Autofluorescent Red Blood Cells in Protoporphyrria

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To the Editor: An 8-y-old Japanese boy was referred to our hospital because of a 3-y history of photosensitivity. He had experienced several episodes of painful swelling without vesicles on his face, hands, and feet after sunlight exposure. On physical examination, several shallow scars were observed on his face. Evaluation of peripheral blood by fluorescence microscopy revealed autofluorescent red blood cells. The increased level of total erythrocyte protoporphyrin (1,329 µg/dL) on biochemical analysis led us to diagnose protoporphyrria. Genetic analysis was not performed because of the lack of consent from the patient and his parents. Avoidance of sunlight and use of protective clothing, including long sleeves and hats, alleviated his symptoms. Protoporphyrria is the most common type of porphyria in childhood and is characterized by intense burning sensation on the skin after sunlight exposure. Total erythrocyte protoporphyrin level of more than five times the upper limit of normal is diagnostic of this condition [1]. By utilizing the fluorescence emission property of protoporphyrin, erythrocyte fluorescence test can effectively differentiate protoporphyrria from the other causes of photosensitivity [2]. Repeated phototoxic reactions in protoporphyrria commonly

induce chronic skin lesions, such as wax-like scarring on the face or knuckle lichenification. Because visible light is the major cause of photosensitivity, transparent sunscreens are insufficient in preventing symptoms in these patients [1]. In the present case, avoidance of sunlight and use of protective clothing alleviated the patient's symptoms.

Declarations

Conflict of Interest None.

References

1. Bissell DM, Anderson KE, Bonkovsky HL. Porphyria. *N Engl J Med*. 2017;377:862–72.
2. Lau KC, Lam CW. Automated imaging of circulating fluorocytes for the diagnosis of erythropoietic protoporphyrria: a pilot study for population screening. *J Med Screen*. 2008;15:199–203.

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