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PILI TORTI—CONGENITAL AND ACQUIRED

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Abstract. Pili torti or twisted hairs is considered a rare, congenital, probably inherited disorder of the hair follicle. It may be associated with other ectodermal and neurological abnormalities. Acquired pili torti as a result of cicatricial alopecia has not been recognised. The purpose of this paper is to report five cases of congenital pili torti and four cases of acquired pili torti seen by the authors. An attempt is made to classify the various distinct clinical syndromes in which pili torti may be found.

Pili torti is a rare abnormality of the hair. Clinically, it has a peculiar frizzy character (small tight curls) and is abnormally fragile and consequently broken off or short. Microscopically it is twisted about its own axis in a varying degree, from 90° to 360°. A certain twisting of the hair around its own axis is occasionally found in normal hair (7). The twisting of hair in pili torti may resemble that of monilethrix when viewed in a two-, but not in a three-dimensional plane (3, 18) and some cases diagnosed monilethrix were in fact pili torti (6).

Pili torti was first noted by Schutz in 1900 (21) who referred to it in a discussion on monilethrix. Ronchese in 1932 (18) and Galewsky in 1932 (10) independently and at the same time described the condition more fully and applied to it the terms pili torti (twisted hair) and trichotortosis. Since then, some eighty cases have been described in the world literature and various associated abnormalities have been reported. Ectodermal dysplasias associated with pili torti were stressed, such as widely spaced teeth and enamel hypoplasia (3), follicular hyperkeratosis (11), ichthyosis (2, 8, 13, 20, 24) and dystrophic nails and corneal opacities (9). Beare (4) described a variant of the condition associated with mental deficiency. Menkes et al. (14) and Aguilar et al. (1) reported retardation of growth and focal cerebral and cerebellar degeneration in association with pili torti and used the term "kinky hair disease". Pollitt et al. (15) reported sibs with hair shaft changes in the form of pili torti and trichorrhexis nodosa and who had gross mental retardation and low cystine content of the hair shaft, but normal levels of amino acids in both plasma and urine. More recently, sensory neural hearing loss has been reported (5, 16, 17).

Our intention is to add 5 new cases of pili torti to the literature, to emphasize the occurrence of acquired pili torti in cicatricial alopecia, and to summarize the four clinical conditions in which pili torti has been found.

CASE REPORTS

1. *C.M.* (Figs. 1 and 2). A girl of 3 was referred to the Bridgend General Hospital (South Wales) in 1967. She had had abnormal hair since birth. Her hair did not grow longer than 6 cm, and was dry, straight and flat, and unmanageable. She was a twin; her birth weight was 3 lb 12 oz. She started teething at the age of 10 months, and walking at 13 months. Apart from being of small stature, her development had been normal. Her binovular twin sister had normal hair. Both sisters were fair-skinned, and became easily sunburnt. There was no consanguinity in the family and there was no family history of abnormality involving hair, nails or teeth, nor mental defect or deafness.

On examination she was a fair-skinned girl with blond, fine, dry and short scalp hair. In reflected light the hairs had a peculiar spangled glint. There were no bald patches. Eyebrows, eyelashes and hair on the limbs were normal. Teeth and nails were normal. There was no hearing defect and the child was mentally quite alert. Microscopic examination of the hair shaft showed sudden twists through 180° at irregular intervals (Fig. 3). Urinary aminoacid chromatogram was normal. Urinary porphyrins were present in normal amounts and phenylalanine was not detected. Her twin sister and mother were examined and found to have no abnormal hairs.

