Erythroderma with Scaling Patches

Ming-Hsien Lin  Julia Yu-Yun Lee  Mei-Hui Yang  Sheau-Chiou Chao

CASE REPORT

A 16-month-old Taiwanese girl presented with generalized erythroderma covered by fine, translucent scales at birth in 2003. The baby was born preterm with birth weight of 2645 g. She was the second child of clinically normal parents. Intrauterine fetal death occurred during the first gestation of her mother due to placental abruption. On examination, the body weight was 7.3 kg. She manifested persistent erythroderma and continuous peeling of the skin. Sparse polycyclic patches with double-edged scaling at the borders were present on the extremities (Fig. 1A). The hair of the scalp, eyebrows and eyelashes were short and of irregular length. The nails were normal. Hair from the scalp was clipped for microscopic examination (Fig. 1B). Eosinophilia (16.5%) was found and serum IgE level was within normal limits. Multiple-antigen simultaneous test (MAST) showed 4+ positive reaction toward egg white and milk.

Fig. 1
(A) polycyclic patches with double-edged scaling at the borders (B) hair.

Fig. 2
Automated sequencing of the SPINK5 gene. (A) A homozygous change from A to T in the first position of codon 754 (2260 A>T), predicting a substitution of lysine by premature termination codon (K754X) in the proband. (B) The same nucleotide substitution in her parents. (C) Sequence from normal population.
DIAGNOSIS: Netherton’s Syndrome

The 33 exons of the SPINK5 gene were amplified by PCR using previously described primers. Automated sequencing showed a homozygous mutation, 2260 A>T (K754X) in exon 24 in the SPINK5 in the proband (Fig. 2A). The mutation was also found in the parents (Fig. 2B). This mutation is expected to result in premature termination codon of translation.

DISCUSSION

Netherton’s syndrome (NS) is a rare, autosomal recessive disease characterized by congenital ichthyosiform erythroderma (CIE), ichthyosis linearis circumflexa (ILC), trichorrhexis invaginata (TI) and atopic diathesis. Infants with NS usually present with erythroderma covered by fine, translucent scales, which can be difficult to distinguish clinically from erythrodermic psoriasis, nonbullous congenital ichthyosiform erythroderma, or other infantile erythrodermas. The ichthyosis gradually evolves into ILC in the majority of patients. Hair shaft abnormalities usually develop later during infancy and early childhood. In approximately 20% of patients, NS may be complicated by progressive hypernatremic dehydration, disturbed thermoregulation, enteropathy, bronchopneumonia, sepsis or failure to thrive. These complications can be fatal. However, diagnosis is often delayed until the pathognomonic TI or ILC appears and is recognized clinically.

The NS gene locus has been localized on chromosome 5q32 and pathogenic mutations were identified within the SPINK5 (serine protease inhibitor, Kazal-type 5) gene. The SPINK5 encodes the serine protease inhibitor LEKTI (lymphoepithelial Kazal-type-related inhibitor), which may function in anti-inflammatory and/or anti-microbial protection of mucous epithelia.

Treatment with acitretin and PUVA are effective in NS. Bland emollients, antihistamines to relieve itching, and antibiotics when indicated are also helpful. Tacrolimus ointment had been reported with satisfactory result. Regular monitoring of the serum tacrolimus concentration is still essential in this setting. Keratolytic agents containing salicylic acid and urea should be avoided at least in the first few years of life, because they can severely impair skin barrier function which could result in excessive penetration and absorption of drugs.

Children with NS are often misdiagnosed as having atopic dermatitis or ichthyosis. Excessive systemic absorption of therapeutic agents interrupted skin-barrier could complicate NS. Thus infants born with exfoliative erythroderma should be examined and followed up carefully for ILC and TI. The availability of prenatal molecular diagnosis can offer an important option to families with NS.

ACKNOWLEDGMENT

This study was supported by grant NSC91-2314-B006-113 from the National Science Council, Taiwan, Republic of China.

REFERENCES