CASE REPORT

Monilethrix. Observation of rare hair dysplasia in two siblings. Clinical and Dermoscopic Report

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ABSTRACT

Monilethrix is an autosomal dominant genetic hair dysplasia, rarely recessive. It is considered as a congenital pathology of the hair, having as its main characteristic a very altered hair shaft in its shape. Two cases from our file are presented: two brothers treated in 2009. Their clinical and dermatoscopic features are reviewed.

INTRODUCTION

Monilethrix is an autosomal dominant genetic process, rarely recessive, with high penetration and variable expression, belonging to the congenital hair strands with deformed hair shaft as their main characteristic.

It is usually recognized by the rolling alternation of elliptic nodules and dystrophic narrowings giving an appearance of "rosary beads or pearl necklace", with a tendency to rupture in the internodular areas, resulting in the presence of short hairs and hyperkeratosis with perifollicular erythema.

Diagnosis is usually clinical and trichoscopy allows the diagnosis to be easily confirmed. The clinical picture tends to improve in adolescence.[1]

CLINICAL CASES

There are two patients, the older is a 17-year-old female and the younger, her 14-year-old brother. (Picture 1)

The siblings report that the clinical picture, when it comes to the elderly patient, initiates from birth, indicating that from day 3 of her birth she presented with hair loss, reappearing when she was 13.

The brother presented with a similar clinical picture but it initiated when he was 1 or 2. The family was not able to accurately specify its chronology though. They indicated that the hair started to growth again at approximately the same age as his sister. What they do ensure is that there was never a need to cut their hair. The patient had received multiple treatments with vi-

tamins, various shampoos and antifungal treatments. For specific reasons, they could not confirm any medical family history.

Upon examination of the scalp and hair of the sister, a thin hair was found, especially in the frontal hair implantation line and the anterior area of the scalp (Photo No.2). The same characteristics were observed in the frontoparietal area of both sides (Photo No.3).

Trichoscopy was performed, which shed light on the presence of elliptic nodules, intermittent constrictions, perifollicular scales and angled hairs (Photo 4-5). Erythema and perifollicular scales are also observed (photo 6).

The brother presented similar clinical and trichoscopic characteristics (less intense). Partial frontoparietal alopecia (photo 7), follicular papules at the level of the nape of the neck (photo 8) and the same trichoscopic structures, as in the case of his sister (photo 9).

The clinical picture suggested the diagnosis of monilethrix and trichoscopy confirmed it. The biopsy that was performed on the sister was not useful because she reported a "Normal-looking scalp."

She received daily treatment with 5% Minoxidil lotion, and after two or three months, hair growth and a moderate increase in hair density were observed. An 8 month follow up was done. After such period of time, contact with both siblings was lost.

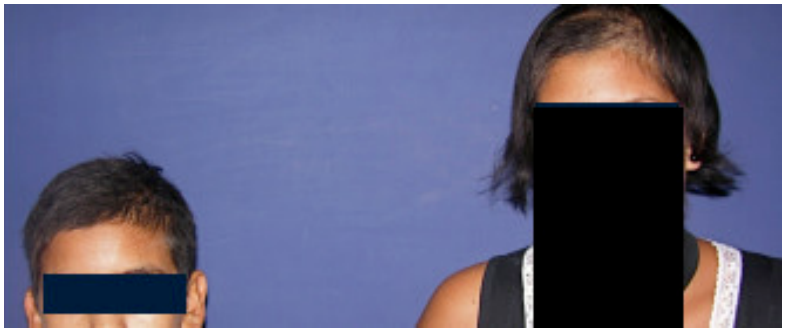

Photo 1. Two siblings showing frontoparietal alopecic areas.

