TaqI RFLP in human adenylate kinase-1 (AK1) gene region on chromosome 9

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DESCRIPTION OF PROBE: 3.25 kb BamHI fragment (hAK1B3.25) spanning exon 2 to exon 5 of the adenylate kinase-1 gene was derived from a human genomic clone (lambda hAK1-1, kindly provided by Dr. A. Nakazawa) isolated from Maniatis' library.

POLYMORPHISM: Fragment hAK1B3.25 detects two polymorphic bands, 6.4 kb and 5.5 kb, in TaqI digested human genomic DNA. The polymorphic TaqI site appears to lie within the 3' untranslated region of the AK1 gene (1).

ALLELE FREQUENCIES: Estimated from 100 unrelated Caucasians

	<u> 5126 (</u> KD)	rrequenc
A1	6.4	0.79
A2	5.5	0.21

ABSENCE OF POLYMORPHISMS: Digests of human DNA from 9 or more unrelated Caucasians showed no polymorphisms with the following restriction endonucleases: BamHI, BglII, BstEII, EcoRI, HindIII, MboI, MspI, PstI, PvuII, RsaI, SacI and XbaI.

CHROMOSOME LOCALISATION: The AK1 gene maps to human chromosome 9 at q34 (2).

MENDELIAN INHERITANCE: Co-dominant inheritance was demonstrated using 8 families.

PROBE AVAILABILITY: Subclone phAK1B3.25 is available for linkage studies from NTBH (Calgary).

COMMENTS: The probe detects a strong constant band of 1.3 kb and two weaker bands of 0.3 kb and 1.4 kb.

REFERENCE: 1. Nakazawa, A., Japan, personal communication.
2. Ferguson-Smith et al. Hum. Genet. 34:35-43, 1976.

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