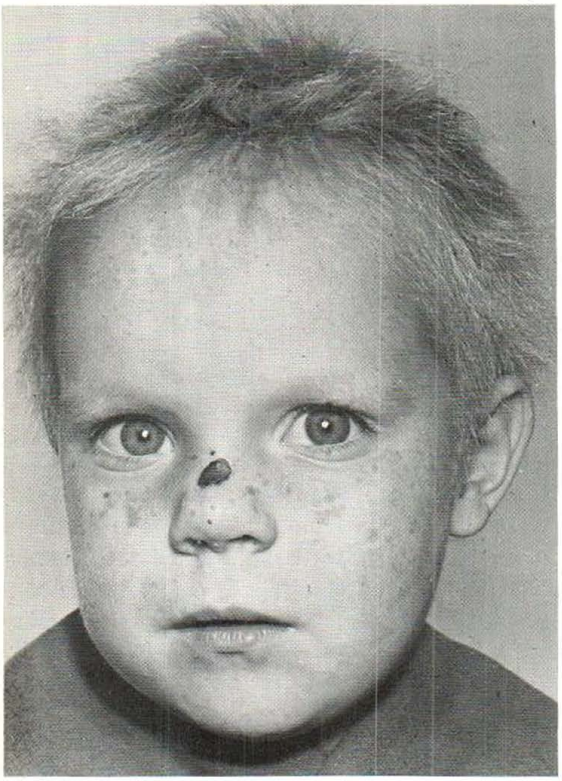

Fig. 1

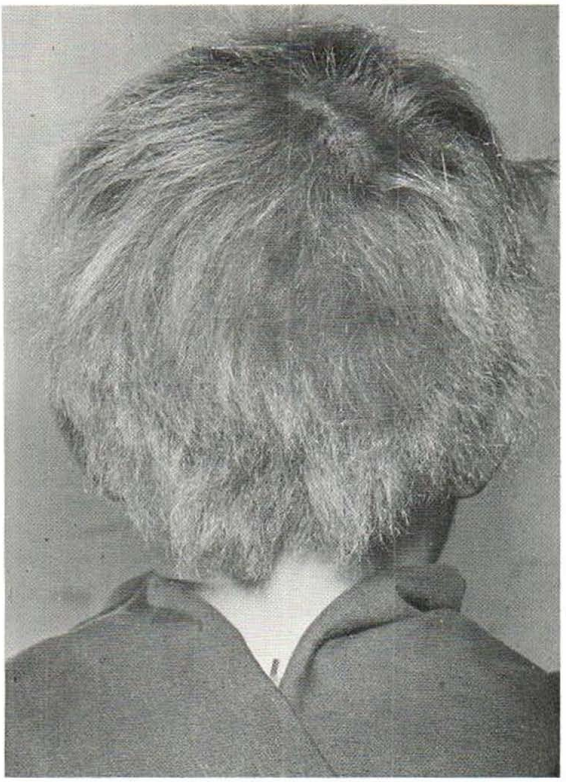

Fig. 2

Figs. 1 and 2 (Case 1). Pili torti (Ronchese type) showing thin blond frizzy hair.

2. *S. M.* A girl of 9 was referred to El-Hussein University Hospital in Cairo, Egypt, in 1970 complaining of extreme shortness and brittleness of the hair of the scalp. There was no family history of abnormality concerning hair, nails, or teeth, nor any mental defect or deafness. Also, there was no consanguinity in the family.

On examination, the scalp was covered with black hair which was short, dry thin, sparse and clung closely to the scalp. The length of the hair did not exceed 4 cm. Eyebrows, eyelashes and hair on the limbs were normal. Teeth and nails were normal and examination of other system showed no abnormality.

Microscopic examination of the hair shaft showed the hairs to be twisted on the long axis of the shaft at irregular intervals (Fig. 4). The hair shaft between the twists were normal in appearance. Trichorrhexis nodosa (Fig. 5) was also present. Urinary aminoacid chromatogram was normal.

