A glycine to serine polymorphism in the C-propeptide of the human type II procollagen

H.Vissing*, M.Rasmussen, M.D'Alessio, B.Lee, C.Dobkin¹ and F.Ramirez
Brookdale Center for Molecular Biology, Mt. Sinai Center Medical Center, New York, NY 10029 and ¹Institute for Basic Research, NYC, NY, USA

Source/Description: Amplification of exon 51 of the type 2 procollagen gene was carried out by the polymerase chain reaction (PCR) using oligonucleotide 5'-TTGAATGCCAGCTCCCA-GGCCT-3' as the 5' primer and 5'-CGACGGACACGGTTTG-ACGGA-3' as the 3' primer. The amplified product was subsequently digested with PvuII and analysed on a 3% Nusieve/1% SeaKem agarose gel.

Polymorphism: The polymorphic alleles differ by a single base A to G change in exon 51 and results in a glycine to serine change at amino acid 191 in the C-propeptide of type II procollagen (1). The base change A to G can be typed by the presence of a PvuII site.

Frequency: Studied in 26 unrelated caucasians.

Glycine^(PvuII+) — 0.84

Serine^(PvuII-) — 0.16

Chromosomal Localisation: 12q14.3 (2).

Mendelian Inheritance: Codominant segregation was demonstrated in one three generation family.

Other Comments: The PCR was carried out by denaturing at 94°C for 1.5 minutes, annealing at 55°C for 2 minutes and extending at 72°C for 2 minutes. The PCR product is a 338bp fragment which in the presence of the glycine codon can be cleaved by PvuII into two fragments of 184bp and 154bp.

Acknowledgements: This work was supported from grants from the National Institute of Health (AR-38648 and HD-22657) and the March of Dimes Birth Defects Foundation (1-1042).

References: 1) Vissing, H. *et al.* (1989) *J. Biol. Chem.* **264**, 18265-18267. 2) Law, M.L. *et al.* (1986) *Ann. Hum. Genet.* **50**, 131-137.

* To whom correspondence should be addressed

Dinucleotide repeat polymorphism at the D18S34 locus

James L.Weber* and Paula E.May
Marshfield Medical Research Foundation, 510 North St. Joseph Avenue, Marshfield, WI 54449, USA

Source/Description: A human genomic *AluI* fragment was cloned into mp10 and selected by hybridization to poly(dC-dA)·poly(dG-dT). The cloned fragment was designated Mfd26. Sequencing of Mfd26 provided the information necessary for polymerase chain reaction primer synthesis. The clone length was 223 bp, and the predicted length of the amplified fragment was 115 bp.

Primer Sequences: CAGAAAATTCTCTCTGGCTA (CA strand); CTCATGTTCTGGCAAGAAT (GT strand).

Frequency: Estimated from 108 chromosomes of unrelated CEPH family grandparents (Caucasians). PIC = 0.78.

Allele(bp)	Frequency	Allele(bp)	Frequency
119	0.01	109	0.20
117	0.03	107	0.06
115	0.07	105	0.11
113	0.28	103	0.01
111	0.22		

Chromosomal Localization: Assigned to chromosome 18 using DNA templates isolated from panels of somatic cell hybrids.

Mendelian Inheritance: Co-dominant segregation was observed in 15 two generation families.

Other Comments: Conditions for the amplification reactions were as described in the reference except that samples were processed through 27 temperature cycles consisting of 1 min at 94°, 2 min at 55° and 2 min at 72°. Sizes of the alleles were determined by comparison to mp8 DNA sequencing ladders and were the averages of the sizes of the GT-strand and CA-strand bands. The dinucleotide repeat sequence in Mfd26 was of the form (AC)₂₈A. The sequence of Mfd26 has been submitted to GenBank.

Acknowledgements: This work was supported by the Marshfield Clinic and NIH grant GM41773.

References: Weber, J.L. and May, P.E. (1989) *Am. J. Hum. Genet.* **44**, 388-396.

* To whom correspondence should be addressed