QUIZ SECTION

MJS PUBLISHING

A Seven-year-old Girl with Exfoliative Erythroderma: A Quiz

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A 7-year-old Vietnamese girl was admitted to the department of dermatology with exfoliative erythroderma. She was born at term after an uneventful pregnancy to healthy unrelated parents. In the absence of a clear diagnosis, her desquamation and erythroderma were treated empirically with topical emollients. She did not exhibit growth retardation or have a history of recurrent skin, upper respiratory tract, or gastrointestinal tract infections. There was no family history of similar skin and hair disorders. On dermatological examination, she had a disseminated rash characterized by serpiginous patches with collarettes of double-edged scales, resembling ichthyosis linearis circumflexa (Fig. 1). The scalp hair, eyelashes, and eyebrows were normal. Trichoscopic examination of the scalp hair did not reveal trichorrhexis invaginata. Blood analysis found elevation of total IgE to 1,537 UI/mL (normal range < 100 UI/mL) and eosinophilia. RIDA qLine Allergy Panel 1–4 using specific IgE antibodies was strongly positive to cow's milk and eggs. Other serological tests were in the normal range. The skin biopsy showed hyperkeratosis, acanthosis, and a well-developed granular layer. A mild dermal inflammatory infiltrate was present (Fig. 2).

What is your diagnosis?

1: Congenital ichthyosiform erythroderma
2: Netherton syndrome
3: Omenn syndrome
4: Ichthyosis with hyper IgE syndrome

See next page for answer.

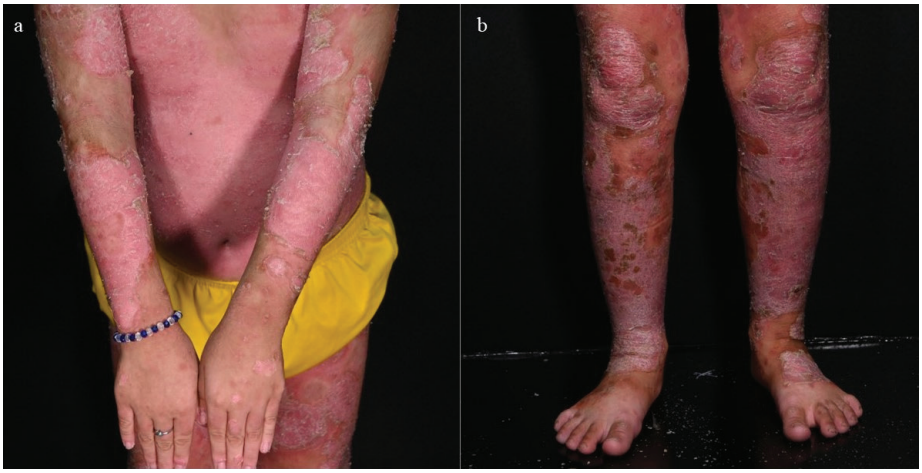

Fig. 1. Clinical photo: a disseminated rash characterized by serpiginous patches with collarettes of double-edged scales, resembling ichthyosis linearis circumflexa.

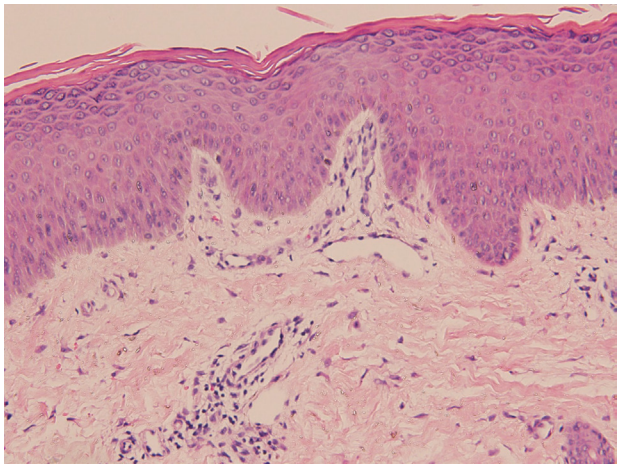

Fig. 2. Histology showed hyperkeratosis, acanthosis, and a well-developed granular layer. A mild dermal inflammatory infiltrate was present (haematoxylin and eosin staining; x 100).

ANSWERS TO QUIZ

A Seven-year-old Girl with Exfoliative Erythroderma: A Commentary

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Diagnosis: **Netherton syndrome**

Netherton syndrome is an autosomal recessive form of ichthyosis, caused by mutations in the *SPINK5* gene. The syndrome includes ichthyosiform dermatosis, hair shaft defects, and atopic manifestations (1).

There are 2 variants of skin manifestations: congenital ichthyosiform erythroderma, and ichthyosis linearis circumflexa (2).

Trichorrhexis invaginata is clinically characterized by thin, spiky, and brittle hairs. Trichoscopy revealed typical characteristics of bamboo hair and golf-tee hair. Atopic manifestations are constant, which include atopic dermatitis-like lesions with constant pruritus, asthma, allergic rhinitis, urticaria, angioedema, food and airborne allergies, eosinophilia, and elevated serum IgE (3).

To further characterize the condition of this patient, mutational analysis of the *SPINK5* gene was undertaken by the Sanger technique. All 33 exons and exon-intron boundaries of the *SPINK5* gene were amplified by PCR. We detected a heterozygous mutation consisting of the splicing variant in intron 21 (IVS21+1G > T or c.2015+1G > T) and a variant in exon 32 (c.3211C > T: p. Arg1071Cys). The genetic analysis of her parents was normal. The hair and skin examinations of her parents did not reveal any impairment.

The c.2015+1G > T is a novel variant that has not been reported in the literature, capable of inducing mis-splicing of mRNA transcript. A functional study of this mutant might help to further elucidate the pathogenic mechanism of SPINK5. The other variant, c.3211C > T (rs374003659), is a variant of unknown significance with a very low frequency of the T allele in the general population (0.000032) and has a PolyPhen-2 score of 1.0, suggesting that this variant might play a causative role in the disease.

There are several therapeutic options for the treatment; however, the outcome is variable (2, 4, 5).

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IRB approval status: The parents signed the consent form.

The authors have no conflicts of interest to declare.

REFERENCES

1. Kilic G, Guler N, Ones U, Sekerel BE, Adalioglu G, Danisman N. Netherton syndrome: report of identical twins presenting with severe atopic dermatitis. Eur J Pediatr 2006; 165: 594. https://doi.org/10.1007/s00431-006-0141-0
2. Leung AK, Barankin B, Leong K. An 8-year-old child with delayed diagnosis of Netherton syndrome. Case Rep Pediatr 2018; 2018: 1–4. https://doi.org/10.1155/2018/9434916
3. Hovnanian A. Netherton syndrome: skin inflammation and allergy by loss of protease inhibition. Cell Tissue Res 2013; 351: 289–300. https://doi.org/10.1007/s00441-013-1558-1
4. Roda A, Mendonça-Sanches M, Travassos AR, Cabete J, Tellechea O, João A. Infliximab therapy for Netherton syndrome: a case report. JAAD Case Rep 2017; 3: 550–552. https://doi.org/10.1016/j.jdcr.2017.07.019
5. Maatouk I, Moutran R, Tomb R. Narrowband ultraviolet B phototherapy associated with improvement in Netherton syndrome. Clin Exp Dermatol 2012; 37: 364–366. https://doi.org/10.1111/j.1365-2230.2011.04231.x