TaqI RFLP at 19q13.1 identified by the anonymous DNA sequence p5B18 [D19S28]

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Source/description: A 0.5 kbp EcoRI/PstI DNA segment subcloned in pSP64 (Promega Biotec) and derived from a human der (19q) specific lambda EMBL3 library (Smeets et al., in prep).

Polymorphisms: TaqI (TCGA) detects a two allele polymorphism with bands at either 2.7 kbp (A1) or 1.7 kbp (A2) and no constant bands.

Not polymorphic for BglII, EcoRI, EcoRV, MspI, PvuII, RsaI and SacI, when tested on chromosomal DNA of 10 unrelated individuals.

Frequency: Studied for TaqI alleles in 41 unrelated Caucasians: 2.7 kbp allele (A1) 0.55 and 1.7 kbp allele (A2) 0.45.

Chromosomal localisation: Localised at 19q13.1 by Southern blot analysis using a panel of human-rodent somatic cell hybrids containing unique segments of 19q (Schonk et al. 1989).

Mendelian inheritance: Co-dominant segregation demonstrated in 16 myotonic dystrophy (DM) families. Linked to the DM gene at recombination frequency 11.8 cM with lod score 2.92 (Brunner et al., submitted).

Probe availability: Available, contact B. Wieringa.

References: Schonk et al. (1989) Genomics 3, in press.

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