

SECTION EDITOR: WALTER W. TUNNESSEN, JR, MD

## Picture of the Month

Lisa N. Gelles, MD

**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**).

*From the Division of Dermatology, Children's Medical Center, and the Section of Dermatology, Department of Medicine, Wright State University School of Medicine, Dayton, Ohio.*



Figure 1.



Figure 2.



Figure 3.

# 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.

**P**ili torti, a rare hair shaft abnormality first described by Ronchese,<sup>1</sup> 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.<sup>2-4</sup> 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.<sup>5</sup> While pili torti is frequently an isolated finding, associated problems such as dental abnormalities, nail dystrophy, corneal opacities, keratosis pilaris, and ichthyosis have been reported.

Late-onset pili torti was originally described by Beare.<sup>6</sup> 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 sensorineural deafness. Menkes kinky-hair disease, a disorder in intestinal copper transport, is characterized by pili torti, depigmented hair, severe neurological abnormalities, bony changes, and early death. Other associations with pili torti include deafness and hypogonadism (Crandall syndrome), and basal cell carcinoma and follicular atrophoderma (Bazex syndrome). Twisted hairs have also been seen in hypohidrotic ectodermal dysplasia, pseudomonilethrix, and trichothiodystrophy in association with other characteristic findings.

Acquired pili torti may occur at the edge of areas of cicatricial alopecia in patients with discoid lupus ery-

thematosus, pseudopelade, congenital erythropoietic porphyria, and systemic sclerosis.<sup>3</sup> Pili torti has also been described in some patients treated with retinoids.<sup>7</sup>

### MICROSCOPIC FINDINGS

On light microscopic examination, the hair shaft is flattened and twisted around its long axis. Most common are 180° twists; however, 90° and 360° twists also occur. Multiple broken hairs and trichorrhexis nodosa (fractured hair with the ends stuck together resembling broomsticks) are often found. In classic pili torti, twists occur in groups of 4 or 5 and are irregularly distributed throughout the hair shaft.

### MANAGEMENT

There is no satisfactory treatment for pili torti. It is important to counsel parents of children with classic pili torti that the hair may become less fragile with age. They should also be instructed to avoid trauma to the hair. This patient's condition improved slightly after sleeping on a satin pillowcase.

### 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.

Accepted for publication September 18, 1998.

I thank Dr David Mirkin for the photography.

Reprints: Lisa N. Gelles, MD, Section of Dermatology, Wright State University, PO Box 927, Dayton, OH 45435.

### REFERENCES

1. Ronchese F. Twisted hairs (pili torti). *Arch Dermatol Syphilol.* 1932;26:98-109.
2. Laub D, Horan R, Yaffe H, Rand R, Baden H. A child with hair loss. *Arch Dermatol.* 1987;123:1071-1074.
3. Kurwa AR, Abdel-Aziz AH. Pili torti: congenital and acquired. *Acta Derm Venereol Suppl (Stockh).* 1973;53:385-392.
4. Lyon JB, Dawber RPR. A sporadic case of dystrophic pili torti. *Br J Dermatol.* 1977;96:197-198.
5. Telfer N, Cutler TP, Dawber RPR. The natural history of dystrophic pili torti. *Br J Dermatol.* 1989;120:323-325.
6. Beare JM. Congenital pilar defect showing features of pili torti. *Br J Dermatol.* 1952;64:366-372.
7. Dawber RPR. An update of hair shaft disorders. *Dermatol Clin.* 1996;14:753-772.