
A BglI polymorphism in the type VI ($\alpha 3$) procollagen gene [COL6A3]

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SOURCE AND DESCRIPTION OF CLONE: The probe P24 is a 1.5 kb cDNA cloned in the EcoRI site of pUC19 (1).

POLYMORPHISM: Allelic BglI fragments of 6.7 kb (A1) and 6.0 kb (A2) are observed.

FREQUENCY A1: 0.61 (30 unrelated Caucasians)
 A2: 0.39

NOT POLYMORPHIC FOR: BamHI, EcoRI, EcoRV, HindIII, PstI, PvuII, RsaI, StyI or XbaI (in 6 unrelated individuals).

CHROMOSOMAL LOCALIZATION: Assigned to 2q37 by somatic cell hybrid analysis and in situ hybridization (2).

MENDELIAN INHERITANCE: Co-dominant segregation observed in one three-generation family. (See figure below.)

PROBE AVAILABILITY: Contact Dr. Mon-Li Chu.

REFERENCES

1. M.-L. Chu et al. (1987) Eur. J. Biochem. 168:309-317.
2. D. Weil et al. (1988) Am. J. Hum. Genet. 42:435-445.

ACKNOWLEDGEMENT: Supported by NIH grants AR38923 and AR38912.