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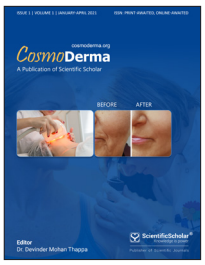

Spot the Diagnosis

A rare necklace of pearls

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Quick Response Code:

A 4-year-old girl child, 2nd by birth order, presented with hair loss all over scalp and easily breakable hair. Condition was observed by parents since infancy and no family members were affected with similar condition. Examination revealed diffuse thinning of hair all over the scalp along with thinning of eyebrows, multiple broken hair at different level, and keratotic follicular papules over scalp and nape of the neck [Figure 1]. Hair pull test was strongly positive. Hair mount showed normal hair; however, on trichoscopic examination, we could see beaded appearance with multiple broken hair stubs and alternate bands of thickening and thinning at regular interval [Figure 2]. What is the diagnosis?

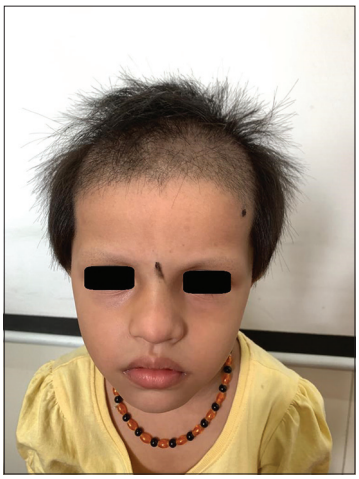

Figure 1: Diffuse thinning of hair over scalp and eyebrows.

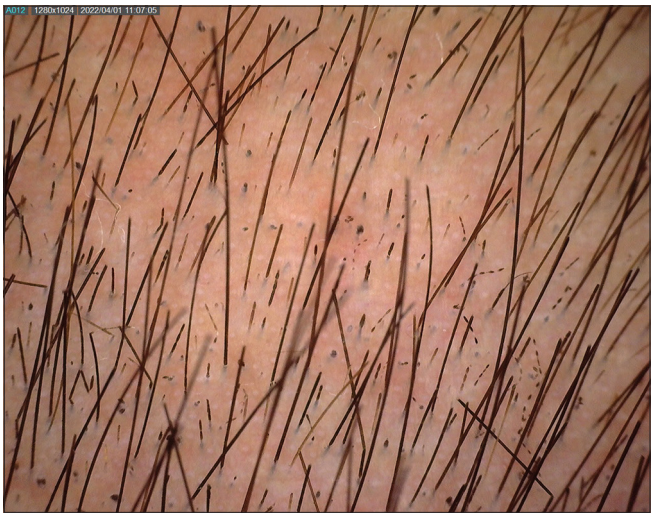

Figure 2: Dermoscopy revealed beaded appearance of hair with broken hair stubs.

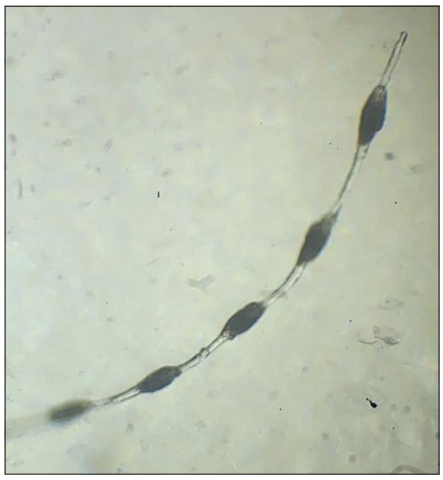

Figure 3: ×10 Hair mount showing hair shaft with nodes and internodes at regular interval (Under light microscopy X 10).

Answer: Monilethrix

Monilethrix is a rare genodermatosis of hair shaft which usually shows autosomal dominant inheritance with variable expression. There is mutation in hair keratin gene hHb1, hHb3 and hHb6 or KRT81, KRT83, or KRT86. There can be mutation in desmoglein 4 gene, where autosomal recessive inheritance is seen.[1]

The patient usually presents in the early childhood with diffuse alopecia, very short hair which are fragile and lustreless hair with easy pluckability. Keratotic follicular papules are seen, mainly over occipital area.

Trichoscopy reveals nodes and internodes placed at regular interval giving typical necklace appearance to a hair shaft with multiple broken hair stubs at variable length. Nodes represents normal hair caliber while internodes are the area of defect.[2]

Light microscopy of hair mount usually shows regularly placed nodes and internodes. However, in our case, not all hair was affected with the condition, only fine hair showed the characteristic morphology. The first hair mount showed normal hair. However, when we did hair mount under trichoscopy guidance by plucking out affected hair, we could see characteristic morphology [Figure 3]. The case was diagnosed based on history, clinical examination, trichoscopic, and hair mount findings. However, genetic testing was not done in the patient due to cost concerns.

There is no cure for this disorder.[3] Main modality of management is avoiding trauma, friction, chemical application, and mechanical damage. Patients show improvement with topical 2% minoxidil, acitretin, griseofulvin, biotin, iron supplements, and N-acetyl cysteine with high recurrence rate.[3,4] Oral minoxidil is a promising therapeutic option.[5]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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