Tetranucleotide repeat polymorphism at the human beta-actin related pseudogene 2 (ACTBP2) detected using the polymerase chain reaction

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Source/Description: The polymorphic repetitive sequence (AAA-G)₁₁ AA (AAAG)₁₅ is located in the 5' flanking sequence of human beta-actin pseudogene 2, beginning at base 176 (1). The sequence was identified from a search of the EMBL and GenBank DNA sequence databases (GenBank V00481). The predicted amplified sequence length is 519 bp.

Primer Sequences:

5'GAGAGAGAGAAAGGAAGGAAGG3'

5'ATGGGGTACTTCAGAGTCAGG3'

Frequency:

Allele	Size (bp)	Frequency
A1	519	0.14
A2	513	0.24
A3	501	0.32
A4	485	0.08
A5	467	0.22

Heterozygosity 68% (estimated using 40 unrelated Caucasian individuals)

Mendelian Inheritance: Observed in CEPH families 1029 (Utah pedigree K1345) and 982 (Utah pedigree K1331).

Other Comments: 30 cycles of PCR were performed (2) using 200–300 ng of genomic DNA and 150 ng of each primer in a final reaction volume of 50 µl. The reaction mix contained a final magnesium concentration of 2.5 mM, and 10% dimethyl sulphoxide (DMSO). Primers were designed using the program PRIMER (3).

Chromosomal Localization: 5.

References: 1) Moos, M. and Gallwitz, D. (1983) *EMBO J.* **2**, 757–761. 2) Weber, J.L. and May, P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396. 3) Lowe, T., Sharefkin, J., Yang, S.Q. and Dieffenbach, C.W. (1990) *Nucl. Acids Res.* **18**, 1757–1761.

Dinucleotide repeat polymorphism in the human alpha-cardiac actin gene, intron IV (ACTC), detected using the polymerase chain reaction

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Source/Description: The polymorphic sequence (TG)₂₅ lies in intron IV, 61 bases 5' from the start of exon 5 of the human alpha-cardiac actin gene (1). The sequence was identified from a search of the EMBL and GenBank DNA sequence databases (GenBank J00072). The predicted amplified sequence length is 233 bp.

Primer Sequences:

5'TCAGAGACAAATGGTGACAGC3'

5'GACATTGTTGGCATACAGG3'

Frequency:

Allele	Size (bp)	Frequency
C1	239	0.57
C2	221	0.20
C3	219	0.23

Heterozygosity 41% (estimated using 40 unrelated Caucasian individuals)

Mendelian Inheritance: Observed in CEPH families 1029 (Utah pedigree K1345) and 982 (Utah pedigree K1331).

Other Comments: 30 cycles of PCR were performed (2) using 200–300 ng of genomic DNA and 150 ng of each primer in a final reaction volume of 50 µl. The reaction mix contained a final magnesium concentration of 2.0 mM, and 10% dimethyl sulphoxide (DMSO). Primers were designed using the program PRIMER (3).

Chromosomal Localization: 15q11-qter.

References: 1) Hamada, H., Petrino, M.G. and Kakunaga, T. (1982) *Proc. Natl. Acad. Sci. USA* **79**, 5901–5905. 2) Weber, J.L. and May, P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396. 3) Lowe, T., Sharefkin, J., Yang, S.Q. and Dieffenbach, C.W. (1990) *Nucl. Acids Res.* **18**, 1757–1761.