Pili Torti: Clinical Findings, Associated Disorders, and New Insights Into Mechanisms of Hair Twisting

Paradi Mirmirani, MD; Sara S. Samimi, MD; Eliot Mostow, MD, MPH

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The estimated time to complete this activity is 1 hour.

GOAL

To understand pili torti to better manage patients with the condition

LEARNING OBJECTIVES

Upon completion of this activity, you will be able to:

1. Distinguish pili torti from other hair shaft disorders.
2. List conditions frequently associated with pili torti.
3. Explain the pathophysiologic mechanisms that can lead to pili torti.

INTENDED AUDIENCE

This CME activity is designed for dermatologists and general practitioners.

CME Test and Instructions on page 148.

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Dr. Mirmirani is a dermatologist, The Permanente Medical Group, Kaiser Permanente Vallejo Medical Center, California; Assistant Clinical Professor, Department of Dermatology, University of California, San Francisco; and Adjunct Professor, Department of Dermatology, Case Western Reserve University, Cleveland, Ohio. Dr. Samimi was a medical student and currently is an intern, University of Pennsylvania, Philadelphia. Dr. Mostow is Head of the Dermatology Section and Professor, Northeastern Ohio Universities College of Medicine, Rootstown.
Correspondence: Paradi Mirmirani, MD, Kaiser Permanente Vallejo Medical Center, Department of Dermatology, 975 Sereno Dr, Vallejo, CA 94589 (Paradi.Mirmirani@kp.org).

Pili torti is a hair shaft disorder characterized by hair that does not grow long and is easily broken; the hair often has a coarse or spangled appearance. A diagnosis is made by light microscopy of flattened hair twisted 180° along its axis. Although pili torti may be isolated, it is commonly associated with other congenital defects and therefore, if identified, further evaluation for possible neurologic deficits and ectodermal disorders is an important part of the clinical evaluation.

Alterations of the inner root sheath likely lead to the abnormal molding and twisting of the hair shaft. More recent research suggests that these alterations may occur in the face of mitochondrial dysfunction and may be influenced by the presence of reactive oxygen species.

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Case Report

A 19-month-old boy who was otherwise healthy presented with hair that was short and brittle since birth (Figure 1). On physical examination, he had short, brown, spangled hair that was coarse and broken along the occipital and temporal rim, though it was average in density. A tug test produced short broken hair segments; a hair mount was performed and light microscopy showed hair twisted 180º along its axis, confirming the diagnosis of pili torti (Figure 2). Genetic evaluation showed no signs of any identifiable syndromes. Hearing tests revealed minor left ear deficit and follow-up was recommended.

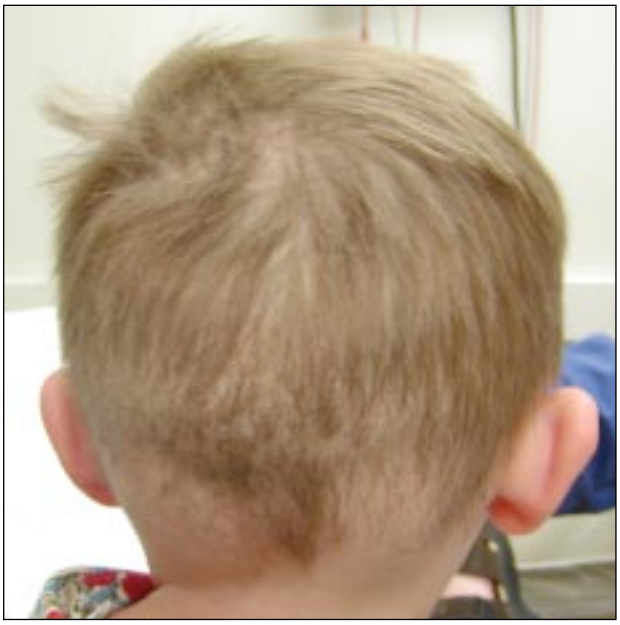

Figure 1. Short, brown, spangled hair that was coarse and broken along the occipital and temporal rim.

Figure 2. Light microscopy showing hair twisted 180° along its axis (original magnification ×10).

Comment

Pili torti (*pili* meaning hair; *torti* meaning twisted in Latin) is a condition in which the hair shaft is flattened at irregular intervals and twisted 180º along its axis, with each twist being 0.4 to 0.9 mm in width and occurring in groups of 3 to 10.[1,2] The twisted hair is brittle and easily broken.[3] Scalp hair tends to be sparse, blond, and strikingly spangled due to the unequal reflection of light from twists in the hair; patchy alopecia and coarse stubbles typically are seen in the occipital and temporal areas due to friction.[1] Pili torti also can involve the eyebrows and eyelashes. To diagnose pili torti, the hair that is easily broken off of the distal hair shaft is placed on a glass slide and viewed using light microscopy. The characteristic narrow and numerous twists distinguish this disorder from typical curly hair and from other hair shaft disorders that can cause hair fragility and breakage (Table 1). Considering that twisted hairs commonly are seen in healthy scalps and to a minor degree in other conditions of the scalp and hair, a substantial area of involvement of twisted hair should be present to invoke a true diagnosis of pili torti.[4] There is no treatment for pili torti, but it tends to improve after puberty. Recommendations to patients should include gentle hair care.

