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Diagnosis of erythropoietic protoporphyria with severe liver injury: A case report

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Abstract

BACKGROUND

Porphyria is a rare disease with complex classification. Erythropoietic protoporphyria (EPP) is an autosomal recessive inherited disease, and most caused by mutations in the *FECH* gene. EPP combined with liver injury is rarer.

CASE SUMMARY

This paper reports a case of EPP which was admitted to the hospital with "abnormal liver function to be investigated" and diagnosed by repeated questioning of medical history, screening of severe liver injury caused by common causes, and sequencing of the whole exon genome of the second generation. Through diagnosis and treatment of this patient to summarize clinical characteristics of EPP with liver injury, and put forward clinical

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