A new polymorphic probe on chromosome 3p: λ LIB18-88 (D3S1093)

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Source/Description: λ LIB18-88 is a 2.1 kb EcoRI fragment isolated from a Charon 21A human chromosome 3 library (1).

Polymorphism: MspI digestion of genomic DNA and hybridization with the probe detects a two-allele polymorphism: 14.0 kb (A1) and 9.6 kb (A2).

Frequency: Estimated from 120 unrelated Caucasians.

A1: 0.41

A2: 0.59

Frequency of Heterozygosity: 0.48.

Not Polymorphic For: BamHI, BglII, DraI, EcoRI, HindIII, HinfI, PvuII, RsaI and TaqI.

Chromosomal Localization: Using a somatic cell hybrid panel (2) which was based on linkage groups anchored to physically localized markers, the probe was assigned to the short arm of chromosome 3 between 3p14-p21.

Mendelian Inheritance: Mendelian inheritance has been demonstrated in a two generation von Hippel-Lindau disease (VHL) family.

Probe Availability: Available for collaboration.

Acknowledgement: This project has been funded at least in part with Federal funds from the Department of Health and Human Services under contract number N01-CO-74102.

References: 1) American Type Culture Collection No. 57717, National Lab ID Code LA03NS01. 2) Lerman, M., Latif, F., Glenn, G. *et al.* (1991) Isolation and regional localization of a large collection (2,000) of single copy DNA fragments on human chromosome 3 for mapping and cloning tumor suppressor genes. *Human Genetics* **86**, 567-577.

Detection of a TaqI polymorphism within the human TPA gene (PLAT)

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Source/Description: A 971 bp PCR-amplified genomic DNA fragment of the human TPA gene. The sequences of the primers were as follows with the corresponding nucleotides of the TPA gene (1) indicated in parentheses: 5'-TTACAGCCATGCTG-ATGGC-3' (27187-27205), 5'-ACTGGCGTCAGTGCCTG-A-3' (28141-28158).

Polymorphism: TaqI identifies a two allele polymorphism with bands at either 2.7 kb (C1) or 2.3 kb (C2), as well as a monomorphic band at 0.5 kb.

Frequency: Estimated from 23 unrelated Caucasians.

C1: 2.7 kb allele: 0.41

C2: 2.3 kb allele: 0.59

Not Polymorphic For: BamHI, ClaI, SacI, SphI, XhoI.

Chromosomal Location: Chromosome 8, bands 8p12-q11.2 (2).

Mendelian Inheritance: Co-dominant segregation shown in three families of German origin.

PCR Conditions: PCR was performed using a Perkin Elmer Cetus Kit employing 0.5 μ g of genomic DNA under the following conditions: 35 cycles of 30-s denaturation at 94°C, 40-s annealing at 58°C, and 180-s elongation at 72°C. The elongation product was purified by spin dialysis in a Centricon-30 microconcentrator (Amicon). 100 ng were used for nick translation.

Other Comments: The TPA cDNA identifies the same polymorphism together with four invariable bands.

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References: Degen, S.J.F. *et al.* (1986) *J. Biol. Chem.* **261**, 6972-6985. 2) Yang-Feng, T.L. *et al.* (1986) *A. J. Hum. Genet.* **39**, 79-87.

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