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Detection of a high mutation frequency in exon 12 of the porphobilinogen deaminase gene in patients with acute intermittent porphyria

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Abstract

Direct cDNA sequencing was performed on asymmetrically amplified transcripts from the porphobilinogen deaminase (PBG-D) gene of thirteen unrelated individuals with acute intermittent porphyria. Four different mutations and a polymorphic site were detected in exon 12 of the gene, four being the result of single base substitutions and one being caused by dinucleotide deletion. All of these mutations are located in domain 3 of the PBG-D molecule, with the single base substitutions affecting the hydrophobic interfaces between domains 1 and 3. The dinucleotide deletion results in a frame-shift producing a premature stop codon.