Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Dermatology

Trichoscopic Signs of Netherton Syndrome: A Comprehensive Case-Based Insight

Rasha Moumna[1*], Meryem Marhaoui[1], Ghita Filali Baba[1], Laila Benzekri[1], Nadia Ismaili[1]

[1]Dermatology Department, Ibn Sina University Hospital, Rabat, Morocco

DOI: https://doi.org/10.36347/sjmcr.2025.v13i04.028 | **Received:** 09.03.2025 | **Accepted:** 20.04.2025 | **Published:** 22.04.2025

***Corresponding author:** Rasha Moumna, MD
Dermatology Department, Ibn Sina University Hospital, Rabat, Morocco

Abstract | **Case Report**

Netherton syndrome (NS) is a rare autosomal recessive genodermatosis characterized by a triad of ichthyosis linearis circumflexa, atopic diathesis, and distinctive hair shaft anomalies. Its diagnosis is often delayed, particularly in early infancy, due to incomplete or nonspecific clinical presentations. Among the cutaneous manifestations, hair shaft anomalies tend to present early and provide highly specific diagnostic clues. We report the case of a six-month-old girl presenting with persistent pruritic and scaly dermatitis, associated with short, dull scalp hair and sparse eyebrows. Trichoscopic examination revealed a rich combination of hair shaft anomalies, including trichorrhexis invaginata (bamboo hairs), matchstick hairs, golf tee hairs, pili torti, and trichorrhexis nodosa. These findings, although underdocumented in the literature, are highly specific and collectively supported the diagnosis of NS in the absence of ichthyosis linearis circumflexa or initial genetic confirmation. This case highlights the essential role of trichoscopy as a rapid, non-invasive diagnostic tool in infants with unexplained xerosis, flexural dermatitis or erythroderma. Early recognition of NS through its trichoscopic features can guide appropriate management and timely referral for genetic counseling, even in the absence of full phenotypic expression.

Keywords: Netherton Syndrome**,** Trichoscopy**,** Hair Shaft Anomalies**,** Trichorrhexis Invaginata**,** Pediatric Genodermatoses.

INTRODUCTION

Netherton syndrome (NS) is a very rare and potentially severe genodermatosis inherited in an autosomal recessive pattern. Despite a better understanding of its clinical features, making an accurate and timely diagnosis remains challenging, especially in the early stages of the disease when the manifestations are subtle or nonspecific.

Trichoscopy has emerged as a simple, rapid, and non-invasive tool that can confirm the diagnosis of NS by identifying its characteristic features. While trichorrhexis invaginata is the most recognized hair shaft anomaly in NS, other features have been reported as highly specific to the diagnosis, yet remain widely underdocumented.

Here, we report a case of NS with subtle clinical manifestations, in which trichoscopy proved essential in uncovering the diagnosis by revealing the full spectrum of hair shaft anomalies.

CASE REPORT

A six-month-old girl, born to a second-degree consanguineous marriage, with a history of atopic diathesis and recurrent upper respiratory tract infections, presented with a persistent, pruritic, and scaly dermatosis evolving over the past four months. Dermatological examination revealed diffuse xerosis and erythematous scaly patches, predominantly affecting the trunk and flexural areas, especially the neck folds and diaper region (Figure 1).

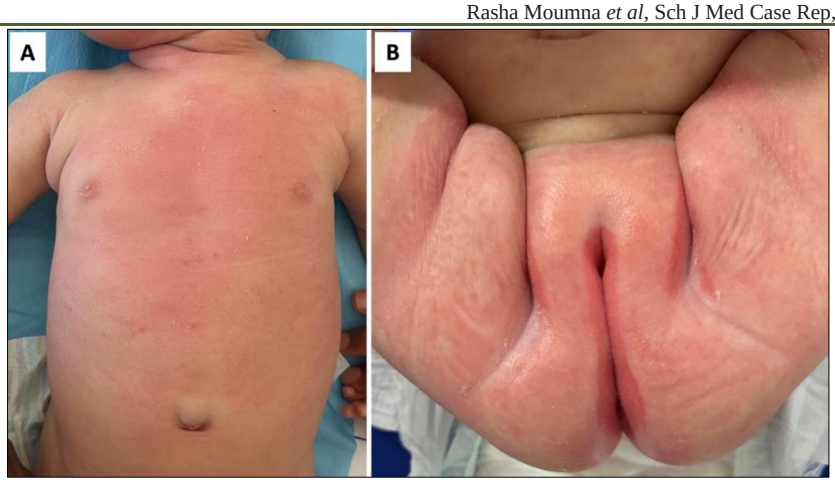

Figure 1: Clinical features on admission: (A) Diffuse xerosis and erythematous, scaly patches over the trunk. (B) Well-demarcated erythema with fine scaling involving the intertriginous areas of the diaper region.

Initial differential diagnoses included atopic dermatitis, psoriasis, and seborrheic dermatitis. However, the presence of short, dull hair and sparse eyebrows (Figure 2) prompted a trichoscopic evaluation.