3. *R. M.* A man of 23 was referred to the Cardiff Royal Infirmary (South Wales) in 1966. He had noticed a patch of baldness in the scalp for 6 months. Scalp hair had been short since childhood, but he had never seen patches of baldness. He shaved infrequently, but did not have any abnormality of nails or teeth (Figs. 6 and 7). Examination showed diffuse thinning of the scalp hair with an area of alopecia on the vertex and occipital region. The skin of the scalp showed slight atrophy at the site of alopecia. The rest of the scalp was covered with thick jet-black hair, broken at various lengths from the

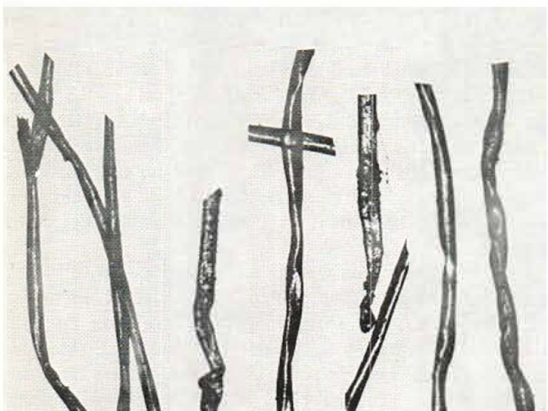

Fig. 3. Microscopical examination of the hairs from 5 patients, showing varying degrees of twists.

root. Gross abnormality of the hair shafts was evident by their spangled appearance in reflected light. Eyebrows and eyelashes were very sparse. Beard hairs were only present on the lips and chin and the rest of the beard skin felt smooth. There was complete loss of hair in the axillae and on arms and legs. Nails, teeth, and examination of other systems showed no abnormality. Microscopic examination (Fig. 8) showed the hair shafts to be flattened and twisted. At irregular intervals it was seen to have a number of closely-set twists. Mechanical manipulation showed a tendency for the shaft to fracture at the sites of multiple twists (Trichoptilosis). Some of the hairs pulled out were in the anagen phase. The hair root with root sheath still attached to it showed the twists inside the follicle.

4. *N. N.* Sister of R. M. aged 28, was called to be examined as she was known to have alopecia and hair abnormality. She was found to have areas of alopecia affecting the frontal and occipital regions. She had had wiry unmanageable hair since childhood. The scalp hair showed the same clinical features as her brother, being thick, jet-black and wiry, and in reflected light showed a spangled appearance. Some of the hairs were broken off short. There were no eyebrows and very few eyelashes; there was neither body hair nor axillary hair. A few wiry black pubic hairs were present. Teeth and nails were normal. Examination of other systems showed no abnormality. Microscopic examination of hair from the scalp showed changes similar to those of her brother. One of the pubic hairs examined microscopically also showed twists of pili torti. Both patients were investigated for aminoaciduria but no abnormality was detected on urinary chromatography. The family history revealed abnormal hair and patchy alopecia in several members (Fig. 9). She also has a mongol child with normal hair.

5. *B. B.* A woman aged 33 who had an area of alopecia on the vertex of her scalp since childhood, but this had become more marked since the age of 14. On examination she was found to have ichthyosiform erythroderma with an aera of alopecia on the vertex. Large numbers of hairs surrounding the area of alopecia were shorter, darker and coarser than the rest of the scalp hair. A few hairs examined microscopically showed multiple twists through 180° (Fig. 3). Some of the hair roots with root sheath still attached showed sudden twists at the proximal end. There was no family history of ichthyosis or hair abnormality.

Recently, changes of pili torti were noted in the presence of cicatricial alopecia. The following cases demonstrate such presence (Fig. 3).

6. *A. F.* A woman aged 58 who has systemic lupus erythematosus with discoid lesions on the scalp and right cheek. The scalp lesion has produced an area of cicatricial alopecia on the vertex. Some of the hairs surrounding the area of alopecia were short, thick, and wiry. Microscopic examination of these hairs showed similar changes of pili torti with flattening of the shaft and twists through 180°.

