An Xba I polymorphism 3' to the human erythropoietin (EPO) gene

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SOURCE AND DESCRIPTION OF CLONE: E49F¹ consists of a 1.4 kb EPO cDNA insert in the Eco RI site of pSP65. A 0.6 kb Bst EII-Stu I fragment which contains only 5'-untranslated and amino-acid coding sequences was isolated for use as a hybridization probe (BS probe).

POLYMORPHISM: After Xba I digestion and Southern blotting, the BS probe hybridizes to a 10 kb genomic DNA fragment when the polymorphic Xba I site is present and a 12 kb fragment when the site is absent (see figure). An invariant 1.3 kb fragment is also detected (not shown).

FREQUENCY: The frequency of the presence [f(+)] and absence [f(-)] of the polymorphic Xba I site were determined by analysis of DNA from 63 unrelated individuals.

Population	Chromosomes Examined	f(+)	f(-)
Black	38	0.16	0.84
Caucasian	88	0.23	0.77
Total	126	0.21	0.79

NOT POLYMORPHIC FOR: Bam HI (5 unrelated individuals tested).

CHROMOSOMAL LOCATION: Mapped to 7q11-q22, by analysis of somatic cell hybrids and *in situ* hybridization.²

MENDELIAN INHERITANCE: Demonstrated in 19 nuclear families.

PROBE AVAILABILITY: Contact Dr. C. Shoemaker, Genetics Institute, Cambridge, MA.

OTHER COMMENTS: The Xba I polymorphism is in linkage disequilibrium with a known Hind III polymorphism.² On all chromosomes analyzed (14 individuals), both sites were either present or absent. Double digests using Bam HI (B), Hind III (H) and Xba I (X) allowed mapping of sites in the figure (polymorphic sites are designated by arrows with closed circles).

REFERENCES: 1) Jacobs, K. *et al.* (1985) *Nature* **313**:806-810; 2) Law, M.L. *et al.* (1985) *Proc. Natl. Acad. Sci. USA* **83**:6920-6924.

ACKNOWLEDGEMENTS: Plasmid E49F was the generous gift of Dr. Charles Shoemaker. G.L.S. was supported by National Institutes of Health grant 2T32-GM07471.