Congenital erythropoietic porphyria: a case report.

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Congenital erythropoietic porphyria is a rare autosomal recessive disorder of heme synthesis resulting from deficiency of uroporphyrinogen III synthase (UROIIIS). It is the most severe porphyria. The clinical manifestations are markedly variable due to the different mutation in the UROIIIS gene. We recently diagnosed a case of congenital erythropoietic porphyria. A 9-year-old boy presented with recurrent ulcers on the skin especially dorsum of the hands and feet since aged 3. The physical examination revealed ulcers on the dorsum of the feet, mutilation of the fingers, fluorescent erythrodontia, and darkening and hypertrichosis of the sun exposed area. Laboratory findings showed mild hemolysis, red urine, increased serum alkaline phosphatase level, and fluorescence of the red blood cell and urine. The histopathology was consistent with porphyria. The urine and plasma porphyrin levels confirmed the diagnosis of congenital erythropoietic porphyria. The administration of oral ultracarbon and topical zinc oxide has been tried.

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