

A dinucleotide repeat polymorphism in the gene for the γ subunit of the human Fc ϵ receptors (FLER16)

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Source/Description: A (CA)₇(TA)(CA)₁₅(TA)₇(GA)₁₆ repeat was identified in the first intron of the gene for the human Fc ϵ receptor γ subunit at position 1575 (EMBL accession no. J05285). This repeat is located at 1039 in Figure 1 of (1), where +1 corresponds to the beginning of the start codon ATG. Primers that uniquely amplified this repeat were designed for the polymerase chain reaction. The predicted length was 153 bp.

Primer Sequence:

GAT-TTT-ACA-TAT-ATA-TCT-CTA-TC (CA strand);
CAG-AGT-CC-T-ACC-TTC-CTA-A (TG strand).

Frequency: Estimated from 51 parents of CEPH families. The calculated heterozygosity was 0.70.

Allele	bp	Frequency	Allele	bp	Frequency
A1	165	0.020	A6	155	0.137
A2	163	0.010	A7	153	0.128
A3	161	0.088	A8	151	0.500
A4	159	0.029	A9	149	0.049
A5	157	0.029	A10	143	0.010

Chromosomal Localization: The gene for the γ subunit of the Fc ϵ receptor has been assigned to chromosome 1q23 by *in situ* hybridization (2).

Mendelian Inheritance: Co-dominant segregation was observed in 3 informative CEPH families.

PCR Conditions: Polymerase chain reactions were carried out in a 12.5 μ l volume containing 100 ng genomic DNA, 200 μ M each dGTP, dCTP and dATP; 12.5 pmole of one unlabelled primer; 12.5 pmoles of a gamma ³²P-ATP end-labelled primer (0.75uCi) and 0.625 units Taq polymerase for 30 cycles as follows: 94°C for 1 min, 53°C for 1 min, and 72°C for 1 min. The reaction products were analyzed on a 6% denaturing polyacrylamide gel and visualized by autoradiography.

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Dinucleotide repeat polymorphism at the human erythropoietin receptor locus (EPOR) at 19p13

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Source/Description: A compound tetranucleotide and dinucleotide repeat (GGAA)₁₄-AG-(GA)₂₂ is present in the promoter region of the human erythropoietin receptor gene, beginning at base -563 from the start codon ((1) GenBank accession no. M77244).

Primer Sequences:

5' GCCTGGGTGACAGAGCAACAC 3' (GA strand)
5' CCTGGCACATAGCGAACATTC 3' (TC strand).

Allele Frequency: Estimated from 160 chromosomes of unrelated CEPH parents. Observed heterozygosity = 87%

Allele	Size (bp)	Frequency	Allele	Size (bp)	Frequency
A1	268	0.01	A9	250	0.04
A2	264	0.02	A10	248	0.23
A3	262	0.01	A11	246	0.08
A4	260	0.08	A12	244	0.13
A5	258	0.01	A13	242	0.01
A6	256	0.06	A14	240	0.09
A7	254	0.01	A15	236	0.04
A8	252	0.18			

Alleles found in four reference CEPH parents are: 133101 = A6, A14; 133102 = A6, A10; 134501 = A8, A10; 134502 = A6, A8.

Chromosome Localization: The EPOR locus has been assigned to 19p13 by *in situ* hybridization of the EPOR cDNA to metaphase chromosomes (2).

Mendelian Inheritance: Co-dominant inheritance was observed in one two generation family (17 individuals) and one three-generation family (40 individuals).

PCR Conditions: The PCR reactions are carried out using the Perkin-Elmer 9600 in 10 μ l volumes containing 10 ng of DNA, 4 ng of each primer and 0.33 units of Taq polymerase (Boehringer Mannheim). The buffer contains 10 mM Tris-HCl (pH 8.3), 1.5 mM MgCl₂, 50 mM KCl, 0.01% gelatin, and 200 μ M each dNTP. Amplification is 94°C for 5 min, followed by thirty cycles of 94°C, 30 sec; 60°C, 1 min; 72°C, 30 sec; with a final extension at 72°C for 5 min. The TC primer shows less background than the GA primer when kinased. The PCR products are fractionated on 4% denaturing polyacrylamide gels.

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