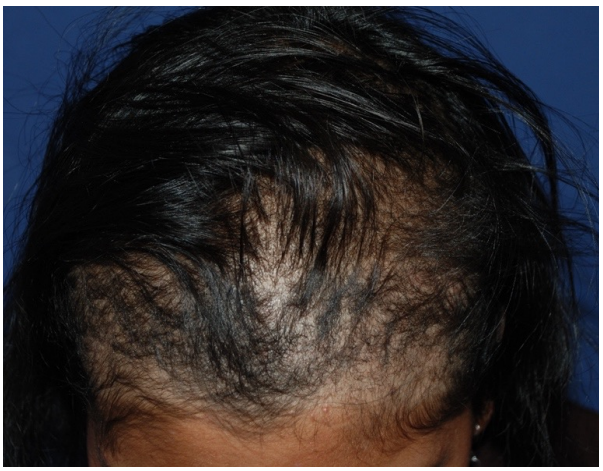

Photo 2. Sister. Partial alopecia is observed at the implantation site and anterior side of the scalp.

Photo 3. Sister. Hair thinning in the frontoparietal areas on both sides.

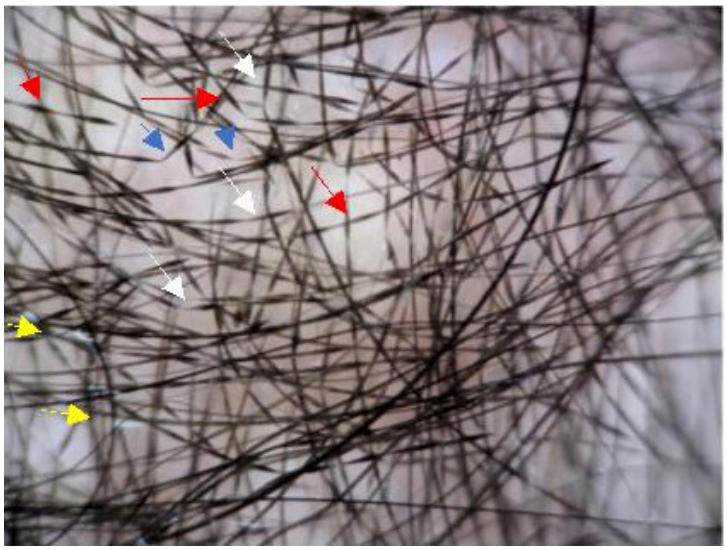

Photo 4. Trichoscopy. Elliptical nodules (red arrow). Intermittent constrictions (white arrow). Perifollicular scales (yellow arrow). Angled hair (blue arrow).

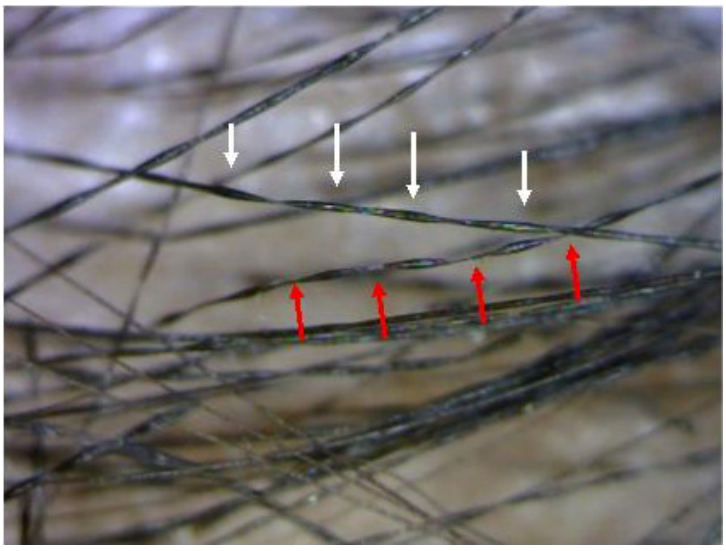

Photo 5. Presence of elliptical nodules (white arrow) and constrictions (red arrow).

