An Eco R1 polymorphism of a human platelet factor 4 (PF4) gene

Cynthia Guzzo, Mark Weiner, Eric Rappaport, Paul LaRocco, Saul Surrey, Mortimer Poncz and Elias Schwartz

Departments of Pediatrics, Human Genetics and Dermatology, University of Pennsylvania School of Medicine, and Division of Hematology, The Children's Hospital of Philadelphia, Philadelphia, PA 19104, USA

SOURCE AND DESCRIPTION OF CLONE: Two partial length human cDNA clones (410 and 600 bp) isolated from a human erythroleukemia (HEL) cell cDNA library were used for detection of genomic DNA fragments. The probe for chromosomal location studies using *in situ* hybridization was a 1.4 kb Pst fragment isolated from a PF4 genomic clone, inserted into M13mp18, and linearized with Eco R1.

POLYMORPHISM: EcoR1 (G/AATTC) identifies an invariant band at 10.0 kb and a simple polymorphism with a band at 5.0 kb or 3.1 kb.

FREQUENCY: We studied 25 persons (21 Caucasians, 2 Asians, and 2 blacks);

5.0 kb allele: 0.44

3.1 kb allele: 0.56

NOT POLYMORPHIC FOR: Bgl I (3), Bgl II (3), Bcl I (3), Bal I (2), Xba I (8), Hind III (10), Bam HI (10), Taq (5), Sst I (3), Kpn I (2).

CHROMOSOMAL LOCATION: The probe has been localized to the region 4q11-q21 by *in situ* hybridization².

MENDELIAN INHERITANCE: Demonstrated in one family (see pedigree below). The father is homozygous for the 3.1 kb band, the mother homozygous for the 5.0 kb band, and the child is heterozygous.

PROBE AVAILABILITY: Available for collaborative studies. Contact M. Poncz.

REFERENCES: 1) Poncz M, *et al.*: Cloning and characterization of platelet factor 4 cDNA derived from a human erythroleukemia cell line, Blood (in press); 2) Griffin CA, *et al.*: Human platelet factor 4 gene is mapped to 4q11-q21, Cytogenetics and Cell Genetics (submitted).

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