

What Is Your Diagnosis?



A 4-year-old girl presented with a 2-year history of extremely dry hair. She had alopecia from birth to 2 years of age when hair growth began. She had normal intelligence and did not have ocular, nail, or skeletal abnormalities. Physical examination revealed thick, wavy, coarse, unmanageable hair.

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The authors report no conflict of interest.

The Diagnosis: Uncombable Hair Syndrome (Pili Trianguli et Canaliculi)

Uncombable hair syndrome is a rare hair shaft defect first described in 1973 by Dupre et al¹ who named it *cheveux incoiffables*. This condition is characterized by dry, frizzy, rough hair that cannot be combed or brushed (Figure 1).² Scalp hairs are arranged in bundles³ and the hair has a glistening appearance (spun glass hair).⁴ Usually, only scalp hair is involved; facial hair, eyebrows, and body hair are unaffected.⁴ Typically, the entire scalp is affected; however, localized involvement also has been described.⁵ The quantity of hair, rate of growth, and fragility usually are normal.³ Autosomal-dominant, autosomal-recessive, and sporadic cases have been reported. There is no gender predilection⁴ and onset may be during infancy or puberty.⁶ Alterations in combability usually present in the first year of life with the first terminal hair growth or after normal hair growth. The hair often becomes drier and lighter in color, curlier, and progressively uncombable.⁴

The pathogenesis of uncombable hair syndrome remains unclear. Amino acid and copper metabolism are thought to be normal in patients with uncombable hair syndrome.⁶ However, abnormal keratinization of the inner root sheath may cause an irregularly shaped hair shaft. Histopathologic studies have demonstrated focal asymmetry of the hair bulb and lateral loss of the hair matrix. This asymmetric matrix abnormality manifests as a longitudinal groove and may result in the clinical phenotype of these patients.⁴

Differential diagnosis of uncombable hair syndrome includes loose anagen hair syndrome; woolly hair; pili torti et canaliculi; Rapp-Hodkin ectodermal dysplasia; progeria; and ectrodactyly-ectodermal dysplasia-clefting syndrome.⁷ Diagnosis of uncombable hair syndrome is based on clinical presentation and examination of hair with both light microscopy and scanning electron microscopy. The hair shaft may be normal or have a longitudinal groove on light microscopy (Figure 2).⁶ This longitudinal groove in the hair shaft is not specific to uncombable hair syndrome and also may be seen in several types of ectodermal dysplasia.⁸ Scanning electron microscopy can demonstrate depressions along the length of the hair shaft that resemble canals (Figure 3). Definitive diagnosis of uncombable hair syndrome is based on cross-sectional examination revealing triangular-, oval-, and kidney-

bean-shaped hair shafts on scanning electron microscopy (Figure 4).⁹

Uncombable hair syndrome has been associated with abnormalities of the teeth, nails, and sweat glands.⁷ Ocular abnormalities include retinal pigmentary dystrophy and juvenile onset cataracts.⁴



Figure 1. Thick, wavy, coarse, unmanageable hair.



Figure 2. Light microscopy demonstrated grooved hair (H&E, original magnification $\times 40$).

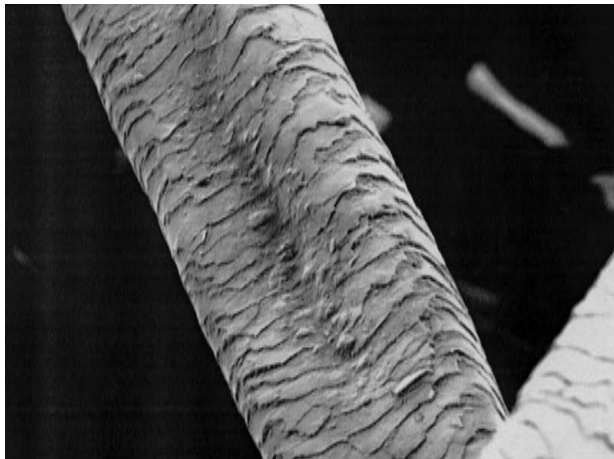


Figure 3. Scanning electron microscopy revealed longitudinal groove along the length of the hair shaft (original magnification $\times 1000$).

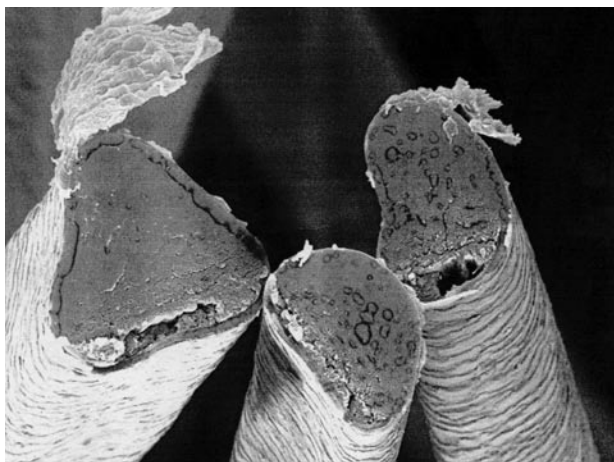


Figure 4. Cross-sectional examination revealed triangular-, oval-, and kidney bean-shaped hair shafts on scanning electron microscopy (original magnification $\times 750$).

Dental anomalies can consist of enamel hypoplasia, enamel defects, and oligodontia. Abnormalities of the digits such as syndactyly, polydactyly, and brachydactyly also can be found.⁴ Angel-shaped phalangoeipiphyseal dysplasia also has been demonstrated in a few patients.⁷

Treatment of uncombable hair syndrome is limited.⁴ However, the condition usually improves with time without treatment and may spontaneously regress. Increased hair length often improves manageability and improvement has been noted with biotin in some patients.⁴

Accurate diagnosis of uncombable hair syndrome (pili trianguli et canalculi) is essential to determine other associated conditions and syndromes and provide accurate prognostic information.

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