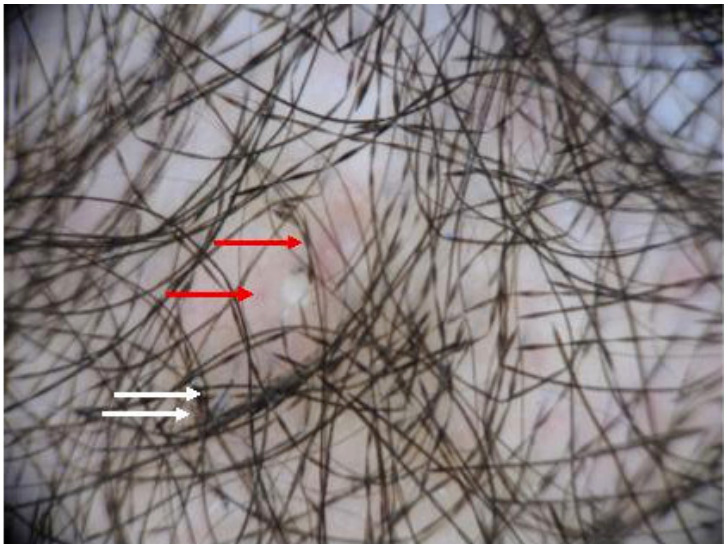

Photo 6. Presence of scales (white arrow) and perifollicular erythema (red arrow).

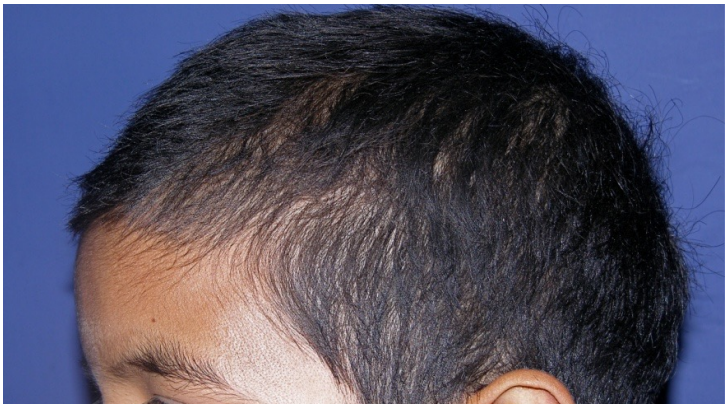

Photo 7. Brother Light hair in the frontoparietal area.

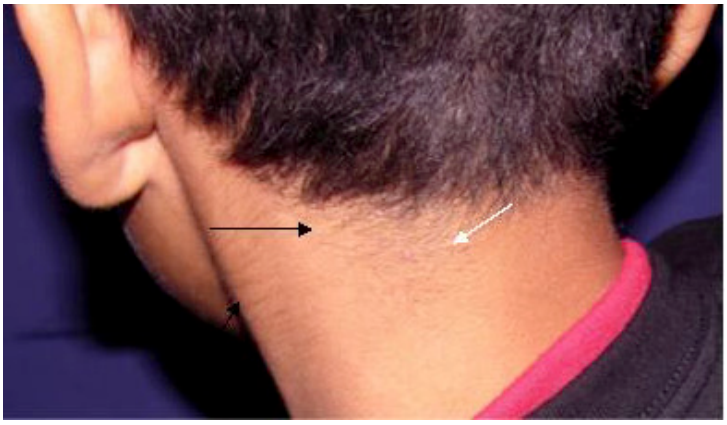

Photo 8. Follicular papules at the level of the nape of the neck.

