A PvuII polymorphism near the 5' end of the type II procollagen gene [COL2A1]

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SOURCE AND DESCRIPTION: The probe WV38 is a 4.0 kb EcoRI/BamHI fragment from the 5' end of the human pro $\alpha 1$ (II) collagen gene (1).

POLYMORPHISM: Allelic PvuII fragments of 1.8 kb (A1) and 0.95 kb/0.85 kb (A2) are observed. (See figure below).

FREQUENCY: A1: 0.21 (50 unrelated Caucasians)
 A2: 0.79

NOT POLYMORPHIC FOR: BamHI, BglI, EcoRI, HindIII, HinfI, MspI, PstI, Styl, or TaqI (in 6 unrelated individuals).

CHROMOSOMAL LOCALIZATION: Assigned to 12q14.3 (2, 3).

MENDELIAN INHERITANCE: Co-dominant segregation observed in three three-generation families and three two-generation families.

PROBE AVAILABILITY: Available.

REFERENCES:

1. F.O. Sangiorgi et al. (1985) Nucl. Acids Res. **13**:2207-2225.
2. C. Huerre-Jeanpierre et al. (1986) Am. J. Hum. Genet. **38**:26-37.
3. M.L. Law et al. (1986) Ann. Hum. Genet. **50**:131-137.

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