Self-Resolving Uncombable Hair-Like Syndrome

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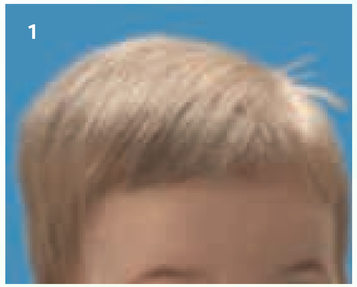
History of Present Illness: The patient presents with stiff, rough, and shiny blond hair affecting over 75% of the scalp. At birth, the patient's hair laid flat and smooth. At around age two, patient experienced diffuse hair loss without known triggers. Hair regrew with stiff and rough texture with inability to comb flat. From around age three until present, patient's hair has continued to gradually return to original state over approximately nine months. Pediatric dermatology also evaluated patient.

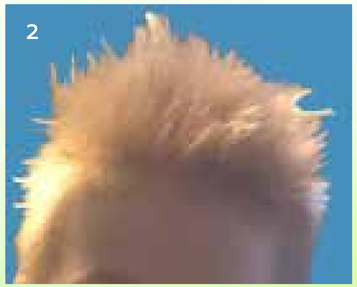
Medical History/Surgical History: Seasonal allergies

Physical Examination: Rough blond upright hair over the frontal, parietal, and crown of scalp. Negative hair pull test.

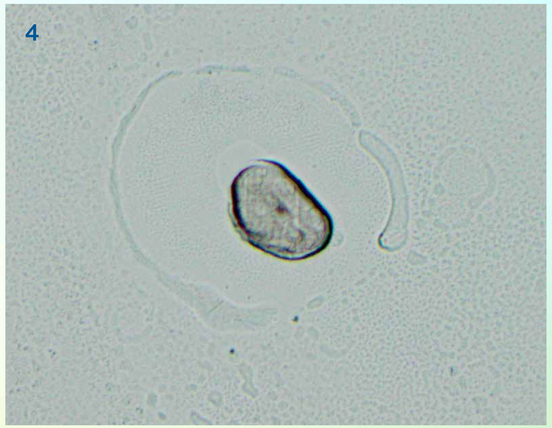
Studies: Genetic testing not obtained due to insurance coverage issue.

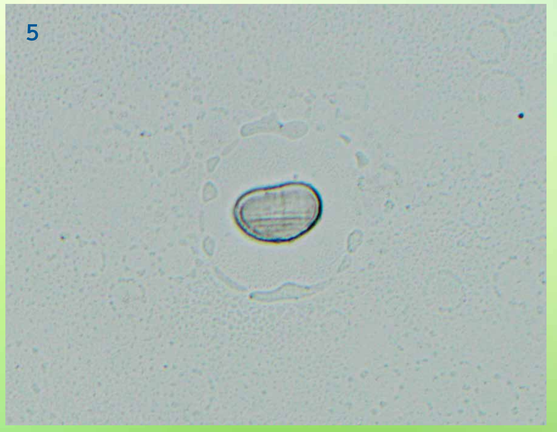
Biopsy: *Advanced Dermatology Associates* (1/03/2020) Mid scalp and right mid scalp hair snippings: Reniform-shaped hair shafts intermixed with oval and circular hair shafts.





Figures 1, 2, and 3. Age 1.5, age 2.5, and age 3.5





Figures 4 and 5. (Frozen cross-section 10x). Reniform-shaped hair shafts intermixed with oval hair shafts.

Diagnosis: Self-Resolving Uncombable Hair-Like Syndrome

Uncombable hair syndrome, also known as cheveux incoiffables, "spun-glass hair," and pili triangulate et canaliculi, is a rare structural hair abnormality. The condition was initially described in 1973 by Dupre and later by Stroud and Mehregan. In 1977, the histopathological changes were detailed in 10 patients and named pili triangulate et canaliculi.

Uncombable hair syndrome typically occurs in infancy. Blond or silver hair is frizzy and unmanageable with inability to comb hair flat on scalp. Hair may grow at a normal or decreased rate. It is suggested that misshapen dermal papillae alter the shape of the inner root sheath into a triangular form either due to premature keratinization and/or cell-cell junctional disruption of the inner root sheath.

Sporadic and familial cases have been reported. Homozygous or compound heterozygous mutations have been identified in three genes that encode hair shaft formation – peptidylarginine deiminase 3 (PADI3), transglutaminase 3 (TMG3), and trichohyalin (TCHH). More recently, a heterozygous variation in phosphoinositide-specific phospholipase C delta 1 subunit (PLCD1) has been reported.

The gold standard for diagnosis is scanning electron microscope. Recently, an easier and more cost-effective method via frozen section was reported. This new method utilizes Mohs micrographic surgery sectioning techniques. Histopathologically, there is a longitudinal groove along the entire hair shaft which corresponds to a triangular or kidney-bean shaped hair shaft on horizontal cross-section.

Uncombable hair syndrome normally resolves spontaneously and gradually around puberty. The resolution typically occurs over a period of many years. One case report described a self-resolving case that occurred within approximately nine months and without a known trigger. Management includes gentle hair care without excessive hair pulling or chemical treatment. Some have found improvement with biotin 0.3 mg orally three times daily.

Although not directly associated, it is recommended to examine patients with uncombable hair syndrome for signs of ectodermal dysplasia. Case reports have associated uncombable hair syndrome with phalangoepiphyseal dysplasia, retinal dystrophy, juvenile cataract, and bracydactyly.

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