Dinucleotide repeat polymorphism at the PENK locus

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Source/Description: A $(dC\text{-}dA)_n \cdot (dG\text{-}dT)_n$ sequence was found within an intron of the human proenkephalin gene by computer search of GenBank (Accession number K00489). Polymerase chain reaction primers were selected from the sequence to give an amplified fragment with predicted length of 79 bp. The pair of primers was designated Mfd31.

Primer sequences: TAATAAAGGAGCCAGCTATG (CA strand); ACATCTGATGTAAATGCAAGT (GT strand).

Frequency: Estimated from 106 chromosomes of unrelated CEPH family grandparents (Caucasians). PIC = 0.43.

Allele (bp)	Frequency	Allele (bp)	Frequency
83	0.01	77	0.02
81	0.42	75	0.01
79	0.54		

Chromosomal Localization: Litt *et al.* mapped PENK to chromosome 8. This assignment was confirmed 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 first 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 at PENK was of the form $(AC)_{13}A$.

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References: 1) Weber,J.L. and May,P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396. 2)Litt,M., Buroker,N.E., Kondoleon,S., Douglass,J., Liston,D., Sheehy,R. and Magenis,R.E. (1988) *Am. J. Hum. Genet.* **42**, 327–334.

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Dinucleotide repeat polymorphism at the D2S72 locus

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Source/Description: A human genomic *Alu*I fragment was cloned into mp10 and selected by hybridization to poly(dC-dA)·poly(dG-dT). The cloned fragment was designated Mfd36. Sequencing of Mfd36 provided the information necessary for polymerase chain reaction primer synthesis. The clone length was 238 bp, and the predicted length of the amplified fragment was 165 bp.

Primer sequences: AGCTATAATTGCATCATTGCA (CA strand); TGGTCTATAACTGGTCTATG (GT strand).

Frequency: Estimated from 120 chromosomes of unrelated CEPH family grandparents (Caucasians). PIC = 0.71.

Allele (bp)	Frequency	Allele (bp)	Frequency
173	0.02	165	0.20
171	0.14	163	0.30
169	0.02	161	0.01
167	0.31	159	0.01

Chromosomal Localization: Assigned to chromosome 2 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 Mfd36 was of the form $(AC)_{15}AT(AC)_6A$. The sequence of Mfd36 has been submitted to GenBank.

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

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