Table SI. Primers for PCR and subsequent Sanger sequencing.

Gene, exon	Detected variants	Forward, 5'-3'	Reverse, 5'-3'	Tm	Fragment length
LDLR, 2	c.100T>G p.(Cys34Gly)	CAGTTTCTGATT CTGGCGTTG	ATCATGCCCAA AGGGGACTC	60	197
LDLR, 4	c.316_328delCCCAAGACGTGCT p.(K107Rfs*95) c.325T>G p.(Cys109Gly) c.401G>C (p.Cys134Ser) c.433_434dupG p.(Val145Glyfs*35) c.552T>G p.(Cys184Trp) c.616A>C (p.S206R)	CAGTGCTGGGA AATGTGTACA	CACCTAAATCA CTGCATGTCC	60	664
LDLR, 5	c.798T>A p.(Asp266Glu)	CACACTCTGTC CTGTTTTCCAG	AGAATGGGGGAT GGAAAACCAG	60	191
LDLR, 6	c.887G>A p.(Cys296Tyr) c.888C>A p.(Cys296*) c.938 G>A p.(Cys313Tyr)	TGAATCCATTTG CATGCGTT	CTACAGCACTCA TGTCTCAGT	60	308
LDLR, 6	spl Intron 6 c.940+1_ c.940+4 delGTGA (g.18154_18157delGTGA)	ATGAGTGCCAA GCAAACTGA	GGCAGAGTGGA GTTCCCAA	58	285
LDLR, 7	c.986G>A p.(Cys329Tyr) c.1048C>T p.(Arg350*)	AGAGTGACCAG TCTGCATCC	ACTGAGGCATGA GGGGTTTG	60	232
LDLR, 8	c.1186G>C p.(Gly396Arg)	CTCCGTCTCTAG CCATTGGG	ATGAAACTCCCC CACCACTG	57	255
LDLR, 8	spl Intron 8 c.1186+1G>T	GTTGGGTTCCCG TGGTGAAT	TGTCCCTGAAAG GAAAGCCG	60	595
LDLR, 9	c.1202T>A p.(Leu401His) c.1277 T>C (p.Leu426Pro)	AGGCTAGCCTTA TGCTCCTAG	AGGAGCCCTCAT CTCACCTG	60	464
LDLR, 10	c.1478_1479delCT p.(Ser493Cysfs*42)	GAATGATCTGCA GGTGAGCG	GTGGATACGCAC CCATGAAC	58	338
LDLR, 11	c.1684_1691delTGGCCCAA p.(Pro563Hisfs*14)	GGGGTTCCCAGC AGGACTAT	CTCTCCAATGGG CAGGTAGG	62	285
LDLR, 12	c.1730G>C p.(Trp577Ser) c.1775G>A p.(Gly592Glu)	CAGCACGTGACC TCTCCTTAT	GCCACCTAAGTG CTTGCATC	60	218
LDLR, 15	c.2230C>T p.(Arg744*)	TAGGCGCACACC TATGAGAAG	TCGTGACCAAAA TGTTCGTGG	60	317
APOB, 26	c.9175C>T p.(Arg3059Cys)	TGTTCTCGTTGTT TCCAGCAGA	GCATGGCACTGT TTGGAGAAG	60	312
APOB, 26	c.10580G>A p.(Arg3527Gln) c.10580G>T p.(Arg3527Leu)	ATATGCGTTGGA GTGTGGCT	ACCGCTAAAGGA GCAGTTGA	60	265
APOB, 29	c.13480_13482delCAG p.(Gln4494del)	TGACTGTGGTTG ATTGCAGC	CTTTCTGCCACTG CTCAGGA	58	238
ABCG5, 10	c.1336C>T p.(Arg446*)	CGAGTCCCACTA GCTCCAT	ACTGACTGTTCT GAAACCTGC	58	237
ABCG8, 7	c.1083G>A p.(Trp361*)	CGAGGGGTGATC AGCATTGT	ATAGGGGTGTTCT GTGTCGC	60	139
ABCG8, 11	c.1629G>T p.(Arg543Ser)	AGCCTCATCATC ACCAGGAG	GAAAGCAGGCTT CATCCAGT	60	453
PCSK9, 9	c.1486C>T p.(Arg496Trp)	ACTGTATGGTCA GCACACTCG	CCTCTAGGAAAC CCTTCCCG	60	361
LIPA, 8	c.894G>A p.(Q298=)	ACATGAACCCCA AATGCACTC	TTGCACCTCCTTC ATATGGAC	60	315

PCR reactions were performed with Taq AB PCR master-mix (Alkor Bio Group).

