NASZA DERMATOLOGIA Online
OUR DERMATOLOGY Online

Case Report

Monilethrix - Case report of a rare disease

Tasleem Arif[1,2], Imran Majid[3], Mir Laieq Ishtiyaq Haji[2], Nuzhat Samoon[2,4]

[1]Department of Dermatology, STD & Leprosy, District Hospital Baramullah, Kashmir, India, [2]Directorate of Health Services, Jammu and Kashmir, India, [3]CUTIS Skin and Laser Institute, Srinagar, Kashmir, India, [4]Department of Pathology and Transfusion Medicine, District Hospital Baramullah, Jammu and Kashmir, India.

Corresponding author: Dr. Tasleem Arif, MBBS, MD, E-mail: dr_tasleem_arif@yahoo.com

ABSTRACT

Monilethrix is a rare genetic disorder of hair characterized by beaded appearance of the hair shaft leading to hair fragility and patchy dystrophic alopecia. In this disorder, the hair shaft has alternate widenings (nodes) and constrictions (internodes) that lead to fracture of hair shaft and varying degree of alopecia. We report an eight year old Kashmiri boy who presented with diffuse hair loss since infancy. As monilethrix is a rare disease entity which prompted us to report this case.

Key words: Alopecia; beaded appearance; hair fragility; monilethrix

INTRODUCTION

The word Monilethrix is derived from Latin 'monile' meaning 'necklace' and the Greek 'thrix' meaing 'hair'. It's a structural hair shaft disorder characterized by a beaded appearance due to the presence of elliptical or fusiform nodes which have a diameter of normal hair. These nodes are medullated and separated by areas of constrictions called as internodes which lack medulla. These unmedullated internodes are the sites of hair breakage leading to dystrophic alopecia [1]. The first case of monilethrix was described by Walter Smith in 1879. However, the term 'monilethrix' was coined by Radcliff Crocker [2]. Monilethrix is transmitted as an autosomal dominant trait with high penetrance but variable expressivity. However, there are sporadic cases of autosomal recessive inheritance reported for this disease [3].

CASE REPORT

An 8-year-old male child born of a nonconsanguineous marriage presented to us in the outpatient department (OPD) with a chief complaint of diffuse hair loss since seven months of his age. There is increased fragility and inability to grow long hair over the scalp. He had normal hair initially after which it got replaced with short and sparse hair. The parents revealed that the hair broke when it reached a certain length of few centimeters. There was no such history in any of his family members.

Macroscopic inspection of the scalp showed sparse short stubby hair of few centimeters in length with no signs of scarring (Fig.1). The nape of the neck showed multiple prominent horny follicular papules with hair emerging from the summit of these papules. His eyebrows and eyelashes were normal. On light microscopic examination hair shafts revealed the characteristic alternating fusiform or spindle shaped swellings (nodes) and constrictions (internodes) which confirmed the diagnosis of monilethrix in our patient.

Developmental examination of the child was normal. His mental and physical growth was normal. There was no nail, dental, sweat gland, otorhinolaryngological or other systemic abnormality. Routine laboratory screening was unremarkable. His serum copper and ceruloplasmin levels were normal.

DISCUSSION

The cause of monilethrix remains unclear. Several Genetic studies have suggested that monilethrix

How to cite this article: Arif T, Majid I, Ishtiyaq Haji ML, Samoon N. Monilethrix - Case report of a rare disease. Our Dermatol Online. 2015;6(1):46-48.
Submission: 15.10.2014; **Acceptance:** 04.12.2014
DOI: 10.7241/ourd.20151.11

is likely caused by a hair keratin mutation. Some researchers have suggested mutations in the human hair basic keratins hHb1 and hHb6 in this disorder [4]. The most common mutation is the E413K mutation in hHb6 [5]. Autosomal dominant monilethrix is caused by mutations in hair keratin genes KRT81, KRT83, or KRT86, whereas in autosomal recessive form, mutations in the desmoglein-4 gene (DSG4) have been reported [6]. Defects in the cortical keratins lead to wrinkling of the cells at the internodes and make them susceptible to fracture while at nodes, the growth is normal [7].

Monilethrix occurs mainly on the scalp which can be widespread or circumscribed. The eyelashes, eyebrows, axillary, pubic, and limb hair are affected occasionally. The increased fragility of the hair shaft to premature weathering results in its inability to attain a normal length. The condition persists throughout life but may resolve spontaneously in some cases [2,8]. Monilethrix is usually associated with keratosis pilaris presenting as horny keratotic papules. In our case there were also keratotic horny papules on the nape of the neck but the skin biopsy was not taken.

