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**Case Report**

# Seven Year old Saudi Female with Monilethrix

**Saleh Mesfer Alghamdi, Hamad Alfahaad*, Abdullah Moshrif Aladnan****

Department of Dermatology, Khamis Mushait General Hospital, Aseer Region, Saudi Arabia.
* Department of Dermatology, College of Medicine, Najran University, Najran, Saudi Arabia.
** Department of Dermatology, King Fahad Hospital, Albaha, Saudi Arabia.

**Abstract** Monilethrix is a rare genetic hair disorder known as beaded or nodal hair disease as described by Walter Smith in 1897. It is characterized by hypotrichoses of the scalp of all races and both sexes. It is inherited as autosomal dominant even though many sporadic cases and autosomal recessive inheritances have been reported. Affected individuals usually have keratosis pilaris (KP) and koilonychia. Juvenile cataracts, mental and physical retardation, epilepsy, syndactyly, and tooth and nail abnormalities rarely reported. This case reports a 7-year-old female, who manifested the symptoms of monilethrix –Syndrome. The patient has a brittle and sparse and did not grow to extended length. Combing was abandoned as many hairs were broken easily. On examination, most of the hairs were broken and had a striated appearance, bilateral superciliary madarosis. Her nails and teeth were normal and no ophthalmological abnormality. The occipital scalp and nape of neck showed asymptomatic keratotic papules, suggestive of keratosis pilaris. Microscopic examination revealed the characteristic uniform beading of the hair nodes at the distance of 0.5mm, thus confirming the diagnosis of monilethrix. This case is reported for its rarity and occurrence in the Middle Eastern population.

***Key words***
Monilethrix, Trichorrhexis nodosa, Sabouraudi syndrome.

## Introduction

Monilethrix is a rare inherited disorder of the hair. It is also known as beaded or nodose hair disease. This condition was described by Walter Smith in 1897.[1] The term is derived from Latin and Greek words monile, meaning necklace, and thrix, meaning hair respectively, which aptly describes the regular beaded appearance of the hair in this condition. Radcliff coined the term monilethrix,[2] which is also known as Sabouraudi syndrome after the comprehensive description of the disease by the syndrome's namesake.[3]

Monilethrix is a congenital hypotrichoses condition of the scalp and is found in all races and both sexes. It is inherited as autosomal dominant even though many sporadic cases and autosomal recessive inheritances have been reported. The defect appears to be in chromosome 12q13.[4] The hair follicles may become filled with defective keratin and the root sheaths are imperfect.[5] In addition, the internodal hair shaft has defective keratin and breaks easily, giving the appearance of alopecia. The thinner internodal shaft is abnormal, but the nodes evidence normal hair growth. The inter node is characterized by wrinkling and invagination of outer cuticle cells, absence of medulla cytoplasmic vacuolation and abnormal tonofilaments.[6] The scalp hairs usually appear normal immediately after birth but later start to break off, mainly from the occiput. Apart from the scalp hairs, eyebrows, eye lashes and body hair that includes pubic and axillary hair may also be affected.[7] These hairs are short, sparse, dry and fragile. They have a uniform beaded

**Address for correspondence**
Dr. Hamad Alfahaad
College of Medicine, Najran University,
King Abdulaziz Road, Najran, Saudi Arabia.
P.O. Box 1988.
Email: hamadyam@gmail.com.

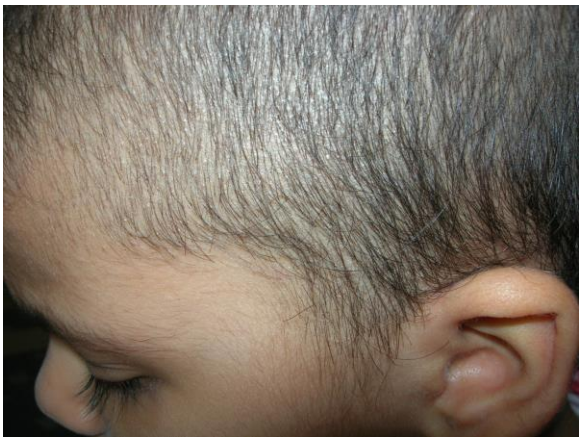

**Figure 1** Hypotrichosis of scalp hairs which are broken with a striated appearance and partial hair loss in the lateral eyebrow.

