Dinucleotide repeat polymorphism in the human tubulin alpha 1 (testis specific) gene (TUBA1)

S.Todd and S.L.Naylor

Department of Cellular and Structural Biology, The University of Texas Health Science Center at San Antonio, 7703 Floyd Curl Drive, San Antonio, TX 78284, USA

Source/Description: A (GT)_n repeat sequence is located at position 837 of the human tubulin, alpha 1 (testis specific) gene on chromosome 2q (1). PCR primers have been designed (TUBA1GT1 5'CGGAAGTGTAGGCTCCAAAAGGGT3' and TUBA1GT2 5'GACCATCGGTACCCTCACAAAGTG3') to amplify a 295 bp region containing the polymorphic repeat.

Frequency: Twenty unrelated individuals demonstrated four alleles. The observed heterozygosity was 45%.

Allele	Frequency
A1	0.03
A2	0.56
A3	0.30
A4	0.11

Chromosomal Localization: The human tubulin, alpha 1 (testis specific) (TUBA1) gene has been localized to chromosome 2q (2).

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

Other Comments: The PCR reactions were performed on 200 ng genomic DNA with 10 ng ³²P-labeled TUBA1GT1 primer and 10 ng unlabeled TUBA1GT2 primer. These reactions were carried out as described previously (3). The amplified products were run on a 5% polyacrylamide, 32% formamide sequencing gel at 80 watts constant power for approximately 4 hours in 1×TBE buffer.

References: 1) Hall, J.L. and Cowan, N.J. (1985) *Nucl. Acids Res.* **13**, 207–223. 2) Gerhard, D.S. and Bruns, G. (1985) *Cytogenet. Cell Genet.* **40**, 639–640. 3) Theune, S. *et al.* (1991) *Genomics* **9**, 511–516.

A PvuII RFLP at the HOX 1.4 homeobox locus (HOX1D)

J.Goto, D.A.Figlewicz, M.Lutchman, F.Ruddle¹ and G.A.Rouleau

Centre for Research in Neurosciences, McGill University, 1650 Cedar Avenue, Montreal, Quebec H3G 1A4, Canada and ¹Departments of Biology and Human Genetics, Yale University, New Haven, CT 06511, USA

Source/Description: pG31H980 is a 980 bp HindIII fragment cloned into pGEM vector from the human genomic phage clone 31A (1).

Polymorphism: pG31H980 reveals a two-allele restriction fragment length polymorphism with bands at 4.0 kb (A1) and 2.1 kb (A2) on Southern blot analysis of human genomic DNA digested with PvuII.

Frequency: Estimated from 28 unrelated Caucasians (56 chromosomes).

A1 (4.0 kb) = 0.96

A2 (2.1 kb) = 0.04

Not Polymorphic For: BamHI, BclI, BglI, BglII, BstEII, EcoRI, EcoRV, HincII, HindIII, HinfI, MspI, RsaI, SacI, StuI and TaqI in a screen of five unrelated Caucasians.

Chromosomal Localization: Mapped to 7p13-p15 by Southern blot hybridization to a panel of human/mouse somatic cell hybrids and in situ hybridization (1).

Mendelian Inheritance: Codominant segregation is demonstrated in four two- or three-generation families.

Probe Availability: Contact F.Ruddle.

Other Comments: Final filter washes in 0.1×SSC/0.1% SDS at 65°C.

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Reference: 1) Ferguson-Smith *et al.* (1989) *Genomics* **5**, 250–258.