Case study

Neterthon syndrome revealed by recurrent skin infections: a case report

ABSTRACT

Netherton syndrome (NS) is a rare autosomal recessive genodermatosis caused by pathogenic variants of the SPINK5 gene. It is characterized by a triad of atopic terrain, circumflex linear ichthyosis that may be confused with atopic dermatitis, and hair shaft abnormalities.

The prognosis can be severe in newborns with life-threatening complications and high postnatal lethality, skin manifestations and hair shaft abnormalities persist throughout life, symptoms decrease with age and growth improves during the second year of life.

We report the case of an infant of 3 months followed for cutaneous xerosis, hospitalized for repeated cutaneous infection, trichoscopy and skin biopsy have allowed the diagnosis of netherthon syndrome without associated immune deficiency, with a good evolution under antibiotic therapy , administration of immunoglobulin and local care.

Key words: neterthon syndrome, genodermatosis, ichthyosis, trichoscopy.

1. INTRODUCTION:

Netherton syndrome (NS) is a rare autosomal recessive genodermatosis with an incidence of 1 case per 200,000 newborns.
Characterized by the triad of congenital ictyosiform erythroderma often of neonatal onset, a characteristic capillary anomaly (Trichorrhexia invaginata) and atopic manifestations.
It is a potentially fatal syndrome, frequent complications include hypernatremic dehydration, recurrent infections, diarrhea and intestinal malabsorption. The course of the disease is heterogeneous.

2. PRESENTATION OF CASE:

This is a 3 months old infant, unique in its fritory, first degree consanguinity, father followed for asthma controlled under background treatment, pregnancy followed carried out at term, high way delivery for breech presentation, birth weight 2900 g, no perinatal suffering, fall of the cord at 7 days of life, vaccination in progress, exclusive breast feeding, good psychomotor development.
Follow-up for skin xerosis that appeared at the age of 2 months, symptomatic treatment (skin emollient).
The patient presented in our service, febrile at 39 degrees under antipyretic, tonic and reactive, general state preserved, no sign of septic shock, with a staturopunderal delay at -2 DS, weight: 3500 g, height: 55 cm, PC: 41 cm, no dehydration folds.
Physical examination revealed diffuse skin lesions, predominantly on the face, with thick yellow-brown scaly patches with polycyclic and serpiginous edges on an oozing and hematous background, associated with purulent pustules predominantly on the forehead [Fig. 1], the eyebrows were sparse, especially on the lateral parts, as well as short, sparse and brittle hair, mainly in the frontal and lateral regions [Fig. 2]

