An insertion deletion polymorphism associated with C-FES

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Source/Description: C-FES genomic DNA was used as a probe to isolate a homologous clone (CFES/S117) from a human genomic DNA cosmid library. The genomic DNA, inserted in the BamHI site of vector c2RB, contains 42 kb human genomic DNA which can be excised with EcoRI.

Polymorphism: Insertion-deletion type polymorphism reveals polymorphism with BamHI, BglII, and MspI. Complete linkage disequilibrium between the polymorphic systems. With BglII there are 5 constant fragments of 13, 8.5, 7.8, 2.3, and < 1.

Enzymes Tested But Not Polymorphic: EcoRI, HincII, HindIII, PstI, RsaI and TaqI.

Frequency: Studied in 80 Caucasians (BglII).

Allele D1: 6.5 kb 0.28

Allele D2: 6.2 kb 0.78

Heterozygosity/PIC: 0.44/0.36.

Mendelian Inheritance: Autosomal inheritance confirmed in 3-generation CEPH families.

Chromosome Localization: Maps to 15q (lods > 6.0) consistent with 15q25–15qter localization of C-FES (1).

Probe Availability: Freely available.

Other Comments: Probe contains repetitive DNA and requires hybridization with excess unlabeled human DNA (2).

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KpnI RFLP in the human histidine-rich glycoprotein gene

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Source and Description: Probe BCH contains a 2.1 kb cDNA copy of the human HRG gene inserted in pGEM-I (1).

Polymorphism: After KpnI digestion of genomic DNA the probe detects two allelic fragments of 19.3 kb and 16.8 kb.

Frequency: Estimated from 24 unrelated Caucasians:

19.3 kb allele (A1) 0.61

16.8 kb allele (A2) 0.39

Not Polymorphic For: AluI, BclI, DraI, EcoRV, HaeIII, HincII, MboII, NdeI, NsiI, RsaI, SacI, SspI, StyI, XbaI, XmnI and see (2).

Chromosomal Localisation: Chromosome 3 (2).

Mendelian Inheritance: Co-dominant segregation demonstrated in 4 families (25 informative meioses).

Probe Availability: The cDNA of HRG was a generous donation from Dr. T. Koide, Department of Biochemistry, Niigata University School of Medicine, Niigata 951, Japan.

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