The first cases of pili torti published by Ronchese[5] in 1932 described the isolated finding of twisted hairs in 2 young sisters with blond hair. Since then, pili torti has been linked with a wide array of congenital defects, including leukonychia, dental abnormalities, keratosis pilaris, dystrophic nails, and ichthyosis.[4] A review of the Online Mendelian Inheritance in Man® (OMIM) identified 19 entries for pili torti. The more common congenital defects and syndromes associated with pili torti, including Beare syndrome,[6] Björnstad syndrome,[7] Menkes syndrome,[8,9] Rapp-Hodgkin syndrome,[10] trichodysplasia-xeroderma,[11] and trichothiodystrophy (photosensitive),[12] are discussed in Table 2

Table 1.

Hair Shaft Disorders With Fragility and Breakage

Congenital
Monilethrix (beaded hair)
Trichorrhexis invaginata (bamboo hair)
Trichorrhexis nodosa
Trichothiodystrophy
Acquired
Bubble hair
Trichorrhexis nodosa

(refer to OMIM for a more exhaustive list of disorders in which pili torti has been reported). Pili torti also can be acquired, as seen with administration of oral retinoids[13] and in cicatricial alopecia,[14] but the abnormality is localized or patchy rather than diffuse. The wide range of disorders associated with pili torti suggests that a number of pathophysiologic mechanisms can lead to the common final pathway of irregular shaping of the hair shaft. Morphologic studies of the hair shaft have shown no abnormalities of keratin in the hair cortex. Instead the twisted hair is likely due to irregularities in the inner root sheath, which may induce an uneven molding of the hair shaft.[3] In acquired pili torti, it is presumed that perifollicular fibrosis causes rotational forces that distort the hair follicle.[4]

Studies detailing the pathogenesis of pili torti and sensorineural hearing loss have shed new light on possible mechanisms of hair twisting. Although initially reported by Björnstad in 1965,[15] it was not until 2007 that a defect of the *BCS1L* gene was identified.[16] Mutations of the *BCS1L* gene affect mitochondrial respirasomes (the basic unit for respiration in human mitochondria) and lead to production of reactive oxygen species. *BCS1L* mutations previously had been reported to cause 2 pediatric syndromes with profound multisystem organ failure: mitochondrial complex III deficiency[17] and GRACILE (growth retardation, amino aciduria, cholestasis, iron overload, lactic acidosis, and early death) syndrome.[18] However, the specific mutation in *BCS1L* that leads to the highly restricted pili torti and sensorineural hearing loss results in a much more mild functional disruption of mitochondria. Björnstad syndrome seems to illustrate the exquisite sensitivity of ear and hair tissues to mitochondrial function, particularly the production of reactive oxygen species.[16]

In 1962, Menkes et al[9] described a family of 5 boys with pili torti, early growth retardation, and neurodegeneration.[8] This X-linked recessive disorder was subsequently found to be due to a mutation of the ATPase, Cu(++) transporting, alpha polypeptide gene, *ATP7A*.[19] It had been presumed that because normal copper transportation is essential to formation of disulfide bonds in hair keratin, defective keratin production was the cause of twisted hair. However, because impaired copper transportation also is known to lead to mitochondrial dysfunction and the production of reactive oxygen species,[20] the cause of pili torti in Menkes syndrome may be similar to Björnstad syndrome.

When evaluating a child with abnormal hair, a hair mount can provide a rapid diagnosis in the office. If pili torti is identified, further evaluation for neurologic deficits, ectodermal disorders, and hearing loss is an important part of the clinical evaluation. Alterations of the inner root sheath likely lead to the abnormal molding and twisting of the hair shaft. More recent research suggests that these alterations may occur in the face of mitochondrial dysfunction and may be influenced by the presence of reactive oxygen species.

Table 2.

Pili Torti and Commonly Associated Findings

Disorder	Inheritance	Associated Findings
Congenital		
Isolated		
Early onset (Ronchese)	AR, AD, sporadic	Thin, blond, fragile hair[5]
Late onset (Beare syndrome)	AD	Jet black scalp hair with sparse body and beard hair, development of patchy alopecia after puberty[6]
Associated With Neurologic Changes		
Björnstad syndrome	AR, AD	Sensorineural hearing loss and pili torti[7]
Menkes syndrome	X-linked recessive	Early growth retardation, abnormal hair, and focal cerebral and cerebellar degeneration[8,9]
Associated With Ectodermal Dysplasia		
Rapp-Hodgkin syndrome	AD (variable expression), sporadic	Anhidrotic ectodermal dysplasia, cleft lip, cleft palate, and pili torti; hypodontia, fingernail abnormalities, and hypospadias also are features in males[10]
Trichodysplasia-xeroderma	AD	Dry skin and abnormal hair findings, including alopecia, hypotrichosis, pili torti, and trichorrhexis nodosa[11]
Trichothiodystrophy (photosensitive)	AR	Ichthyotic skin, physical and mental retardation, brittle hair (including pili torti), photosensitivity[12]
Ronchese variants	AR, AD, sporadic	Early onset pili torti and other findings that may include leukonychia, dental abnormalities, keratosis pilaris, dystrophic nails, and ichthyosis[4,5]
Acquired		
Oral retinoids		Dry skin and pili torti; resolution with cessation of medication[13]
Cicatricial alopecia		Residual hairs at the center of a scarred plaque become twisted[14]

Abbreviations: AR, autosomal recessive; AD, autosomal dominant.

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