A 4-YEAR-OLD girl had poor hair growth since birth. At birth her hair was coarse and kinky, and it fell out when she was 8 weeks old. Within months, some hair grew on her central scalp; however, she remained bald in the occipital and temporal areas for more than 1 year. By 2 years of age she had brittle hair of variable lengths throughout her scalp. Hairs were shortest in areas of greatest friction, and her mother noted that they broke easily even with gentle handling. The child required few haircuts.

With the exception of recurrent otitis media, she was otherwise healthy. There was no known family history of problems with the hair or skin. The child’s growth and development were normal. Hearing evaluations revealed no abnormal findings.

On physical examination, she seemed healthy and had blonde hair that shimmered in reflected light. Her hair was coarse, fragile, and ranged in length from approximately 1 to 7 cm. Broken hairs were most prominent in the occipital region (Figure 1). Her eyebrows were sparse. Her nails, teeth, eyes, and skin were normal. Light microscopic examination of cut hairs from the child’s scalp was performed (Figure 2 and Figure 3).

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Denouement and Discussion

**Pili Torti**

*Figure 1. The scalp hair is sparse, brittle, and irregular in length, particularly in the occipital region.*

*Figure 2 and Figure 3. On light microscopy, the hair shaft demonstrates pronounced twisting, which is typical of pili torti.*

Pili torti, a rare hair shaft abnormality first described by Ronchese, is characterized by abnormal flattening and 180° twisting of the hair around its long axis. This disorder has been classified into 4 types: classic early onset (Ronchese type), late onset (Beare type), syndrome associated, and acquired.

**CLINICAL MANIFESTATIONS**

Children with the classic type of pili torti may present with abnormal hair at birth, but more commonly they develop the hair abnormality between 2 months and 2 years of age. Affected children have fragile, poorly manageable, brittle hair that shimmers in reflected light. Eyebrows and eyelashes are frequently involved. Clinical severity is highly variable. While more severely affected individuals have diffuse stubble throughout the scalp, those with milder forms of the disorder have patchy alopecia most prominent in areas affected by friction. A small percentage of patients are so mildly affected that they seem to have normal hair.

Pili torti is most commonly inherited as an autosomal dominant trait, although autosomal recessive and sporadic transmission have been reported. Most children with pili torti are girls with blond hair that is lighter than that of their unaffected family members. The hair tends to become less fragile with age, particularly after puberty, although some patients are severely affected throughout life. While pili torti is frequently an isolated finding, associated problems such as dental abnormalities, nial dystrophy, corneal opacities, keratitis pilaris, and ichthyosis have been reported.

Late-onset pili torti was originally described by Beare. Persons with this autosomal dominantly inherited condition have alopecia of the scalp and face, and body hair that develops after puberty. Early breakage of eyebrows and eyelashes is often the presenting sign. Most patients studied have had jet-black hair and a mental deficiency.

Several syndromes have been associated with pili torti. In Bjornstad syndrome, an autosomal dominantly inherited condition, pili torti occurs in association with sensorineurial deafness. Menkes kinky-hair disease, a disorder in intestinal copper transport, is characterized by abnormal flattening and 180° twisting of the hair around its long axis. This disorder has been classified into 4 types: classic early onset (Ronchese type), late onset (Beare type), syndrome associated, and acquired.

**DIFFERENTIAL DIAGNOSIS**

Pili torti must be differentiated from other disorders that produce brittle hair. Monilethrix (beaded hair), trichothiodystrophy (sulfur-deficient hair), and trichorrhexis nodosa (noded hair) all are characterized by hair loss and brittleness. With the exception of trichothiodystrophy, which requires examination under polarized light, these conditions may be differentiated by light microscopic examination of the hair shaft. Children with the “uncombable hair syndrome” (pili trianguli et canaliculi) may have hair with a spangled appearance in reflected light similar to that of pili torti, but this condition is easily distinguished by the uncombable appearance of the hair and the normal hair density.

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