

Rapid and Easy Diagnosis of Netherton Syndrome with Dermoscopy

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Background: Netherton syndrome is a rare autosomal recessive disease demonstrating ichthyosis linearis circumflexa, atopic findings, and hair shaft anomalies. Trichorrhexis invaginata is the pathognomonic hair shaft anomaly seen in this syndrome.

Objective: In recent years, hair shaft anomalies have been described as “matchstick” and “golf tee” signs. We present a patient with Netherton syndrome diagnosed by the presence of matchstick and golf tee hairs in addition to trichorrhexis invaginata.

Contexte: Le syndrome de Netherton est une maladie autosomique récessive rare, qui se manifeste par une ichthyose linéaire circonflexe, des signes atopiques et des anomalies de la tige pileaire. La trichorrexie invaginée est l'anomalie pileaire pathognomonique du syndrome.

Objectif: Au cours des dernières années, des expressions telles que «tige d'allumette» ou «té de golf» ont été employées pour décrire les anomalies de la tige pileaire. Sera exposé dans l'article un cas de syndrome de Netherton dans lequel la présence de tiges pileaires en «tige d'allumette» ou en «té de golf», outre celle de la trichorrexie invaginée, a signé le diagnostic.

Case Report

A 30-month-old male presented with recurrent widespread pruritic skin lesions responding to topical steroids. There was no history of repeated infections. He was the first-born child of a mother and father who were second-degree cousins. There was no family history of atopy.

A dermatologic examination revealed widespread ichthyosis and typical lesions of ichthyosis linearis circumflexa on the extremities demonstrating a linear and tram-track appearance (Figure 1). A physical examination revealed weight and height below the third percentile. Routine blood biochemical values were normal except for slight eosinophilia and increased total IgE. Immunoglobulins, immunization responses, flow cytometry of peripheral lymphocytes, growth hormone and cortisol values, and celiac antibodies were normal. Skin-prick testing was

not done due to the severe pruritus, leading to the use of antihistaminics. Food-specific IgE demonstrated increased egg white-specific IgE. Dermoscopy of eyebrow and scalp hair revealed “matchstick” and “golf tee” hair in addition to trichorrhexis invaginata (Figure 2 and Figure 3), and a diagnosis of Netherton syndrome was made. Topical emollients and steroids and removal of egg white from the diet were advised. Two weeks later, the skin lesions had almost completely regressed.

Discussion

Netherton syndrome is an autosomal recessive disorder first described by Netherton in 1958.¹ It is characterized by ichthyosis, atopic manifestations, and hair shaft abnormalities (trichorrhexis invaginata).² In recent years, several mutations in the *SPINK5* gene encoding LEKTI (lympho-epithelial Kazal type-related inhibitor) have been reported in these patients,³ and abnormality in cutaneous expression of LEKTI has been described.⁴ Clinically, the hair appears sparse, dull, brittle, and short.² Microscopically, the distal portion of the hair shaft invaginates into the proximal portion, forming a “ball in cup” appearance. Being pathognomonic for Netherton syndrome, these hairs are termed “bamboo hair” or “trichorrhexis invaginata.”⁵ The hairs of patients break easily when exposed to weak traction because of a reduced number of disulfide bonds. This

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Figure 1. Ichthyosis linearis circumflexa on the arm.

suggests a reduced crosslinkage of keratin structures and a weaker coherence of cortical cells.⁶ When hair breaks at the point of invagination, the proximal shaft with its cup-shaped end remains. This appearance is named “golf tee,” a term coined by de Berker and colleagues in 1995.⁵ de Berker and colleagues also reported that hair shaft anomalies may be visualized in eyebrow hair, a valuable method in diagnosis.⁵ In addition, Powell and colleagues evaluated three cases with Netherton syndrome and found that hair shaft anomalies such as trichorrhexis invaginata and “golf tee” were more frequent in eyebrow hair compared to scalp hair.⁷ The golf tee appearance has been described by other authors as well.^{2,6,8}



Figure 2. Trichorrhexis invaginata or bamboo hair, temporal scalp ($\times 20$ original magnification).

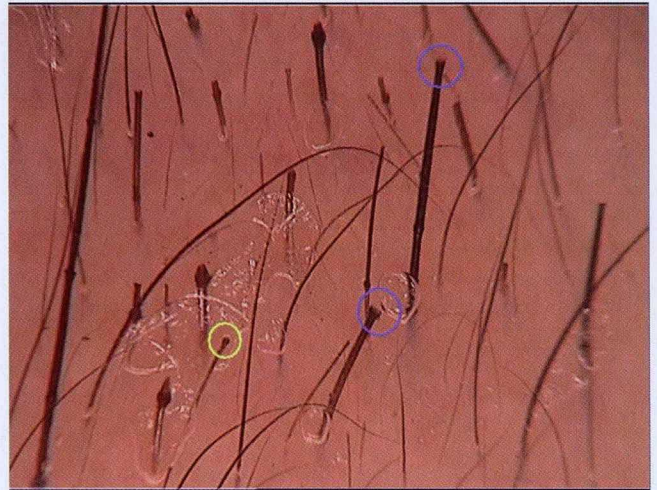


Figure 3. Matchstick and golf tee hair, eyebrow ($\times 20$ original magnification).

Goujon and colleagues introduced the term “matchstick” to describe a broken hair shaft with a bulging end.⁸ Neri and colleagues demonstrated that all three findings could be visualized in eyelashes.⁶

Although a single bamboo hair is sufficient to establish the diagnosis in Netherton syndrome, this pathognomonic hair feature is not easy to find. Several hair samples might be necessary before obtaining the diagnostic specimen.⁷ Moreover, hair growth is sparse, and hair shaft abnormalities are not usually detectable in the first months of life in affected children and increase with age. Despite identification of the *SPINK5* gene, the costs of molecular and genetic tests limit its use in diagnosis. Videodermoscopy is a very handy tool in rapidly visualizing abnormal hairs in affected children without causing any trauma.⁶

It is not exactly known how the matchstick sign is formed. We believe that the proximal hair shaft invaginates into the distal hair shaft, and when breakage occurs, the bulging end of the proximal hair shaft remains.

Conclusion

Hair shaft anomalies should be searched for by dermoscopy in the eyebrows and eyelashes of patients with Netherton syndrome. The recently described matchstick and golf tee signs render faster and easier diagnosis.

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