Human TaqI RFLP recognized by neurofilament gene probes

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SOURCE AND DESCRIPTION OF CLONES: (a) 1.2 kb EcoRI mouse cDNA clone (NF68) corresponding to the coding sequence for the 68 kd neurofilament light chain gene [1] and (b) 915 bp PvuII/AvaII fragment (NFL915PA) from 3' region of human light-chain neurofilament clone (NF-L)[2].

POLYMORPHISM: Two polymorphic bands 3.6 kb and 2.8 kb, in TaqI digested human genomic DNA are recognized both by the mouse and human clones. The polymorphic TaqI site is located approximately 2.5 kb 3' of the human NF-L gene.

FREQUENCY: Based on a sample of 80 unrelated Caucasian individuals, the frequency of the 4.2 kb allele was 0.62 and of the 3.4 kb allele was 0.38.

MENDELIAN INHERITANCE: Co-dominant inheritance was demonstrated in 12 families, with a total of 49 individuals.

NOT POLYMORPHIC FOR: ApaI, BamHI, BglII, BstEII, EcoRI, MboI, StuI and SstI, based on panels of 9 unrelated Caucasian individuals.

CHROMOSOMAL LOCALISATION: The human neurofilament light chain gene has been localised to chromosome 8[2].

PROBE AVAILABILITY: Mouse cDNA clone was supplied by Dr. N.L. Cowan [1]. Human genomic clone was provided by Dr. J-P. Julien [2].

REFERENCES: 1. Lewis and Cowan, J. Cell Biol. 100:843-850, 1985.
2. Julien et al., Biochim. Biophys. Acta 909:10-20, 1987.

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