Netherton Syndrome: Diagnosis made easy with Trichoscopy

Samira Zobiri, Hanane Touati*

Mustapha Hospital, Dermatology Department, Algiers, Algeria

***Corresponding author:**

Samira Zobiri

Mustapha Hospital, Dermatology Department, Algiers, Algeria, Tel: 00213664746628

Email: samira_zobiri@yahoo.fr

Received : February 26, 2024

Published : April 10, 2024

ABSTRACT

Netherton syndrome is a rare autosomal recessive disorder. Congenital ichthyosiform erythroderma (CIE) or ichthyosis linearis circumflexa (ILC), hair shaft abnormalities, and atopic diathesis (elevated serum IgE) characterize it. Netherton syndrome is usually misdiagnosed as atopic dermatitis due to the presence of eczematous skin lesions and allergic problems. We report a case of a twelve-month-old girl misdiagnosed as atopic dermatitis in whom trichoscopy has found bamboo hair aspect making Netherton syndrome diagnosis formal.

Keywords: Trichoscopy, bamboo hair, trichorrhexis invaginata, Netherton syndrome

INTRODUCTION

Netherton syndrome is a rare autosomal recessive disorder described by Comel (1949) and Netherton (1958). This genodermatosis is classically distinguished by a triad of clinical manifestations: congenital ichthyosiform erythroderma [1], hair shaft abnormalities, and immune dysregulation such as atopy. Netherton syndrome is characterized with a specific hair shaft alteration, identified as trichorrhexis invaginata or bamboo hair with an invagination of the hair shaft [2,3]. An early diagnosis is crucial to start the correct management of these patients. In recent years, the application of trichoscopy has increased in identifying scalp hair shaft disorders, making it a simple tool for an early diagnosis of Netherton syndrome.

We report a case of a twelve-month-old girl misdiagnosed as atopic dermatitis in whom trichoscopy has found bamboo hair aspect making Netherton syndrome diagnosis formal.

CASE PRESENTATION

A twelve-month-old girl referred to our clinic with perioral erythema-squamous lesions, circinate erythematous-squamous lesion with double squamous border, severe generalized cutaneous xerosis and permanent moderate itching. Review of her medical record showed that she was born at the 37th week of gestation after a pregnancy of healthy related parents, the medical record showed that she had also a respiratory distress with broncho-pulmonary infection and

DOI: https://doi.org/10.35702/Derm.10013

the presence of skin lesions tagged as seborrheic dermatitis, eczema and nappy rash. She benefited from a sweat test, zinc level test, heart ultrasound, autoimmunity assessment and rhinofibroscopy, all of this exam returned normal. She was treated with topical steroids and emollient.

During the following months, the desquamation resolved, but ultimately the patient developed generalized, pruritic, erythematous lesions and she was referred to our clinic.

On physical examination, her skin was dry, and there were erythematous scaly patches on the abdomen, face, and extremities (Figures 1-2).

She presented short, brittle, greyish-black hair, with non-scarring area of alopecia, eyebrows showed short, brittle hairs and alopecic area (Figure 3). Trichoscopy found the pathognomonic aspect of trichorrhexis invaginata or bamboo hair aspect (Figures 4,5). Blood testing found hypereosinophilia 1830 element/uL, hyper IgE: 744UI/ml. Emollients were prescribed in association with topical corticosteroids for short periods.

DISCUSSION

Netherton syndrome is a rare congenital skin disorder that is inherited in an autosomal recessive pattern. It is a triad of congenital ichthyosiform erythroderma, trichorrhexis invaginata (TI), and an atopic diathesis [3,4]. It is characterized as premature desquamation of the stratum corneum and impairment of the skin barrier [5].

Mutations in the serine protease inhibitor (SPINK5), a gene located on chromosome 5q31-32 [6,7], result in increased activity of epidermal proteases which cause desquamation.

Each SPINK5 mutation leads to a different length of LEKTI protein, resulting in genotype/phenotype correlations in cutaneous severity, susceptibility to atopic dermatitis [4], and growth retardation, skin infection, increased stratum corneum protease activities [3], and elevated kallikrein levels in the stratum corneum. The prevalence of this condition is estimated to be 1/200.000 [8,9].

Congenital ichthyosiform erythroderma is the generalized erythroderma and desquamation present at birth [1]. It evolves into a migratory, serpiginous, erythematous, patches with double-edged scales at the periphery. This Ichthyosis linearis circumflexa waxes and wanes throughout the patient's life and is accompanied by pruritus [10].

Patients with Netherton's syndrome have sparse hair, which is dry, short, spiky, and brittle. The diagnosis of Netherton's syndrome may be definitely established by identifying at least one hair shaft with trichorrhexis invaginata [11].

The term trichorrhexis invaginata is derived from the Greek trichos, meaning hair, and rhexis, meaning rupture and "invagination" of hair shafts within themselves at the keratinization zone [12].

Trichorrhexis invaginata, also called bamboo hair, is an abnormality in which the hair shaft invaginates on itself at several points along the shaft. It occurs because of an intermittent keratinizing defect of the hair cortex [12,13].

Incomplete conversion of the sulfhydryl –SH group onto S-S disulfide bonds in the protein of the cortical fibers leads to cortical softness and subsequent invagination of the fully keratinized distal hair shaft into the softer, abnormally keratinized proximal hair shaft. Intussusception of the distal hair shaft into the proximal hair shaft results in a distinctive ball-and-socket hair shaft deformity [14].

On low-magnification trichoscopy, this appears as multiple small nodules spaced along the shaft at irregular intervals [15].

High-magnification trichoscpy shows an invagination of the distal portion of the hair shaft into its proximal portion, forming a "ball-in-cup" appearance, which is considered pathognomonic of Netherton's syndrome [16].

Occasionally, a cupped proximal hair ends in which the distal end has fractured might be seen. This abnormality is often referred to as "golf tee hairs" [17].

Recently, "matchstick" hairs were described in a patient with Netherton's syndrome [18]. These hairs are visible on a handheld dermoscope as short hair shafts with a bulging tip and are equivalent to golf tee hairs visible at higher magnifications.

Several authors have indicated that trichorrhexis invaginata and golf tee hairs are easiest to find on trichoscopy of the eyebrow area [13,17], because the number of lesions per millimeter of hair shaft is 10 times higher in the eyebrow area than the scalp in patients with Netherton's syndrome [18].

Trichorrhexis invaginata often improves with age and can

sometimes completely subside. Differential diagnosis of small dark nodules in hair axis also includes trichorrhexis nodosa, monilethrix and black piedra [19]. Other hair anomalies, such as pili torti, trichorrhexis nodosa, and helical hairs, may be found in patients with Netherton's syndrome but are not specific to the disease. The differential diagnosis includes Omenn syndrome, generalized seborrheic dermatitis, erythrodermic psoriasis, staphylococcal scalded skin syndrome and non-bullous ichthyosiform erythroderma [20,21].

In our case, Netherton syndrome was misdiagnosed as atopic dermatitis, prompt diagnosis was easily done using trichoscopy [15].

CONCLUSION

Netherton syndrome is a rare disease, which presents serious complications in the neonatal period due to dehydration, hypothermia, weight loss, respiratory infection and sepsis.

Trichoscopy visualizing TI and golf tee hair is painless, non-invasive diagnostic tool, accessible and precise in evaluating erythroderma and ichthyosis in infants and children.

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Citation: Zobiri S, et al. (2024). Netherton Syndrome: Diagnosis made easy with Trichoscopy. Dermis. 4(2):13.

DOI: https://doi.org/10.35702/Derm.10013