StuI RFLP in the human ABL locus

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Source/Description: A 3.6 kb EcoRI fragment (lambda ABL3) containing portions of exons 7 and 8 and the intervening intron of the ABL oncogene was isolated from a chromosome 9-specific library (ATCC # LA09NS01) using a 1.7 kb Abelson murine leukemia virus probe (v-abl, ONCOR, Inc.). A single copy 1.8 kb BglII fragment (ABL3) was derived from the original lambda clone and detects the StuI RFLP.

Polymorphism: The 1.8 kb BglII probe detects polymorphic fragments of 7.2 kb and 4.2 kb in StuI digested genomic DNA.

Allele Frequencies: The allele frequencies were estimated from 26 unrelated caucasians.
Allele #1 7.4 kb .06
Allele #2 4.0 kb .94

Enzymes not detecting Polymorphisms: The following enzymes were found not to detect polymorphisms using at least 10 unrelated individuals: MspI, RsaI, TaqI.

Chromosomal Location: The ABL oncogene has been mapped to chromosome 9q34 (1).

Mendelian Inheritance: Codominant inheritance.

Availability: The plasmid pABL3 is available from Dr M.Smith's laboratory at U.C. Irvine.

Comments: The 1.8 kb BglII probe fragment has been subcloned in pUC19 at the HincII site to create plasmid pABL3. A double digest with EcoRI and HindIII will excise the 1.8 kb probe intact. No probe prehybridization is necessary. Precise location of the probe fragment within the ABL oncogene was aided by published restriction data of the ABL locus (2).

References:1. Jhanwar,S.C. *et al* (1984) *Cytogenetics and Cell Genetics* **38**, 73–75. (2) Shtivelman,E. *et al* (1985) *Nature* **315**, 550–554.

An additional HindIII polymorphism at the coagulation factor XIIIA locus

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Source and Description of Clone: A 543 bp cDNA insert was isolated from a human placenta library constructed in EcoRI-cleaved λgt10 (1). It covers from the aminoacid 361 to the aminoacid 541 of the human coagulation factor XIIIa (FXIIIA) subunit and it is adjacent to a 1161 bp cDNA segment that already detects several RFLPs (2).

Polymorphism Described: HindIII identifies a 2 allele polymorphism (H1: 5.4 kb; H2: 3.7 kb) and three invariant bands of 5.7, 2.5 and 2.3 kb.

Frequency: estimated from 28 unrelated Caucasians.
H1: 0.14 ± 0.05
H2: 0.86

Not Polymorphic for: AvaII, BclI, BglI, BglII, MspI, PstI, and TaqI with DNA from 13, 7, 13, 14, 7, 12 and 7 unrelated Caucasian individuals, respectively.

Chromosomal Localization: to bands p24–p25 on chromosome 6 (3).

Mendelian Inheritance: Co-dominant segregation has been observed in 3 two-generation families.

Probe Availability: Contact Dr U.Grundmann, Berhingwerke, AG, Marburg.

Other Comments: This RFLP was observed under wash stringency of 0.3× SSC at 65°C.

References: (1) U.Grundmann *et al.* (1986) *Proc. Natl. Acad. Sci. USA* **83**, 8024–8028. (2) P.G.Board *et al* (1988) *Am. J. Hum. Genet.* **42**, 712–717. (3) P.G.Board *et al.* (1988) *Cytogenet. Cell Genet.* **48**, 25–27.