GENETICS AND GENODERMATOSES

A CASE OF GENERALIZED PUSTULAR PSORIASIS WITH A DEFICIENCY OF IL-36RN (DITRA)

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Background: Generalized pustular psoriasis (GPP) is a rare, life- threatening disease characterized by repeated attacks of diffuse, pustular skin eruptions with general symptoms including fever, peripheral blood neutrophilia
and elevated C- reactive protein (CRP) serum levels.
Generalized pustular psoriasis (GPP) is an inflammatory skin disorder which occurs in patients with or without psoriasis vulgaris (PV) and is characterized by sudden fevers and extensive erythemas with pustules and edema, sometimes with life-threatening circulatory and/or respiratory disturbances. Those symptoms develop recurrently and therefore lifelong treatment is often required to manage disease activity. Mutations in IL36RN have been identified in familial GPP and in sporadic cases of GPP. Cases with IL-36RN mutations have been named as deficiency of IL-36RN (DITRA).

Observation: A 6-yr boy was admitted to the hospital because of "repetitive diffuse pustules for 4 years." The patient has a 23-yr sister without similar episode. No family history of PV and eczema. Physical examination showed diffuse erythema with pustules on the scalp, thinning hair. Large erythema with papules and yellow scaly sputum on the face, trunk and limbs, pustules on the limbs, and yellowing, thickening and deformation of the toenail deck. Skin biopsy suggested GPP. High-throughput sequencing of all exons showed IL36RN C.115+6T>C homozygous mutation, IL36RN C.227C>T heterozygous mutation by the patient, and parents and sister were C.115+6T> C heterozygous mutation.

Key message: Since the discovery of IL36RN as a gene responsible for GPP, mutations of IL36RN have now been reported in more than 70 cases of GPP worldwide. Three major variant types (c.115+6T > C, c.140A > G, c.227C > T) were identified in China, among which c.115+6T > C exhibited the highest frequency (55.81%) and possibly those are founder mutations. In contrast to GPP+PV, IL36RN mutations possibly play a more important role in the development of GPP alone.