

---

**An RFLP in the gene for the human pro-alpha 2 chain of Type V collagen (COL5A2)**

---

Robin C.Schwartz, Alexandra Liddell, Francesco Ramirez<sup>1</sup> and Petros Tspouras

---

University of Connecticut Health Center, Department of Pediatrics, Farmington, CT and <sup>1</sup>SUNY Health Science Center at Brooklyn, Department of Microbiology and Immunology, Brooklyn, NY, USA

---

**SOURCE/DESCRIPTION:** DMC-2, a 3.5 kb genomic DNA clone of the COL5A2 gene inserted into pBR322. This Hind III fragment contains the last exon and the 3' flanking sequence of the COL5A2 gene.

**POLYMORPHISM:** MspI identified a simple two allele polymorphism with a band at either 13kb or 9kb.

**FREQUENCY:** 74 unrelated individuals from the CEPH (Dausset, J.) panel were studied.

13kb allele (A1): 0.149 ± 0.06

9kb allele (A2): 0.851 ± 0.06

**NOT POLYMORPHIC FOR:** EcoRI, HindIII, BamHI, TaqI, PstI, SstI, PvuII.

**CHROMOSOMAL LOCALISATION:** 2q24.3→q31 (Emanuel. et. al. 1985)

**MENDELIAN INHERITANCE:** Autosomal dominant inheritance was demonstrated in 4 kindreds with a total of 50 individuals.

**PROBE AVAILABILITY:** No restrictions. Contact F. Ramirez.

**OTHER COMMENTS:**

**REFERENCE:** 1. Dausset, J. Presse Med (1986) 15, 1801-1802.  
2. Emanuel, B.S., et. al. Am J. Hum Genet (1985) 37, 873-882.

**ACKNOWLEDGEMENTS:** This study was supported in part by a grants to P.T. #AR- 1221 and to F.R. #AR- 38648 from the National Institutes of Health.