NcoI and TaqI RFLPs for human M creatine kinase (CKM)

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SOURCE/DESCRIPTION:

Probe pHMCKUT contains a 135 bp cDNA fragment inserted into pGEM 3. The probe corresponds to nucleotides 1201 to 1336 located in the 3' untranslated region of human M creatine kinase. The probe is specific for human M creatine kinase [EC 2.7.3.2] and does not hybridize to human B creatine kinase sequences.

POLYMORPHISM:

NcoI identifies a two allele polymorphism of a band at either 2.5 kb or 3.6 kb. TaqI identifies a two allele polymorphism at either 3.8 kb or 4.5 kb.

FREQUENCY:

NcoI studied in 49 unrelated individuals:
4.5 kb allele: 0.33
3.8 kb allele: 0.67

TaqI studied in 40 unrelated individuals:
2.5 kb allele: 0.23
3.6 kb allele: 0.78

NOT POLYMORPHIC FOR:

AccI, AvaI, AvaII, BamHI, BanII, Bsp1286, BstI, BspMI, Asp718, DdeI, DraI, EcoRV, FnuDII, HinfI, HindIII, HpaII, HphI, NdeI, NheI, PstI, SauI, Sau96I, ScrFI, SphI, SspI, SpeI SstI, XbaI, XmnI

CHROMOSOMAL LOCALISATION:

Human M creatine has been localized to chromosome 19q (2)

MENDELIAN INHERITANCE:

Autosomal codominant inheritance was shown in six informative Caucasian families.

PROBE AVAILABILITY:

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REFERENCE:

(1) Perryman et al. (1986) Biochem. Biophys. Res. Comm. 140: No. 4, 981-989.
(2) Nigro et al. (1987) Am. J. Hum. Genet. 40: 115-125.

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