Two polymorphisms for the human hepatic lipase (HL) gene

C.Heinzmann, J.Ladiaz¹, S.Antonarakis¹, A.Diep, M.Schotz² and A.J.Lusis

Department of Medicine and Microbiology and Molecular Biology Institute, UCLA, Los Angeles, CA 90024,

¹Department of Pediatrics, Johns Hopkins Hospital, Baltimore, MD 21205 and ²Division of Research, VA Wadsworth Medical Center, Los Angeles, CA 90273, USA

SOURCE AND DESCRIPTION OF CLONE: A 1.65 kb ECORI cDNA insert corresponding to the full length hepatic lipase (HL) sequence isolated from a rat liver cDNA library (1).

POLYMORPHISMS: Two polymorphisms detected by HL:

HindIII: allele 1: 3.7 kb allele 2: 2.7 kb and 1 kb
invariant bands: 12.5 kb, 7 kb and 2 kb
MspI: allele 1: 1.3 kb allele 2: 1.1 kb
invariant bands: 6.2 kb, 0.5 kb and 0.3 kb

FREQUENCY: HindIII among 81 unrelated individuals

allele 1: 0.85 allele 2: 0.15

MspI among 63 unrelated individuals

allele 1: 0.5 allele 2: 0.5

NOT POLYMORPHIC FOR: PvuII, PstI (18 unrelated individuals), KpnI (10 individuals), HinfI, AluI, EcoRV, BamHI, BglII, SstI, EcoRI, HaeIII (5 individuals)

CHROMOSOMAL LOCALIZATION: Chromosome 15q21 by somatic cell and in situ hybridization (2).

MENDELIAN INHERITANCE: Segregation was demonstrated in one family of 19 individuals.

PROBE AVAILABILITY: Requests for probe to M. Schotz at above address.

REFERENCES: 1) M. Komaromy and M. Schotz, PNAS 84, 1526-1530 (1987).

2) R. Sparkes et al., Genomics 1, 138-144 (1987).

ACKNOWLEDGEMENTS: Funded by grant HL 27-481 from the United States Public Health Service. AJL is an established investigator of the American Heart Association.