

Table S1 : List of primers for PCR amplification of *LDLR*, *PCSK9*, exon 26 of *APOB* and *APOE* genes. And list of primers for splicing reporter minigene assay and Sanger sequencing of the RT-PCR products.

LDLR OMIM#606945	Forward		Reverse	Length
Promotor-F	TCAGTGTCTATTAGGTGATTTGGAATAAC	Promotor-R	TGGCTTCCCGCGATTG	551pb
Exon1-F	TCCTCCCCCTGCTAGAAACC	Exon1-R	CCAGCCGTTTGGGAAGG	351pb
Exon2-F	TTTTCCCATACCCAGAGAGTC	Exon2-R	TCATTCTCTCCCCACCTCCTAAT	651pb
Exon3-F	TCAGTGGGTCTTTCTTTGAGTG	Exon3-R	AGCGGCTTGCCAGTGTGT	452pb
Exon4-F	GAGGAAACTGAGGCACCGAG	Exon4-R	TCCACTTCGGCACCTAAATCA	651pb
Exon5-F	GCAAAAAGGCCCTGCTTCTTT	Exon5-R	GCAAGCAGCAAGGCACAGA	251pb
Exon6-F	TTTGCATGCGTTCTTATGTGAAT	Exon6-R	TCTGCAAGCCGCCTGC	256pb
Exon7-F	CCTGGCACCCACCATTCTAC	Exon7-R	CCATGTCTCAGGAAGCGCAGA	751pb
Exon8-F	GGTCCCGTGGTGAATGATG	Exon8-R	GAGCCCTCAGGAGCAAACAG	451pb
Exon9-10-F	GGTGCCTCCTCTGGCTCAG	Exon9-10-R	TTCTGCTCCCTCCATTCC	651pb
Exon11-F	CCTGAGCCTGGCTGTTTCTTC	Exon11-R	GCTTGTCCCAGAGCCACCT	301pb
Exon12-F	TGGAGAGGGCGTCACAGG	Exon12-R	TGCGTTCATCTTGGCTTGAGT	401pb
Exon13-14-F	TGGCCTGTGTCTCATCCCA	Exon13-14-R	CAAGCCCGGTGCTGATGT	651pb
Exon15-F	CTTTCGTCATTAGGCACACA	Exon15-R	TTGCAAAGAGGGCAAGAAGT	451pb
Exon16-F	CCGAATTGAGTCTACAACCT	Exon16-R	GAGGCTATTCCACAGCACGG	501pb
Exon17-F	CGCAAGGCGATCTCTAAACAA	Exon17-R	CCGCTGACATTCTGAATGAGC	451pb
Exon18-F	GCAGGAGGCTACCAGGCAG	Exon18-R	CATTGCATGGGCACTGTCC	451pb

APOB OMIM#107730	Forward		Reverse	Lenght
Ex26-3500-F	CATCTGTCATTGATGCACTGCA	Ex26-3500-R	GGCTTTGCTTGTATGTTCTCCG	650pb

PCSK9 OMIM#607786	Forward		Reverse	Length
Exon1-F	TGAACTTCAGCTCCTGCACA	Exon1-R	GCGAACCTTCCCCTGAATA	591pb
Exon2-F	CCTGAATGGCACATTTGAAAG	Exon2-R	TGCTCAATACATACTTGCTGTCC	542pb
Exon3-F	TGTTTGCTGCTGTCCAAATG	Exon3-R	GGCAGAGCAAATGGATTCAG	243pb
Exon4-F	CAGACTCGGTGAGCTGTGAA	Exon4-R	AGGGAGGGGACAGTGACAG	644pb
Exon5-F	GCTTCCACAGACAGGTAAGCAC	Exon5-R	GGGTTCTTCATCTGCACTCG	583pb
Exon6-F	TTCGCAGCAGCATTCCAC	Exon6-R	TCCAAAGCCAGAAGGGTTC	482pb
Exon7-F	AGTCAGATTTTCCTTAGGAGGG	Exon7-R	ACTGAGTGCCTTGAAGGCAC	476pb
Exon8-F	TGTCTGTGTGCACGTGTGTT	Exon8-R	CAGACAAGGGAGAAGGGAGA	378pb
Exon9-F	GTAAGGAGGATGACGCCACC	Exon9-R	AAGAGCTGGAGTCTGGAGGAT	466pb
Exon10-F	AGCTCCTTGTCCCAGAAG	Exon10-R	GAGTATGGAAGTCAAGTCAGGCTTGAG	448pb
Exon11-F	GGCTCAGAGAGGTTGAATGG	Exon11-R	GCATCTACCTGGCAAACCG	534pb
Exon12-F	TGGTAGGCATCTGTCTATCTCC	Exon12-R	GAAGCATCCCCATCCCC	597pb

APOE OMIM#107741	Forward		Reverse	Length
PROMOTOR-F	GCCCCCTCTCCAGATTACAT	PROMOTOR-R	GCTGGGGCTGAGTAGGACT	356pb
Exon1-F	GGGAGAACAGCCACCTCGT	Exon1-R	CCCCTGGCTCCCAGTTATG	450pb
Exon2-F	GAGTTAAATAGGGAATGGTTGG	Exon2-R	TGAGGATGAGAGGGGGAGGAG	293pb
Exon3-F	TCCAAAGAAGCATTGTGGAGCA	Exon3-R	GCCAGGAGTCAGAAATGGGAAGA	442pb
Exon4a-F	GGCCTGGGTCTCTGCTGGTT	Exon4a-R	TTCTGCAGGTCATCGGCATC	773pb
Exon4b-F	GAAGGCCTACAAATCGGAACTGG	Exon4b-R	GGCCAGCAGATGCGTGAAAC	840pb

List of primers for minigene splicing assay

Name	Sequence	Position	Analysed variant of <i>LDLR</i>
RLDL Exon3 L	CAAAGACAGGATTGGCAAGG	c.191-379	c.313+5G>T
RLDL Exon3 R	CGGAAGAGGCTTGGTATGAG	c.313+146	
RLDL Exon 4 L	AGCAGTGCTGGGAAATGTG	c.314-200	c.686A>T
RLDL Exon 4 R	TTTCTTGGCATGTTGTTGG	c.694+119	
RLDL Exon15 F	CTCCAAGGTCATTTGAGAC	c.2141-350,	c.2311G>A
RLDL Exon15 R	CAAGTGAGAGAAGGTCAGCA	c.2311+300,	

Vector exonic primers for RT-PCR

Name	Sequence	Vector exon	
Alpha2-3globine F	CAACTTCAAGCTCCTAAGCCACTG	exons 2-3	from: D. Baralle, J Med Genet 2005; 42:737-748
B2 fibronectine R	GGTCACCAGGAAGTTGGTTAAATCA	exon 4	