
An RFLP close to the human collagen I gene COL1A1

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SOURCE/DESCRIPTION: NST70 a 1.4kb EcoRI fragment subcloned into Bluescript M13 phagemid from a human genomic cosmid library.**POLYMORPHISM:** Rsa I identifies one invariant band at 3.0kb and a dimorphism with a band at either 2.4kb or 2.1kb.**FREQUENCY:** Studied 54 unrelated European Caucasians.2.4kb allele (C2) 0.82 \pm 0.042.1kb allele (C1) 0.18 \pm 0.04**NOT POLYMORPHIC FOR:** Bam HI, Hind III.**CHROMOSOMAL LOCALISATION:** c. 10kb 3' of COL1A1, previously mapped to 17q 21.31 - 2205 (1).**MENDELIAN INHERITANCE:** Codominant segregation observed in 10 pedigrees.**PROBE AVAILABILITY:** Now available for collaboration. Contact B.C. Sykes.**OTHER COMMENTS:** Not to be confused with other Rsa I RFLP in COL1A1 (2). Increases PIC at COL1A1 from 0.43 to 0.64. Provides an additional marker for some forms of osteogenesis imperfecta.**REFERENCE:** 1. E. Retief et al. Hum. Genet. (1985) 69, 304-308.
2. B. Sykes et al. Lancet (1986) *ii*, 69-72.**ACKNOWLEDGEMENTS:** We thank the MRC for support.