TaqI and MspI RFLPs are detected by the human 2,3-biphosphoglycerate mutase (BPGM) cDNA

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Source/Description: The XMEL4 clone is a 1.5 kb cDNA subcloned into the EcoRI site of pBluescript SKII+. XMEL4 was isolated from a human melanoma cDNA library. Sequence analysis showed complete homology between XMEL4 and the 2,3-biphosphoglycerate mutase (BPGM) cDNA sequence (1, 2).

Polymorphism: Both TaqI and MspI detect two-allele polymorphisms when probed with XMEL4.

Frequency: Allele frequencies were determined by typing the 80 parents of the CEPH reference families.

Enzyme	Allele	Size	Frequency
TaqI	1	6.0 kb	0.55
	2	5.5 kb	0.45
MspI	1	7.0 kb	0.83
	2	6.2 kb	0.17

Not Polymorphic For: RsaI, HinfI, PvuII, EcoRI, HindIII, BclI, BglII and SacI (5 unrelated individuals screened for each enzyme).

Chromosomal Localization: BPGM has been assigned to 7q31-q34 (3).

Mendelian Inheritance: Codominant segregation detected in all 40 CEPH families.

Probe Availability: Available from NCD at the above address.

Other Comments: Multipoint linkage analysis of the MspI polymorphism on the 40 CEPH reference families showed that BPGM is located in the interval between MET (7q31) and TCRB (7q35) and that it is tightly linked to D7S93 (theta = 0.03, LOD = 10.5).

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References: (1) Joulin,V. *et al.* (1986) *EMBO J.* **5**, 2275–2283; (2) Sakoda,S. *et al.* (1988) *J. Biol. Chem.* **263**, 16899–16905; (3) Barichard,F. *et al.* (1987) *Hum. Genet.* **77**, 283–285.

A VNTR polymorphism, D1S110, at 1q21 – 1q31

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Source and Description of Clone: pBH516 consists of a 2.2 kb EcoRI-SacI fragment subcloned into pUC19. pBH516 was isolated from a chromosome 1 partial-MboI-digested library in vector EMBL-3 (Lebo *et al.*, 1986). pBH516 was obtained by screening with the core minisatellite in lambda33.6 (Jeffreys, Wilson and Thein, 1985), as part of a search for VNTRs.

Polymorphism: BamHI identifies multiple alleles in the range of 5.5–6.5 kb.

Frequency: pBH516 detects a human VNTR site with many closely spaced alleles. The proportion of CEPH grandparents heterozygous is 0.625. 204 chromosomes were examined.

Not Polymorphic For: EcoRI, HindIII, HinfI, MboI, MspI, PstI, PvuII.

Chromosomal Location: Between ATP1A2 (1q21–q23) and D1S61 (1q25–q31), based on pedigree analysis in the CEPH families.

Mendelian Inheritance: Codominant segregation observed in 6 CEPH families.

Probe Availability: Available without restriction.

Other Comments: Alleles are closely spaced and are best resolved by extended electrophoresis.

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