

**EcoRI RFLP at 19 cen-q13.2 identified by the anonymous DNA sequence pPM6.7 (D19S18)**

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**Source/description:** A 0.75 kbp BamHI/PstI DNA segment subcloned in pGEM4 (Promega Biotec) and derived from a human der 19 specific pCos2 EMBL library (Hulsebos et al., 1986).

**Polymorphisms:** EcoRI (GAATTC) identifies a two allele polymorphism with bands at either 40kbp (A1) or 25 and 15kbp (A2) and an invariant band at 4.5kbp. The probe is also polymorphic for MspI (CCGG) with bands at either 5kbp (B1) or 3.5kbp (B2) and an invariant band at 4.0kbp. Not polymorphic for TaqI, PvuII, PstI, BamHI, RsaI, and HindIII, when tested on chromosomal DNA of 5 unrelated individuals.

**Frequency:** Studied for EcoRI alleles in 44 unrelated Caucasians: 40kbp allele (A1) 0.65; 25kbp/15kbp allele (A2) 0.35. Frequency MspI polymorphism not studied.

**Chromosomal localisation:** Localised at 19cen-q13.2 by Southern blot analysis using a panel of human Chinese hamster cell hybrids containing unique subregional fragments of chromosome 19 (Schonk et al., in prep.).

**Mendelian inheritance:** Co-dominant segregation demonstrated in 12 myotonic dystrophy (DM) families. Linked to the DM gene at male recombination frequency 0.10 (Lod-score=2.62).

**Probe availability:** Available - for collaboration only - from B.Wieringa.

**Reference:** T. Hulsebos, B. Wieringa, R. Hochstenbach, D. Smeets, J. Schepens, F. Oerlemans, J. Zimmer, and H.H. Ropers (1986) Cytogenet. Cell Genet. 43, 47-56.

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