EcoRI polymorphism within the GLI gene (chromosome 12q13.3-14.1)

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Source/Description: pKK36P1 contains a 1.55 kb PstI genomic fragment of GLI cloned into the PstI site of pGEM-3 (1).

Polymorphism: EcoRI identifies a two allele polymorphism (A1: 18 kb; A2: 5 kb).

Frequency: Estimated from 91 individuals.
A1 18 kb (0.72)
A2 5 kb (0.28)

Chromosomal Localisation: Assigned to chromosome 12q13.3-14.1 using a somatic cell hybrid panel and in situ hybridization (1, 2).

Probe Availability: Contact B.Vogelstein.


EcoN I polymorphism in the human cholesteryl ester transfer protein (CEPT) gene

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Source and Description of Clone: cDNA clone containing the coding sequence from amino acid number 9 of mature CETP protein spanning the complete coding sequence to the 3’ untranslated region (1, 2).

Polymorphisms: EcoN I digestion identifies a two allele polymorphism with bands of 9.0 kb and 8.0 kb. In addition, constant bands of 6.0 kb and smaller are noted.

Frequency: Estimated in 8 unrelated Caucasians.
9.0 kb allele: 0.56
8.0 kb allele: 0.44
The heterozygosity index was 49%.


Chromosomal Localization: The CETP gene has been assigned to chromosome 16 q12–16 q21 (3).

Mendelian Inheritance: Codominant segregation of the alleles was documented in one informative family with nine meioses (data not shown).

Probe Availability: The probe was obtained from D.Drayna, Genentech Inc., San Francisco, CA.

Acknowledgement: We thank Tommy F.Hyatt for providing excellent technical assistance.


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