HincII RFLP in the human gene for the heavy neurofilament subunit (NF-H)

G.Lacoste-Royal, M.Mathieu and D.Gauvreau

INRS-Santé, 245 Hymus Boulevard, Pointe-Claire, H9R 1G6, Canada

SOURCE/DESCRIPTION: pJL101 contains a 4.6 kb HindIII fragment derived from a liver genomic library and subcloned into pTZ19 (Lees et al.), that encodes part of the NF-H gene; a 1.4 kb HindIII-BamHI fragment corresponding to part of the first intron was used as a probe.

POLYMORPHISM: HincII identifies a two-allele polymorphism with fragments of either 18 kb or 6.8 kb (Figure 1.)

FREQUENCY: Studied in 43 unrelated Caucasians.
18.0 kb allele: 0.31
6.8 kb allele: 0.69

NOT POLYMORPHIC FOR: BamHI, KpnI, MspI, PstI, RsaI, TaqI.

CHROMOSOMAL LOCALISATION: Localised to 22q (Mattei et al.)

MENDELIAN INHERITANCE: Co-dominant segregation shown in 2 families (12 individuals).

PROBE AVAILABILITY: Contact G. L.-R.

OTHER COMMENTS: Low background under usual stringency conditions.

REFERENCES: Lees, J.F. et al. (1988) The EMBO Journal 7 1947-1955.
Mattei, M.G. et al. (1988) Human Genetics 80 293-295.

ACKNOWLEDGEMENTS: We thank Dr. R. Lazzarini for the use of his NF-H probe and Dr. M. Bonnycastle for collecting some of the blood samples. This work was supported by MRC Canada grant #MA-10027.