“My Daughter Has Thin and Short Hair”

Anissa Zaouak^{a, c} Ehsen Ben Brahim^{b, c} Raja Jouini^{b, c} Houda Hammami^{a, c}
Samy Fenniche^{a, c}

^aDermatology Department, Habib Thameur Hospital, Tunis, Tunisia; ^bAnatomopathology Department, Habib Thameur Hospital, Tunis, Tunisia; ^cResearch Unit “Genodermatoses and cancers” LR12SP03, Tunis, Tunisia

Question

A 16-year-old female presented to our dermatology department for a history of congenital hypotrichosis. She was born from a consanguineous marriage. Her sister and brother also suffered from sparse and fragile hair. Dermatologic examination revealed diffuse hypotrichosis with sparse, short, and broken hair on her entire scalp (Fig. 1). Her hypotrichosis was more pronounced on her occipital scalp. She also had small keratotic papules on her occipital

region. Her eyebrows and eyelashes were not affected. There were no tooth nor nail abnormalities. There was no evidence of mental retardation, epilepsy, or systemic diseases.

Dermoscopic Findings

Dermoscopy of the scalp was performed, showing numerous fragile hairs with uniform elliptical nodes separated by intermittent constrictions, giving hairs a beaded appearance (Fig. 2).

What is your diagnosis?

Fig. 1. Severe hypotrichosis of the scalp.

Fig. 2. Dermoscopy shows hair shaft beading caused by narrow constrictions of the hair separating elliptical nodes (FotoFinder, FotoFinder Systems GmbH, Bad Birnbach, Germany; original magnification: $\times 50$).

Answer: Monilethrix**Clinical Course**

The patient was initially treated with topical minoxidil 5%, applied twice daily for 6 months without significant results. She was advised to avoid trauma to her congenitally abnormal hair.

Discussion

The name monilethrix comes from the Latin word for necklace (monile) and the Greek word for hair (thrix) meaning “beaded hair” [1]. Monilethrix is a rare autosomal dominant disease of the hair shaft characterized by alopecia as a result of hair breakage [2]. Clinically, there is normal hair at birth but, within the first months of life, a characteristic moniliform aspect of the hair develops [3]. Perifollicular erythema and follicular hyperkeratosis are also common. In our patient, only the scalp was affected, but in extensive cases, there may be eyebrow, eyelash, and nail involvement. There is usually intrafamilial variability and the disease may range from almost normal scalp to total alopecia [3]. When performed, as in our case, trichoscopy shows regular variations in the diameter of the hair shaft with elliptical dilations (nodes) and constrictions (internodes) [4]. Rakowska et al. [5] coined the term “regularly bended ribbon sign” in monilethrix, which means hairs with beaded appearance bent regularly at multiple locations. This case is being reported not only for the rarity of the disease but also to highlight the role of trichoscopy as a useful and non-invasive diagnostic technique enabling a rapid and accurate diagnosis of this particular alopecia. Correlation of the characteristic clinical presentation together with typical dermoscopic features could help in avoiding unnecessary scalp biopsies.

Monilethrix (OMIM No. 158000) typically shows an autosomal dominant inheritance pattern with high penetrance but variable expression, as in our patient. More than one gene is involved in monilethrix. In fact, the autosomal dominant type is caused by heterozygous mutations in type II hair keratin genes KRT81 and KRT83 on chr12q13. All these keratins are expressed in the keratin-

izing hair shaft cortex. However, the autosomal recessive form of monilethrix was reported to be caused by mutations in desmoglein 4 (*DSG4*) encoding one of the major components of the desmosome, which is essential for proper cellular adhesion of epithelial cells [6–8].

In monilethrix, there is no genotype-phenotype correlation. This is to say that the same mutation could be expressed by different phenotypes of different severities. This raises the question of other influencing factors of this mutation including the influence of environment factors and other non-genetic factors. In fact, according to Stranger et al. [9], humans are characterized by marked inter-individual differences in the expression levels of their genes.

This particular alopecia can predispose individuals to low self-esteem and a negative body image [10]. Hence, assessing the impact of this genodermatosis on the quality of life is important.

There is no effective treatment currently available for this hair condition. Oral retinoids, griseofulvin, and oral or topical minoxidil may be helpful in rare cases [10, 11]. The most important fact is to identify this congenital alopecia for the patient and his family and for the physician to avoid unnecessary treatment modalities.

In summary, dermoscopy, as illustrated in our case, could improve diagnostic accuracy of hair and scalp disorders.

Statement of Ethics

The patient gave her informed consent to use her photos and details of her case

Disclosure Statement

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Keywords

Monilethrix · Alopecia · Dermoscopy

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