

Identification of mutations in the protoporphyrin oxidase gene and its diagnostic implications in porphyria variegata in Chile

Porfiria variegata en Chile: Identificación de mutaciones en el gen protoporfirinógeno oxidasa y su implicancia diagnóstica

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Variegate porphyria (VP) results from a hereditary deficiency of protoporphyrinogen oxidase (PPOX) that is transmitted in an autosomal dominant fashion. The diagnosis is based on the clinical symptoms and is confirmed biochemically. Sometimes, however, these diagnostic tools reveal limitations in establishing the definitive diagnosis of the prevailing type of acute porphyria. In these patients, molecular genetic analyses can be useful. We performed molecular genetic studies in 13 Chilean families by PCR amplification of the PPOX gene, conformation sensitive gel electrophoresis, and automated DNA sequencing. In five symptomatic patients from different families, respectively, the biochemical data confirmed the diagnosis of VP. In seven other families, however, the biochemical studies were not conclusive. Furthermore, the original biochemical analysis in one clinically severely affected patient from a further family even suggested the diagnosis of erythropoietic protoporphyria (EPP). Besid