

AvaII polymorphism in the human LDL receptor gene

Helen H.Hobbs, Victoria Esser and David W.Russell

Department of Molecular Genetics, University of Texas Health Science Center at Dallas, 5323 Harry Hines Blvd., Dallas, TX 75235, USA

SOURCE/DESCRIPTION: pLDLR3 contains a full-length cDNA corresponding to the human LDL receptor mRNA (1). The vector is pcD minus the SV40 early region promoter (2).

POLYMORPHISM: AvaII identifies a bi-allelic polymorphism (bands of 3.6 and 3.4 kb) for a site within Exon 13 of the LDL receptor gene (3). Co-digestion with a non-polymorphic enzyme such as XbaI which cleaves in intron 12 yields more readily separable bands of 1.9 and 1.7 kb (See Fig.).

FREQUENCY: Studied in 9 American caucasian individuals

AvaII/XbaI 1.9 kb O. 56

AvaII/XbaI 1.7 kb O. 44

NOT POLYMORPHIC FOR: BamHI, BglII, EcoRI, EcoRV, HindIII, KpnI, Scal, SstI, SstII, SphI, TaqI, XbaI, or XhoI. Polymorphisms for PstI (4), PvuII (5,6), and StuI (7) have been described.

CHROMOSOMAL LOCATION: 19p13.1-13.3 (ref. 8)

MENDELIAN INHERITANCE: Co-dominant segregation demonstrated in 1 family (see Fig.).

PROBE AVAILABILITY: pLDLR3 may be obtained from the American Type Culture Collection, 12301 Parklawn Drive, Rockville, MD, 20852. Request strain number 57004.

OTHER COMMENTS: DNA sequence at polymorphic site is GGTC = AvaII or GGTC ≠ AvaII.

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