection and a constellation of ocular, dental and skeletal abnormalities.

Leprechaunism
This entity is characterised by grotesque elfin-like facies with thick large lips, large low set ears, breast enlargement and prominent genitalia. Absence of subcutaneous tissue gives rise to excessive folding of the skin. Generalised hypertrichosis is seen in three quarters of cases and is most prominent on the forehead and cheeks.

Other syndromes that produce hypertrichosis include: Gorlin Syndrome with patent ductus arteriosus, hypoplasia of the teeth, eyes and labia majora and craniofacial dysostosis; Lissencephaly with growth and mental retardation and a smooth brain without sulci and gyri; Rubinstein-Taybi Syndrome with broad thumbs, physical and mental retardation cryptorchardism and characteristic facies; Schinzel-Giedion

Syndrome with club feet, skeletal abnormalities and characteristic facies with redundant folds of skin overlying a short neck; Lawrence Seip Syndrome with congenital lipoatrophy and juvenile onset insulin-resistant diabetes, hyperlipidaemia and hepato-splenomegaly; Hunter's, Hurler's and Winchester's Mucopolysaccharidoses Syndromes; and Foetal alcohol Syndrome with mental and physical retardation, microcephaly, cardiac anomalies, joint defects, unusual facies and capillary haemangiomas.

POEMS syndrome is an acronym for Peripheral neuropathy, Organomegaly, Endocrine dysfunction, Monoclonal gammopathy and Skin changes. The main skin changes include hypertrichosis on the extensor surfaces, malar region and forehead, hyperpigmentation and oedema. Scarring alopecia on the abdomen has been described.

Waardenburg?s syndrome is associated with terminal hair on the tip of the nose and a beard that extends over the entire surface of the cheeks.