A Point to Note in Pili Torti

Sir,

An 18-year-old male patient born of consanguineous marriage presented with dry, lustreless, fragile, brittle, and sparse hairs involving scalp and eyebrows. There was a history of decreased hearing in the patient. His two younger siblings gave a similar history of hair loss and hearing defect. There was no history of delayed milestones. Light microscopy of hairs revealed flattened hair shaft with a twist at an angle of 180°. His hormonal profile, i.e., growth hormone, luteinizing hormone, and serum testosterone were within normal limits. Serum copper and ceruloplasmin levels were normal. Pure tone audiometry (PTA) and brainstem evoked response audiometry (BERA) revealed bilateral moderate to severe sensory neural deafness.

Pili torti is a rare, congenital or acquired autosomal dominant disease, clinically presented as dry, lustreless, fragile, and brittle hairs\(^{[1]}\) and on scalp hair microscopy as flattened hair shaft at irregular intervals and twisted at 180° along its axis. The first case of pili torti was reported in two young blond-haired sisters by Ronchese in 1932.\(^{[1]}\) The condition may be associated with sensory neural deafness.\(^{[2]}\)

An 18-year-old male patient, born of a consanguineous marriage had presented with thinning and loss of hair from scalp and eyebrows since birth. The associated complaints include decreased hearing since 2 years of age. There was a similar history of the hearing defect and hair loss in his younger siblings. There was no history of delayed milestones. Clinical examination showed dry, lustreless, fragile, and brittle hair of scalp and sparse hairs involving eyebrows [Figure 1].

Light microscopy of hairs revealed flattened hair shaft with a twist at an angle of 180° [Figure 2]. Blood investigations revealed growth hormone level to be 0.32 ng/dl, luteinizing

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**Figure 1:** Sparse, dry, lustreless, fragile, and brittle hair involving scalp

**Figure 2:** Pili torti - flattened hair shaft twisted at an angle of 180°
hormone to be 4.07 mIU/ml and serum testosterone to be 724.02 ng/dl. The hormonal essay was within normal limit. Serum copper and ceruloplasmin levels were normal. PTA showed moderate to a severe mixed hearing loss in the right and left ear [Figure 3] and BERA revealed moderate to severe sensory neural deafness [Figure 4].

Figure 3: Pure tone audiometry - moderate to severe mixed hearing loss in right and left ear

Figure 4: Brainstem evoked response audiometry
In our case, since the patient had sensory neural deafness and pili torti, the differential diagnosis were Björnstad syndrome and Crandall syndrome. Crandall syndrome is characterized by pili torti, sensory-neural deafness, and hypogonadism. The growth hormone, testosterone, and luteinizing hormone levels were tested and they were within normal limits. Hence, Crandall syndrome was ruled out thus confirming the diagnosis of Björnstad syndrome.

Till date, approximately only twenty cases of Björnstad syndrome have been reported. Petit et al. reported three cases in one family suggestive of autosomal dominant inheritance.[3] Loche et al. also reported Björnstad syndrome in the family as an autosomal recessive mode of transmission.[4] In this case history of consanguinity of marriage was present with a similar history in siblings representing autosomal recessive inheritance.

The hearing aid was advised to the patient as a part of treatment.

A case of pili torti with decreased hearing should be investigated for Björnstad syndrome and Crandall syndrome. This case is presented for its rarity and classical clinical presentation of Björnstad syndrome.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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