7. *D. M.* A woman aged 72, who had patchy areas of cicatricial alopecia on the scalp. On the vertex was a large area of alopecia with a few hairs still present in the centre. A clinical diagnosis of pseudo-pelade was

Fig. 4. Pili torti. Case 2 showing multiple close-set twist and twists at the proximal end of hair.

conformed by biopsy. The hairs in the centre of th alopecia and some surrounding the cicatrices showe clinical features of pili torti. Microscopic examinatio showed twists of the hair shaft. There was no loss of ha elsewhere.

8. *M. W.* A girl aged 19, a case of congenital erythr poietic porphyria (12) had extensive areas of cicatrici alopecia as a result of chronic blistering. The skin the scalp showed coarse, wavy features of pili torti ar microscopic examination showed the typical twists of th shaft.

9. *J. I.* A woman aged 54, attending the Radcliffe I firmary (Oxford, England) since 1962. She has sy temic sclerosis with changes in the skin, affecting h hands, face and scalp. The scalp shows cicatricial alopec with a few hairs still present. The existing hair sho changes of pili torti. Some of the hairs examined und the microscope showed flattening of the shaft and mu tiple twists through varying degrees.

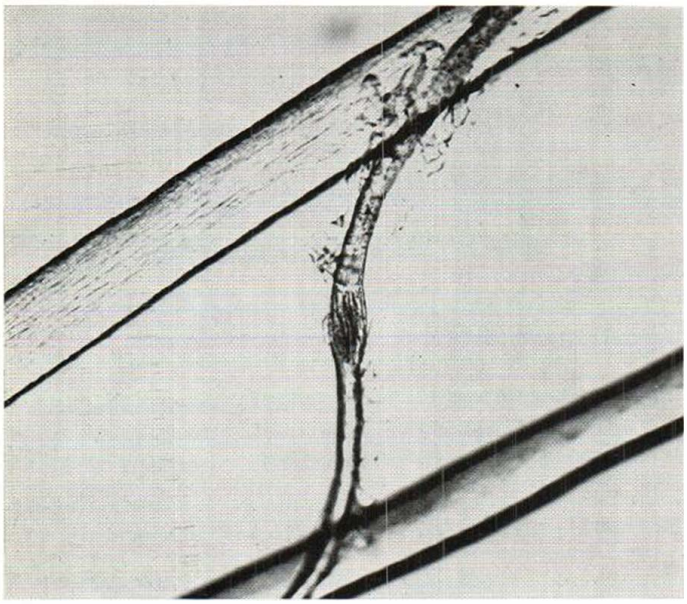

Fig. 5. Trichorrhexis nodosa. Case 2.

COMMENT

In Table I an attempt is made to classify the four distinct clinical syndromes in which pili torti may be found. Pili torti may thus manifest itself either as a developmental abnormality or as an end result of diseases which may alter the structure or function of the hair follicle. The first two cases show the classical features of pili torti as described by Ronchese (18) and Hellier (11). This is a rare developmental defect occurring usually in female children and manifesting itself early in life. The majority of affected children have thin blond hair; when only a part of the scalp is affected, these hairs are of lighter colour than the adjacent normal hair. Intelligence and mental development are usually normal. Associated abnormalities may include pityriasis capitis, enamel hypoplasia with widely spaced teeth (3), nail dystrophy and corneal opacities (9). These associated abnormalities suggest the defect to be purely ectodermal.

The association of pili torti and ichthyosis has been identified and reported (2, 8, 13, 20, 24). This has been shown in our fifth case in which she has ichthyosiform erythroderma and pili torti. The latter was most prominent on the hair surrounding the area of alopecia on the vertex of the scalp. Such a hair defect may be missed entirely unless the examination of the plucked hair is carried out.

