Value of dermoscopy for the diagnosis of monilethrix

T Baltazard MD, F Dhaille MD, G Chaby MD, C Lok MD PhD

Affiliations: Department of Dermatology, Hôpital Nord, University of Amiens, [80000] - Amiens, France

Corresponding Author: Thomas Baltazard, Department of Dermatology, CHU AMIENS, Place Victor Pauchet, 80000 – AMIENS, France, Tel: +33322455841 Fax: +33322456072, Email: baltazard.thomas@chu-amiens.fr

Abstract

Monilethrix is a rare genodermatosis characterized by a hair shaft dysplasia responsible for hypotrichosis. We report the case of a child with monilethrix with no associated cases in the family. Trichoscopy facilitated the diagnosis. A 2-year-old boy presented with diffuse alopecia and persistent fragile hair for several months. Clinical examination revealed alopecia with hairs broken several millimeters from the scalp. Trichoscopy revealed zones of dystrophic constriction of the hair shaft, separated at regular intervals by elliptical nodes of normal thickness, giving a "necklace" appearance. The diagnosis of monilethrix was made on the basis of these specific features. The diagnosis of monilethrix was more difficult to establish in our patient owing to the absence of any familial cases.

Keywords: monilethrix, alopecia, scalp, dermoscopy

Introduction

Monilethrix is a rare hereditary hair shaft anomaly in which affected hairs have a specific beaded morphology. This disease has a predominantly autosomal dominant transmission, a variable penetrance, and clinical presentations of variable severity (dystrophic hair confined to the occiput to total alopecia). It is usually first observed during early childhood. We report a patient with severe monilethrix diagnosed by trichoscopy.

Case Synopsis

A 2-year-old boy was referred to our department with suspected trichotillomania. He presented with diffuse alopecia and persistent fragile hair for several months. He was born at term and exhibited vellus hair at birth. Clinical examination revealed alopecia with hairs broken several millimetres from the scalp (**Figure 1**), particularly in the temporal and occipital regions. The hair was slightly denser at the vertex and pruriginous keratosis pilaris was noted. The nails of the great toes were thin and fragile with discreet koilonychia. The rest of the clinical examination, particularly the eyebrows, eyelashes, and body hair, was normal.

Trichoscopy revealed zones of dystrophic constriction of the hair shaft, separated at regular intervals by elliptical nodes of normal thickness, giving a "necklace" appearance; perifollicular papules were also visible (**Figure 2**). The diagnosis of monilethrix was adopted on the basis of these specific features. Careful examination of the parents did not reveal any signs of monilethrix. Simple surveillance and avoidance of hair trauma were proposed.

Case Discussion

Four genes are associated with monilethrix and are responsible for autosomal dominant transmission of the disease [1, 2]. An autosomal recessive form has also been described and is caused by mutations of gene DSG4 coding for desmoglein 4 [3]. The diagnosis of monilethrix was more difficult to establish in our patient owing to the absence of any familial cases. A de novo mutation could explain why this boy was the only one in the family to be affected by this hair disease [4]. As penetrance of the disease is also variable from one subject to another, the diagnosis may have been missed in one of the two parents despite a thorough examination.

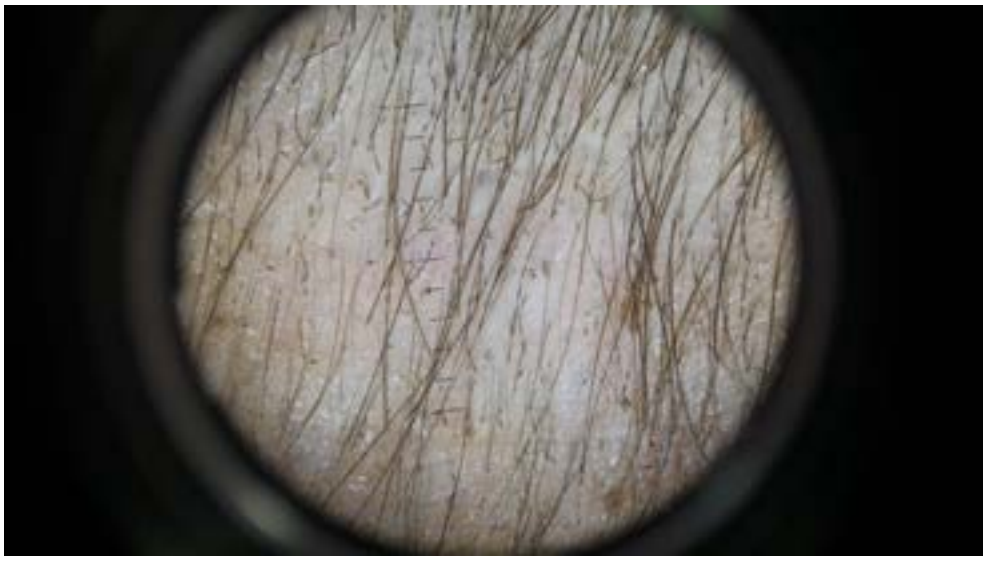

Figure 1. *Rarefaction and shortening of the hairs that are broken several millimetres from the scalp with apparent sparing of the vertex.*

