A PstI polymorphism for the human porphobilinogen deaminase gene (PBG)

Jin-Sung Lee and Maria Anvret*

Department of Clinical Genetics, Karolinska Hospital, 104 01 Stockholm, Sweden

SOURCE/DESCRIPTION : A 1.7 kb Eco RI-Taq I fragment was used as a probe. This fragment was isolated from pUSE 109 containing a 7 kb Eco RI genomic fragment from the human porphobilinogen(PBG) deaminase(EC 4.1.3.8.) gene. (1)

POLYMORPHISM : Pst I digests of human genomic DNA were hybridized to the probe. This presented a two allele polymorphism with bands of 2.2 kb(A1), 1.4 kb(A2) and constant bands of 1.6 kb, 0.7 kb.

FREQUENCY : As studied in 37 unrelated European Caucasians :
A1 (2.2 kb) : 0.58
A2 (1.4 kb) : 0.42

NOT POLYMORPHIC FOR : Eco RI , Hind III , Bgl II ,
Bam HI , Hinf I , Taq I

CHROMOSOMAL LOCALISATION : 11, q23-qter region (2)

MENDELIAN INHERITANCE : Co-dominant segregation in 3 unrelated families.

PROBE AVAILABILITY: Information from M.Goossens (1)

OTHER COMMENTS : The previous published Msp I polymorphism was also detected within this genomic fragment and revealed similar results as Llewellyn,D.H.,et al.(1)

REFERENCE : (1) Llewellyn,D.H.,Kalsheker,N.A.,Elder,G.H.,et al.(1987) Nucleic Acid Res.15,1349. (2) Wang,A.L.,Arredondo-Vega,F.X.,et al.(1981) Proc.Nat.Acad.Sci.(USA) 78,5734-5738

*To whom correspondence should be addressed