

# Higher Responsiveness to Rosuvastatin in Polygenic versus Monogenic Hypercholesterolaemia: A Propensity Score Analysis.

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**Table S1.** Description of LDLR and APOB mutations in patients with monogenic hypercholesterolemia.

Gene	cDNA Change	Protein Change	Number of Affected Proband	ACGS Category
APOB	c.10580G > A	p.(R3527Q)	7	5
LDLR	c.666C > A	p.(Cys222*)	1	5
LDLR	c.(?-187)_(190+1_191+1)del	Promoter - exon 2 deletion	2	5
LDLR	c.(67+1_68-1)_(313+1_314-1)del	Exon 2 - exon 3 deletion	1	5
LDLR	c.2056C > T	p.(Gln686*)	1	5
LDLR	c.(313+1_314-1)_(1186+1_1187-1)dup	Exon 4 - exon 8 duplication	4	5
LDLR	c.693C > A	p.(Cys231*)	1	5
LDLR	c.526_533dup	p.(Asp178Glufs*31)	1	5
LDLR	c.1141G > T	p.(Glu381*)	2	5
LDLR	c.1775G > A	p.(Gly592Glu)	11	4
LDLR	c.798T > A	p.(Asp266Glu)	2	4
LDLR	c.1117G > T	p.(Gly373Cys)	1	4
LDLR	c.100T > G	p.(Cys34Gly)	1	4
LDLR	c.2000G > A	p.(Cys667Tyr)	1	4
LDLR	c.662A > G	p.(Asp221Gly)	4	4
LDLR	c.2483delA	p.(Tyr828Phefs*101)	1	4
LDLR	c.265T > C	p.(Cys89Arg)	1	4
LDLR	c.283T > C	p.(Cys95Arg)	1	4
LDLR	c.591C > G	p.(Cys197Trp)	1	4

<i>LDLR</i>	c.530C > T	p.(Ser177Leu)	1	4
<i>LDLR</i>	c.1790C > A	p.(Thr597Asn)	1	4
<i>LDLR</i>	c.191-1G > A	p.?	1	4