Biallelic DNA polymorphism of an alpha-tubulin gene family member on chromosome 12 [TUBA/MspI/2.2;2.0 kb]

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SOURCE AND DESCRIPTION OF CLONE: mHL1301, a 1132 bp PstI insert into M13, subcloned from a DMSO-induced HL60 (promyelocytic leukemia cell line) cDNA library. Clone contains part of alpha-tubulin gene, including a novel 5' untranslated region (265 bp) and 867 nucleotides of the coding sequence that differs from previously analyzed human alpha-tubulin genes by more than 50 silent nucleotide substitutions.

POLYMORPHISM: MspI digestion identifies approximately 22 invariant bands and a two-allele polymorphism with variant bands at 2.2 (Band O) and 2.0 (Band P) kb. (Figure 1).

FREQUENCY: Studied in 80 persons of mainly Caucasian (Northern European) ancestry. PIC: 0.359. Heterozygote frequency: 0.425.
2.2 kb allele (A1) 0.625
2.0 kb allele (A2) 0.375

NOT POLYMORPHIC FOR: EcoRI (9 persons), BamHI (9 persons) or TaqI (19 persons), including Caucasians, Blacks, Orientals and AmerIndians.

CHROMOSOMAL LOCALIZATION: The polymorphic fragments were localized to chromosome 12 using a panel of 18 mouse/man hybrids. In addition, invariant bands M and N are on chromosomes 11 and 12, respectively. Extensive cross-hybridization to mouse genes prevented chromosomal assignments of other fragments.

MENDELIAN INHERITANCE: Segregation was tested in 27 large three-generation linkage reference families and was compatible with codominant Mendelian inheritance (Figure 1).

PROBE AVAILABILITY: Available from W. Salser.

COMMENTS: When a very strong hybridization signal is obtained, a third fainter variant band is sometimes seen at 1.8 kb. and appears to cosegregate with the 2.2 kb band. The 2.0 kb band (Band P) remains faint in A1/A1 homozygotes indicating a fragment of similar size to A2 but of lower homology.

REFERENCE: Davis, R.C., Thomason, A.R., Fuller, M.L., Slovin, J.P., Chou, C-C, Chada, S., Gatti, R.A. and Salser, W. mRNA species regulated during the differentiation of HL-60 cells to macrophages and neutrophils. *Devel. Biol.* **119** (1987) 164-174.

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