
A three allele restriction fragment length polymorphism within the human *Col2A1* gene

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SOURCE/DESCRIPTION: Human *COL2A1* clone p β HColIIf, corresponds to the 7.3kb *EcoRI* fragment contained in plasmid pIs10 of Sangiorgi et al. (1,2).

POLYMORPHISM: Three common alleles, detected by *HinfI*, are probably caused by variable number tandem repeats. Invariant bands are observed at 420bp, 780bp, 1,100bp and 1350bp. Some individuals have no additional bands (null allele). The other alleles are at 1.9kbp (B allele) and 2.3kbp (A allele).

FREQUENCY: Genotype frequencies observed in 49 Caucasian individuals. A/A = 0.31; A/Null = 0.10; B/B = 0.08; B/Null = .10; A/B = .33; Null/Null = .88. Allele frequencies for 98 alleles: Null = 0.18; A = 0.52; B = 0.30.

NOT POLYMORPHIC FOR: *BamHI*, *EcoRI*, *PvuII* and *TaqI*,

CHROMOSOMAL LOCALISATION: Located in *Col2A1* coding region on chromosome 12q131-12q132 (2).

MENDELIAN INHERITANCE: Demonstrated in one extended family.

PROBE AVAILABILITY: Freely available.

REFERENCES:

1. Strom CM and Upholt WB. *Nucleic Acids Res.* 12:1025-1038, 1984.
2. Sangiorgi et al., *Nucleic Acids Res.* 13:2208-2225, 1985.

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