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Case Report

Netherton's Syndrome, an uncommon genodermatosis

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Abstract Netherton's syndrome (NS) is the most common of the multisystem ichthyosiform syndromes, and comprises an icthyosiform dermatosis with a variable erythroderma, hair shaft defects and atopic features. Mutation in type 5 (SPINKS) gene has been identified in several NS patients. We report a young male who had perioral erythema, ichthyosiform lesion and hair abnormalities. He was diagnosed on clinical as well as histological ground.

Key words
Netherton syndrome, ichthyosis linearis circumflexa, trichorrhexis nodosa, trichorrhexis invaginata.

Introduction

Netherton's syndrome (NS) is an autosomal recessive multisystem ichthyosiform syndrome. Mutation in SPINKS[1] gene leads to intermittent disruption of epidermal and hair shaft maturation and keratinization. Electron microscopy shows premature lamellar body secretions and abnormal lipid processing in intercellular space.[2] This leads to an increase in epidermal DNA synthesis and turnover. The clinical features include generalized erythroderma of variable intensity, at or shortly after birth. Common complications in the neonatal period and infancy are temperature instability, skin and systemic infection and hypernatremia. A rare late complication is dilated cardiomyopathy.[3] During childhood approximately half of patients develop lesion of ichthyosis linearis circumflexa (ILC) on the trunk and limbs. A typical ILC lesion is an erythematous exfoliating or scaly, annular or polycyclic, flat patch with an incomplete advancing double edge of peeling scale.[4] Rarely pustular lesions are superimposed. Between acute attacks, the skin may look normal many patients have perioral erythema and peeling. Chronic blepharitis and ectropion may lead to keratitis. Hair defects include trichorrhexis invaginata, trichorrhexis nodosa and pili torti.[5] Scalp, eyebrows, eyelash and body hair remains sparse, slow growing, lusterless and brittle. Topical steroids are ineffective. Topical or systemic antibiotics are required for skin and respiratory infection. PUVA therapy has produced variable results.[6]

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Case report

A 15-year-old boy presented with erythematous, exfoliating polycyclic patches on arms, legs, foot and nape of neck (**Figure 1**). Since the age of 5-month perioral erythema with some peeling was present. Scalp hair was sparse, brittle and lusterless

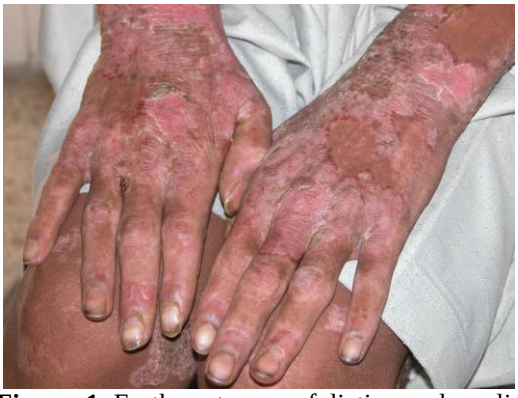

Figure 1 Erythematous, exfoliating polycyclic patches on hands.

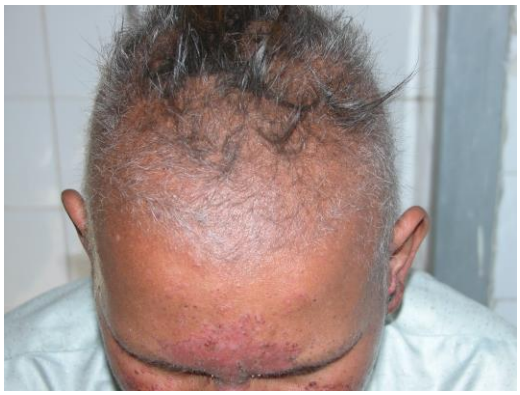

Figure 2 Sparse scalp hair.