Table SII. Phenotypic characteristics of mutation-positive adult patients.

A, Patients with mutations in the *LDLR* gene

Patient ID	Age	Sex	Medical history of ASCVD	Family history	Xanthomas	Max TC, mmol/l	LDL-C, mmol/l	Mutation	Variant ID
27	66	F	MI at the age of 57, CAD	Yes	None	10	Nd	Missense Exon 2 c.100T>G p.(Cys34Gly)	rs879254405
1	56	F	MI at the age of age 53	Yes	Yes	10.3	Nd	Frameshift Exon 4 c.433_434dupG p.(Val145Glyfs*35)	Novel
33	29	F	No	Yes	Yes	10.8	8.6	Missense Exon 5 c.798T>A p.(Asp266Glu)	rs139043155
60	36	F	Increased IMT	Yes	Yes	12	8.5	Missense Exon 6 c.887G>A p.(Cys296Tyr)	rs879254707
9	31	M	CAD from the age of 29	Yes	Yes	10.3	Nd	Nonsense Exon 6 c.888C>A p.(Cys296*)	rs879254708
38	43	M	Increased IMT	Yes	Yes	11.9	7.5	Missense Exon 7 c.986G>A p.(Cys329Tyr)	rs761954844
39	67	F	Carotid artery stenosis, 30%-40%	Yes	Yes	14.7	12	Missense Exon 7 c.986G>A (p.Cys329Tyr)	rs761954844
45	36	M	Increased IMT	Yes	Yes	13.5	10.9	Nonsense Exon 7 c.1048C>T p.(Arg350*)	rs769737896
32	29	M	Anterior descending artery stenosis, 30%	No	Yes	9.7	8.5	Missense Exon 8 c.1186G>C p.(Gly396Arg)	Novel
59	32	F	Carotid artery stenosis, 30%-40%	No	Yes	16	Nd	Missense Exon 9 c.1202T>A p.(Leu401His)	rs121908038
35	42	M	Hypertension	Yes	Yes	9.8	7.7	Frameshift Exon 10 c.1478_1479delCT p.(Ser493Cysfs*42)	rs869025453
36[a]	70	F	MI at the age of 55, CAD	Yes	Yes	12	Nd	Missense Exon 12 c.1775G>A p.(Gly592Glu)	rs137929307
69	49	M	MI at the age of 47, carotid artery stenosis, 50%	Yes	None	10.7	Nd	Nonsense Exon 15 c.2230C>T p.(Arg744*)	rs200793488

B, Patients with mutations in the *APOB* gene

30	39	F	Carotid artery stenosis 30%-40%	No	None	11.2	9.8	In-frame deletion c.13480_13482delCAG p.(Gln4494del)	rs562574661
31	65	F	CAD, hypertension	No	None	17	9	In-frame deletion c.13480_13482delCAG p.(Gln4494del)	rs562574661
58	59	F	MI at the age of 56	No	Yes	12.9	Nd	c.10580G>T p.(Arg3527Leu)	rs5742904

Table SII. Continued.

C, Patients with mutation in the *ABCG5*/*ABCG8* genes

Patient ID	Age	Sex	Medical history of ASCVD	Family history	Xanthomas	Max TC, mmol/l	LDL-C, mmol/l	Mutation	Variant ID
18	50	F	MI at the age of 47, obesity, hypertension, stable angina	Yes	Yes	9.9	6.1	Nonsense *ABCG5* c.1336C>T p.(Arg446*)[b]	rs199689137
2	36	M	CAD, MI (age 30), hypertension, obesity, fatty liver disease acanthosis nigricans	Yes	None	Nd	Nd	Missense Ex11 *ABCG8* c.1629G>T p.(Arg543Ser)	rs201690654

D, Patient with variant in the *PSCK9* gene (unknown significance)

Patient ID	Age	Sex	Medical history of ASCVD	Family history	Xanthomas	Max TC, mmol/l	LDL-C, mmol/l	Mutation	Variant ID
70	47	F		No	None	Nd	Nd	c.436G>A p.(Asp146Asn)	Novel

ASCVD, atherosclerotic cardiovascular disease; CAD, coronary artery disease; HC, hypercholesterolemia; IMT, intima-media thickness; LDL-C, low density lipoprotein cholesterol; MI, myocardial infarction; Nd, no data; TC, total cholesterol. [a]Patient also had a *ABCG8* c.1921G>A variant p.(Ala641Thr) of uncertain significance, which was found in the Northwest Russia 694 exomes dataset with a minor allele frequency $2.8x10^{-3}$ (GnomAD AF 10^{-3}). [b]The *ABCG5* c.1336C>T p.(Arg446*) variant was found in the Northwest Russia 694 exomes dataset (19) with MAF $9,5x10^{-4}$ (GnomAD AF $1,8x10^{-4}$).

