Uncombable hair syndrome

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Abstract
Uncombable hair syndrome is a rare structural anomaly of the hair shaft with variable penetrance (MIM 191480). It may become first apparent from 3 months to 12 years of age. The hair is normal in quantity and is usually silvery-blond or straw-colored. It is disorderly, it stands out from the scalp, and cannot be combed flat. The underlying structural anomaly is longitudinal grooving of the hair shaft, which appears triangular or kidney-shaped in cross section. Affected individuals often have a negative family history, though the characteristic hair shaft anomaly can be demonstrated in asymptomatic family members by scanning electron microscopy. To be clinically apparent, 50 % of hairs must be affected by the structural abnormality. Spontaneous improvement often occurs in later childhood. Pili canaliculi have also been described associated with features of ectodermal dysplasia, and are a constant, though less pronounced finding, in the loose anagen hair syndrome. Autosomal-dominant inheritance is likely. The stiffness of the uncombable hair has been explained by the triangular form of the hair shaft in cross section. It has been suggested that the condition may result from premature keratinization of the inner root sheath. Diagnosis is suspected at clinical examination and confirmed through visualization of the hair shaft anomaly.

Keywords: uncombable hair syndrome, spun-glass hair, pili trianguli et canaliculi, loose anagen hair syndrome, ectodermal dysplasia, Bork syndrome, angel-shaped phalango-epiphyseal dysplasia

Disease name and synonyms

- Uncombable hair syndrome
- Spun-glass hair (Stroud and Mehregan, 1973)
- Syndrome des cheveux incoiffables (Dupré *et al.*, 1973)
- Pili trianguli et canaliculi.

Definition

Uncombable hair syndrome is a distinctive hair shaft defect characterized by disorderly hair that stands out from the scalp and cannot be combed flat. The underlying structural hair shaft anomaly is longitudinal grooving, which appears triangular or kidney-shaped in cross section.

Differential diagnosis
Clinical appearance is usually characteristic, though unruly hair may also be due to pili torti, isolated or syndromic, to twisting dystrophy (pili torti et canaliculi) observed in Marie Unna type hereditary hypotrichosis or in ectodermal dysplasias, and to wooly hair or diffuse partial wooly hair. The features in scanning electron microscopy are distinctive.

Frequency
It is an infrequent disease.

Clinical description
Uncombable hair may become first apparent from 3 months to 12 years of age. The hair is normal in quantity, and is usually silvery-blond or straw-colored. The hair stands out from the scalp, and its wild disorderly appearance totally resists any effort to control it with brush or comb. In some cases this effort leads to hair breaking, though increased fragility is not a constant feature. The abnormality usually affects all scalp hair, though localized pili trianguli et canaliculi have been described (Ravella *et al.*, 1987). Similar changes are also a constant, though less pronounced, finding in loose anagen hair syndrome (Boyer *et al.*, 1996). The eyebrows and eyelashes are normal. In later childhood a considerable degree of spontaneous improvement may occur.

Management including treatment
Treatment in children is usually not necessary and unsuccessful, although positive response to biotin has been reported (Shelley and Shelley, 1985). The use of shampoo with zinc pyrithione may improve the clinical aspect of the hair due to its moisturizing effect, and so should extensive use of hair conditioners. The disorder tends to ameliorate spontaneously with age.

Etiology
Autosomal dominant inheritance is likely. The stiffness of the uncombable hair has been explained by the triangular form of the hair shaft in cross section. It has been suggested that the condition may result from premature keratinization of the inner root sheath, which itself conforms in configuration to the abnormal outline of the hair shaft, and forms an irregular, rigid tube that then alters the shape of the emerging hair shaft.

Diagnostic methods
Diagnosis is suspected at clinical examination and confirmed through visualization of the hair shaft anomaly. Diagnosis of pili trianguli et canaliculi is difficult to make with a light microscope, though moving of the micrometer gently will allow one to appreciate the canalicular formation along the entire length of the hair. Under polarized light the hair shaft has a characteristic homogeneous band on one edge caused by the shadow thrown as light passes over the longitudinally grooved hair. Examination of cross sections of hairs embedded in paraffin can be helpful by demonstrating the triangular or kidney-shaped cross section of hair shafts (Shelley and Shelley, 1985). Scanning electron microscopy may be necessary to confirm the diagnosis.

Genetic counseling
It is usually not indicated, unless associated features of ectodermal dysplasia, or angel-shaped phalango-epiphyseal dysplasia are present, which is uncommon. Recognition of phalango-epiphyseal dysplasia is important, since this particular type of phalanx may be associated with multiple epiphyseal dypslasia, particularly of hips, with risk of secondary degenerative osteoarthritis in adulthood (Fritz and Trüeb, 2000).

Unresolved questions
Distinction between pili trianguli et canaliculi and pili torti et canaliculi
It is not clear whether the hair abnormalities in all cases were pili trianguli et canaliculi.
Associated features, such as atopic dermatitis, angioma, café-au-lait spots, alopecia, and other signs of ectodermal dysplasia, have been reported in some cases (Braun-Falco *et al.*, 1982). It is not clear whether the hair abnormalities in all cases were.
Uncombable hair associated with other clinical signs
Uncombable hair has been described in association with congenital hypotrichosis, juvenile cataract, retinal pigmentary dystrophy, oligodontia, and brachymetacarpy (Bork *et al.*, 1987). The condition was inherited as an autosomal dominant trait (Bork syndrome, MIM 191482).

- Silengo *et al.* (1993) reported an isolated case with additional findings.
- De Jong et al. (1990) reported an isolated case of uncombable hair associated with crystalline cataract.
- Fritz and Trüeb (2000) reported an isolated case of uncombable hair associated with brachydactyly due to angel-shaped phalango-epiphyseal dysplasia.

The interrelationship of uncombable hair with features of skeletal and ophthalmologic abnormalities remains to be elucidated.

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