A TaqI DNA polymorphism in the human cathepsin B gene (CTSB)

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Source/Description: Probe pCB1 contains 627 bp of 3' cDNA of cathepsin B and ~880 bp untranslated region (1).

Polymorphism: A two-allele polymorphism with bands at 6.2 kb (A) and 5.4 kb and 0.8 kb (B) is detected after hybridization with human genomic DNA digested with TaqI.

Not Polymorphic For: BamHI and RsaI.

Frequency: 24 unrelated Caucasians (North America) were studied.

A allele: 0.77

B allele: 0.23

Chromosomal Location: Cathepsin B (CTSB) has been reported to 8p22 or 13q14 (2). Our preliminary data confirms the chromosome 8 location (Fong *et al.*, in preparation).

Mendelian Inheritance: Codominant segregation in 3 families with 2 generations.

Other Comments: High stringency wash of the blots in 0.1×SSC/0.1% SDS at 65° shows no significant background. Two additional constant bands at 2.4 kb and 0.5 kb were identified. The digests were run on a 1.0% agarose gel on a constant field electrophoresis at 60 V.

Probe Availability: Available on request from Wang-Ting Hsieh or Dunne Fong.

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References: 1) Fong, D. *et al.* (1986) *Proc. Natl. Acad. Sci. USA* **83**, 2909-2913. 2) Bouyon, C. *et al.* (1989) *Cytogenetics and Cell Genetics* **HGM10**, 966.

pDP1007, detects an X polymorphism with HindIII in negroids at the ZFX locus

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Source and Description of Clone: pDP1007 is a genomic 1.3kb HindIII fragment subcloned into pUC13 from the phage clone λOX82 (1). λOX82 is derived from interval 1A2 of the human Y chromosome (1). The probe also hybridizes to a locus (ZFX) at Xp21.3 on the human X chromosome (2).

Polymorphism: HindIII digests reveal a simple 2 allele X-specific polymorphism with band sizes 19kb and 14kb, and a constant Y-specific band at 1.4 Kb.

Frequency:

Allele	Size (kb)	Negroid	Caucasoid
A1	19	0.72	1.00
A2	14	0.28	0.00

Studied 41 caucasoid and 36 negroid unrelated X chromosomes.

Not Polymorphic For: BamHI, BglI, BglII, EcoRI, MspI, PstI, SstI, TaqI, XbaI.

Chromosomal Localization: The ZFY and ZFX genes have been assigned by *in situ* hybridization to Yp11.32 and Xp21.3 respectively (2).

Mendelian Inheritance: Polymorphism follows an X-linked segregation pattern.

Probe Availability: Probe obtained from D. Page, Whitehead Institute for Biomedical Research, Cambridge.

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References: 1) Page, D. C. *et al.* (1987) *Cell* **51**, 1091-1104. 2) Muller, G. and Schempp, W. (1989) *Hum. Genet.* **82**, 82-84.

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