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Image Article

Erythropoietic Protoporphyria

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Introduction

A 41-year-old woman presented with a 5-year history of recurrent erosion and erythema on her face, neck, and extremities associated with headache, fatigue, needle prickling pain, and numbness in both hands. In the meantime, she has experienced repeatedly abdominal pain, dark red colored bloody stool, and had both cerebral infarction and cerebral hemorrhages for one time. She was diagnosed with systemic amyloidosis at other hospitals but no relevant evidence could be found. Physical examination was notable for erosion and atrophic scars on the face (Figure 1), neck, anterior chest wall, both hands, and thighs. A Wood's lamp examination illuminated areas of yellow fluorescence on her face (Figure 2). On further history, she reported the disease got worse in summer and every menstrual period. Porphyria was suspected and whole exome sequencing (WES) showed the IVS3-48T/C heterozygote, then a diagnosis of erythropoietic protoporphyria porphyria was made [1]. She was treated with oral hydroxychloroquine and beta carotene and also advised with avoiding sunlight exposure. After 2 months of treatment, the skin lesions abated and the symptoms were relieved. Another Wood's lamp examination showed that the original fluorescent areas had disappeared.



Figure 1: Physical examination was notable for erosion and atrophic scars on the face.

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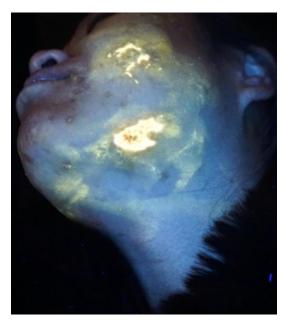


Figure 2: A Wood's lamp examination illuminated areas of yellow fluorescence on her face

Erythropoietic protoporphyria porphyria is difficult to diagnose, while delay in treatment would severely damage patients' quality of life and social activities [2]. If a patient complains of recurrent skin lesions at sun exposure areas, unexplained abdominal pain, and psychosomatic manifestations, we need to be aware of the possibility of porphyria. We should also bear in mind the utility of Wood's lamp examination in diagnosing cutaneous porphyria and it may be related to the severity of the disease process [3].

Conflict of Interest Disclosures: No disclosures were reported.

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Reference

- Berroeta L, Man I, Goudie DR, Whatley SD, Elder GH, et al. (2007) Late presentation of erythropoietic protoporphyria: case report and genetic analysis of family members. Br J Dermatol. 157:1030-1.
- Bissell DM, Anderson KE, Bonkovsky HL (2017) Porphyria. N Engl J Med 377: 862-72.
- Dyer JM, Foy VM (2022) Revealing The Unseen: A Review of Wood's Lamp in Dermatology. J Clin Aesthet Dermatol 15:25-30.

Volume 8; Issue 4