BclI RFLP of the plasminogen activator inhibitor type 2 gene (PLANH) on chromosome 18q21-q23

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Source and Description of Clone: The probe, pPAI-2/ED 1.6, contains the first 1.6 kilobases of plasminogen activator inhibitor type 2 cDNA (1). pPAI-2/ED 1.6 was subcloned from pBTA447 as an EcoRI/DraI restriction fragment into pUC19. The insert may be excised from pPAI-2/ED 1.6 by digestion with EcoRI and HindIII.

Polymorphism: BclI identifies two alleles:

Allele B1 — 4.8 kb

Allele B2 — 3.9 kb

Constant bands at 6.2, 3.6 and 2.1 kb.

Frequencies: In 70 unrelated Caucasian individuals:

Allele B1 (4.8 kb): 0.26

Allele B2 (3.9 kb): 0.74

Not Polymorphic For: BglII, PstI.

Chromosomal Localisation: The PLANH2 gene was localised to 18q21–23 by *in situ* hybridization (2).

Mendelian Inheritance: Mendelian inheritance was demonstrated in four two-generation families (11 meioses scored).

Probe Availability: pBTA447 can be obtained upon request from Biotech Australia Pty Ltd, PO Box 20, Roseville NSW, 2069, Australia.

Other Comments: PLANH2 cDNA contains an *Alu* repeat in the 3' untranslated region (1). A PLANH2 cDNA probe which does not contain this repeat can be excised by digestion of pBTA447 with EcoRI and DraI.

Acknowledgement: This work was supported by the Queensland Cancer Fund.

References: 1)Antalis *et al.* (1988) *Proc. Natl. Acad. Sci. USA* **85**, 985–989. 2) Ye *et al.* (1989) *J. Biol. Chem.* **264**, 5495–5502.

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Two MspI RFLPs at the D17S258 locus

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Source/Description: A 4.9 kb EcoRI fragment from cosmid 1516 was subcloned into the EcoRI site of pTZ19R (p1516-R4).

Polymorphism: p1516-R4 identifies a two allele polymorphism with MspI (A1: 3.3 kb, A2: 2.4 kb) with constant bands of 1.8 kb, 1.05 kb and 0.88 kb.

Frequency: (determined in the designated number (N) of unrelated Caucasians)

p1516-R4 (N = 14)

A1: 0.46

A2: 0.54

Not Polymorphic For: *RsaI*, *PvuII*, *PstI*, *TaqI*, *BglII*, *EcoRI* in a panel of 8 unrelated individuals.

Chromosomal Localization: c1516 was assigned to chromosome 17p11.2 using a chromosome 17-somatic cell hybrid panel (1). This localization is based on the absence of c1516 in hybrid DH110-D1 retaining a del(17)(p11.2p11.2).

Mendelian Inheritance: Codominant segregation of this RFLP has been observed in two French-Acadian kindreds (58 individuals).

Probe Availability: Contact ATCC.

Other Comments: Preassociation with human placental DNA is required prior to hybridization at normal stringency (65°C, 1 M Na⁺) (1). A *HindIII* RFLP identified by c1516 has been previously described (1).

Acknowledgements: Supported by an MDA Task Force grant, a Baylor Mental Retardation Research Center (HD24064–02) grant, an Advanced Technology Program from the state of Texas and NIH grant RO1 NS-27042 to J.R.L. and P.I.P. BF is the recipient of an MDA postdoctoral fellowship.

References: 1) Patel, P.I. *et al.* (1990) *Am. J. Hum. Genet.* **46**, 801–809.

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