Dinucleotide insertion/deletion polymorphism in intron 50 of the COL2A1 gene

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Source/Description: Amplification by the polymerase chain reaction (PCR) was carried out using oligonucleotides 5'-CCTGCAGGAGACTACTGGATTGACC-3' and 5'-AAGGTCATCTGGACGTTGGCAGTGT-3'. The amplified product was analyzed by electrophoresis through a 6% polyacrylamide gel.

Polymorphism: The two alleles differ by the presence or absence of the dinucleotide CT (nucleotides 3399–3400, ref. 1) in intron 50: 5'...TCCTCAA[CT]CTTTCTTT...3'

Frequency: Estimated from 70 chromosomes of unrelated CEPH parents (Caucasians).

Allele (bp)	Frequency
L1 594	.91
L2 592	.09

Chromosomal Localization: 12q13.1–13.2 (ref. 2).

Mendelian Inheritance: Co-dominant segregation was observed in one 3-generation family and two 2-generation families.

Other Comments: The PCR conditions were 40 cycles of 94°C for 1 minute, 55°C for 1 minute, and 72°C for 1.5 minutes. Heterozygous individuals demonstrate a third band upon electrophoresis of amplified DNA, which consists of heteroduplexes.

Acknowledgements: This work was supported in part by grants from the Human Growth Foundation, the NIH (HD22657), and an Arthritis Foundation Postdoctoral Fellowship to G.E.T.

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A common polymorphism in the human arylsulfatase B (ARSB) gene at 5q13-q14

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Source/Description: Sequencing of full-length cDNAs encoding human ARSB (1) revealed a G or A at nucleotide position 1072 which predicted a Val or Met at codon 358.

Polymorphism Frequency: The frequencies of the (A1) 358-Val and (A2) 358-Met alleles in PCR-amplified genomic DNA from 110 unrelated Caucasian individuals by dot-blot hybridization were 0.62 and 0.38, respectively.

Protocol: To detect the nucleotide 1072 polymorphism, a 121 bp ARSB genomic fragment is amplified using sense (5'-GTG-GGCTTTGTGGCAAG-3') and antisense (5'-TTGTGCCATT-GGTGTGT-3') primers. Dot-blot hybridization is performed using allele-specific oligonucleotides for (A1) 358-Val (5'-CAA-CACTCGTGAAGCTG-3') and for (A2) 358-Met (5'-CAACA-CTCATGAAGCTG-3'), which are hybridized at 42°C and washed at 51 or 53°C, respectively.

Chromosomal Location: The human ARSB gene has been localized to the chromosomal region 5q13-q14 (2).

Other Comments: The deficient activity of ARSB results in mucopolysaccharidosis type VI (Maroteaux-Lamy disease). This polymorphism may serve as a useful marker for identification of mutant alleles in patients with mucopolysaccharidosis type VI, as a marker for linkage analysis of other disease causing genes localized to this region, and for the orientation of other polymorphic variants and/or sequence tagged sites in the chromosomal region 5p11-q13.

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Acknowledgements: This work was supported in part by a grant (5 RO1 DK 25759) from the National Institutes of Health and by a General Research Center grant (RR-00071) from the National Institutes of Health.

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