

Bamboo Hair Syndrome or Netherton Syndrome - A Case Report

Puja Balpande¹, Puja Balpande²

Department Of Dermatology, LMH, Nagpur, Maharashtra, India

Department Of Dermatology, LMH, Hingna Road, Digdoh Hills, Nagpur, Maharashtra India

Abstract: Netherton Syndrome (AKA- Bamboo Hair Syndrome) is a non treatable, Autosomal Recessive Disorder of infancy and childhood, so has no sex predilection.

Family history may reveal consanguinity.

There is triad -

A] Ichthyosiform linearis circumflexa

B] Hair Shaft Defect like trichorrhexis invaginata &

C] Atopic Diathesis.

Caused by mutation in Serine Protease Inhibitor Kazal type 5 gene (SPINK5), which is predominantly expressed in the lamellar granules of epithelia and lymphoid tissue.

I. Introduction

Bamboo Hair Syndrome or Netherton Syndrome (NS) is a rare hereditary Autosomal Recessive multi systemic disorder which presents primarily with generalised erythroderma at birth or soon after⁽¹⁾. With a incidence of 1/200,000⁽²⁾. NS should be invariably thought of in patients with generalised erythroderma and scaling resembling congenital ichthyosiform erythroderma, or continuous peeling of the skin⁽³⁾.

Other common Systemic 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⁾ on microscopic examination. The third characteristic feature of NS is an imbalance of the immune system. Serum IgE level is drastically increased⁽⁶⁾. Treatment is symptomatic and consist of topical emollients, keratolytics, tretinoin and corticosteroids, alone or in combination. There are variable results with PUVA therapy⁽⁷⁾.

Case Report:

A 7 ½ year old male child first issue of third degree consanguineous marriage presented with scaling all over face, bilateral upper limb and abdomen since 15 days after birth. There was history of itching associated with it on and off. History of growth retardation and low IQ with history recurrent cough and cold was present. No history of similar complaints were present in siblings. On cutaneous examination revealed scaly hyperpigmented patches over face (forehead, periorbital, perioral) [Figure 1a, 1b], bilateral upper and lower limb, back and abdomen (ichthyosiform linearis circumflexa) with in between islands of normal skin was present.



Figure 1a: Scaly hyperpigmented patches over face (forehead, periorbital, perioral)



Figure 1b: Scaly hyperpigmented patches over face (forehead, periorbital, perioral)

On scalp thick scales were present [Figure 2].



Figure 2: Thick Scales present on scalp.

Genu Valgus deformity was present [Figure 3].



Figure 3: Genu Valgus Deformity

On laboratory investigation Serum IgE 836.8IU/ml was present. No eosinophilia was present. Bone age was 4-5 years of age.

Skin Biopsy showed epidermal parakeratosis, acanthosis, elongation of rete ridges and spongiosis. Dermis showed dense inflammatory infiltrate of lymphocytes and neutrophils. [Figure 4]



Figure 4: Epidermal parakeratosis, acanthosis, elongation of rete ridges and spongiosis. Dermis shows dense inflammatory infiltrate of lymphocytes and neutrophils

On hair mount, bamboo hair were not seen as shown below. [Figure 5]



Figure 5: Shows no bamboo hair on hair mount.

Gene mapping could not be done due to financial constraints.

II. Conclusion

On the basis of history and investigations diagnosis of Netherton's Syndrome was kept. Patient was treated with emollients, antihistaminics and supplemental multivitamins. We present this case due to rarity of the disorder. The bamboo hair appearances on hair mount is pathognomonic of this disease. However, studies suggest this feature might improve as the age advances, as was the case in this patient. Early identification and nutritional supplements can improve quality of life and prevent growth retardation.

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