Read Item - Congenital Hypotrichosis and Atrichia

Author: RD Sinclair
Date: 16/05/2000
Publisher/Journal:
Keywords: Congenital Hypotrichosis hereditary

Abstract: Doctor's resource on Congenital Hypotrichosis and Atrichia

Congenital Hypotrichosis and Atrichia

Definition
There are several apparently distinctive genotypes, that share as a common phenotype a total and permanent absence of hair or atrichia congenita..

Congenital hypotrichosis is a less severe form of the same process, where hair is present, but is diffusely thinned. Scarring may or may not be present. It may occur as an isolated defect or as one manifestation of a number of syndromes.

Epidemiology
Rare.

Aetiology
Most cases of atrichia congenita and congenital hypotrichosis are autosomal recessive and are not associated with any other developmental abnormalities. Several autosomal dominant pedigrees have also been described in the literature with subtle differences

Pathogenesis
The hair loss is either due to follicular agenesis or programmed destruction. Loss within the first year is probably due to an abnormality that affects the unsynchronised post-natal pelage (that develops at approximately 12 months of age), but spares the first two synchronised hair malts that begin in utero at 26 weeks and 38 weeks gestation (and is complete by 12 weeks of age). The explanation for atrichia congenita developing later in childhood, the so called acquired form is unclear, but may involve two (or more) successive gene mutations akin to KnudsonÕs hypothesis for the development of retinoblastoma *.

Clinical features
In atrichia congenita the child may be either completely bald at birth; or the normal coat of lanugo hair may be present and only gradually lost. It may take up to 5 years before all this hair disappears.

In some of the autosomal recessive (AR) pedigrees of atrichia congenita, lanugo hair is normal at birth, but is soon shed and never replaced. Hair follicles are absent, but sebaceous glands are normal in number, albeit small. In other AR pedigrees the children are bald from birth.

Atrichia with Papular cysts is a variant of autosomal recessive atrichia congenita, where in addition to scalp hair loss, numerous pin-head sized, white, smooth epidermoid cysts develop around the time of puberty on the cheeks.

In autosomal dominant (AD) atrichia congenita the hair is normal until about the age of 5 years when it is replaced by short, sparse hair of a lighter colour. Diffuse thinning of the scalp hair tends to progress, such that by the late-teens very few hairs remain on the scalp .

The related condition of Madarosis is the congenital absence of eyebrows. Inheritance is autosomal dominant and within a family the scalp hair may or may not also be affected.

Atrichia congenita associated with atrophoderma vermiculata and epidermoid cysts is probably an autosomal dominant condition and histology shows scarring alopecia.

Hereditary hypotrichosis may also occur as an isolated anomaly, present either from birth or first noticed around the age of 3. The severity of the hypotrichosis is extremely variable. Most cases are sporadic, and scarring is not a prominent feature.

Investigation
A scalp biopsy may be useful to exclude alopecia areata and for prognostic reasons.

Diagnosis
The absence of ichthyosis and the normal nails, teeth and sweat glands seen in the various forms of atrichia congenita separates them from ectodermal dysplasias with atrichia or hypotrichosis . Alopecia totalis rarely occurs in the first year of life.

Prognosis
The hair loss is usually permanent.

Treatment
Wigs are the only appropriate treatment as there is no donor hair available for transplantation, and there is no medical therapy. Psychological support of the parents initially, and later the child is important.

Key Points
Total baldness, occuring either at birth or delayed for up to 10 years occurs either as part of an ectodermal dysplasia or as an isolated defect not associated with nail, teeth, sweating or other developmental defects. These isolated developmental alopecias, known as atrichia congenita exist in both autosomal dominant and autosomal recessive forms, and may rarely be associated with papular epidermoid cysts and atrophoderma vermiculata. Body hair is generally normal, but the eyebrows may be lost in Madarosis

Congenital hypotrichosis occurs as an isolated defect of variable severity and inheritance. Some hair remains in this condition.

*In 1971 Knudson proposed an hypothesis explaining the inheritance of bilateral retinoblastoma that is
now readily accepted. Development of this malignancy requires two successive gene mutations: the first is
inherited as an autosomal dominant trait in the germ line and consequently is present in all somatic cells;
the second is somatic, occurring in a retinal cell. Since the second change is a random one the
retinoblastoma may then occur in only one eye or not at all. Sporadic cases require two mutations in the
somatic cell line to initiate the tumour. As this is unlikely to happen in both eyes, such cases are always
unilateral and since the germ cells are unaffected, it is not hereditable.