

A case of Netherton's syndrome with cerebral infarction

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Netherton's syndrome, a rare congenital disease of childhood, is characterized by variable cutaneous erythematous eruptions with different manifestations. A five-year-old boy, who presented with ichthyosis linearis circumflexa, atopic manifestations and pili torti, had spastic hemiparesia due to cerebral infarction. Netherton's syndrome can easily be misdiagnosed as Leiner's disease, generalized psoriasis or nonbullous congenital ichthyosiform erythroderma, especially in the neonatal period, because of its nonspecific clinical and histological features. Pediatricians should consider this syndrome in the differential diagnosis of the generalized erythematous skin disorders of childhood associated with various abnormalities.

Key words: erythematous eruption, atopic manifestation, hair shaft keratinization defect, autosomal recessive inheritance, spastic hemiparesia.

Netherton's syndrome is a rare congenital disease characterized by hair shaft keratinization defects, atopic manifestations and variable erythematous skin eruptions. Here we present a case of Netherton's syndrome with spastic hemiparesia due to cerebral infarction.

Case Report

A five-year-old boy, born to consanguineous parents who were cousins, was admitted to our hospital with erythematous skin lesions in July 1999.

Because of generalized exfoliative erythroderma in the neonatal period, he was hospitalized and treated with antibiotics against suspected sepsis.

Physical examination revealed bilateral allergic conjunctivitis and spastic hemiparesia on the right side of the body with augmented deep tendon reflexes. There was no abnormality in the other organ systems.

Dermatological examination showed generalized erythematous eruptions (Fig. 1a) and some annular erythematous plaques with a double-edged scaly rim on the extremities (Fig. 1b). Thick, adherent and yellow scales covered especially the scalp hair and forehead. There was some sparse and fragile hair on the vertex. Polarization microscopy of scalp hair showed small fractures and pili torti (Fig. 2).

Complete blood counts, urinalysis, erythrocyte sedimentation rate, liver function tests, renal function tests, electrolytes, total proteins, and amino acid profile were normal. Total IgE level was 2,000 IU/L (normal range: 0-100 IU/L).

Magnetic resonance (MR) imaging of the brain showed encephalomalasia with multiple cysts secondary to cerebral infarction as a result of occlusion of the arteria cerebri media (Fig. 3).

The dermatopathologic examination revealed marked papillomatosis. The stratum corneum was thick and the granular layer decreased. This biopsy specimen was reported as psoriasiform dermatitis.



Fig. 1a. Generalized erythematous skin eruptions of the patient.



Fig. 1b. Characteristic annular erythematous plaques with a double-edged scaly rim on the extremities of the patient.



Fig. 2. Pili torti is seen on the polarization microscopy.



Fig. 3. T1 weighted magnetic resonance imaging of the brain showed encephalomalacia with multiple cysts secondary to cerebral infarction.

Discussion

Netherton's syndrome is a rare and severe autosomal recessive disease characterized by hair shaft keratinization defects, atopic manifestations and variable erythematous eruptions ranging from congenital ichthyosiform erythroderma to ichthyosis linearis circumflexa (ILC)¹⁻³.

Netherton's syndrome causes severe neonatal disease which can be easily misdiagnosed as Leiner's disease, generalized psoriasis or nonbullous congenital ichthyosiform erythroderma because of the nonspecific histological features^{2,4}. The patients develop generally migrating erythematous patches with a double-edged scaly rim, one of the characteristic signs of ILC, after two years of age³. Infrequently, hypernatremia, aminoaciduria, mental deficiency, neurological defects, delayed growth, recurrent infections and hyper- or hypoglobulinemia may be detected in the neonatal period¹.

The patient had a history of neonatal sepsis. Spastic hemiparesia was detected in his neurological examination. MR imaging of the brain revealed cerebral infarction due to occlusion of the arteria cerebri media with unknown etiology.

The histopathologic features of ILC are frequently indistinguishable from psoriasis or seborrheic dermatitis⁵. Yoshiike et al.⁶ detected many clinical, morphological and biochemical similarities between ILC and psoriasis. It is believed that Netherton's syndrome is a hyperproliferative situation with a reduced epidermal transit time. The biopsy specimen of the patient was reported as psoriasiform dermatitis.

Trichorrhexis nodosa, pili torti, monilethrix, angle-bent hair and trichorrhexis invaginata may develop in patients as hair shaft defects of the syndrome¹. The polarization microscopy showed pili torti.

Atopic manifestations can be seen in the syndrome⁷. Allergic conjunctivitis was observed in the patient's physical examination, and total IgE rate was elevated. Topical tretinoin, local corticosteroid, emollients and cyclosporine have been used in the treatment of Netherton's syndrome. However, no single treatment showed sufficient beneficial results^{1,8}. A relative reduction in the severity of the lesions was achieved with cyclophosphamide therapy⁹. Low-dose retinoid therapy and PUVA seem to also be effective¹⁰⁻¹².

The patient's parents refused all treatment modalities aside from topical emollients.

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