Monilethrix - Case report of a rare disease

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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’ meaning ‘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...
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 shows 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].

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