A DNA polymorphism with KpnI of the human liver-type phosphofructokinase (PFKL) gene

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SOURCE AND DESCRIPTION OF CLONE: The 3.3 kb genomic BamHI fragment of human liver phosphofructokinase (PFKL) gene cloned in the BamHI site of pGEM-3 to produce clone pG-PFKL 3.3 was used as a hybridization probe.

POLYMORPHISM: After digestion with KpnI and Southern blotting, the PFKL probe hybridizes to a 1.7 kb DNA fragment (allele A1) when the polymorphic KpnI site is present and a 1.9 kb fragment (allele A2) when the site is absent (see Figure).

FREQUENCY: The frequencies of alleles A1 and A2 determined by analysis of DNA from 82 unrelated Caucasian individuals were: A1=0.35, A2=0.65.

NOT POLYMORPHIC FOR: ApaI, AluI, AvaI, BalI, BclI, BglI, BglII, ClaI, DdeI, DraI, EcoRI, EcoRV, HhaI, HincII, HindIII, HpaI, MspI, NciI, NcoI, PvuII, SalI, SmaI, SstI, TaqI, XbaI, XhoI, XmnI (five unrelated individuals tested).

CHROMOSOMAL LOCATION: Chromosome 21q22.3.

MENDELIAN INHERITANCE: Demonstrated in 40 nuclear families.

PROBE AVAILABILITY: Contact Dr. Yoram Groner, Department of Virology, Rehovot, Israel.

OTHER COMMENTS: Polymorphism information content is 0.35.

REFERENCES: 1) Levanon, D. et al (1987) Biochem. Biophys. Res. Commun. **147**: 1182-1187; 2) Cox, D.R. et al. (1984) Cytogenet. Cell. Genet. **37**: 441; 3) Van Keuren, M. et al. (1986) Hum. Genet. **74**: 34-40.

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