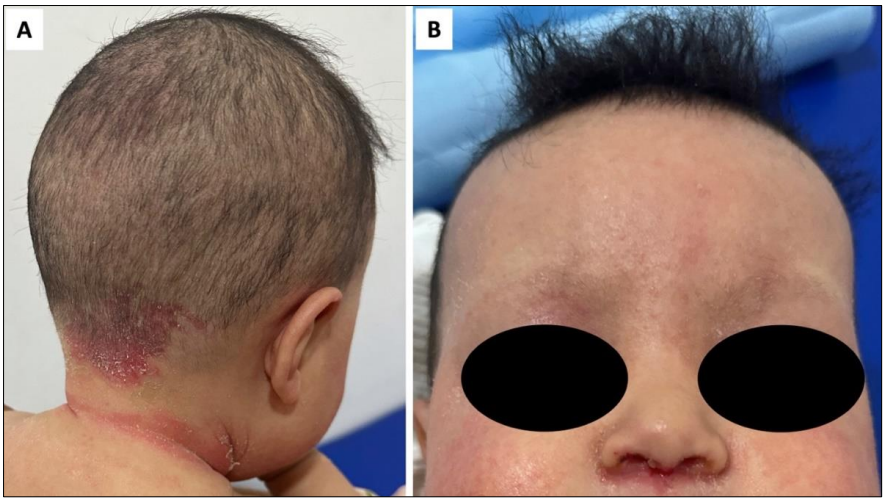

Figure 2: Erythematous and scaly plaques on the neck folds (A), associated with short, dull hair and sparse eyebrows (B)

Trichoscopy revealed numerous trichorrhexis invaginata, forming the characteristic bamboo hairs. Additional findings included multiple golf-tee and matchstick hairs, along with occasional pili torti and trichorrhexis nodosa (Figure 3). These findings strongly supported the diagnosis of Netherton syndrome. The patient was referred for genetic counseling and multidisciplinary follow-up.

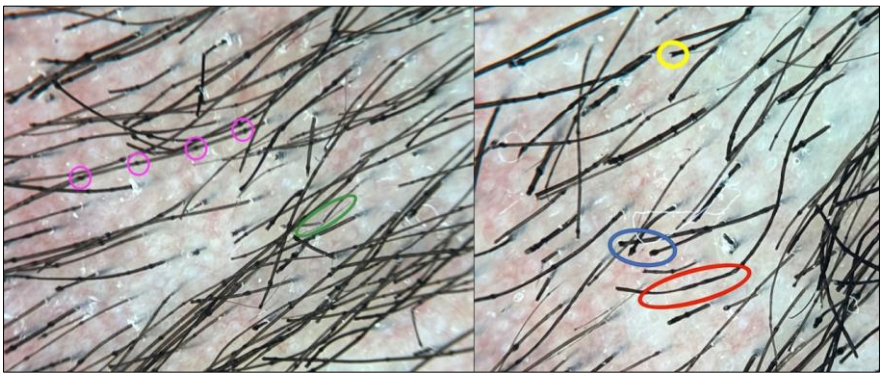

Figure 3: Trichoscopic examination showing multiple trichorrhexis invaginata (pink), forming bamboo hairs, along with matchstick hairs (blue), golf tee hairs (yellow), trichorrhexis nodosa (green), and pili torti (red)

DISCUSSION

Netherton syndrome (NS) is a rare and potentially severe autosomal recessive genodermatosis caused by mutations in the SPINK5 gene, which encodes the serine protease inhibitor LEKTI. This dysfunction leads to a cascade of proteolytic activity, compromising the epidermal barrier and resulting in marked skin inflammation and increased allergen permeability. Clinically, NS is characterized by the classical triad of ichthyosis linearis circumflexa (ILC), hair shaft anomalies, and atopic diathesis [1].

Beyond the dermatologic manifestations, other clinical features have been described in NS, including intellectual disability, neurologic involvement (such as seizures or spastic diplegia), delayed growth, and recurrent infections affecting the skin, eyes, or respiratory tract. Immunologic imbalances, including hypo- or hypergammaglobulinemia, may further contribute to this increased susceptibility to infections [2].

Diagnosis remains challenging, often delayed by incomplete, atypical, or fluctuating presentations. In particular, ILC—the most recognizable cutaneous feature of NS—is highly variable and may not manifest until later in infancy. In the early months of life, clinical signs are typically subtle and nonspecific, ranging from isolated xerosis and flexural erythema to erythroderma. These manifestations are often misdiagnosed as more common inflammatory dermatoses such as atopic dermatitis, seborrheic dermatitis, or psoriasis, thereby contributing to diagnostic delay [2–5].

In contrast, certain hair shaft anomalies are highly specific to NS and can be detected very early in the disease course. Affected individuals often present with short, brittle hair and sparse eyebrows. The pathognomonic hair shaft anomaly in NS is trichorrhexis invaginata (TI), which results from intermittent cortical keratinization defects. It is characterized by the invagination of the distal portion of the hair shaft into its proximal segment, resulting in a distinctive 'ball-and-socket' appearance. Under trichoscopy, these appear as multiple small nodular swellings regularly distributed along the hair shaft, creating the typical "bamboo hair" appearance [2-6].

