

Pst I RFLP close to the LDL receptor geneH.Funke¹, J.Klug¹, P.Frossard², R.Coleman² and G.Assmann^{1,3}

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SOURCE/DESCRIPTION: Synthetic 45 base oligodeoxynucleotide representing a sequence from the 18th exon of the LDL receptor gene (see availability section).

POLYMORPHISM: Pst I two allele polymorphism located approximately 1.7 kb downstream from the 3' end of the 18th exon.

FREQUENCY: In 35 randomly selected students the following allelic frequencies were determined:

3.4 kb fragment	0.6
2.85 kb fragment	0.4

NOT POLYMORPHIC FOR: In 10 individuals no polymorphisms were detected with XmnI, EcoRI, BamHI, HindIII, BglII, StuI, TaqI, MspI, and PvuII.

CHROMOSOMAL LOCALISATION: The LDL receptor gene has been assigned to p13.1-p13.3 on chromosome 19 (1).

MENDELIAN INHERITANCE: Codominant segregation has been observed in 3 families (17 individuals).

PROBE AVAILABILITY: Probe was chemically synthesized starting with nucleotide 4744 and ending with nt 4788 as numbered in (2).

OTHER COMMENTS: The localisation of cutting sites outside exon 18 was done in relation to the PstI site in exon 18 (see figure).

REFERENCE: 1. Lindgren, V. et al. (1985) PNAS 82, 8567-8571
2. Yamamoto, T. et al. (1984) Cell 39, 27-38

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Fig.: Partial restriction map from the 3' end of the LDL receptor gene. Oligodeoxynucleotide position is represented as a bar. * indicates the polymorphic site.