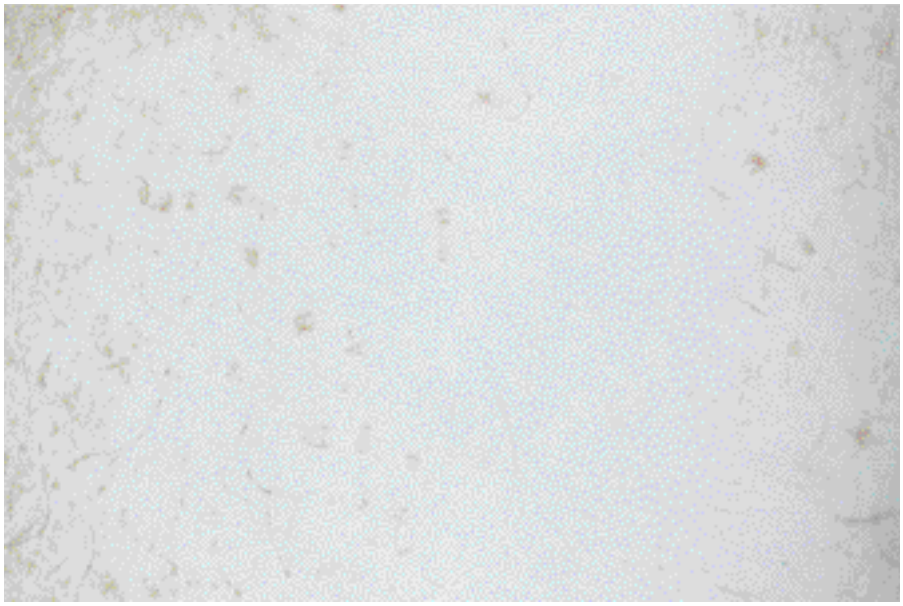
# What Is Your Diagnosis?



A 44-year-old man presented with brittle scalp hair of varying lengths.

PLEASE TURN TO PAGE 453 FOR DISCUSSION

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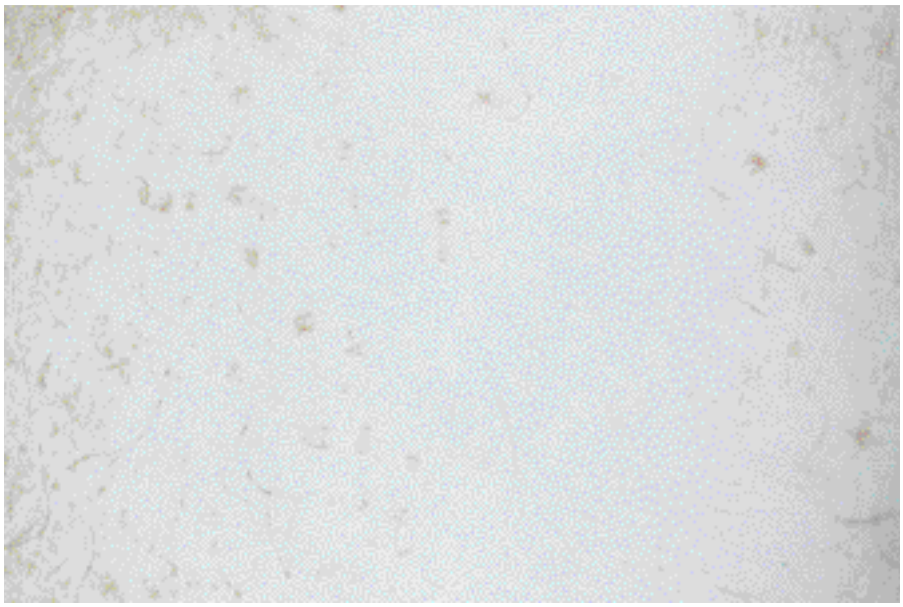
# The Diagnosis: Monilethrix



Our patient has had monilethrix since early childhood and noted some improvement in his early twenties. His maternal grandmother, 2 sons, and brother also have this condition, but one of his sons and his brother are only mildly affected. His mother and sister do not have the condition. Multiple patches of keratosis pilaris also were noted on physical examination.

The authors report no conflict of interest.

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This condition was first described by Walter Smith in 1879 and was later termed *monilethrix*.[1-3] *Monile* is Latin for necklace, and *thrix* is Greek for hair.[1,3,4] It is a rare, autosomal-dominant hair shaft disorder, with incomplete penetrance and variable expressivity.[1,2,4-6] Some autosomal-recessive cases have been reported, and infrequent reports of monilethrix occurring in association with other medical conditions have been published.[1,4]

Monilethrix can occur at any age but is more common in early childhood.[1,2] Alopecia is often a presenting symptom because the condition causes affected hairs to become brittle, dry, and lusterless,

with a tendency to fracture spontaneously from mild trauma.[1,4] Although scalp hair is predominately affected, all body hair may be involved in severe cases.[1,6] Other associated symptoms include follicular papules and soft or fragile nails.[5,7] Keratosis pilaris, when present on the back of the neck, is an especially helpful indicator of the diagnosis.[1] Up to 90% of patients with monilethrix have follicular keratosis.[5] Note the patient's flexural involvement.

Clinical suspicion and family history of monilethrix aid in the diagnosis, but confirmation is made by microscopic study of affected hairs, which demonstrate a beaded appearance under light microscopy. These beads (or nodes) are spaced up to 1 mm apart by thin internodal areas.[8] The nodes are elliptical and regular.[1,8] On cross section of affected hair, the nodes are more than twice the diameter of the internodes.[4] The internodes appear as tapered, nonmedullated, untwisted constrictions.[1] Nodes often show wear of the cuticle, while interruption in the scale at the internode corresponds with the longitudinal ridging or fluted appearance.[1,8,9] The grooves become progressively distinct toward the terminal ends of the hair.[1,8,9] The internodes are the point of weakness, and breakage at the internodes results in failure of affected hairs to attain normal lengths. This phenomenon accounts for the presentation of alopecia in patients with monilethrix.[1,4]

Healy et al[10] were the first to release results of genetic studies that mapped a link between monilethrix and the type II keratin on chromosome 12q13. The specific location of the structural protein defect are mutations in the hHb1 and hHb6 genes of type II hair keratin.[5,6] However, another study has reported cases without a link to these genes, indicating genetic heterogeneity.[11]

Pseudomonilethrix can be differentiated from monilethrix on light microscopy by the presence of irregular nodes, hair shaft depressions, irregular twists in the shafts, and breaks in normal-appearing shafts with brushlike ends. On physical examination, an absence of keratosis pilaris is noted. Some conditions are thought to result from trauma because the symptoms commonly resolve when vigorous hair dressing ceases.[1] In other cases, the appearance of beading is an artifact of hair compression during collection and examination.

There is no specific or effective treatment for monilethrix. In some patients, the condition tends to improve with age, pregnancy, or after puberty, while in others it persists throughout their lifetime.[1,2,5] Some lengthening of the hair can be achieved by avoiding trauma to the hair.[1] Improvement has been reported in isolated cases with oral retinoids, pregnancy, griseofulvin, L-cystine, and topical tretinoin.[1,3,7,12] Spontaneous improvement also has been reported.[2,3]

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