



GENETICS AND GENODERMATOSES

THE FIVE-YEAR-FOLLOW-UP OF THREE JAPANESE PATIENTS WITH INCOMPLETE ERYTHROPOIETIC PROTOPORPHYRIA

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Background: Erythropoietic protoporphyria (EPP) is an inherited cutaneous porphyria caused by both the partial deficiency of ferrochelatase (FECH) and the existence of IVS3-48C in trans to a mutated FECH allele. Recently, we reported three Japanese patients with mild phenotype of EPP who have the homozygous IVS3-48C polymorphism without causative FECH gene mutations. We hypothesized that a slight decrease in the FECH activity caused by the homozygous IVS3-48C polymorphism would result in a mild clinical appearance of EPP via the slight increase of protoporphyrin. This condition is referred to as incomplete EPP. We herein report the five-year follow-up of three Japanese patients with incomplete EPP.

Observation: We previously diagnosed three patients with mild photosensitivity to have incomplete EPP. At the time of the diagnosis, these patients were a 6-year-old girl (patient 1), a 7-year-old girl (patient 2) and a 9-year-old boy (patient 3), and the levels of protoporphyrin in the erythrocytes were 115 µg/dl, 103 µg/dl and 122 µg/dl (normal: 30-86 µg/dl), respectively. They had all been born of non-consanguineous relationships. We proposed that these patients to avoid exposure to sunlight and have observed their clinical symptoms and protoporphyrin levels every year for the past five years. Consequently, they have shown increases in the protoporphyrin levels over these five years. Patient 3 occasionally suffered from painful photosensitivity. However, in patients 1 and 2, the frequency and severity of photosensitivity gradually decreased as they grew older, although they were unable to completely avoid sun exposure and sometimes showed mild sunburn.

Key message: The findings on the present study suggest that the clinical appearance of incomplete EPP patients may improve with age. Although the mechanism underlying the improvement in their incomplete EPP symptoms remains unclear, we speculate that a slight suntan may be associated with the improvement in their photosensitivity.

