P-024 - NEUROVISCERAL PAIN IN ERYTHROPOIETIC PROTOPORPHYRIA-RELATED HEPATOPATHY: REPORT OF A BRAZILIAN CASE

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INTRODUCTION: Erythropoietic Protoporphyrin (EPP), an autosomal recessive disorder, is the most common porphyria in childhood. About 90% of the cases are caused by a loss-of-function variant and a low-expression variant in the FECH gene, reducing ferrochelatase activity to 10-30%. Protoporphyrin IX, a lipid-soluble molecule, is the main toxic substrate in EPP, causing immediate photosensitivity. Mainly produced in bone marrow, it progressively accumulates in liver, leading to liver failure in 5% of the cases. Whereas the photosensitivity is the major concern, primarily hepatic presentation has been reported. OBJECTIVES: to report one case of protoporphyrin hepatopathy. MATERIALS AND METHODS: case report. RESULTS: a 14 year-old Brazilian girl was evaluated during a second consecutive episode of opioid-dependent abdominal pain, elevated transaminases, mild jaundice, autonomic signs and red urine. She had no medical issues but mild photosensitivity since childhood. Infectious and structural causes were excluded. Semi-quantitative porphobilinogen (PBG) test was positive and the findings were interpreted as an acute porphyria. The patient slowly progressed to discharge after standard acute attack management, but was hospitalized three more times in a 6-month interval. Meanwhile, hepatic biopsy showed ductular reaction with brown pigment deposits in biliary canaliculi and PBG and aminolevulinc acid (ALA) quantitative tests had normal results, leading to a revised approach. Plasmatic porphyrins demonstrated an extremely high protoporphyrin level (207 mg/dl; reference range: 0-1). Next-generation sequencing detected a previously undescribed likely pathogenic variant (p.Gln122Argfs*23) and the known intronic polymorphism c.315-48T>C, in heterozygosis in FECH. Segregation analysis wasn’t performed. Light exposure avoidance and beta-carotene were prescribed. Subsequent extensive approach to exclude differential diagnosis of abdominal pain, including magnetic ressonance, digestive endoscopy, colonoscopy and electroencephalogram, showed no abnormalities. A 7-month later comparative hepatic biopsy is ongoing. CONCLUSIONS: neurovisceral pain, the remarkable finding in acute porphyrias due to ALA (and possibly PBG) accumulation, was described in a few reports of advanced protoporphyrin hepatopathy. There’s no demonstration of those substrates’ excess in the benign course of EPP. Since sequential liver and bone marrow transplantations are expected to achieve better outcomes in protoporphyrin hepatopathy, in the present case, the recurrent abdominal pain requires careful evaluation to management decision.