EcoRI polymorphism in the human third complement component (C3) gene

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Source/Description: A cDNA fragment, pC3.11 (1) was used as probe.

Polymorphisms: *EcoRI* cleavage of genomic DNA reveals a 2 allele polymorphism with band sizes of 5.2 and 5.6 kb. Invariant bands of 11.5, 11.0, 9.0 and 3.0 kb were also present.

Frequency: Estimated from 35 unrelated Caucasians.

	Allele	Frequency
D1	5.2	0.67
D2	5.6	0.33

Chromosomal Localisation: The human C3 gene had been assigned to chromocome 19 p13.3-p13.2 (2).

Mendelian Inheritance: Co-dominant inheritance was documented in one family with 8 individuals.

Probe Availability: Contact Dr. G.Fey (Scripps Clinic, La Jolla, CA, USA).

Acknowledgement: We thank Dr.G.Fey for providing the cDNA probe, pC3.11.

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MspI and DraI polymorphisms at the ERBA beta locus on chromosome 3p

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Source/Description: The plasmid pBH302 carries a 2.0 kb BamHI insert in pGem 1. This fragment was subcloned from Lambda PGE, which was isolated by screening a human genomic bank with ERBA beta sequence pheA4. The 2 kb fragment carries a short exon in the 5′ untranslated region of ERBA beta which is identical to the published sequence (1).

Polymorphism: A HindIII polymorphism with two allelic fragments 7.0 kb (A1) and 5.5 kb (A2) was described (2). MspI digestion of genomic DNA and hybridization with the probe detects two alleles with fragments of 3.5 kb (B1) and 2.0 kb + 1.0 kb (B2). DraI digestion of genomic DNA and hybridization with the probe detects two alleles with fragments of 2.0 kb (C1) and 1.8 kb (C2).

Frequency: 90 unrelated Caucasians were studied for the MspI polymorphism. 3.5 kb allele (B1) 0.48, 2.0 kb + 1.0 kb allele (B2) 0.5265 unrelated Caucasians were studied for the DraI polymorphism. 2.0 kb allele (C1) 0.46, 1.8 kb allele (C2) 0.54.

Not Polymorphic For: EcoRI, TaqI, PvuII, BamHI, BglII, RsaI, HinfI.

Chromosomal Localization: ERBA beta has been mapped to two regions on chromosome 3. To 3p21.33–3p22 by Drabkin *et al.* (3) and to 3p24.2–3p24.3 (Albertson *et al.*) (4).

Mendelian Inheritance: Co-dominant segregation has been demonstrated in two three generation families with 14 individuals and 27 individuals for the MspI polymorphism, and in one three generation family with 14 individuals for the DraI polymorphism.

Linkage Disequilibrium: There is evidence for linkage disequilibrium between the MspI and DraI polymorphisms (−42, $p < 0.005$) (5).

Probe Availability: Available for collaboration. Contact W.E.C.Bradley.

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