

BglII polymorphism at the human erythropoietin gene

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Source and Description: A monkey erythropoietin (epo) cDNA probe containing sequences from amino acid residue 16 of the mature protein to near the 3' end of the mRNA was used (1).

Polymorphism: BglII detects a two allele polymorphism with bands at 12 kb (A1) and 8 kb (A2) and an invariant band at 4.5 kb.

Frequency: Estimated in 30 unrelated individuals. A1, 0.85; A2, 0.15.

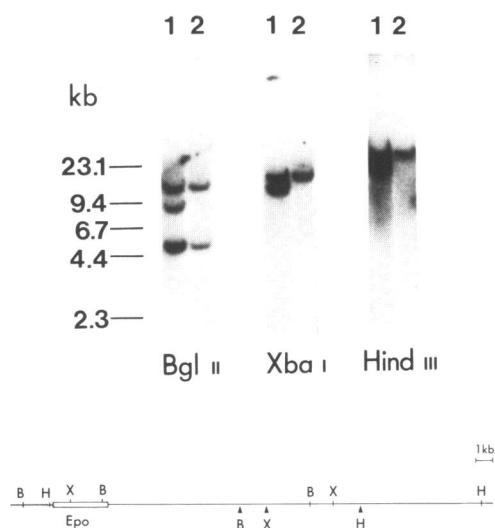
Chromosomal Localization: 7q21 (2).

Probe Availability: Contact F-K Lin (1).

Other Comments: In all individual samples tested the BglII (B) polymorphism and two previously described (2, 3) XbaI (X) and HindIII (H) polymorphisms were either all present or all absent. The positions of the restriction sites in the two alleles (polymorphic sites are indicated by the closed triangles) are suggestive of an approximately 4 kb insertion in A1 or deletion event in A2 3' to the epo gene and 5' to the polymorphic BglII site.

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References: 1) McDonald, J.D. *et al.* (1986) *Mol. Cell. Biol.* **6**, 842–848. 2) Law, M.L. *et al.* (1985) *Proc. Natl. Acad. Sci. USA* **83**, 6920–6924. 3) Semenza, G.L. *et al.* (1987) *Nucl. Acids Res.* **15**, 6768.



SacI identifies an additional RFLP at the D11S12 locus

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Source/Description: The 5.5 kb insert of plasmid pADJ762 was used as a hybridization probe (1). The same polymorphism is also detected by a 0.9 kb EcoRI/SacI subfragment of the above mentioned clone.

Polymorphism: SacI digestion identifies a biallelic RFLP with bands of 1.8 (B1) and 1.7 (B2) kb. In addition, the 5.5 kb probe recognizes two constant fragments of 7.6 and 2.0 kb.

Frequency: Analysed in 45 unrelated Italian Caucasians.

Allele	Frequency
B1	0.64
B2	0.36

Not Polymorphic For: AvaII, BamHI, BglII, EcoRI, HincII, HindIII, PstI, PvuII, RsaI, XbaI (each enzyme tested on at least eight unrelated individuals).

Chromosomal Localization: D11S12 was assigned to chromosome region 11p15.5 (2).

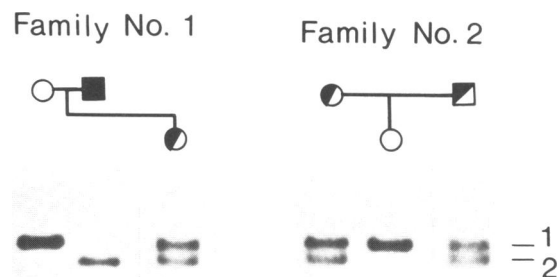
Mendelian Inheritance: Co-dominant segregation was observed in three two-generation families.

Probe Availability: Available through ATCC.

Other Comments: pADJ762 was previously reported to detect RFLPs for BclI, MspI and TaqI (1).

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References: 1) Barker, D. *et al.* (1984) *Am. J. Hum. Genet.* **36**, 1159–1171. 2) Mannens, M. *et al.* (1987) *Hum. Genet.* **75**, 180–187.



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