A MspI polymorphism for the human porphobilinogen deaminase gene

David H.Llewellyn[1,2], Noor A.Kalsheker[1], George H.Elder[1]*, Paul R.Harrison[2], Stany Chretien[3] and Michel Goossens[3]

[1]Department of Medical Biochemistry, University of Wales College of Medicine, Heath Park, Cardiff CF4 4XN, [2]Beatson Institute for Cancer Research, Bearsden, Glasgow, UK and [3]INSERM U-91 Hôpital Henri Mondor, 94010 Creteil, France

SOURCE/DESCRIPTION: A 7 kb EcoRI fragment of the human porphobilinogen (PBG) deaminase (EC 4.1.3.8) gene isolated from an adult human genomic DNA library.

POLYMORPHISM: Hybridisation to MspI (CCGG) (BRL Ltd) digested human genomic DNA identifies a 2 allele polymorphism with bands of 3.0 kb (A1) or 2.2 kb (A2) and invariant bands of 1.1, 0.95, 0.8 and 0.5 kb.

FREQUENCY: Studied in 16 unrelated European Caucasians:
3.0 kb (A1) allele : 0.47
2.2 kb (A2) allele : 0.53

NOT POLYMORPHIC FOR: Not extensively investigated; Taq I, Bgl II, Pvu II in at least 6 unrelated individuals.

CHROMOSOMAL LOCALISATION: 11, q 23 - qter region (1).

MENDELIAN INHERITANCE: Co-dominant segregation in 5 unrelated families.

PROBE AVAILABILITY: Information from M. Goossens at above address.

OTHER COMMENTS: Requires high stringency hybridisation and post-hybridisation washing procedures. Polymorphism only very weakly detected by full-length cDNA for PBG deaminase mRNA from human thalassaemic spleen(2).

REFERENCE: (1) Wang, A.L., Arredondo-Vega, F.X., Giampetro, P.F., et al (1981) Proc.Nat.Acad.Sci.(USA) 78, 5634-5738. (2) Raich, N., Romeo, P.H., Dubart, A., et al (1986) Nucleic Acids Res. 14, 5955-5968.

ACKNOWLEDGEMENTS: Supported by an MRC project grant to G.H.E., N.K., P.R.H.

*To whom correspondence should be addressed