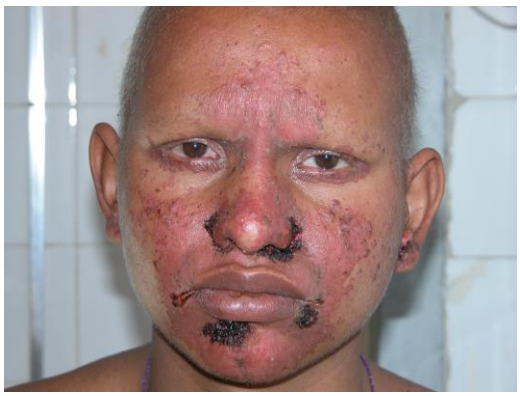

Figure 3 Spars eyebrows and eyelashes, bilateral ectropion.

(**Figure 2**). Eyebrows and eyelashes were also sparse. Bilateral ectropion was present (**Figure 3)**. Some pustular lesion and ulceration on sole were present. Examination of hair under light microscopy showed trichorrhexis nodosa (TN), trichorrhexis invaginata (TI) [**Figure 4**] and pili torti. Histopathology showed

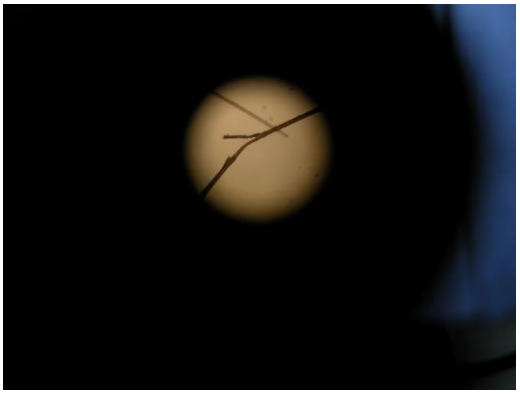

Figure 4 Microscopic examination of hair, showing trichorrhexis invaginata.

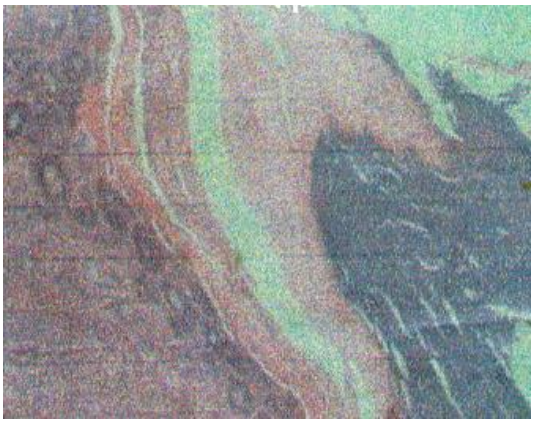

Figure 5 Hyperkeratosis and mega granules.

hyperkeratosis, prominent granular cell layer with mega keratohyaline granules with some psoriasiform changes as shown in **Figure 5**. Clinical examination and result of histopathology confirmed the diagnosis of Netherton's syndromes.

Discussion

Netherton's syndrome is an autosomal recessive syndrome in which mutation in SPINK[5] gene occurs. It is a triad of ILC, hair abnormalities and atopy.[4] In our patient there were erythematous, polycyclic lesions with some scaling and lichenification, as well as, hair shaft abnormalities. Ectropion and perioral erythema was also present. All the three types of hair abnormalities related to this syndrome, were present in this

patient, like TI, TN and pili torti. Some pustules were also present. But in our patient there was no history of atopy. Diagnosis was by triad of congenital erythroderma. poor hair growth and failure to thrive. Collodion membrane is not a feature of this syndrome. In neonatal and infancy period common complications are temperature instability, skin and systemic infection. Hypernatremic dehydration and dermopathic enteropathy are also common. A rare late complication is dilated cardiomyopathy.[3] Not all patients have ILC like lesions. In some patients ichthyosiform erythroderma was also present.[7] There were 2 case reports of squamous cell carcinoma developing in NS.[8,9]

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