Dinucleotide repeat polymorphism at the human cardiac beta-myosin gene

Mihael H.Polymeropoulos, Hong Xiao, Denise S.Rath and Carl R.Merril

National Institute of Mental Health Neuroscience Center, St. Elizabeths Hospital, Room 131, 2700 Martin Luther King Avenue, Washington, DC 20032, USA

Source/Description: The polymorphic (GT)_n repeat begins at base pair 1526 of the human cardiac beta-myosin gene (HMSYHCO1) on chromosome 14 (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as described previously (2). The predicted length of the amplified sequence was 91 bp.

Primer sequences:

CTGCATCTGAGCATATGGGA (GT strand); CATTGAGATATGCAGGCTT (AC strand).

Frequency: Estimated from 48 chromosomes of unrelated individuals.

Heterozygosity Index = 66%. PIC = 0.60.

Allele (bp)	Frequency	Allele (bp)	Frequency
B1 102	0.02	B4 94	0.42
B2 98	0.10	B5 92	0.06
B3 96	0.38	B6 90	0.02

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

Chromosomal Localization: HMSYHCO1 has been assigned to chromosome 14q11.2-q13 (1).

Other Comments: The PCR reaction was performed on 80 ng of genomic DNA using 100 pmoles of each oligonucleotide primer. The samples were processed as described (3) except that the denaturation cycle at 94°C was extended to 1.4 minutes. The dinucleotide repeat was based on a (GT)₁₅ sequence.

References: 1) Cox,D.W. *et al.* (1989) *Cytogenetics and Cell Genetics* **51**, 980. 2) Weber,J.L. and May,P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396. 3) Weber,J.L. *et al.* (1990) *Nucl. Acids Res.* **18**, 4637.

Dinucleotide repeat polymorphism at the human ATP synthase beta subunit gene (ATPSB)

Mihael H.Polymeropoulos, Denise S.Rath, Hong Xiao and Carl R.Merril

National Institute of Mental Health Neuroscience Center, St. Elizabeths Hospital, Room 131, 2700 Martin Luther King Avenue, Washington, DC 20032, USA

Source/Description: The polymorphic (GT)_n repeat begins at base pair 345 of the human ATP synthase beta subunit (ATPSB) gene (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as described previously (2). The predicted length of the amplified sequence was 111 bp.

Primer sequences:

CTGGGACTACTGGCACATG (GT strand); GGCAA-CGTGGTGAAACCTT (AC strand).

Frequency: Estimated from 48 chromosomes of unrelated individuals.

Heterozygosity Index = 60%. PIC = 0.54.

Allele (bp)	Frequency
A1 117	0.02
A2 115	0.21
A3 113	0.21
A4 111	0.56

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

Chromosomal Localization: ATPSB has been assigned to chromosome 12p13-qter (3).

Other Comments: The PCR reaction was performed on 80 ng of genomic DNA using 100 pmoles of each oligonucleotide primer. The samples were processed as described (4) except that the denaturation cycle at 94°C was extended to 1.4 minutes. The dinucleotide repeat was based on a (GT)₁₁ sequence.

References: 1) Ohta,S. *et al.* (1988) *J. Biol. Chem.* **263**, 11257–11262. 2) Weber,J.L. and May,P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396. 3) Neckelmann,N. *et al.* (1989) *Genomics* **5**, 829–843. 4) Weber,J.L. *et al.* (1990) *Nucl. Acids Res.* **18**, 4637.