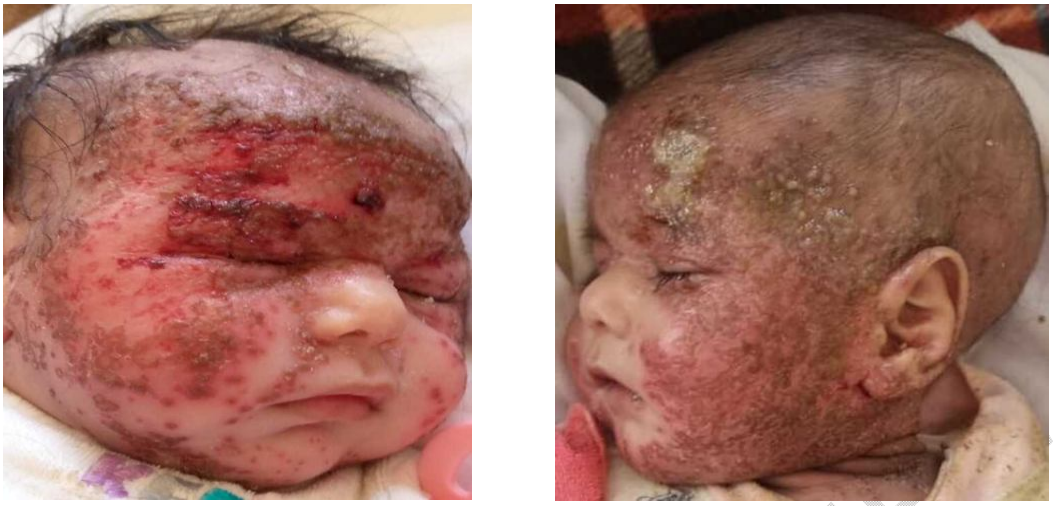

Fig.1 (a, b): Ichthyosiform erythroderma pre-dominant in the face

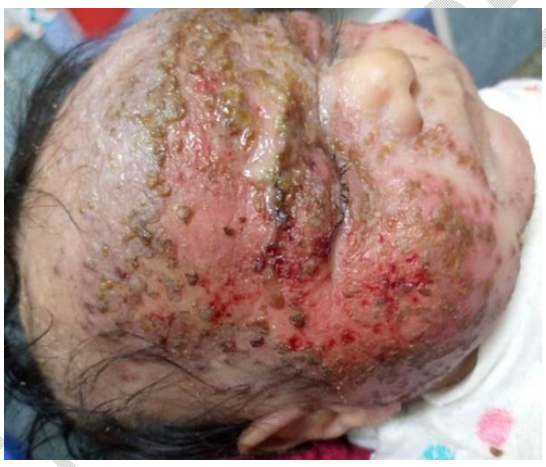

Fig.2: Sparse eyebrows, with a short sparse and brittle hair mainly in the frontal and lateral regions

The patient was hospitalized, the paraclinical workup found a neutrophil hyperleukocytosis at 20.8 10^3/ µl with a hypereosinophilia at 1800/ µl, a C-reactive protein at 160 mg/l, with an elevated level of IgE, a congenital and acquired immune deficiency workup realized returned in the norm.
Hair trichoscopy revealed the presence of bamboo hairs with the appearance of golf hairs: the trichorrhexis invaginata [Fig. 3].

Fig.3: Trichorrhexis invaginata (bamboo hair) in Netherton syndrome.

Skin biopsy revealed hyperkeratosis, acanthosis and focal hypergranulosis with a superficial perivascular lymphocytic infiltrate.
Patient put on intravenous antibiotics (protected amoxicillin) for 10 days, local treatment with low potency topical corticosteroids and emollients and the administration of immunoglobulin, with good clinical evolution.

3. DISCUSSION:

Netherton syndrome SN is a rare autosomal recessive cornification disorder, the involvement of the SPINK5 gene in Netherton disease was proven in 2000 [1], the mutations found are numerous, the coding polymorphisms of SPINK5 are involved in the pathogenesis of atopic dermatitis and asthma [2]. Clinically, SN is characterized by the classic triad of congenital ichthyosiform erythroderma, associated with a specific hair shaft anomaly called trichorrhexis invaginata ("bamboo hair") and atopic dermatitis [3].
Clinical manifestations may begin at birth or after a free interval of a few weeks. The skin manifestations are diverse: diffuse xerosis, erythema and lichenification at the flexural areas, urticaria and congenital ichthyosiform erythroderma. The most severe cases may have a collodion membrane at birth.
The hair is short, weak, sparse and dull ; Trichoscopy or light microscopic examination on the hair or better on the eyebrows [4], reveals the pathognomonic sign called trichorrhexis invaginata, which may be absent in the first years of life [5], golf hairs resulting from the breakage of the hair shaft at the invagination leaving the part of the shaft in the shape of a cup can also be observed, Another hair shaft abnormality that can be encountered is the match sign, which consists of a broken hair shaft with a bulging tip [6], pili torti, trichorrhexis nodosa and helical hairs are other hair shaft abnormalities reported in NS [7].
Other clinical manifestations of neterthon syndrome include allergic manifestations such as cow's milk allergy or asthma which remains frequent. Chronic non-infectious diarrhea, and neurological deficits may be associated [7, 8]. Complications include hypernatremic dehydration, impaired thermoregulation, growth retardation, infections and sepsis [8, 9].
The first line of treatment is topical therapy, emollients are recommended at least twice a day preferably after bathing, antiseptics are recommended 2-3 times a week.
Topical corticosteroids, topical calcineurin inhibitors and therapies include narrow band UVB phototherapy, as well as administration of immunoglobulin are recommended in the short term and in localized areas during flares [10]. Follow-up of patients with SN should include regular dermatological examinations because of the increased risk of skin cancer [10].

4. CONCLUSION:

Neterthon syndrome remains a rare disease, the diagnosis of which can be delayed because the atopic features and skin manifestations can be confused with atopic dermatitis, the histological examination of the hair allows to make the diagnosis.
The early diagnosis of the disease allows an adapted management, thus improving the short and long term prognosis.

5. REFERENCES:

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