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Netherton Syndrome, a Case Report and Review of Literature

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Netherton Syndrome (NS) is a rare hereditary autosomal recessive multisystem disorder which presents with generalized erythroderma at birth or soon after^[1]. Its incidence is estimated to be 1/200,000^[2]. NS presents in most (but not all) patients with generalized erythroderma and scaling resembling congenital ichthyosiform erythroderma, or continuous peeling of the skin^[3]. Other common features of the disease are enteropathy, hypoalbuminemia, aminoaciduria, mental retardation, growth retardation, and immunologic abnormalities^[4]. NS presents almost

with a specific hair shaft abnormality known as "bamboo hair"^[5]. The third characteristic feature of NS is an imbalance of the immune system. Serum level of IgE is markedly elevated^[6]. Treatment is symptomatic such as topical emollients, keratolytics, tretinoin and corticosteroids, alone or in combination. PUVA therapy has produced variable results^[7]. We present a case of NS with a positive familial history, admitted due to failure to thrive and erythroderma and severe diarrhea. The diagnosis was based on clinical as well as histological findings.

A 63-day-old boy, born premature at 1850 g to non consanguineous parents was admitted due to severe failure to thrive and diarrhea and generalized erythroderma and scaling. There was a history of one missed conception and previous child who succumbed to death at seventh month due to ichthyosiform disease and recurrent infection without any specific diagnosis. The weight gain was only 200 grams in 2 months. He was irritable and the skin examination showed generalized erythroderma covered by fine, translucent scales on extremities and scalp. This case was examined with colonoscopy and active colitis was diagnosed. Blood culture was positive for *staphylococcus aureus*. His hair fall was significant at admission (Fig. 1). The typical characteristic of bamboo hair was determined (Fig. 2). The laboratory findings during hospitalization showed mild hypernatremia (serum sodium=150), direct hyperbilirubinemia (Total bilirubin=9, direct bilirubin=6.2), elevated hepatic enzymes (aspartate aminotransferase =140, alanine aminotransferase=54), hypothyroidism (thyroid stimulating hormone =13, thyroxine=7) and C-Reactive protein=4+. He did not tolerate breast milk; therefore we started a specific hypoallergenic formula (Neutramigen) parallel with parenteral nutrition. Unfortunately, he succumbed to death a month later due to infection. His irritability might be related to universal pruritis whereas generalized sweeping with paraffin relieved this symptom. Sepsis was diagnosed by positive blood culture for *Staphylococcus aureus*. His antibody screen was almost normal except for IgE level which was slightly elevated (IgE=7.6 mg/dl). Our patients had sparse, brittle and markedly thin hair alongside trichorrhexis nodosa (bamboo hair). Typically,

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Fig. 1: Generalized erythroderma and hair fall in Netherton Syndrome



Fig. 2: Bamboo hair

diagnosis was delayed until the appearance of the pathognomonic hair shaft anomaly. Rectal endoscopic biopsy showed active colitis compatible with dermopathic enteropathy reported by other authors. Diagnosis was made by triad of congenital erythroderma, bamboo-like growths in the hair shaft and failure to thrive due to prolonged diarrhea.

In summary, children with ichthyosiform dermatosis, diarrhea and growth failure may have underline disease such as NS. We would like to highlight the importance of recognition of growth failure and severe diarrhea and malnutrition in these children as well as prompt treatment of any infections.

Key words: Netherton Syndrome; Failure to Thrive; Trichorhexis Invaginata; Erythroderma

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