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Late Diagnosis of Netherton Syndrome: A Moroccan Case Report

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Abstract

A shortage of the LEKTI protein results from mutations in the *SPINK5* gene (1), which causes Netherton syndrome, a rare autosomal recessive genodermatosis. Trichorrhexis invaginata, atopic symptoms, and congenital ichthyosiform erythroderma are the three clinical characteristics that define this illness. We describe the case of a Moroccan woman, age 26, who had a history of misdiagnosed atopic dermatitis and who subsequently developed alopecic patches, polycyclic scaling, and global erythroderma. The diagnosis of Netherton syndrome was confirmed by trichoscopic examination, which showed the pathognomonic feature of trichorrhexis invaginata. After receiving low-dose acitretin and emollients, the patient's condition significantly improved. This example demonstrates the difficulties in diagnosing Netherton syndrome and stresses the value of trichoscopy and early detection to avoid delays in the proper course of treatment. Dermatologists need to be more attentive in order to diagnose patients accurately and provide the best care possible.

Keywords

Netherton syndrome; *SPINK5* mutation; Trichorrhexis invaginata; Ichthyosis linearis circumflexa; Acitretin.

Introduction

A serious hereditary skin condition called Netherton syndrome is brought on by mutations in the *SPINK5* gene, which is found on chromosome 5q32 [1]. The LEKTI protein, a serine protease inhibitor necessary for preserving epidermal homeostasis [2], is encoded by this gene. Loss of LEKTI function [3] causes unchecked protease activity, which damages the skin barrier and causes persistent allergy symptoms, chronic inflammation, and an increased risk of infection [4]. The typical trio of congenital ichthyosiform erythroderma or ichthyosis linearis circumflexa, trichorrhexis invaginata (sometimes called bamboo hair), and severe atopic diathesis, which includes increased IgE levels, food allergies, and asthma, is how Netherton syndrome manifests clinically. Netherton syndrome is frequently misdiagnosed, which causes delays in receiving the right therapy, because of its varied presentation and similarities to other dermatological disorders such psoriasis and atopic dermatitis. To enhance patient outcomes and avoid problems, early detection and precise diagnosis are crucial [5].

Case Report

Our dermatology department received a referral for the evaluation of a Moroccan woman, age 26, who had severe erythroderma and intractable pruritus. She had been treated for atopic dermatitis since she was a baby, according to her medical history, and topical corticosteroids had only partially helped. When a skin biopsy was done at age 20, the results first pointed to Darier disease. Topical retinoid therapy, however, did not result in any appreciable improvement. The patient was examined and found to have generalized erythroderma with ichthyosis linearis circumflexa-like polycyclic, serpiginous scaling. She also had short, brittle hair and widespread alopecia. Trichorrhexis invaginata, the defining characteristic of Netherton syndrome, was confirmed by trichoscopic examination, which showed many nodules along the hair shafts. Ige levels (1,200 IU/mL) were significantly higher in laboratory tests, but other metabolic parameters were within normal ranges.

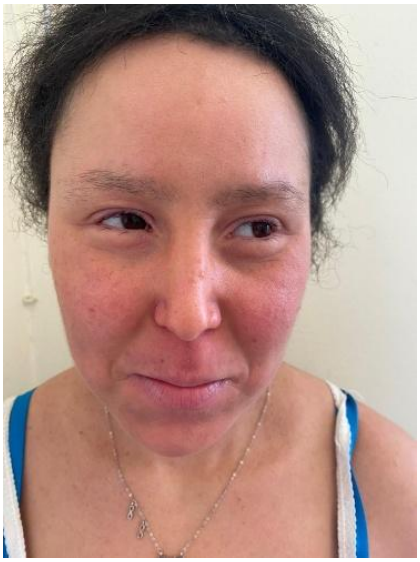

Figure 1: Generalized erythroderma with polycyclic scaling in a 26-year-old patient with Netherton syndrome.

Figure 2: Trichoscopic image showing trichorrhexis invaginata (bamboo hair), pathognomonic for Netherton syndrome.

Netherton syndrome was conclusively diagnosed based on the trichoscopic and clinical results. Due to resource constraints, genetic testing to detect *SPINK5* mutations were not carried out, despite the fact that it would have offered additional proof. In addition to rigorous emollient therapy and wet wrap bandages, the patient was initiated on a low-dose regimen of acitretin (10 mg daily). Her skin condition significantly improved over the next few months, exhibiting less itching and scaling. But the structural abnormalities of the hair continued, indicating that this illness is chronic.

Discussion

As demonstrated by our example of a 26-year-old Moroccan lady who was first misdiagnosed with atopic dermatitis and Darier disease, Netherton syndrome (NS) is a complex genodermatosis that presents major diagnostic and treatment hurdles. The pathogenesis of the illness is caused by mutations in SPINK5 that result in LEKTI deficiency. This leads to unchecked kallikrein activity, which interferes with the function of the epidermal barrier and causes severe atopy, recurrent infections, and chronic inflammation. Although trichoscopy reveals pathognomonic trichorrhexis invaginata, and genetic testing confirms SPINK5 mutations, the diagnosis is still difficult because of phenotypic overlap with common dermatoses. While new treatments like dupilumab show promise for reducing inflammation and pruritus, albeit with varied results, management necessitates a multidisciplinary strategy that weighs the advantages of low-dose systemic retinoids for scaling against their tendency to worsen skin fragility [5]. The condition's systemic signs - including growth retardation, hypernatremic dehydration in newborns, and immunodeficiency - further complicate management, underlining the necessity for comprehensive surveillance and supportive therapies. In order to better characterize the spectrum of this rare disorder and optimize therapeutic strategies, future directions will concentrate on creating biomarkers for disease activity, investigating gene-editing technologies, and setting up international registries. It will be emphasized that early detection and customized intervention are crucial for improving patient outcomes [6].

Conclusion

This instance emphasizes how crucial it is to keep a close eye out for Netherton syndrome in patients who exhibit anomalies of the hair shaft and refractory erythroderma. Trichoscopy and clinical correlation

provide early diagnosis, which is crucial for starting the right treatment and enhancing quality of life. Since this ailment is uncommon, it is essential that pediatricians and dermatologists become more aware of it in order to prevent delays in diagnosis. In order to properly treat the varied manifestations of this severe condition, future research should concentrate on increasing therapy choices, especially focused biologic medicines.

Conflict of Interest

None declared.

Ethical approval

Obtained from the institutional review board.

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