A variant of the above was first noted by Ullmo (23) and its features stressed by Beare (4). The inheritance seems to be that of an autosomal dominant type and the abnormality can be detected in several generations of the family with about equal incidence in males and females. Whereas the Ronchese type presents in infancy as diffuse alopecia and tends to improve by the onset of puberty, Beare's type tends to manifest itself as patchy alopecia after puberty, although the hair abnormality may have existed since early life. Ullmo's and Beare's patients had thick jet-black hair with absence or sparcity of eyelashes, eyebrows and body hair. All these features were shown by N. N. and R. M. (cases 3 and 4) and were reported to be present in several members of the family (Fig. 9). These patients also show mental subnormality although it was not obvious in our patients.

The third type of defect is associated with retardation of growth and neurological abnormalities such as nerve deafness (5, 16, 17) and cerebral and cerebellar degeneration (1, 14). In this type, the hair is kinky and dark from birth and later tends to become lighter in colour. In these children the hairs showed a microscopic picture of a combination of pili torti, monilethrix and

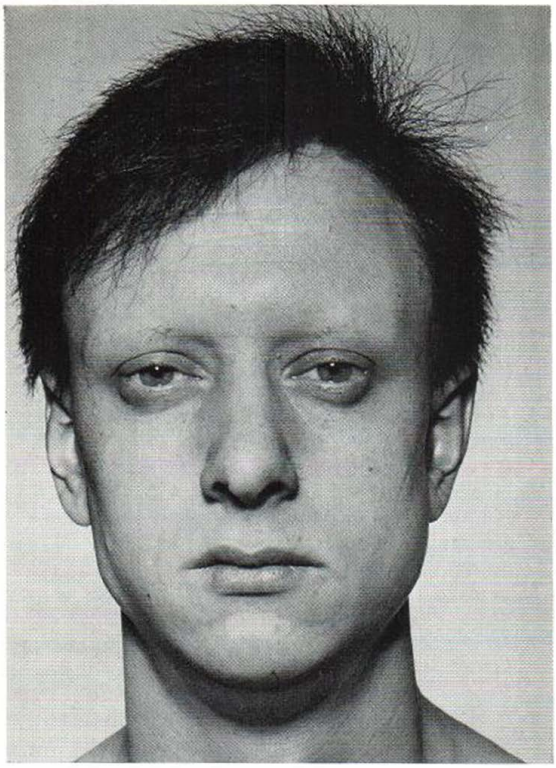

Fig. 6 (Case 3). Pili torti (Beare type). Absence of eyebrows and eyelashes.

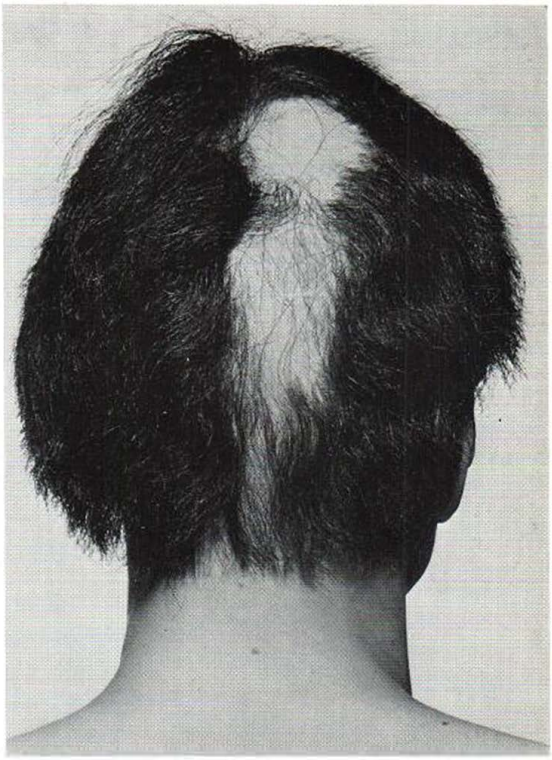

Fig. 7 (Case 3). Pili torti (Beare type). Patchy alopecia with thick black hair showing spangled appearance in reflected light.

trichorrhexis nodosa. The inheritance in these cases seemed to be sex-linked and recessive with appearances of the disease in male siblings. Some of these children have shown aminoaciduria suggesting the existence of a metabolic defect.