The diagnosis of monilethrix can be confirmed on light microscopy which shows alternating wider elliptical nodes and narrower internodes. At the fragile internodes, There is premature weathering and breakage. Transmission microscopy of the internode showes deviation in the axis of the macrofibrils within the cortical cells and disorganized globular inter-macrofibrillar cystine rich material. Dermoscopy can be used as a rapid diagnostic tool for monilethrix which may show hair shafts with uniform spindle shaped nodes and intermittent constrictions (internodes); hairs bent at multiple locations in a regular fashion and a tendency to shaft fracture at the sites of constrictions [9,10]. However, in our case dermoscopy was not done due to the unavailability of the same as the case was diagnosed in a peripheral hospital.

There is no specific treatment for this condition. Avoiding trauma is the primary goal in managing this condition. The various activities which cause an increased susceptibility to fracture of the hair shafts including dyeing, bleaching, curling, etc. should be avoided. Some improvement has been reported with griseofulvin, iron supplementation, oral retinoids and topical minoxidil in isolated cases [11,12].

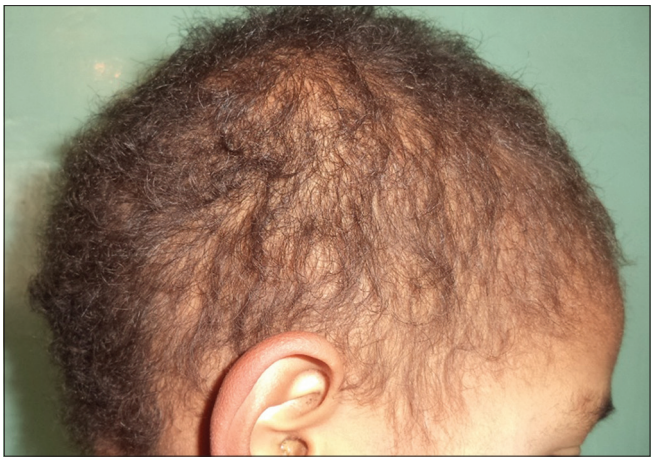

Figure 1: Monilethrix in an 8 year old boy. There is sparse short stubby hair of few centimeters in length over the whole scalp

Though, there are many reports of use of minoxidil in monilethrix but the improvement seems to be temporary and reversible. Rossi et al treated four cases of monilethrix with topical minoxidil 2%. The dosage was kept 1 ml day and night for a duration of one year. There was an increase of normal hair shafts after one year. They concluded that topical minoxidil 2% can be a good therapeutic option for Monilethrix [13].

CONSENT

The examination of the patient was conducted according to the Declaration of Helsinki principles. Written informed consent was obtained from the patient's caregivers for publication of this case report and any accompanying images.

REFERENCES

1. Lencastre A, Tosti A. Monilethrix. J Pediatr. 2012;161:1176.
2. Smith WG. A rare nodose condition of thehair. BMJ. 1879;11:291-6.
3. Zlotogorski A, Marek D, Horev L, Abu A, Ben-Amitai D, Gerad L, et al. An autosomal recessive form of monilethrix is caused by mutations in DSG4: clinical overlap with localized autosomal recessive hypotrichosis. J Invest Dermatol. 2006;126:1292-6.
4. Winter H, Vabres P, Larregue M, Rogers MA, Schweizer J. A novel missense mutation, A118E, in the helix initiation motif of the type II hair cortex keratin hHb6, causing monilethrix. Hum Hered. 2000;50:322-4.
5. Muramatsu S, Kimura T, Ueki R, Tsuboi R, Ikeda S, Ogawa H. Recurrent E413K mutation of hHb6 in a Japanese family with monilethrix. Dermatology. 2003;206:338-40.
6. Farooq M, Ito M, Naito M, Shimomura Y. A case of monilethrix caused by novel compound heterozygous mutations in the desmoglein 4 (DSG4)gene. Br J Dermatol. 2011;165:425-31.
7. Djabali K, Panteleyev AA, Lalin T, Garzon MC, Longley BJ, Bickers DR, et al. Recurrent missense mutations in the hair keratin gene hHb6 in monilethrix. Clin Exp Dermatol. 2003;28:206-10.

8. Solomon IL, Green OC. Monilethrix. N Engl J Med. 1963;269:1279-85.
9. Gummer CL, Dawber RPR, Swift JA. Monilethrix: an electron microscopic and electron histochemical study.Br J Dermatol. 1981;105:529-41.
10. Wallace MP, de Berker DA. Hair diagnoses and signs: the use of dermatoscopy. Clin Exp Dermatol. 2010;35:41-6.
11. Bhatt TA. Monilethrix. Egyptian Dermatol Online J. 2010;6:17.
12. Karaman GC, Sendur N, Basar H, Bozkurt SE. Localized monilethrix with improvement after treatment of iron deficiency anemia. J Eur Acad Dermatol Venerol. 2001;15:362-4.
13. 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.

Source of Support: Nil, **Conflict of Interest:** None declared.