A PstI polymorphism detected by a genomic clone at the human slow troponin T (TNNT1) gene locus

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Source/Description: The clone MSL-G-14p2 is an 800 bp fragment subcloned into the SmaI site of the pUC18 vector (Pharmacia, NJ). The subclone was derived from a phage clone MSL-G-14 which was isolated by screening a human genomic library in lambda EMBL3 A with a 241 bp cDNA clone of human slow troponin T gene (1, 2).

Polymorphism: The probe MSL-G-14p2 identifies a PstI two allele RFLP. Polymorphic bands are at 4.2 kb (A1) and 3.2 kb (A2).

Frequencies: Allele frequencies calculated from 30 unrelated Caucasian chromosomes are 0.40 for the 4.2 kb fragment and 0.60 for the 3.2 kb fragment. The heterozygosity is 0.48 with a PIC of 0.40.

Not Polymorphic For: SstI, NcoI, HincII, StuI, HinfI, BglI, TaqI, PvuII.

Chromosomal Localization: The probe has been previously localized to 19q13.3-q13.4 (1).

Mendelian Inheritance: Co-dominant segregation demonstrated in 3 myotonic dystrophy families. Linkage data will be reported elsewhere (Samson *et al.*, in preparation).

Availability: Available for collaboration. Contact J.R. Gilbert.

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Dinucleotide repeat polymorphism at the human tissue plasminogen activator gene (PLAT)

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Source/Description: The polymorphism is a CA repeat (1, 2) of the form: (CA)₁₅C(TC)₁₀ and starts at position 16,911 (intron 1) of the human PLAT gene on chromosome 8p12-q11.2 (3). The polymorphism can be typed using the polymerase chain reaction (PCR) as described previously (4). The predicted size of the amplified sequence was 111 bp.

Primer Sequences:

GCCTGGACAACATAGAGAAACC (CA strand);
ACTTCAGGCATGTGCCACTG (TG strand).

Frequency: Estimated from 236 chromosomes of unrelated CEPH (Paris) individuals and 54 chromosomes of other unrelated individuals. Observed heterozygosity = 77%.

Allele (bp)	Frequency	Allele (bp)	Frequency
D1 149	0.003	D2 141	0.010
D3 139	0.003	D4 137	0.003
D5 133	0.007	D6 131	0.024
D7 129	0.048	D8 127	0.132
D9 125	0.024	D10 123	0.073
D11 121	0.007	D12 119	0.003
D13 117	0.145	D14 115	0.073
D15 113	0.418	D16 111	0.010
D17 109	0.007	D18 105	0.010

Mendelian Inheritance: Co-dominant segregation was observed in 40 CEPH (Paris) families.

Chromosomal Location: The human PLAT gene has been assigned to chromosome 8p12-q11.2 (3).

Other Comments: The PCR reaction was performed as described previously (4) with inclusion of formamide to a final concentration of 5% in the reaction mix. GenBank accession no. K03021.

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