

# Uneven Hair Breakage in a 17-Year-Old Girl

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A 17-year-old white girl presented for evaluation of “hair loss” that had begun approximately 1 year prior to presentation. Pertinent patient history included emotional stress and use of heating tools to straighten and blow dry her hair, with reported use of 3 times per week. She had no other significant medical history, and she took no medications.

## Physical examination

The patient appeared comfortable and well-nourished, and no signs of developmental delay were apparent. Examination of the right side of the scalp revealed mild distal hair breakage with multiple small nodes on the proximal hair shaft (Figure 1). Hair in this area was also significantly shorter than the surrounding hair (Figure 2). The remainder of the scalp, head, and neck examinations were unremarkable. A focused review of systems, including immunologic, endocrine, and integumentary,



**Figure 1.** Distal hair breakage with multiple small nodes on the proximal hair shaft.

disclosed no abnormal findings. The patient also denied any history of psychological disorders or inability to resist the urge to pull out hair.



**Figure 2.** Decreased hair length in area of breakage.

**Based on the patient's presentation, what is the most likely diagnosis?**

- A. Monilethrix
- B. Trichorrhesis invaginata
- C. Trichorrhesis nodosa
- D. Trichotillomania

**Answer: C. Trichorrhesis nodosa**

The patient lacked features indicative of a genetic cause of the hair breakage. Although she did not use styling tools excessively, her occasional use, combined with the clinical presentation and otherwise inconsequential findings, made acquired trichorrhesis nodosa (TN) the most likely diagnosis. Nonpolarized dermoscopy showed a pattern similar to that of a fraying paintbrush, confirming the diagnosis.

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The authors report no relevant financial relationships.

## DISCLAIMER:

The authors report that informed patient consent was obtained for publication of the images used herein.

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## Discussion

Trichorrhhexis invaginata (TI) is a hair shaft abnormality resulting from intermittent areas of fully keratinized hair invaginating weakened areas with incomplete keratinization. This process occurs at several points along the hair shaft, creating a “bamboo-like” appearance.<sup>1,2</sup> Visualization with trichoscopy demonstrates small nodules along the hair shaft. These points of intussuscepted hair are more susceptible to breakage. TI can arise sporadically but is most often observed in association with Netherton syndrome, a rare autosomal recessive condition characterized by ichthyosiform eruptions and atopy.<sup>3,4</sup> The ectodermal features of Netherton syndrome more commonly present in female patients, classically those ranging from 1 to 5 years of age.<sup>2</sup> TI typically presents in infancy, and in this case, the patient’s age at onset and lack of atopic features make TI an unlikely diagnosis.

Monilethrix is a rare disorder of the hair shaft that often results from heterozygous mutations in the hair-specific keratin genes, *KRT81*, *KRT83*, and *KRT86*. These hair-specific keratin gene mutations are often inherited in an autosomal-dominant fashion. Individuals with monilethrix present with regular thinning of the hair shaft, which causes the hair shaft to resemble beads on a string.<sup>5</sup> Ultimately, the thinning of the shaft increases fragility of the hair, which often results in patchy, dystrophic alopecia. The typical onset of monilethrix is a few months after birth in early childhood.<sup>6</sup> Some data suggest that in a subset of women, the damaged hair may regrow during puberty or during pregnancy. This suggests that hormones may play a role in improving monilethrix.<sup>5</sup> Given that this patient presents at age 17 with worsening hair breakage and no prior history of hair thinning in childhood, it is unlikely that she has monilethrix.

Trichotillomania is a form of noncicatricial alopecia characterized by repetitive pulling of one’s hair that results in significant psychological distress or functional impairment. Trichotillomania is a psychiatric condition listed in the “Obses-

sive-Compulsive and Related Disorders” chapter of the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition.<sup>7</sup> Trichotillomania is more common in women and typically has an onset between 10 and 13 years of age. While the cause of trichotillomania is likely multifactorial, it is thought that there may be a genetic basis to the illness. This genetic basis is supported by a study showing a significantly higher concordance rate of trichotillomania in monozygotic twins (38.1%) when compared with dizygotic twins (0%). Often, individuals with trichotillomania tend to have comorbid body-focused conditions, including nail biting, nail picking, and skin picking.<sup>8</sup>

Ultimately, trichotillomania typically presents as patchy areas of alopecia with no evidence of inflammation.<sup>9</sup> The area of alopecia may contain many broken hairs or hairs regrowing at different lengths.<sup>10</sup> The presence of small nodes in the hair shaft of this patient makes the diagnosis of trichotillomania unlikely. Additionally, the lack of psychiatric history and denial of hair pulling during examination further lowers trichotillomania on the list of differential diagnoses.

TN is a hair shaft disorder characterized by weakened points called nodes that lead to fragility and result in premature breakage.<sup>11</sup> Individuals with this disorder exhibit fractured sites along the hair shaft that resemble the fraying pattern of paint brushes when visualized on trichoscopy.<sup>6</sup> TN can be acquired or congenital. Harsh styling practices that involve heat, excessive brushing, chemicals, or hair traction can lead to development of this disease. Congenital causes include genetic conditions, such as Menkes disease, Kabuki syndrome, and argininosuccinic aciduria.

The gold standard for diagnosis of acquired TN is scanning electron microscopy, although this study is not feasible in most clinical settings. Providers may opt for the use of dermatoscope with nonpolarized light alongside meticulous history taking.<sup>12</sup>

The management of TN is achieved by protecting the hair shaft through utiliza-

tion of a healthy hair care regimen. This regimen can involve the cessation of heat and chemical use and excessive brushing while implementing protective styles that refrain from hair traction. Cleansing and conditioning the hair can also have a substantial impact on TN reversal. Use of shampoos that contain amphoteric or nonionic surfactants tend to have a less damaging effect than their anionic counterparts.<sup>13</sup> Conditioners mimic the moisturizing role of sebum and will counteract the damaging effect of brushing. The “soak and smear” method is also recommended to patients, which involves applying an occlusive moisturizer or oil (such as olive oil or coconut oil) to damp, and conditioned hair.<sup>13</sup> Acquired TN is likely to self-resolve with a consistent and healthy hair care routine that incorporates such practices as those stated above.

## Conclusion

This case demonstrates the importance of a detailed history and thorough physical examination to rule out diseases that have a presentation that is similar to that of TN. Acquired TN is a reversible hair shaft disorder that occurs via mechanical and chemical damage. Treatment of this disorder focuses on preserving the hair shaft through avoidance of the offending agents in conjunction with a nourishing regimen.

## Patient outcome

The patient was reassured that her hair shaft disorder was not a sign of any serious underlying health conditions, and that acquired TN may take anywhere from 2 to 4 years to fully resolve. Additionally, the patient was instructed to avoid shampoos containing sulfates and sulfonates and to use a detangling leave-in conditioner after shampooing. Her hair should be allowed to air-dry whenever possible, and tight ponytail or updo hairstyles should be avoided while the hair is wet.

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