

Detection of four mutations in six unrelated South African patients with acute intermittent porphyria

[P M Ong¹](#), [W G Lanyon](#), [R J Hift](#), [J Halkett](#), [M R Moore](#), [C S Mgone](#), [J M Connor](#)

Abstract

We have screened the hydroxymethylbilane synthase cDNA from six South African patients with acute intermittent porphyria, using a combination of chemical cleavage mismatch analysis and direct sequencing of asymmetrically amplified PCR products. Four mutations were detected, a novel T insertion (771insT) and three missense mutations (R26H, R116W and R173Q). The 771insT mutation produces a stop codon, thirty-three codons downstream and a loss of approximately 20% of the protein is predicted. The R116W mutation, which was found to have a high prevalence in the Dutch population, was detected in three unrelated South African patients.