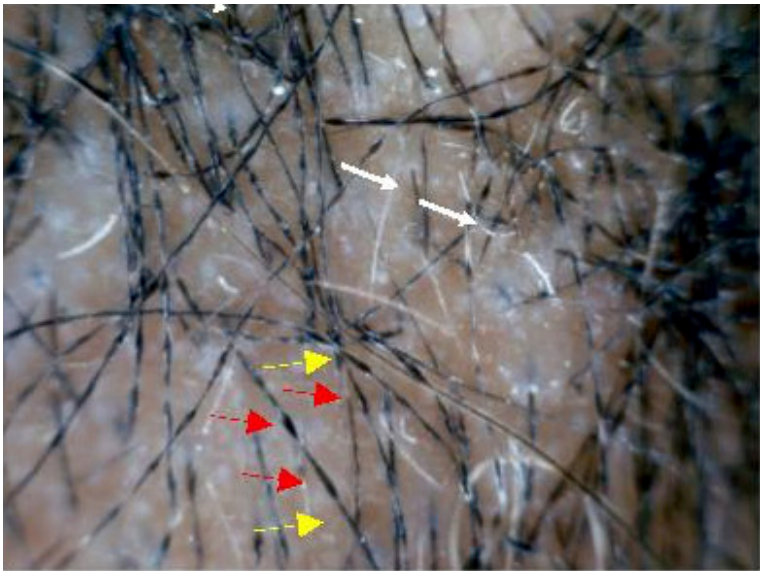

Photo 9. Presence of nodules and constrictions. Perifollicular scales and keratosis. Elliptical nodules (red arrow). Constrictions (yellow arrows). Broken hairs (white arrow).

DISCUSSION

Monilethrix is an autosomal dominant hereditary hair dysplasia with mutations in the genes KRT81, KRT83 and KRT86, associated with trichokeratin or autosomal recessive with mutations in the gene of desmoglein 4 (DSG4) with malformation in the desmosomes of the hair shaft.[2] It is characterized by thinning of the hair shaft, leading to an increase of hair fragility causing residual alopecia. The process can be very variable clinically, from patients with few appreciable lesions, to severe alopecia.[3] "De novo" mutations may explain the existence of unique patients with this clinical picture in the family.[4]

Onset usually occurs during the first few months of age with diffuse progressive hypotrichosis, and red, brittle hair. It manifests throughout the scalp but the most affected area is the occipital region, which involves body hair, eyebrows and eyelashes, and can be accompanied by abutment keratosis. There is rarely nail or dental involvement, mental retardation and cataracts.

Differential diagnosis must be made with pseudomonilethrix., in which the alteration lies in the thickenings that actually correspond to flattenings of the hair shaft, while the internodes represent the normal hair shaft caliber, and are associated with a compulsive habit of biting one's own hair.

Monilethrix improves with age. Procedures that accelerate hair breakage are not recommended.[5]

The diagnosis is based on clinical characteristics, light microscopy examination and trichoscopy. In the latter, zones of dystrophic constriction of the hair shaft can be observed, separated at regular intervals by elliptical nodules of normal thickness, giving a "collar sign" appearance. There is also evidence of perifollicular papules and erythema and presence of peripillary sheaths or cysts. Likewise, there are broken hairs that, according to Castañeda et al,[7] are evidenced confocal microscopy.[4,6,7] Nodules seem to represent normal growth and the constrictive areas are characterized by the wrinkling of the

cortical cells, leading to fragile hair, and the absence of the marrow.[8] Scarring alopecia is deemed as possible.[9]

Regarding the evolution of the process, most patients' hair loss persists with few alterations throughout their lives. However, spontaneous improvement or complete resolution during pregnancy has been reported. In general, there is no cure for this pathology pillar. Minoxidil, both oral at low doses and topical, has been use in some cases with positive response.

Etretinate reduces hair loss, but does not improve hair fragility or density.[10]

CONCLUSION

Observation chart (infrequent) Discoveries of new genetic mutations have led to the description of intrafamily and interfamily clinical forms or the manifestation of cases without a history of novo mutations. Trichoscopy has become an important means of diagnosis. Treatments are not very efficient, achieving only partial improvements with the aforementioned therapies.

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