A Taq I polymorphism for the human platelet glycoprotein IIIa gene (GP3A)

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SOURCE AND DESCRIPTION OF CLONE: A cDNA clone containing a 4.0 kb GPIIIa insert was isolated from a human erythroleukemia (HEL) cell cDNA library (1). A 2.3 kb Eco RI fragment, representing the 5' portion of this insert, was used as a probe for these Southern blot experiments.

POLYMORPHISM: Taq I (T/CGA) identifies invariant bands of 5.0, 3.5, 1.3 1.1, and 0.8 kb and a simple polymorphism with a band(s) at 8.0 kb or 4.8 and 3.2 kb.

FREQUENCY: We studied 17 persons (11 Caucasians, 3 Asians, and 3 blacks); 8.0 kb allele: 53%. 4.8 and 3.2 kb allele: 47%.

NOT POLYMORPHIC FOR: Bam HI (15), Bcl I (7), Eco RI I (15), Kpn I (13), Pst I (13), Sst I (13).

CHROMOSOMAL LOCATION: The GPIIIa gene has been localized to the region 17q21⇒22 by somatic cell hybrid and *in situ* hybridization studies (2).

MENDELIAN INHERITANCE: Demonstrated in one family. The father (I-2, below) is homozygous for the 8.0 kb band, and the mother (I-1) is heterozygous for the 8.0/4.8 and 3.2 kb bands. Three offsprings (II-2, II-3, and II-5) are homozygous for the 8.0 kb band, while two (II-1 II-4) are heterozygous for 8.0/4.8 and 3.2 kb bands.

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

REFERENCES: 1) Butler-Zimrin A, Eisman RL, Vilaire G, Schwartz E, Bennett JS, Poncz M: The structure of platelet glycoprotein IIIa. A common subunit for two different membrane receptors, (submitted); (2) Sosnoski DM, Emanuel BS, Hawkins AL, van Tuinen P, Nussbaum RL, Kao F-T, Schwartz E, Bennett JS, Phillips D, Fitzgerald LA, Poncz M: The chromosomal localization of the vitronectin and fibronectin receptors alpha subunits and of platelet glycoprotein (GP) IIb and IIIa demonstrate close proximity only for GPIIb and GPIIIa, (submitted).

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