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MONILETHRIX PRESENTATION IN THE ABSENCE OF INHERITANCE: A UNIQUE CASE REPORT

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Abstract

Monilethrix is a rare inherited hair disorder characterized by fragile, beaded hair shafts that break easily, resulting in sparse or patchy hair growth. We present the case of a four-year-old Indian girl with complaints of sparse, coarse, easily pluckable hair over the occipital and biparietal area of the scalp since the age of two. Clinical examination revealed characteristic features of monilethrix. The diagnosis was further confirmed by trichoscopy and microscopy. The patient was initiated on 2% minoxidil for management. This case highlights the importance of recognizing the clinical and diagnostic features of monilethrix for accurate diagnosis and management due to its rare presentation and sporadic nature.

INTRODUCTION

Monilethrix is a rare disorder of hair shaft which is characterized by fragile, beaded hair shafts, leading to patchy hair loss and sparse growth mostly on the occipital scalp as it is more exposed to friction. It often starts in infancy or childhood and has a distinctive appearance under microscopic examination.

Monilethrix is an autosomal dominant disorder with high penetrance but variable expressivity. There are only handful of documented case reports of Monilethrix that show a lack of inheritance pattern or familial history, which poses an interesting diagnostic challenge for medical professionals and researchers.

This case report highlights the clinical presentation of a four years old girl and sheds light on the diagnostic challenges and management strategy employed in addressing this uncommon hair disorder.

Case report

A four year old Indian girl was brought to the dermatology out patient department by her parents with the complaints of sparse hairs present over the occipital and biparietal area of the scalp since 2 years. They also complained that the hairs looked coarse and were easily pluckable. There was no history of consanguineous marriage between the parents and there was no history of similar complaints in the parents or siblings.

Cutaneous examination showed fine, sparse and rough hair appearing burnt at the ends. They were present over the occipital and biparietal area of scalp, with no sign of scarring

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of the scalp skin. The length of the hair was relatively short and they were easily pluckable. There were also multiple skin coloured asymptomatic keratotic papules present over the occipital area. (Figure 1-3)

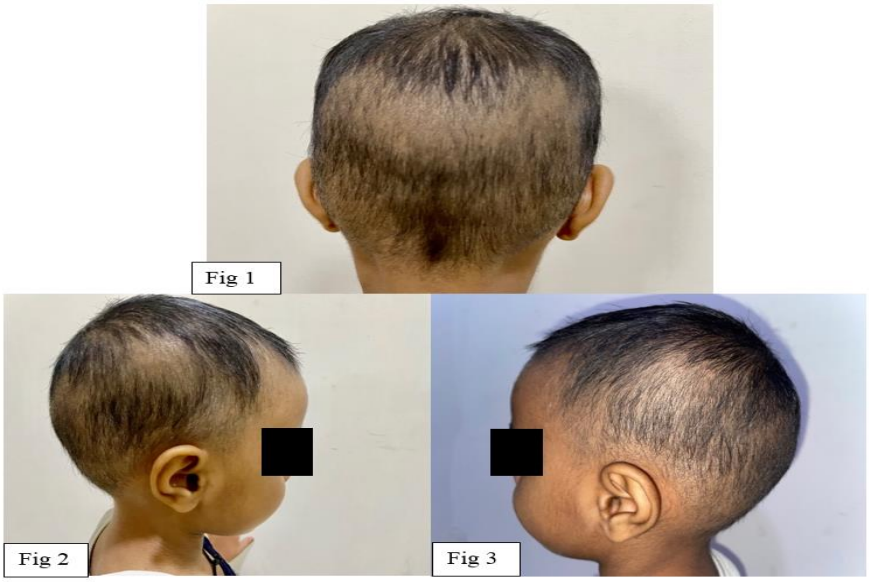

Figure 1,2,3: Diffuse hypotrichosis and coarse hair particularly in the occipital and biparietal area of the scalp.

Hair of eyebrows, eyelashes, axillary, pubic and body were normal. There was no nail, dental, or sweat gland abnormality found.

