Netherton Syndrome: Considering the Rare in a Nonspecific Presentation

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BACKGROUND

Netherton syndrome is an autosomal recessive hereditary ichthyosis and is identified according to clinical course, histopathology, biochemical markers, and genetic mutations.[1] The mutation is in SPINK5 which encodes a protease inhibitor that regulates desquamation.[2] The clinical picture involves a triad of scaly skin, abnormal hair, and atopic features. Infants are usually born with ichthyosiform erythroderma which may persist or transform into scales characteristic to this condition: ichthyosis linearis circumflexa. These lesions are described as serpiginous erythematous plaques with a double-edged scale.[3] Examination of the hair and eyebrows may reveal trichorrhexis invaginata or "bamboo-hair".[4] Under light microscopy, this finding resembles bamboo due to the distal part of the hair shaft falling into the proximal aspect, ultimately making the hairs prone to breakage.[5]

Inherited ichthyoses can be further categorized into syndromic and non-syndromic subtypes. Syndromic involves other organ systems, while non-syndromic presents as an isolated cutaneous manifestation. Due to the clinical presentation of this patient and the history of unaffected parents, an autosomal recessive inheritance pattern was believed to be most probable.

CASE DESCRIPTION

A nine-day-old female, born at 34 weeks gestation (complicated by no prenatal care), presents with severe skin peeling and diffuse erythema.

Physical Exam: Growth parameters for birth weight, length, and head circumference were <1%. Copious green drainage from the bilateral eyes was noted and mucous membranes were intact. Diffuse erythema with superficial desquamation was noted of the face, ears, neck, trunk, and upper/lower extremities (Figures 1 and 2). There were superficial erosions on the buttocks and labia majora and whitish material in the skin folds of the neck, axilla, and groin.

Medical course: Hypocalcemia was noted on a chemistry panel. Complete blood cell count showed anemia and mild eosinophilia. Cultures of the bilateral eye drainage were positive for Staphylococcus epidermidis and Escherichia coli (E. coli). Blood, sputum, and fungal cultures along with HSV/VZV PCR were all negative. Maternal labs showed positivity for amphetamines and methamphetamines. Antibiotics were initiated, and initial management focused on preventing complications from insensible water loss (IV fluids, Petrolatum application).

Figure 1: Anterior areas of diffuse erythema and superficial desquamation of newborn at initial presentation.

Figure 2: Posterior areas of diffuse erythema and superficial desquamation at initial presentation.

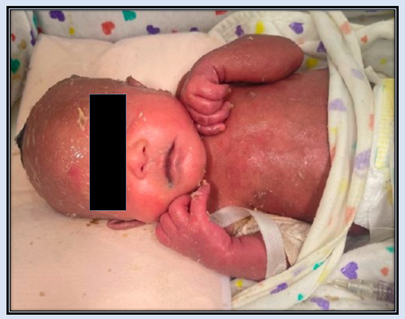

Figure 3: Gradual improvement of erythema and decrease in desquamation occurring throughout hospital course.

Figure 4: Gradual improvement of erythema and decrease in desquamation occurring throughout hospital course.

** Mother consented to photo documentation for educational purposes.**

CASE DESCRIPTION

Erythema and erosions initially improved but diffuse plates of desquamation recurred with cessation of Petrolatum application. Initial diagnosis of staphylococcal scalded skin syndrome was made; however, the newborn continued to demonstrate poor weight gain, desquamation with thick scales, and scalp alopecia, so differential included autosomal recessive congenital ichthyoses. A molecular genetics panel was performed, and the report demonstrated a homozygous SPINK5 mutation which confirmed Netherton syndrome.

With continued skin care management, there was gradual improvement of the erythema and a decrease in the superficial scales (Figures 3, 4). Once settled in an open-air crib, the patient demonstrated new growth of brows, lashes, and hair with no presence of scaling or rashes.

DISCUSSION

The variation and evolution of cutaneous findings makes Netherton syndrome a difficult diagnosis. Thus, a high index of suspicion is needed in those with a mild clinical picture as approximately 20% of neonates or infants with ichthyosiform erythroderma will have this condition.[5]

Early recognition and management is crucial to prevent complications such as hypernatremic dehydration, infection, and nutritional deficiencies. Pruritus is a general complication, which can lead to lichenification, particularly of the flexures.[6] Adults with Netherton syndrome have an increased risk of developing squamous cell carcinomas and basal cell carcinomas at an early age, therefore routine skin examination is recommended.[7]

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