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Identification of *FECH* gene multiple variations in two Chinese patients with erythropoietic protoporphyria and a review

Key words: Erythropoietic protoporphyria, Chinese patients, Clinical manifestation, Ferrochelatase, Missense mutations

Research Summary

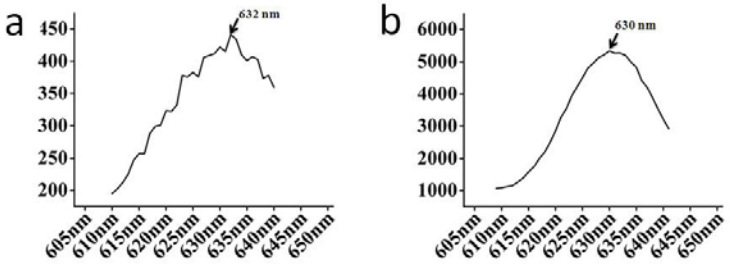
- **In this research, we describe the clinical features of two Chinese patients with EPP, with diagnosis confirmed by the increase of FEP, detection of plasma fluorescence peak at 630-634 nm and analysis of *FECH* gene mutations.**

- **Using gene scanning, we identified a small deletion in the *FECH* gene (c. 973delA) in patient A and a pathogenic *FECH* mutation (c. 1232G>T) in patient B and also observed some nucleotide variations.**

- **The family pedigree of patient A was then established by characterization of the genotype of the patient's relatives.**

Innovation points

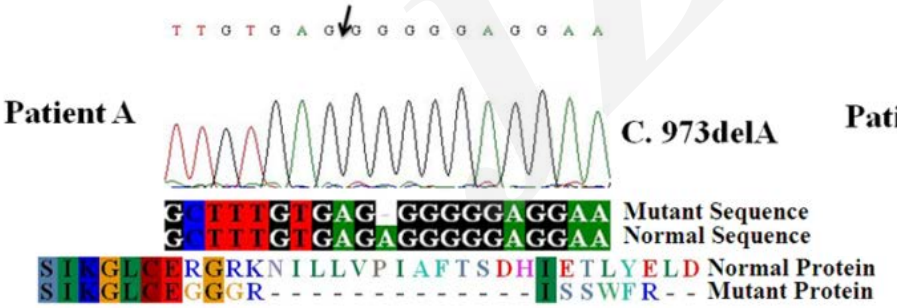
Describe the clinical features

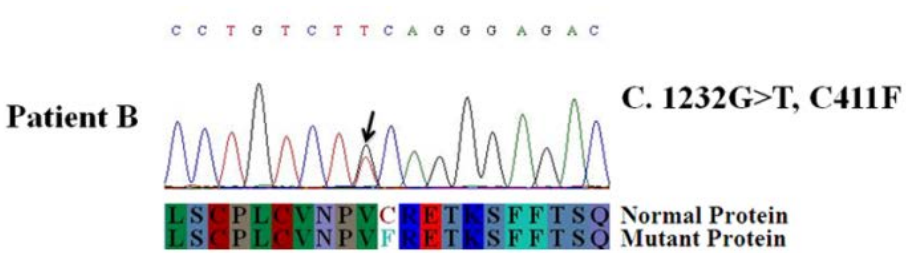


Plasma fluorescent spectrum assay

Chronic skin lesions of the patients

Detect the mutation of *FECH* gene

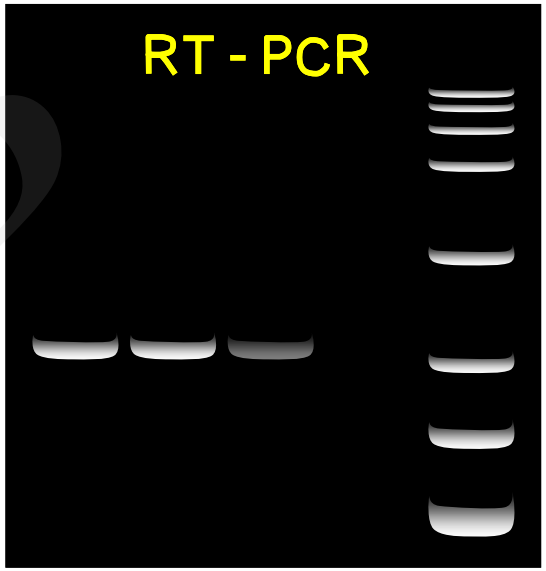




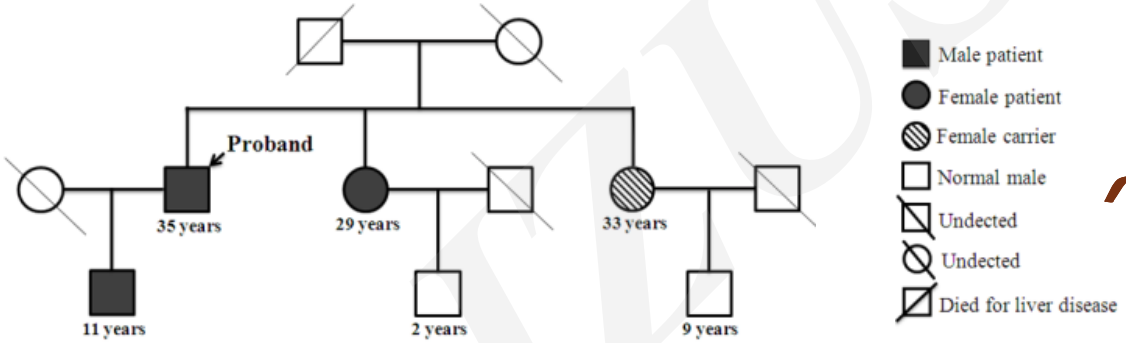
Innovation points

Observe some nucleotide variations

c. 798C>G **c. 921A>G**
IVS 1 as -23C>T **IVS 3 ds +23A>G**
IVS 9 ds +35C>T **IVS3-48 T>C**



Analysis the Family pedigree of the patient A



We confirm the diagnosis of EPP and expand the *FECH* genotypic spectrum