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Case Report

Adult-onset Gunther's disease (congenital erythropoietic porphyria) in a 37-year-old man: a case report

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Abstract A 37-year-old man presented with multiple blisters and ulcerations over the sun exposed skin of 2 years duration. His clinical, radiological, hematological features, fluorescent microscopy of urine and histopathology of subepidermal blister with PAS-positive material deposited in the upper dermis in a perivascular and periadnexal distribution were suggestive of congenital erythropoietic porphyria, was treated with beta carotene and advised to avoid sun exposure by using sun protective clothing. We repot this case because of its rarity.

Key words
Congenital erythropoietic porphyria, adult, photosensitivity.

Introduction

Congenital erythropoietic porphyria (CEP) is one of the rare forms of an intriguing group of metabolic disorders known as porphyria, caused by an autosomal recessive inherited deficiency of the uroporphyrin III cosynthase enzyme. Less than 200 cases have been reported in the literature.[1] The disease usually starts during infancy but occasionally during adulthood. We report a case of CEP with late-onset.

Case report

A 37-year-old man presented with multiple blisters and ulcerations over the sun exposed skin (face, and dorsum of the hands) for last 2 years. He had also history of recurrent attacks of nonspecific blepharitis being treated frequently. Dermatological assessment revealed patchy hyperpigmentation, scarring, erosions, milia affecting face (**Figure 1**) and dorsum of the hand. Hypertrichosis was found on hands, temples, malar region and eyebrows. Nail dystrophy was also observed.

Examination of the eye revealed presence of blepharitis and scarring over the eyelids. Teeth had brown staining (**Figure 1**). X-ray revealed features of decrease bone density.

Clinical evidence of splenomegaly was confirmed by abdominal ultrasonography. Urine fluoresced brilliantly under Wood's light and examination of erythrocytes under fluorescent microscopy demonstrated stable fluorescence. **Table 1** summarized the porphyrin assay. Complete blood count revealed normochromic normocytic anemia with thrombocytopenia (hemoglobin 9.8g/dl, MCV 86.4fl, MCH 28.3pg,

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Figure 1 Patchy hyperpigmentation, scarring, erosions, milia affecting face.

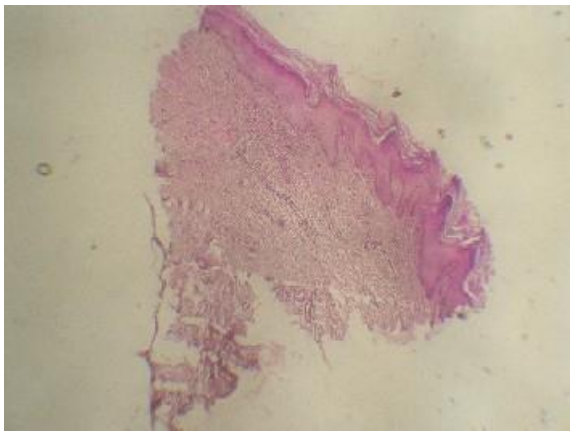

Figure 2 Skin biopsy revealed subepidermal blister with PAS positive material deposited in the upper dermis in a perivascular and periadnaxal distribution.

Table 1 Patient's porphyrin levels in urine blood and erythrocytes.

Substrate	*Porphyrin*	*Level*
Urine (24 hours)	Coproporphyrin	3299 nmol/24hrs
	Uroporphyrin	2566nmol/24hrs
Blood	Copropoophyrin	556nmol/g dry weight
	Protoporphyrin	388mmol/g dry weight
Erythrocytes	Free porphyrin	615 µg/l

MCHC 30.3g/dl, WBC count $8.6x10^3/mm^3$ and platelets $140x10^3/mm^3$). A peripheral blood film revealed poikilocytosis, anisocytosis, and nucleated red cells. The reticulocyte count was 1.7%. Bone marrow was hypercellular with marked megakaryocytic hyperplasia, but no evidence of fibrosis or cellular infiltration. Skin biopsy revealed subepidermal blister with PAS-positive material deposited in the upper dermis in a perivascular and periadnexal distribution (**Figure 2**).

We put the patient on beta carotene and advised to avoid sun exposure by using sun protective clothing.

Discussion

Congenital erythropoietic porphyria is the most severe form of porphyria. The clinical manifestations are markedly variable due to the different mutation in the UROIIIS gene.[2] The nature of the metabolic abnormality is undoubtedly a primary defect of the cosynthase III activity. Some cases of late-onset emphasize the heterogeneity of the disease.[3]

A milder late-onset form, presenting at any age from third decade onward, manifests in a manner similar to porphyria cutanea tarda (PCT) or variegate porphyria (VP), but may also cause thrombocytopenia secondary to hypersplenism.[4] A diagnosis of CEP can usually be suspected on clinical grounds, especially if both the acute and chronic skin changes are found.[5] Erythrodontia of both deciduous and permanent teeth is characteristic.[5]

Adult Gunther's disease is rare and tends to be milder than childhood cases. Photosensitivity, hemolytic anemia and hyperpigmentation are prominent features.[4] Although clinically much less severe, enzyme activity in late-onset CEP is often as low as in childhood form.[6] In the past most patients died by the age of 40 years but improvement in supportive care has improved the prognosis, though the hematological complications may be fatal.[6] Hypersplenism is common.[4,6] Splencetomy may increase erythrocytic life span, reduce porphyrin levels

and increase platelets count in Gunther's disease.

Treatment of CEP is very challenging. Photosensitivity is so severe that photoprotection is crucial. Sun avoidance and use of sun protective clothing are essential. Opaque sunscreen containing titanium dioxide or zinc oxide, possibly with added iron oxide may be of limited value because of the pasty nature of the preparations, hence they are not practical for daily use.[7] Moreover, use of amber window film on home or car window can reduce exposure to Soret band range. The use of oral beta carotene at the doses of 120-180 mg/day has been reported to improve light intolerance in some cases.[8]

Recently allographic bone marrow transplantation from a HLA compatible sibling has emerged and gene therapy has been successfully used '*in vitro*'.[6]

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