

**NcoI RFLP at the creatine kinase-muscle type gene locus (CKMM, chromosome 19)**

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**Source/description:** A 3.2 kbp human genomic DNA fragment (BamHI-Sau3A) of the 3' untranslated and 3' flanking region of the CKMM gene was isolated and subcloned into the BamHI site of vector pSP64 (Promega Biotec).

**Polymorphism:** The CKMM 3'-probe identifies a 2-allele polymorphism with bands at 2.3 and 1.0 kbp (allele A) and 3.3 kbp (allele B). In addition a weak constant 4.2 kbp band is observed.

This probe also detects a 2-allele TaqI RFLP reported previously (1), as either a 4.3 kbp (A) or a 4.2 kbp (B) band.

**Frequency:** Estimated from 30 unrelated Caucasians.

A allele: 0.68

B allele: 0.32

**Not polymorphic for:** BamHI, HindIII, EcoRI, EcoRV, MspI, PstI, PvuII, RsaI, SacI and XbaI.

**Chromosomal location:** The CKMM locus previously has been assigned to 19q13.2-ql3.3 (2).

By Southern blot analysis of human-rodent somatic cell hybrids containing unique subregional fragments of chromosome 19 of man (3) we have assigned the gene to 19q13.2 (J. Schepens et al, in preparation).

**Mendelian Inheritance:** Co-dominant segregation in 8 families with 3 generations.

**Other comments:** Low background/high signal with final wash at 65°C in 0.1 x SSC/0.1% (w/v) SDS. The CKMM locus is closely linked to the Myotonic dystrophy locus (H. Brunner et al, in preparation).

**Probe availability:** On collaborative basis, contact B.W.

**References:**

1. Putney, S. et al., J. Biol. Chem. 250, 14317-14320 (1984).
2. Nigro, J. M. et al., Am. J. Hum. Genet. 40, 115-125 (1987).
3. Hulsebos, T. et al., Cytog. Cell. Genet. 43, 47-56 (1986).

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