RFLPs for the human erythrocyte membrane glycophorin C gene

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SOURCE AND DESCRIPTION OF THE CLONE : pGCF22 is a 0.9 Kb cDNA clone in pKT218 for human erythrocyte membrane glycophorin C (blood group Gerbich antigens) which was isolated from a human liver cDNA library (provided by S.H. Orkin, Children's Hospital Medical Center, Boston) screened with the 0.45 Kb cDNA insert of the pGCW5 clone (Colin et al., 1986). Sequence analysis revealed that the pGCF22 clone encodes amino-acids 10 to 128 of glycophorin C and the entire untranslated 3' end mRNA (unpublished data).

POLYMORPHISM : The probe detects two allele polymorphisms with TaqI (T1 : 9.1 Kb; T2 : 7.6 + 1.5 Kb), PstI (P1 : 1.05 Kb; P2 : 0.9 Kb; invariant bands at 3.6 Kb and 1.3 Kb) and BstXI (B1 : 4.1 Kb; B2 : 2.2 Kb; invariant bands at 2.5 Kb, 1.5 Kb and 1.1 Kb).

FREQUENCY : TaqI (94) alleles T1 : 0.60; T2 : 0.40
PstI (46) P1 : 0.56; P2 : 0.44
BstXI(33) B1 : 0.24; B2 : 0.76
numbers in brackets = unrelated chromosomes studied.

NOT POLYMORPHIC FOR :

BanI, BglII, BstNI, DraI, EcoRI, HindIII, PvuII, SacI, ScaI, StuI, Tth111I, XbaI, XmnI.

Minimum of unrelated chromosomes tested = 10.

CHROMOSOMAL LOCALIZATION : Probe localized to 2q14-q21 by in situ hybridization (Mattei et al., 1986).

MENDELIAN INHERITANCE : The PstI and TaqI RFLPs showed mendelian segregation in one very large three generation family. Although the BstXI RFLP was not tested in families, the blots that revealed it were tested with 10 other probes eliminating the possibility of artefacts due to plasmid contamination or digestion problems.

PROBE AVAILABILITY : available from J.P. Cartron for RFLP studies.

OTHER COMMENTS : There is very strong linkage disequilibrium between the TaqI and PstI RFLPs since only the haplotypes T1P1 and T2P2 were observed in 10 chromosomes tested. There is also linkage disequilibrium between the PstI and BstXI RFLPs; however three haplotypes were observed in 23 chromosomes tested P1B1 (0.23), P1B2 (0.34) and P2B2 (0.43).

REFERENCES : Colin et al., J. Biol. Chem. (1986), 261 : 229-233; Mattei et al., Hum. Genet. (1986), in press.