

## PstI polymorphism at the human P<sub>1</sub>450 gene on chromosome 15

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**Source/Description:** phP<sub>1</sub>450-3', a 1.0 kb EcoRI fragment in pBR322 (Jaiswal *et al.*, 1985).

**Polymorphism:** The probe identifies a two allele polymorphism with bands at either 9.0 kb (A1) or 7.6 kb (A2).

**Frequency:** 20 unrelated healthy Caucasians (North America) and 48 unrelated healthy Norwegians were screened. North Americans: A1: 0.17, A2: 0.83, Norwegians: A1: 0.38, A2: 0.62.

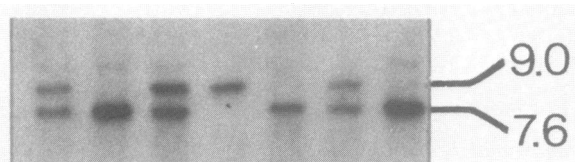
**Chromosomal Localisation:** 15q22 (Hildebrand *et al.*, 1985).

**Mendelian Inheritance:** Codominant segregation was shown in two families.

**Probe Availability:** Contact Dr Nebert.

**Other Comments:** The PstI polymorphism is in strong linkage disequilibrium with the MspI polymorphism (Spurr *et al.*, 1987) described at the same locus ( $\chi^2 = 73.3$ ,  $r = 0.49$ ,  $p < 0.001$ ). We have found no correlation between the PstI polymorphism and lung cancer tested in 65 patients.

**References:** 1) Jaiswal *et al.* (1985) *Nucl. Acids Res.* **13**, 4503-4520. 2) Hildebrand *et al.* (1985) *Biochem. Biophys. Res. Commun.*, 396-406. 3) Spurr *et al.* (1987) *Nucl. Acids Res.* **15**, 5901.



## Two polymorphisms for the human elastin gene

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**Source/Description:** A 1.1 kb EcoRI/BamHI fragment of the 5' end of the elastin cDNA isolated from human fetal aorta cDNA library (1) and subcloned into PUC-8.

**Polymorphism:** HindIII identifies two invariant bands at 18 kb and 12 kb and a simple two allelic polymorphism with a band at either 8 kb (H1) or 5 kb (H2) (in the optimized electrophoresis also the variable 3 kb band can be visualised). EcoRI identifies two invariant bands at 10 kb and 7 kb and a simple two allelic polymorphism with a band at either 10 kb (E1), (comigrates with the constant band) or 3 kb (E2).

**Frequency:**

HindIII	
Studied in 30 unrelated Finnish individuals	
H1 0.8	
H2 0.2	
EcoRI	
Studied in 30 unrelated Finnish individuals	
E1 0.90	
E2 0.10	

**Not Polymorphic For:** BamHI, BglIII, EcoRV, MspI, PstI, PvuII, StuI and TaqI.

**Chromosomal Localization:** The elastin gene has been localised to 2q31-qter by in situ hybridisation (2) and with RFLP-linkage analyses (3).

**Mendelian Inheritance:** Co-dominant inheritance shown in 5 informative Finnish families.

**Probe Availability:** Contact Dr Joel Rosenbloom.

**Other Comments:** The location of these polymorphisms has been confirmed with 250 bp genomic subclone of exon 18 of the elastin gene. Due to the high frequency of Alu-sequences other genomic clones are not highly useful in RFLP-analysis.

**References:** 1) Z.Indik *et al.* (1987) *Proc. Natl. Acad. Sci. USA* **84**, 5680-5684. 2) B.S.Emanuel *et al.* (1985) *Am. J. Hum. Genet.* **37**, 873-882. 3) A.Palotie *et al.* (1989) abstract no. 2063 in *HGM10 Cytogenetic Cell Genet.* **51**, 1057.

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