Monilethrix: a typical case report with microscopic and dermatoscopic findings*

Elisa Fontenelle de Oliveira[1] Ana Luiza Cotta de Alencar Araripe[1]

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Abstract: Monilethrix is a rare hereditary condition generally considered to be an autosomal dominant disorder with variable penetrance. A case of a 6-year-old girl without a familial background for this disease is reported. The diagnosis was made by optic microscopy and dermoscopy. A therapeutic trial with topical minoxidil was conducted.

Keywords: Alopecia; Dermoscopy; Hypotrichosis

The term moniletrix comes from monile (necklace, in Latin) and thrix (hairs, in Greek). It is a rare, hereditary and autosomal dominant condition caused by mutations of the genes encoding trichocytes, hHb1, hHb3 and bHb6 type-2 keratins. It has been proposed that the hHb1 gene mutation produces a milder phenotype. The defect is located on chromosome 12q11-q13.[1] It is characterized by the presence of elliptical nodes and intermittent constrictions on the hair shafts, resulting in hair fragility at those points.[2] Clinical expression varies widely.[1] Normal hair is progressively replaced by abnormal one during the early months of life.[3] Mutations in desmoglein 4 have been found in autosomal recessive forms.[1]

Clinical presentation may be very characteristic, with extremely short, fragile hair emerging from keratotic follicular papules, especially in the occipital region.[4]

In milder forms, it may go unnoticed with few affected follicles. However, in serious forms, the secondary sexual hairs as well as the eyebrows and eyelashes may be compromised.[1]

The hair defect may occur in isolation or be associated with keratosis pilaris, syndactyly, cataracts, dental abnormalities and nail abnormalities.[1,4]

It may cause scarring alopecia.[1]

Trichoscopy shows regular variations in the diameter of the hair shaft with elliptical dilations (nodes) and constrictions (internodes).[4] Fusiform nodes are microscopically seen every 0.7-1 mm. Among the nodes, there is usually no hair marrow (medulla).[5]

Hair shine may show improvements during the summer and with age. The use of topical minoxidil or oral acitretin may be effective in some cases. Initial improvement but with no lasting effect is described with the use of N-acetyl cysteine.[3] Avoiding trauma to the hair is the most effective method in the management of this anomaly.[3]

We report the case of a 6-year-old girl, born of a non-consanguineous marriage. She had two healthy sisters. Her normal hair had progressively been replaced by abnormal one during the early months of life. In fact, she never had a hair cut, due to its easily fragmentation and, consequently short length.

Clinical examination revealed diffuse hypotrichosis of the scalp, as well as coarse hair. Keratotic follicular papules were mainly observed in the occipital region (Figure 1). The diagnosis was confirmed by dermoscopy and optical microscopy, which demonstrated

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[1] Instituto Fernandes Figueira - Fundação Oswaldo Cruz (IFF-FioCruz) – Rio de Janeiro (RJ), Brazil.

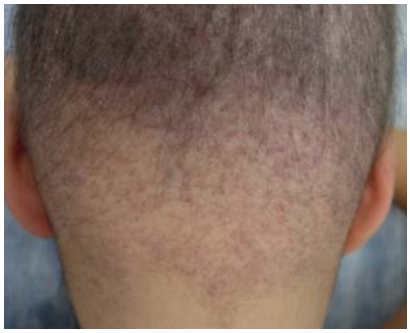

Figure 1: Initial clinical aspect: diffuse hypotrichosis, coarse hair and keratotic follicular papules, especially in the occipital region

Figure 2: Dermoscopic aspect - hair looking like rosary beads, with nodes and constrictions

Figure 3: Optical microscopy of a hair strand- hair looking like rosary beads, with nodes and constrictions

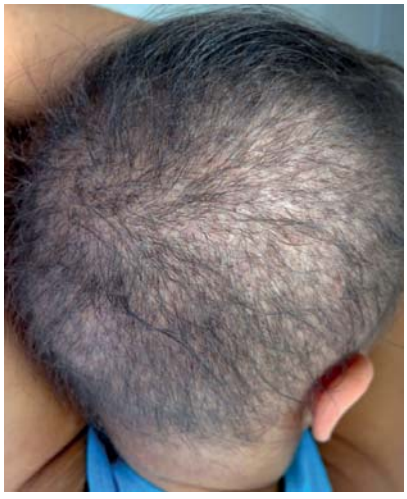

Figure 4: Final clinical aspect – A few months after the use of minoxidil - increase in the length of a few strands of hair

the appearance of "rosary beads", with nodes and constrictions (Figures 2 and 3). She was referred for genetic evaluation and was prescribed minoxidil 2% solution. A few months after, we noticed an increase in the length of a few strands of hair, which did not have the previously described aspect (Figure 4).

Conclusion: Monilethrix is a rare condition that can be identified through characteristic findings. Diagnosis may also be noninvasively made by dermoscopy and optic microscopy of the hair. ❑

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Mailing address:
Elisa Fontenelle de Oliveira
Rua Almirante Guillobel, 32/202 - Lagoa
22471-150 - Rio de Janeiro - RJ
Brazil.
E-mail: elisafontenelle@yahoo.com.br

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