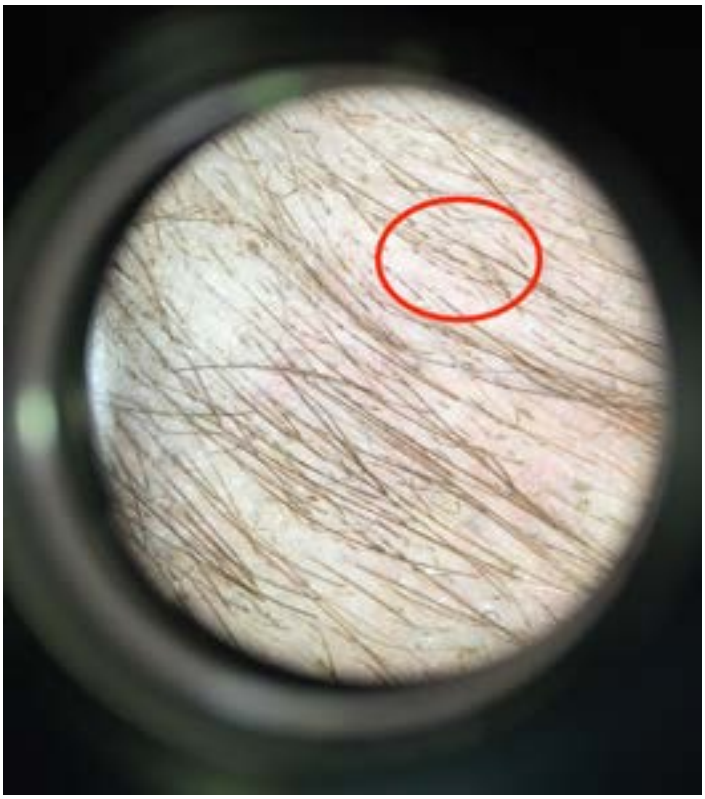

Figure 2. *Dermoscopic (x20) appearance with regular nodes and intermittent constriction along the hair shaft and fracture zones in the narrowed segments, giving a "necklace" appearance (red arrow).*

The diagnosis of monilethrix is essentially clinical, usually confirmed by microscopy or histology showing abnormal hair follicles with alternating constricted and normal portions.

Trichoscopy is much easier and less time consuming than ex vivo microscopic examination; microscopic examination was not performed in this patient. The characteristic trichoscopy features enables one to establish a rapid diagnosis and reveals a characteristic "necklace" appearance of the hair with marked fragility of the hair in the internodal region. In addition, perifollicular keratotic papules may be visualized. The presence of these typical signs in our patient was immediately suggestive of the diagnosis [2, 5]. Regrowth of apparently normal hair at the time of puberty and during summer is classically reported. Treatment with minoxidil and acitretin can be proposed in some cases, but symptoms recur after discontinuation [6, 7]. The avoidance of hair trauma remains the cornerstone of the management of this hair disorder.

Conclusion

Trichoscopy permits a simple, rapid, and non-invasive examination. This is useful for the diagnosis of monilethrix by revealing a "necklace" appearance of the hair and could substitute for ex vivo microscopic examination.

References

1. Ferrando J, Galve J, Torres-Puente M, Santillan S, Nogues S, Grimalt R. Monilethrix: A New Family with the Novel Mutation in KRT81 Gene. Int J Trichology 2012;4:53-5. [PMID:22628999].
2. Oliveira EF, Araripe AL. Monilethrix: a typical case report with microscopic and dermatoscopic findings. An Bras Dermatol 2015;90:126-7. [PMID:25672313].
3. Kato M, Shimizu A, Yokoyama Y, Kaira K, Shimomura Y, Ishida-Yamamoto A et al. An autosomal recessive mutation of DSG4 causes monilethrix through the ER stress response. J Invest Dermatol 2015;135:1253-60. [PMID:25615553].
4. Bindurani S, Rajiv S. Monilethrix with variable expressivity. Int J Trichology 2013;5:53-5. [PMID:23960403].
5. Sharma VK, Chiramel MJ, Rao A. Dermoscopy: A rapid bedside

tool to assess monilethrix. Indian J Dermatol Venereol Leprol 2016;82:73-4. [PMID:26728817].

6. Karincaoglu Y, Coskun BK, Seyhan ME, Bayram N. Monilethrix: improvement with acitretin. Am J Clin Dermatol 2005;6:407-10. [PMID:16343029].
7. Rossi A, Iorio A, Scali E, Fortuna MC, Mari E, Palese E et al. Monilethrix treated with minoxidil. Int J Immunopathol Pharmacol 2011;24:239-42. [PMID:21496408].