Trichoscopy done with ×10 magnification in non-polarized light illumination showed hair shaft with characteristic regularly arranged elliptical nodes separated by internodes, hair shaft of varying length and few broken hair. (Fig 4)

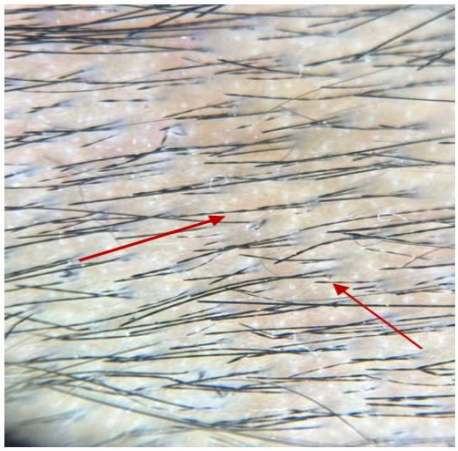

Figure 4: Trichoscopy showing multiple nodes and internodes, various hair thickness, few empty hair follicles.

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Microscopy (10x) of the hair shaft also showed multiple nodes with constrictions in between.

Routine laboratory investigations were within normal limit. (Fig 5)

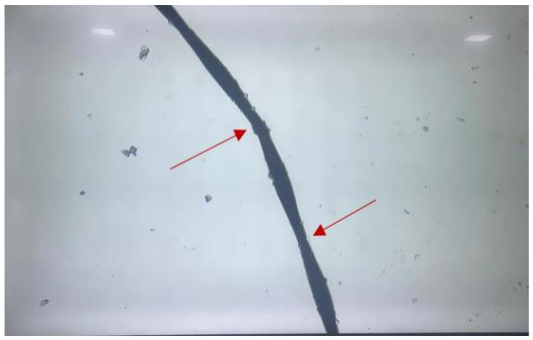

Figure 5: Light microscopy showing constrictions between the nodes.

On the basis of examination, trichoscopy and microscopy, a diagnosis of monilethrix was made.

Patient was started on 2% minoxidil once daily application at night.

DISCUSSION

Monilethrix was first described by Smith in 1879, and later Radcliff Crocker coined the term “monilethrix”, derived from Latin word “monile” meaning “necklace” and Greek word “thrix” meaning “hair.”[1]

It is recognized as a rare genetic disorder caused by mutations in the genes located on chromosome 12q13, which are hHb1, hHb3 and bHb6 type-2 keratins within the hair shaft, resulting in structural fragility and susceptibility to breakage.[2] The autosomal recessive monilethrix is caused by variants in *DSG4*gene, which encodes desmoglein 4 (DSG4).[3]

Globally, the prevalence estimates vary. Some studies indicate an incidence of approximately 1 in 100,000 individuals, making it a relatively uncommon condition.

It affects both males and females, often presenting in childhood or early infancy.

Monilethrix typically manifests as fragile, brittle hair prone to breakage, resulting in short, sparse, and irregularly shaped hair. Under microscopic examination, the affected hair shafts appear as nodes and internodes, resembling a beaded or necklace-like structure. This condition often leads to patchy hair loss, varying in severity from mild thinning to more pronounced areas of baldness, and it can affect different areas of the scalp, eyebrows, eyelashes, pubic, axillary, and general body hair. It is also commonly associated with keratotic papules over the neck and upper trunk, koilonychia and rarely, systemic disturbances such as mental and physical retardation, syndactyly, cataract, teeth, and nail anomalies.[2]

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Diagnosing Monilethrix typically involves a combination of clinical evaluation, examination of the characteristic hair abnormalities under a microscope, trichoscope, and sometimes a skin biopsy for definitive confirmation. On trichoscopy, there is presence of nodes and internodes at regular intervals. Hair shaft appears as beaded caused by the presence of elliptical nodes separated by narrow internodes. Nodes are less than 2 mm apart; usually 0.7–1 mm. The hairs break spontaneously at about 0.5 to 2.5 cm apart. Nodes consist of medulla and have a diameter of normal hair while internodes are devoid of medulla and thus, are the site of fracture. This is also known as “regularly beaded ribbon” sign.[4]

The disease is often life long, but there are numerous cases of affected individuals recovering in adolescent life and during pregnancy. Some cases even report spontaneous resolution.[5]

For managing these patients, avoiding trauma to the hair is the most effective step. Treatment modalities like griseofulvin, retinoids, topical 2% minoxidil, N acetyl cysteine have shown temporary and reversible improvement in few cases.[6] Karincoaglu *et al*. described good results with oral acitretin in a 7-year-old girl.[7] Artificial hair pieces or wigs for better cosmesis can also be considered. But there is still no definitive treatment recognized yet.

CONCLUSION

Monilethrix is a rare condition with characteristic findings. Trichoscopy in our case confirmed the characteristic beaded hair appearance, thereby facilitating a faster and easier diagnosis without the need for invasive procedures like a skin biopsy. We are reporting this case because of its rare presentation and sporadic nature.

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