appearance, and the nodes are less than 2 mm apart; usually 0.7–1mm. the hairs break spontaneously at about 0.5 to 2.5 cm, and the resulting broken short hair has the appearance of having been burnt. 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.[8]

Affected individuals are generally healthy aside from the manifestations described here. Keratosis pilaris (KP) is almost always present, mainly on the occipital scalp and nape of the neck. Sometimes KP is also found on the extensor aspects of limbs. Koilonychia and rare juvenile cataracts are reported as other ectodermal defects. Other rare symptoms include very severe systemic disturbances such as mental and physical retardation, epilepsy, syndactyly, and tooth and nail abnormalities.[9]

Monilethrix can be diagnosed by examining hairs under light microscopy for the characteristic uniform beaded appearance of the hair shaft. Electron scanning microscopy, dermoscopy and video dermoscopy (trichoscopy) have also been used for diagnosis.[10] This disease has to be differentiated from pili torti and trichorrhexis nodosa (Nethorton Syndrome), and a rare syndrome known as pseudomonolethrix has been described where the nodes are irregular with normal hair shaft caliber as an entity in a family of many generations.[11]

There is no effective treatment for this condition, and avoidance of trauma is the most effective way to reduce the breaking of hairs. Topical minoxidil application[12] and oral retinoids[13,14] have been used for this purpose.

**Case Report**

A 7-year-old Saudi girl was brought to our clinic for weak and broken hairs. She was the only child of her parents, who were cousins. None of the other family members of both parents had similar hair loss. She was healthy with no other problems and of normal intelligence. Her scalp hair was apparently normal at birth, but soon had become brittle and sparse and did not grow to extended length. Combing was abandoned as many hairs were broken by the simple task. On examination, she was found to have sparse rough small scalp hairs, which were light brownish in color. Most of the hairs were broken and had a striated appearance, appearing burnt at the ends. She had bilateral superciliary madarosis and no ophthalmological abnormality **(Figure 1)**. Her nails and teeth were normal, but the occipital scalp and nape of neck showed asymptomatic keratotic papules, suggestive of keratosis pilaris **(Figure 2)**. The remainder of her skin and oral mucosa were normal. Routine laboratory investigations, including tests for amino acidurias, ceruloplasmin, copper and zinc levels, were normal. Examination of the hair under low power light microscopy revealed the characteristic uniform beading of the hair nodes at the distance of 0.5mm, thus confirming the diagnosis of monilethrix. Trichorrhexis nodosa (Nethorton's syndrome) was ruled out by the absence of endogenous eczema, erythroderma,

