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A BAM HI RFLP at the human tyrosine aminotransferase (TAT) gene locus at 16q

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SOURCE/DESCRIPTION: BB 0.4, an 0.43 kb Bam HI fragment subcloned into pUC 18, derived from cDNA clone phcTAT2-16 and extending from exon 8 to exon 12 of the human TAT gene (1).

POLYMORPHISM: Bam HI identifies a two allele polymorphism with fragments of 2.5 kb (B1) and 2.2 kb (B2) (Figure). No constant band.

FREQUENCY: Studied in 30 unrelated European Caucasians:

2.5 kb allele (B1)	0.22
2.2 kb allele (B2)	0.78

NOT POLYMORPHIC FOR: Bgl II, Eco RI, Hind III, Msp I, Pst I, Pvu II, Taq I, and Xba I with DNA from 10 unrelated individuals.

CHROMOSOMAL LOCALIZATION: The human TAT gene has been assigned to 16q22.1 (2,3).

MENDELIAN INHERITANCE: Codominant segregation demonstrated in 2 two-generation families, 13 individuals.

PROBE AVAILABILITY: Freely available for linkage analysis. Available for studies on tyrosinemia II on a collaborative basis (contact G.S.).

OTHER COMMENTS: Strong allelic association exists with the previously described Msp I and Hae III RFLPs (1). Approximate haplotype frequencies estimated (4, modified) from phenotypic data of 30 unrelated individuals are: M1B1H1=0.10; M1B1H2=0.12; M1B2H2=0.43; M2B2H2=0.35; all other haplotypes < 0.01. The Bam HI RFLP raises the PIC at the TAT locus from 0.44 (1) to 0.61.

REFERENCES: (1) Westphal, E.M. et al (1988), Hum Genet 79: 260-264; (2) Natt, E. et al (1987), Hum Genet 77: 352-358; (3) Callen, D.F. et al (1988), Genomics 2: 144-153; (4) Ott, J. (1985), Genet Epidemiol 2: 79-84.

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