Tetranucleotide repeat polymorphism at the human aromatase cytochrome P-450 gene (CYP19)

M.H.Polymeropoulos*, H.Xiao, D.S.Rath and C.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 $(TTTA)_n$ repeat begins at the 682 base pair of the human aromatase cytochrome P-450 gene on chromosome 15q21.1 (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as described previously (2). The predicted length of the amplified sequence was 175 bp.

Primer Sequences: GCAGGTACTTAGTTAGCTAC (TTTA strand); TTACAGTGAGCCAAGGTCGT (AAAT strand).

Frequency: Estimated from 46 chromosomes of unrelated individuals. Observed heterozygosity = 91.3%.

Allele (bp)	Frequency	Allele (bp)	Frequency
A1 178	0.02	A4 158	0.24
A2 174	0.37	A5 154	0.28
A3 162	0.09		

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

Chromosomal Localization: The human aromatase cytochrome P-450 gene has been assigned to chromosome 15q21.1 (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 $(TTTA)_8$ sequence.

References: 1) Means,G.D. (1989) *J. Biol. Chem.* **264**, 19385–19391. 2) Weber,J.L. and May,P.E. (1989) *Am. J. Hum. Genet.* **44**, 388–396. 3) Chen,S. *et al.* (1988) *DNA* **7**, 27. 4) Weber,J.L. *et al.* (1990) *Nucl. Acids Res.* **18**, 4637.

* To whom correspondence should be addressed

*Nco*I RFLP in human brain creatine kinase gene (CKBB)

F.T.Dionne*, L.Turcotte, J.Grondin, M.C.Thibault and C.Bouchard
Physical Activity Sciences Laboratory, Laval University, Québec, G1K 7P4, Canada

Source/Description: Probe pHBCK contains a 184 bp Hae III-Hae III cDNA fragment inserted into pGM4Z. This fragment covers the 3′ untranslated region of B creatine kinase gene and is specific for human B creatine kinase as it does not hybridize to human muscle creatine kinase gene (1).

Polymorphism: NcoI identifies a two allele polymorphism with fragments of 4.9 and 5.3 kb and a constant fragment of 11 kb.

Frequency: Studied in 62 unrelated North American Caucasians.

D1 5.3 kb allele: 0.29
D2 4.9 kb allele: 0.71.

Chromosomal Location: The CKBB locus has been mapped to 14q3.2 (2).

Mendelian Inheritance: Co-dominant autosomal segregation was observed in a 2 generation family of 9 members.

Probe Availability: This clone was a generous donation from Dr M.B.Perryman, Dept. Medicine, Baylor College, Houston.

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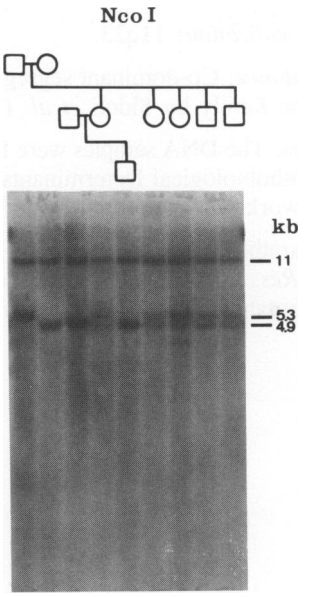
References: 1) Villarreal-Levy *et al.* (1987) *Bioch. Biophys. Res. Comm.* **144**, 1116. 2) Povey *et al.* (1979) *Ann. Hum. Genet.* **43**, 15.



* To whom correspondence should be addressed