The last 4 patients demonstrate the acquired form of pili torti. Schlammadinger's patient (19) developed pili torti after a permanent wave, but he considered it as a congenital tendency to pili torti made more prominent by a permanent wave.

Table I. *Pili torti—congenital and acquired*

	Congenital			
	Ronchese (1932)	Beare (1952)	Monkes et al. (1962)	Acquired
Inheritance	Recessive	Autosomal Dominant	Recessive sex-linked	
Age of onset	Infancy	After puberty	Infancy	Adults
Sex	Female	Male or Female	Male	Female
Scalp	Blond hair with diffuse thinning	Thick black hair with patchy alopecia	Sparse kinky hair	Patchy involvement
Other associated features	1. Keratosis pilaris 2. Widely spaced teeth 3. Enamel hypoplasia 4. Nail dystrophy 5. Corneal opacity 6. Ichthyosis	1. Loss of body hair 2. Mental retardation	1. Retarded growth 2. Nerve deafness 3. Cerebral degeneration 4. Monilethrix 5. Trichorrhexis nodosa	Features of primary disease

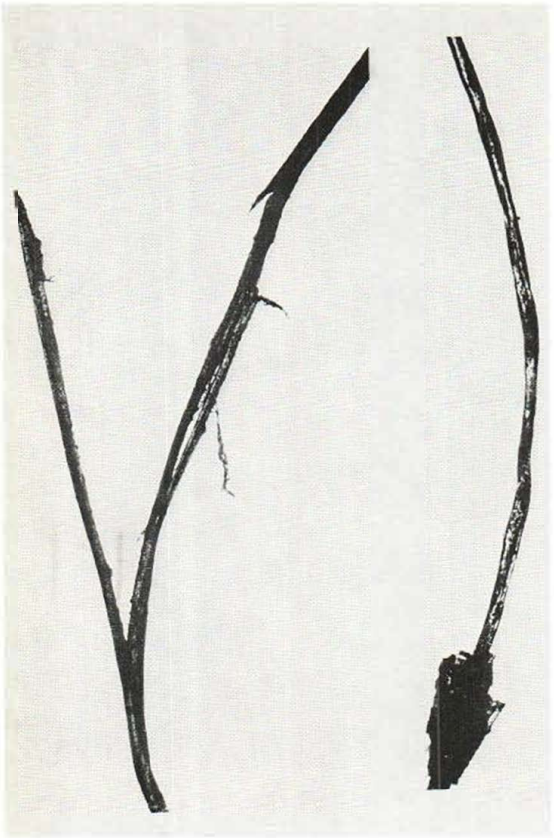

Fig. 8. Microscopic examination of the hair from Cases 3 and 4 showing multiple twists and trichoptilosis.

Scott (22) also reported a patient who developed an area of pili torti after infection of the scalp. This change appears to be secondary to the cicatricial process and may result from distortion of the hair follicle. The acquired form of pili torti is probably a common occurrence but is often unrecognised.

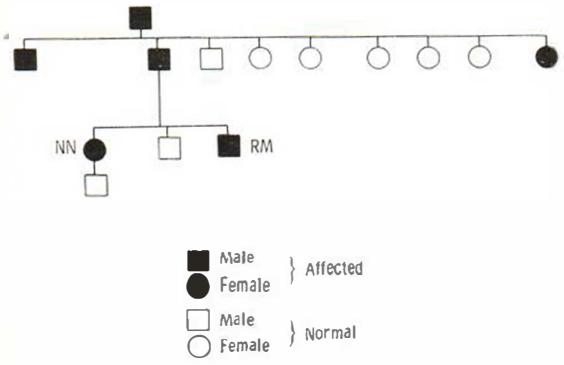

Fig. 9. Pili torti (Beare type)—Family chart.

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