

| | |
|-----------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Title | Dinucleotide repeat polymorphism at the D17S518 locus |
| Authors | Couch, Fergus J.;McCarthy, Tommie V.;Gregg, R. G.;Hogan, K. |
| Publication date | 1991 |
| Original Citation | Couch, F. J., McCarthy, T. V., Gregg, R. G. and Hogan, K. (1991) 'Dinucleotide repeat polymorphism at the D17S518 locus', Nucleic Acids Research, 19(18), p. 5093. doi: 10.1093/nar/19.18.5093 |
| Type of publication | Other |
| Link to publisher's version | https://academic.oup.com/nar/article-lookup/doi/10.1093/nar/19.18.5093 - 10.1093/nar/19.18.5093 |
| Rights | © 1991, Oxford University Press |
| Download date | 2024-03-28 17:28:48 |
| Item downloaded from | https://hdl.handle.net/10468/5043 |

Dinucleotide repeat polymorphism at the D17S518 locus

F.J.Couch^{1, 2}, T.V.McCarthy², R.G.Gregg^{1,*} and K.Hogan¹

¹Department of Anesthesiology, University of Wisconsin-Madison, 600 Highland Avenue, Madison, WI 53705, USA and ²Department of Biochemistry, University College, Cork, Ireland

Source/Description: A human genomic Sau3AI/EcoRI fragment was cloned into pBluescript KS+ and selected by hybridisation to poly(dC-dA).poly(dG-dT). The cloned fragment was designated Wis3. Sequencing was performed to determine primer sequences for the polymerase chain reaction. The predicted length of the amplified fragment was 90 bp.

Primer Sequences:

5' GATCCAGTGGAGACTCAGAG 3' (CA strand);

5' TAGTCTCTGGGACACCCAGA 3' (GT strand).

Frequency: Estimated from 58 chromosomes of unrelated CEPH family members.

Observed heterozygosity = 0.76, PIC = 0.67

| Allele (bp) | Frequency | Allele (bp) | Frequency |
|-------------|-----------|-------------|-----------|
| 100 | 0.017 | 90 | 0.328 |
| 98 | 0.086 | 88 | 0.310 |
| 94 | 0.259 | | |

Chromosomal Localization: Assigned to chromosome 17q11.2-qter by PCR of somatic cell hybrid DNA. Linkage analysis of four CEPH families gave maximum two point LOD scores of 9.82, at $\theta = 0.023$ with NGFR and 6.25, at $\theta = 0.036$ with D17S37.

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

PCR Conditions: Amplification was as follows, denaturation at 94°C (3 min), 32 cycles of denaturation at 94°C (1 min), annealing at 54°C (1 min) and extension at 70°C (1 min) using 0.5 U AmpliTaq, 1×GeneAmp buffer and 200 μM dNTPs (Perkin Elmer-Cetus), 100 ng genomic DNA and 10 pmol of each primer in a volume of 15 μl. Allele sizes were determined by comparison to the cloned sequence on 8% denaturing polyacrylamide gels with autoradiography. The sequenced repeat was (GT)₁₆. The sequence has been submitted to EMBL data Library. Accession number = X60690.

Acknowledgements: This work was supported by a grant from the Department of Veterans Affairs (K.H.), and by EOLAS, the Irish Science and Technology Agency (F.C., T.V.M.).

Dinucleotide repeat polymorphism at the D8S161 locus

F.J.Couch^{1, 2}, T.V.McCarthy², R.G.Gregg^{1,*} and K.Hogan¹

¹Department of Anesthesiology, University of Wisconsin-Madison, 600 Highland Avenue, Madison, WI 53705, USA and ²Department of Biochemistry, University College, Cork, Ireland

Source/Description: A human genomic Sau3AI fragment was cloned into pBluescript KS+ and selected by hybridisation to poly(dC-dA).poly(dG-dT). The cloned fragment was designated Wis2. Sequencing was performed to determine primer sequences for the polymerase chain reaction. The predicted length of the amplified fragment was 157 bp.

Primer Sequences:

5' GATCAAGGAGCATCACATCT 3' (CA strand);

5' TAACATGTCCCCTCATTTGG 3' (GT strand).

Frequency: Estimated from 56 chromosomes of unrelated CEPH family members.

Observed heterozygosity = 0.75, PIC = 0.77

| Allele (bp) | Frequency | Allele (bp) | Frequency |
|-------------|-----------|-------------|-----------|
| 165 | 0.125 | 157 | 0.320 |
| 163 | 0.055 | 155 | 0.160 |
| 161 | 0.055 | 153 | 0.070 |
| 159 | 0.215 | | |

Chromosomal Localization: Assigned to 8q by linkage analysis of CEPH families 1331, 1333 and 1341 which gave maximum two point LOD scores of 12.34 at $\theta = 0$ with D8S34 and 9.93 at $\theta = 0$ with D8S28.

Mendelian Inheritance: Co-dominant segregation was observed in three CEPH families.

PCR Conditions: Amplification was as follows, denaturation at 94°C (3 min), 32 cycles of denaturation at 94°C (1 min), annealing at 51°C (45 sec.) and extension at 70°C (1 min) using 0.5 U AmpliTaq, 1×GeneAmp buffer and 200 μM dNTPs (Perkin Elmer-Cetus), 100 ng genomic DNA and 10 pmol of each primer in a volume of 15 μl. Allele sizes were determined by comparison to the cloned sequence on 8% denaturing polyacrylamide gels with autoradiography. The sequenced repeat was (CA)₂₁. The sequence has been submitted to EMBL data Library. Accession number = X60689.

Acknowledgements: This work was supported by a grant from the Department of Veterans Affairs (K.H.), and by EOLAS, the Irish Science and Technology Agency (F.C., T.V.M.).

* To whom correspondence should be addressed

* To whom correspondence should be addressed