In our patient, trichoscopy revealed a distinctive combination of hair shaft anomalies highly suggestive of NS. In addition to the pathognomonic bamboo hairs, we identified several "golf tee" hairs, which appear as short, broken shafts with a sharply cupped proximal end, resembling the shape of a golf tee. This morphology results from hair breakage occurring at the point of invagination. We also observed multiple "matchstick" hairs, characterized by a rounded or slightly bulging distal tip, resulting from shaft breakage distal to the invagination and preservation of the proximal portion. Both golf tee and matchstick hairs reflect mechanical breakage at points of cortical weakness. Described more recently than trichorrhexis invaginata, these features are now considered as highly specific diagnostic clues and should be systematically assessed when NS is suspected [1-6].

Less frequently reported in NS, but also observed in our case, were pili torti—flat, twisted hair shafts rotated 180° along their axis at irregular intervals—and trichorrhexis nodosa, characterized by longitudinal splitting of the shaft into multiple fine fibers, producing frayed, brush-like ends at the site of breakage. Nodular thickenings may also be visible along the shaft, corresponding to structurally weak points [1, 2]. Although these trichoscopic findings are not specific to NS, as they are commonly seen in other genodermatoses, their presence in association with trichorrhexis invaginata, golf tee, or matchstick hairs helps reinforce the diagnosis [7].

In early infancy, eyebrow and eyelash hairs are often more informative than scalp hairs, as anomalies in these areas tend to appear earlier and in greater density [3-7]. In our case, sparse and brittle eyebrows offered an accessible and high-yield site for examination.

In conventional diagnostic approaches, the identification of hair shaft anomalies in Netherton syndrome relied on light microscopy of plucked hairs, often requiring slide preparation and, in some cases, polarized light to enhance structural detail [3-5]. While effective, these techniques are time-consuming, technically demanding, and may cause discomfort—particularly in infants.

Trichoscopy offers a simple, rapid, and non-invasive alternative. It allows in vivo, real-time visualization of hair shaft anomalies, without the need for hair plucking or slide preparation, making it especially suitable for pediatric patients [7]. This practical advantage reinforces its role as a first-line diagnostic tool in suspected Netherton syndrome.

CONCLUSION

Trichoscopy represents a highly valuable diagnostic tool in Netherton syndrome, particularly in its early stages, when cutaneous signs may be subtle or nonspecific. By revealing characteristic hair shaft anomalies, it facilitates early recognition and helps differentiate NS from more common inflammatory dermatoses encountered in the pediatric population.

In our case, the presence of the full spectrum of trichoscopic signs provided strong diagnostic support and enabled early recognition of NS, despite the absence of ichthyosis linearis circumflexa or genetic confirmation at the time of evaluation. Therefore, based on our experience in our genodermatoses referral center, we recommend the systematic use of trichoscopy in infants presenting with erythroderma, persistent xerosis,

or flexural erythema—particularly when associated with clinical hair findings such as sparse, brittle hair or eyebrows.

Acknowledgements: None. No funding or external assistance was received for the preparation of this manuscript.

REFERENCES

1. Herz-Ruelas, M. E., Chavez-Alvarez, S., Garza-Chapa, J. I., & others. (2021). Netherton syndrome: Case report and review of the literature. *Skin Appendage Disorders*, 7(5), 346–350. https://doi.org/10.1159/000514699
2. Sun, J. D., & Linden, K. G. (2006). Netherton syndrome: A case report and review of the literature. *International Journal of Dermatology*, 45(6), 693–697. https://doi.org/10.1111/j.1365-4632.2005.02637.x
3. Bangaru, H., & Ashwini, D. (2025). Trichoscopy in unveiling the triad of Netherton syndrome. *Clinical Dermatology Review*, 9(1), 76–78. https://doi.org/10.4103/cdr.cdr_42_23
4. Semblano Bittencourt, M. J., Mendes, A. D., Moure, E. R. D., & others. (2015). Trichoscopy as a diagnostic tool in trichorrhexis invaginata and Netherton syndrome. *Anais Brasileiros de Dermatologia*, 90(1), 114–116. https://doi.org/10.1590/abd1806-4841.20153011
5. Small, A. M., & Cordoro, K. M. (2016). Netherton syndrome mimicking pustular psoriasis: Clinical implications and response to intravenous immunoglobulin. *Pediatric Dermatology*, 33(3), e222–e223. https://doi.org/10.1111/pde.12856
6. Akkurt, Z. M., Tuncel, T., Ayhan, E., & others. (2014). Rapid and easy diagnosis of Netherton syndrome with dermoscopy. *Journal of Cutaneous Medicine and Surgery*, 18(4), 280–282. https://doi.org/10.2310/7750.2013.13106
7. Rudnicka, L., Olszewska, M., Waśkiel, A., & Rakowska, A. (2018). Trichoscopy in hair shaft disorders. *Dermatologic Clinics*, 36(4), 421–430. https://doi.org/10.1016/j.det.2018.05.009