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Erythropoietic Protoporphyria

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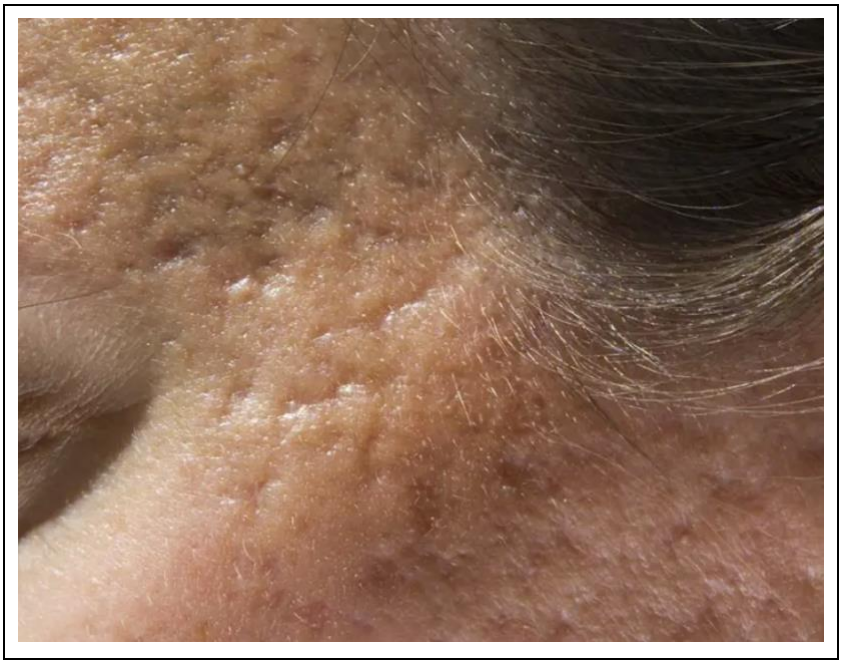

Clinical Image

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 of both hands. In the meantime, she has experienced repeatedly abdominal pain, dark red coloured bloody stool and had both cerebral infarction and cerebral haemorrhages for one time. She was diagnosed of systemic amyloidosis at other hospitals but no relevant evidence could be found. Physical examination was notable for erosion and atrophic scars on face, neck, anterior chest wall, both hands and thighs. A Wood's lamp examination illuminated areas of yellow fluorescence on her face. 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, 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. 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 with 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 disease process [3].

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