Monilethrix, a rare inherited hair shaft disorder in siblings

Sir,

The term monilethrix derives from both the Latin monile (necklace) and the Greek thrix (hair). The hair shaft has a beaded appearance due to the presence of elliptical nodes that have the diameter of normal hair and are medullated, regularly separated by internodes that are narrow, devoid of medulla, and are the site of fracture.[1]

Hair is normal at birth and is replaced with abnormal hair within first few months of life. The hair is beaded and fragile hair that tends to break at points of narrowing, thus leading to short, stubby hair, especially in the sites of friction such as the nape and occipital areas. Follicular keratosis of the affected scalp and keratosis pilaris are also typical. The severity of monilethrix considerably varies even among members of the same family and may range from an almost normal scalp, to total alopecia.[1,2]

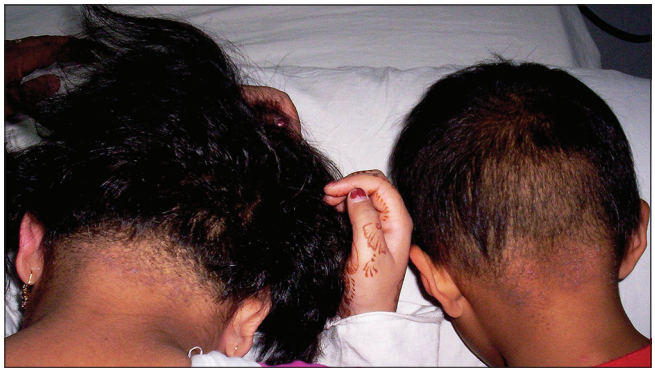

Figure 1: Sparse and short hairs over the occipital area of an 8-year-old boy. Both children having follicular papules over the nape of neck

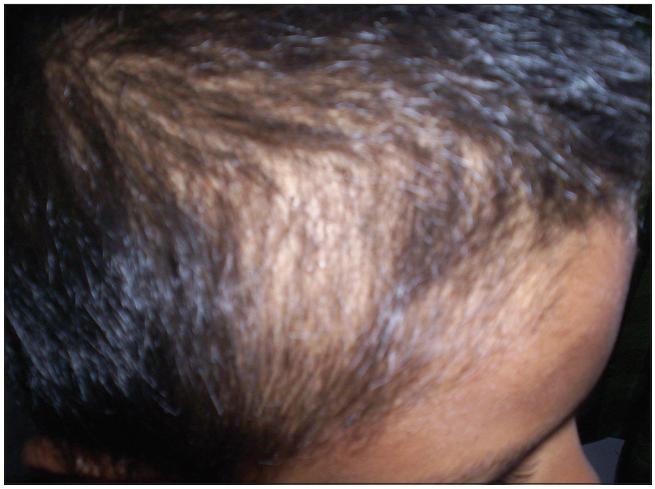

Figure 3: Sparse and short hairs over bitemporoparietal region

In some cases, the eyebrows and eyelashes, pubic and axillary hair, and general body hair may be affected. It may also be associated with koilonychia and rarely, systemic disturbances such as mental and physical retardation, syndactyly, cataract, teeth, and nail anomalies.[3]

An 8-year-old boy, born of a nonconsanguineous marriage, presented with diffuse hair loss after few months of birth. The boy was born at full term after an uncomplicated pregnancy. Hair was easily breakable and also there were tiny discrete lesions over the nape of neck and occipital area. His 10-year-old sister also had complaints of easy breakability of hair along with tiny papules over the nape of neck [Figure 1]. No other family member was having similar complaints.

Macroscopic inspection of the scalp showed fine, short, and sparse hairs over the bilateral parietal and occipital region with no sign of scarring of the scalp skin, but hairs were easily breakable [Figures 2 and 3]. Follicular papules over the occipital and nape of neck region were favoring keratotic pilaris. Examination of occipital hairs by microscopy [Figure 4a and b] revealed elliptical nodes resulting in a beaded appearance of the hair shafts. Developmental examination found his cognitive status to be adequate for age without any signs of mental disorder. There was no nail, dental, or sweat gland abnormality

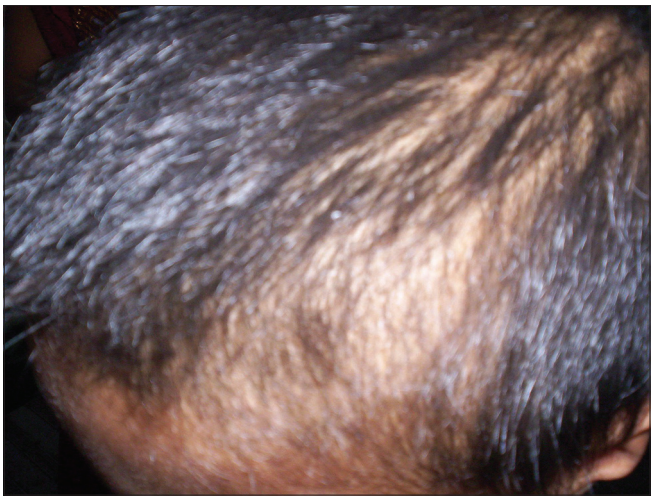

Figure 2: Sparse and short hairs over bitemporoparietal region

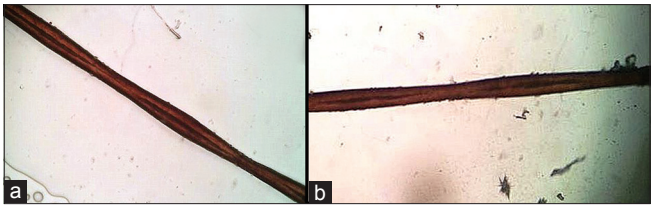

Figure 4: (a and b) ×100 Microscopy of hair shows nodes and inter nodes at regular interval with absence of medulla in intermodal region (a: Boy, b: Girl)

found after thorough physical examination in both children. Routine laboratory screenings was within range in both siblings.

Monilethrix is generally considered to be an autosomal-dominant disorder with variable penetrance. It is caused by mutations of the genes encoding type 2 hair keratins, hHb1 and hHb6. It is mapped to epithelial keratin gene cluster on 12q11-q13. Defective keratins in cortical cells, leading to wrinkling of the cells at internodes and makes them fragile. While nodes represent normal growth.[4] Mutations have been found in desmoglein 4 in the autosomal-recessive form.[2]

The variability of the monilethrix phenotype suggests that other factors also affect gene or disease expression. That was observed in our cases too, both siblings presented with variable severity of the same condition (the boy was affected more than his sister).

Various treatment modalities such as griseofulvin, retinoids, topical 2% minoxidil, and oral N-acetyl cysteine[2,5] have shown temporary and reversible improvement in isolated cases. No effective treatment is currently available.

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