APPENDIX. SUPPLEMENTARY DATA

Table 1 of the supplementary data

Rare variants in candidate genes identified in participants selected in this study

GENE	*SNV*	*Nucleotide*	*Amino acid change*	*Number of carriers*	*Bioinformatic analysis*				*Clin Var*	*Frequency ExAc*[1]	*Frequency 1000 G*[2]
					SIFT[3]	*POLYPHEN-2*[4]	*Mutation Taster*[5]	*PredictSNP*[6]			
LDLR	rs879254375	c.(-135)C>G	NA	1 participant	NA	NA	NA	NA	Pathogenic	-	-
LDLR	rs774467219 rs112029328	c.[274C>G; 313+1G>C]	p.[Gln92Glu;NA]	1 participant	Tolerated (0.15)	Possibly Damaging (0.736)	Deleterious (0.510)	Neutral (0.252)	Pathogenic	0.0009715	0.001
LDLR	rs121908026	c.530C>T	p.(Ser177Leu)	1 participant	Deleterious (0.01)	Probably Damaging (0.999)	Deleterious (0.896)	Deleterious (0.000090)	Pathogenic	8.958e-06	-
LDLR	rs879254692	c.826T>G	p.(Cys276Gly)	1 participant	Deleterious (0)	Probably damaging (0.969)	Deleterious (0.856)	Deleterious (0.000005)	Pathogenic	-	-
LDLR	rs368657165	c.862G>A	p.(Glu288Lys)	1 participant	Deleterious (0.05)	Probably damaging (0.918)	Deleterious (0.714)	Deleterious (0.000018)	Pathogenic	0	-
LDLR	-	c.941-?_1845+?del	Deletion from exon 7 to exon 12	1 participant	-	-	Deleterious (0.999)	Deleterious (0.000008)	Pathogenic		
LDLR	rs773658037	c.1247G>A	p.(Arg416Gln)	1 participant	Deleterious (0.01)	Probably damaging (0.957)	Deleterious (0.806)	(0.000045)	Pathogenic	1.793e-05	0
LDLR	rs755154048	c.1529C>T	p.(Thr510Met)	1 participant	Deleterious (0.02)	Possibly damaging (0.791)	Deleterious (0.679)	Deleterious (0.000013)	Pathogenic/ Likely pathogenic	8.952e-06	-
LDLR	rs781362878	c.1586+5G>A	NA	1 participant	NA	NA	NA	NA	Uncertain/ Pathogenic	1.796e-05	

GENE	*SNV*	*Nucleotide*	*Amino acid change*	*Number of carriers*	*Bioinformatic analysis*				*Clin Var*	*Frequency ExAc*[1]	*Frequency 1000 G*[2]
					SIFT[3]	*POLYPHEN-2*[4]	*Mutation Taster*[5]	*PredictSNP*[6]			
LDLR	rs137929307	c.1775G>A	p.(Gly592Glu)	2	Deleterious (0.01)	Probably damaging (0.925)	Deleterious (0.779)	Deleterious (0.000015)	Pathogenic	8.951e-05	-
LDLR	rs72658865	c.1816G>A	p.(Ala606Thr)	1 participant	Deleterious (0.02)	Possibly damaging (0.5)	Deleterious (0.550)	Deleterious (0.000034)	Likely benign/ Uncertain	8.952e-06	-
PCSK9	rs371488778	c.60_65dup GCTGCT	p.(Leu22_ Leu23dup)	4 participants	NA	NA	NA	NA	Uncertain/ Pathogenic	0.002144	-
PCSK9	rs749573024	c.743G>A	p.(Arg248His)	1 participant	Tolerated (0.19)	Benign (0.001)	-	Neutral	Likely benign	9.099e-06	0
APOB	rs4362589	c.10621A>G	p.(Ile3541Val)	1 participant	Tolerated (0.39)	Benign (0.106)	Neutral (0.024)	Neutral (0.085255)	Unknown	8.99e-06	0
STAP1	rs201996284	c.-60A>G	NA	2 participants	NA	NA	Deleterious	Deleterious	Unknown	-	0
STAP1	rs146545610	c.619G>A	p.(Asp207Asn)	1 participant	Tolerated (0.1)	Benign (0.046)	Neutral (0.289)	Neutral (0.001432)	Unknown	0.0002417	0
STAP1	-	c.803T>C	p.(Ile268Thr)	1 participant	Deleterious (0)	Possibly damaging (0.682)	Deleterious (0)	Deleterious	Unknown	-	-
LDLRAP1	rs121908326	c.605C>G	p.(Ser202Cys)*	1 participant	Tolerated (0.15)	Possibly Damaging (0.736)	Deleterious (0.510)	Neutral (0.251599)	Pathogenic	0.0009715	0.001
LDLRAP1		c.748-7C>G	NA	1 participant	NA	NA	NA	NA	Unknown	-	-
GENE	*SNV*	*Nucleotide*	*Amino acid change*	*Number of carriers*	*Bioinformatic analysis*				*Clin Var*	*Frequency ExAc*[5]	*Frequency 1000 G*[6]
					SIFT[1]	*POLYPHEN-2*[2]	*Mutation Taster*[3]	*PredictSNP*[4]			
APOE	rs745552623	c.335C>A	p.(Ser112Tyr)	1 participant	Tolerated (0.16)	Probably damaging (0.953)	Neutral (0.456)	Neutral (0.280673)	Unknown	1.002e-05	0
APOE	rs121918393	c.460C>A	p.(Arg154Ser)	3 participants	Deleterious (0)	Probably	Deleterious (0.881)	Deleterious (0.000049)	Pathogenic	0.00000	-

						Damaging (0.987)					
APOE	rs769455	c.487C>T	p.(Arg163Cys)	1 participant	Deleterious (0)	Probably damaging (0.993)	Deleterious (0.895)	Neutral (0.003540)	Pathogenic	6.832e-05	0

*This patient carries this variant in heterozygosity.

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