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Sph I restriction fragment length polymorphism on human chromosome 16 detected with an APRT gene probe

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SOURCE/DESCRIPTION: The purified 2.3 kb Bam HI insert fragment from plasmid pAT2.3aprt was used as a probe. This fragment constitutes part, but not all, of the biologically active human aprt gene and contains few or no repetitive elements. (1)

POLYMORPHISM: Sph I identifies a two allele polymorphism with bands at either 8kb or 12kb.

FREQUENCY: As studied in 30 unrelated Caucasians:-

8kb allele (A1)	0.78
12kb allele (A2)	0.22

NOT POLYMORPHIC FOR: Msp I, Sst I, Bam HI, Xho I, Pst I each with a panel of at least 5 individuals.

CHROMOSOMAL LOCATION: The aprt gene has been localised to chromosome 16q24 (2).

MENDELIAN INHERITANCE: Codominant segregation shown in 1 informative 3 generation family of 11 individuals.

PROBE AVAILABILITY: Available from Janet Arrand.

OTHER COMMENTS: This probe also detects a known Taq I polymorphism (1). RFLP's were observed at a stringency of 0.1 x SSC; 1% SDS at 65° using 0.7% gels.

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