Bcl-1 RFLP at the human hepatic lipase gene locus (CIPC)

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Source/Description: Lambda HL2 contains a cDNA corresponding to the human hepatic lipase mRNA from position -8 to the poly A tail (1).

Polymorphism: Bcl-1 detects two invariant bands at 9.7 Kb and 6.6 Kb and a two allele polymorphism with bands at 17.0 Kb (B1) or 12.0 Kb and 5.0 Kb (B2).

Frequency: Studied in 16 unrelated caucasian individuals. The frequency of the F1 allele was 0.47 and of the F2 allele 0.53.

Not Polymorphic For: Apa-1, XBa-1, Bst-1 and see (1).

Chromosomal Localisation: Human hepatic lipase assigned to chromosome 15q21-q23 (1).

Mendelian Inheritance: Co-dominant inheritance demonstrated in one 2 generation family with 5 members.

Probe Availability: Request for probe to LC at the above address.

Reference: 1. Datta,S., Luo,C.C., Li,W.H., Van Tuinen,P., Ledbetter,D.H., Brown,M.A., Chen,S.H., Liu,S.W. and Chan,L. (1988) *J. Biol. Chem.* **263**, 1107-1110.

Acknowledgements: British Heart Foundation, British Diabetic Association and US National Institutes of Health.

A polymorphic cDNA probe on chromosome 17q11.2 located within the NF1 gene [D17S376]

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Source and Description: A 1.2 kb cDNA clone, EVI2B-B (1), obtained by screening a human B-cell cDNA library in λ gt11, with the beginning of pEH1, a jump clone mapping to the von Recklinghausen neurofibromatosis (NF1) region (1). The cDNA insert was subcloned into the EcoRI site of Bluescript.

Polymorphism: After EcoRI digestion of genomic DNA the probe detects two allelic fragments of 9.5 kb and 7.3 kb. No non-polymorphic fragments are detected.

Frequency: Estimated from 51 unrelated Caucasians:
9.5 kb allele (A1) 0.58
7.3 kb allele (A2) 0.42.

Not Polymorphic For: BglII, HindIII, MspI, PstI, PvuII, TaqI and XbaI (studied in 6 unrelated individuals).

Chromosomal Localization: Using a somatic cell hybrid panel the probe was mapped to a 60 kb region of 17q11.2 between two translocation breakpoints in two individuals with von Recklinghausen neurofibromatosis (NF1).

Mendelian Inheritance: Co-dominant segregation demonstrated in 3 three generation families (15 meioses).

Clinical Relevance: Segregation studies in families with von Recklinghausen neurofibromatosis (NF1). The locus EVI2B represented by this cDNA clone is embedded within an intron of the *NF1* gene (2, 3).

Probe Availability: Apply to Francis S. Collins.

Acknowledgements: This work was supported by NIH grant NS23410 to FSC and a fellowship from University of Copenhagen, Denmark, to LBA. RLW and FSC are investigators of the Howard Hughes Medical Institute.

References: 1) Cawthon,R. *et al.* (1990) *Genomics*, (in press).
2) Wallace,M. *et al.* (1990) *Science* **249**, 181-186. 3)
Viskochil,D. *et al.* (1990) *Cell* **62**, 187-192.

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