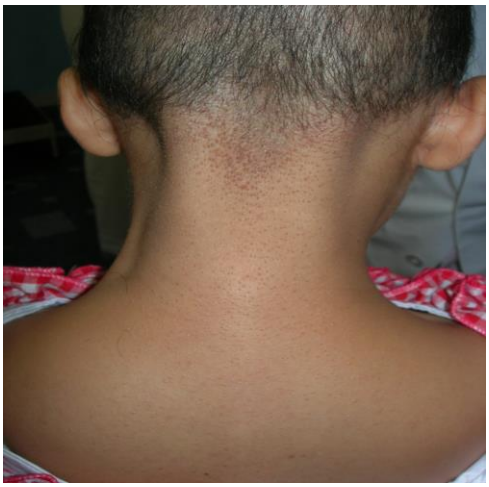

**Figure 2** Keratosis pilaris in the nape of the neck.

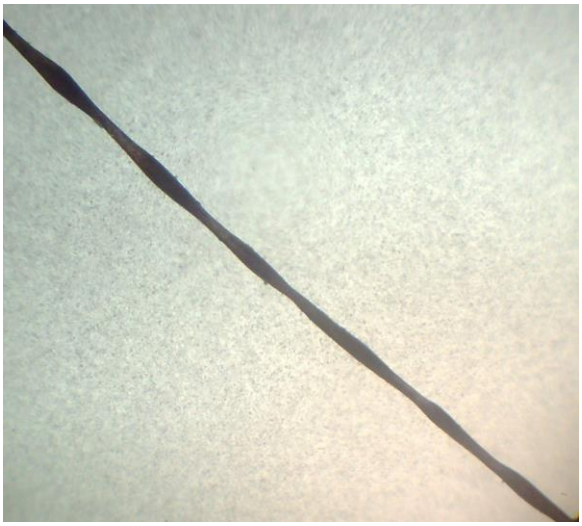

**Figure 3** Light microscopy showing typical beading of hair shaft.

intractable pruritus, increased Eosinophil count in the blood and the typical broken hairs. Pili torti was also ruled out as it manifests with twisted hairs like ribbon: this child had typical nodes in the hair shaft **(Figure 3)**. Her parents were advised of the condition of her hair and the importance of avoiding trauma, vigorous combing and cosmetic hair applications. She was given topical minoxidil 2%.

This case is reported for its rarity and occurrence in the Middle Eastern population.

## References


1. Smith, W. G. A rare nodose condition of the hair. *BMJ*. 1879;**11**:291-6.
2. Croaker, H. R. Diseases of the skin: Their description, Pathology, Diagnosis and Treatment. First Edition, 1888, Publisher H.K. Lewis.
3. Sabouroud, R. Sur Les Cheveux moniliformis (Trichorrhexies et monilethrix). *Annals de Dermatol et de Venereol*.1893;**3**:781-93.
4. Korge, B. P., Hamm, H., Jury, C. S. *et al*. Identification of novel mutations in the basic hair keratin hHb1 and hHb6 in monilethrix: Implication for protein structure and clinical phenotype. *J Invest Dermatol*. 1999; **133**:607-12.
5. De Barker, D. A. R., Dawber, R. P. Variations in the beading configurations in monilethrix. *Pediatr Dermatol*. 1992;**9**:19-21.
6. Ito, M., Hashimoto, K., Katsuumi, K. Pathogenesis of Monilethrix: Computer stereography and electron microscopy. *J Invest Dermatol*. 1990;**95**:186-94.
7. Rarediseases.info.nih.gov/GARD/condition/93/GnA/21427/monilethrix.aspx
8. Solomon, I. L., Green, D. C. Monilethrix. *N Eng J Med*.1963;**269**:1279-85.
9. Erbaqzi, Z.,Erbaqzi, I., Erbaqzi, H. Severe monilethrix associated with intractable Scalp pruritus, subcapsular cataract Brachiocephaly and distinct facial features: a new variant of monilethrix syndrome. *Pediat Dermatol*. 2004;**21**:486-90.
10. Rakowska, A., Slowinska, M., Czuwara, J. Dermascopy as a tool for rapid diagnosis of monilethrix. *J Drugs Dermatol*. 2007; **6**:222-4.
11. Bentley, P. B., Bayles, M. A. A previously undescribed heriditary hair anamoly (pseudomonilethrix) *Br J Dermatol*. 1973; **89**:159-67.
12. Rossi, A., Lorio, A., Scali, E. Monilethrix treated with minoxidil. *Int J Immunopathol Pharmacol*. 2011;**24**:239-42.
13. Karingauglu, Y., Basak, K.C., Muammar, E. S. Monilethrix: Improvement with Acetretin. *Am J Clin Dermatol*. 2005;**6**:407-10.
14. De Berker, D., Dawber, R. P. Monilethrix treated with oral retinoids. *Clin Exper Dermatol*. 1991;**16**:226-28.