A TaqI polymorphism detected by a genomic clone at the locus D5S39 (5q11 – 13)

B.S.Mankoo, G.Melmer, G.Kalsi, R.Sherrington and H.M.D.Gurling

Department of Academic Psychiatry, University College and Middlesex School of Medicine, Riding House Street, London W1N 8AA, UK

Summary: A further RFLP was identified by a phage clone obtained by screening a genomic library with the probe p105–153 (D5S39).

Source/Description: Phage clone 153/06–13.2 was isolated after screening a genomic library made in EMBL-3 (Clontech Laboratories, Inc.) with the probe p105–153.

Polymorphism: TaqI identifies a two allelic RFLP with polymorphic bands at 3.6 kb (D1) and 2.3 + 1.3 kb (D2) (Figure 1).

Frequencies: Studies of 38 unrelated European Caucasians showed allele frequencies of 28 (37%) (A) and 48 (63%) (B).

Not Polymorphic For: BamHI, BclI, BglI, BglII, EcoRI, HaeIII, HincII, HindIII, HinfI, PvuII, RsaI, SacI, Sau3A, StuI, XmnI using DNA from eight unrelated individuals.

Chromosomal Localization: The probe p105–153 has been previously localized to 5q11–13 (1).

Mendelian Inheritance: Codominant Mendelian inheritance was observed in three large families with a total of 33 children.

Availability: The probe is available for collaboration from H.M.D.Gurling.

Other Comments: To block repeated sequences the labeled probe was prehybridized with sonicated human placental DNA (2). Filters were washed at a final stringency of $0.5 \times \text{SSC}/0.1\% \text{ SDS}$ at 65°C .

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References: 1) Alitalo, T. *et al.* (1987) *Cytogenet. Cell. Genet.* **46**, 570. 2) Sealey, P.G., Whittaker, P.A. and Southern, E.M. (1985) *Nucl. Acids Res.* **13**, 1905–1922.

KpnI RFLP at the human muscle-type phosphofructokinase (PFKM) gene locus

F.T.Dionne, J.Groncin and C.Bouchard

Physical Activity Sciences Laboratory, Laval University, Québec G1K 7P4, Canada

Source/Description: PCHPFKM1, a 2.86-kb cDNA clone, contains 76 bp of 5'-untranslated sequence, 2340 bp encoding human muscle PFK, and 399 bp of 3'-untranslated sequence plus a poly(A) tract (1). This clone was a generous donation from Dr Alan McLachlan (1).

Polymorphism: KpnI identifies a two-allele polymorphism with fragments of 17 (allele 1) and 11 and 7.2 kb (allele 2) and a constant fragment of 14 kb.

Frequency: Studied in 34 unrelated Caucasians.

17 kb allele (1): 0.69

11 and 7.2 kb allele (2): 0.31

Not Polymorphic For: AvaII, BamHI, BanI, BglII, DraI, EcoRI, EcoRV, HincII, HindIII, MspI, NcoI, PstI, ScaI, SstI and XmnI with DNA from a panel of 18 subjects.

Chromosomal Location: The PFKM locus has been mapped to 1 cen-q32.

Mendelian Inheritance: Co-dominant autosomal segregation was observed in a 2 generation family of 9 members and in 3 families of 4 members.

Probe Availability: Available from Dr Alan McLachlan (1).

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Reference: 1) Sharma, P.M., Reddy, G.R., Vora, S., Babior, B.M. and McLachlan, A. (1989) *Gene* **77**, 177–183.