Table SIII. Phenotypic characteristics of mutation-positive patients from the children/adolescent group.

A, Patients with mutations in the *LDLR* gene

Patient ID	Age	Sex	Medical history if available	Family history	Xanthomas	TC, mmol/l	LDL-C, mmol/l	Mutation	Variant ID
G29	11	F	HC firstly detected in 9	HC	No	11	9.1	Missense Exon 4 c.325T>G p.(Cys109Gly)	Novel
G36	7	F	Examination due to family history	HC	No	Nd	11.6	Missense Exon4 c.401G>C p.(Cys134Ser)	Novel
G33	18	M	Examination due to family history	HC, CAD	No	8.8	7.2	Missense Exon 4 c.552T>G p.(Cys184Trp)	Reported in LOVD database
G18	6	M	HC firstly detected in 3	HC, CAD	No	10.7	8.6	Missense Exon 4 c.616A>C p.(Ser206Arg)	Novel
G44	11	M	Sinus node disfunction from age 7	HC	No	7.5	5.7	Missense Exon 6 c.938 G>A p.(Cys313Tyr)	rs875989910
G21	19	M	HC firstly detected in 8	HC	No	7.9	5.7	spl Intron 6 c.940+1_c.940+4 delGTGA (g.18154_18157delGTGA)	Novel
G27	21	F	HC firstly detected in 19	HC	No	8	3.7	spl Intron 6 c.940+1_c.940+4 delGTGA (g.18154_18157delGTGA)	Novel
G32	19	M	Examination due to frequent respiratory infections	No	No	10	Nd	spl Intron 6 c.940+1_c.940+4 delGTGA (g.18154_18157delGTGA)	Novel
G41	7	M	HC firstly detected in 3 (cerebrovascular accident with subsequent hemiplegia)	HC	No	6.8	6.3	spl Intron 6 c.940+1_c.940+4 delGTGA (g.18154_18157delGTGA)	Novel
G26	12	M	HC firstly detected in 10, additionally diagnosed with acute lymphoblastic leukemia	HC	No	8.3	5.5	spl Intron 8 c.1186+1G>T	rs730880131
G5	5	M	HC firstly detected in 2, hepatomegaly	HC	No	9.1	7.2	Missense Exon 9 c.1202T>A p.(Leu401His)	rs121908038
G14	15	F	Dyspnea	HC	No	10.4	7.1	Missense Exon 9 c.1202T>A p.(Leu401His)	rs121908038
G30	7	M	HC firstly detected in 5	HC	No	9.5	5.7	Missense Exon 9 c.1277 T>C p.(Leu426Pro)	rs879254851
G17	16	F	Examination due to frequent respiratory infections	No	No	9.6	5	Frameshift Exon 11 c.1684_1691delTGGCCCAA p.(Pro563Hisfs*14)	Novel
G3	11	F	HC firstly detected in 3	HC	No	8.9	5.7	Missense Exon 12 c.1775G>A p.(Gly592Glu)	rs137929307
G22	9	F	HC firstly detected in 6	HC, CAD	No	10.2	6	Missense Exon 12 c.1775G>A p.(Gly592Glu)	rs137929307
G37	6	F	Examination due to frequent respiratory infections	HC	No	Nd	6.9	Missense Exon 12 c.1775G>A p.(Gly592Glu)	rs137929307
G39	8	M	Examination due to family history	HC	No	6.3	4.5	Missense Exon 12 c.1775G>A p.(Gly592Glu)	rs137929307
G49	6	F	Examination due to family history	HC	No	8.5	6.6	Missense Exon 12 c.1775G>A p.(Gly592Glu)	rs137929307
G9	17	M	Secondary cardiomyopathy, HC firstly detected in 12	HC	No	10.1	7.3	Missense Exon 12 c.1730G>C p.(Trp577Ser)	rs138947766

Table SIII. Continued.

B, Patients with mutations in the *APOB* gene

Patient ID	Age	Sex	Medical history if available	Family history	Xanthomas	TC, mmol/l	LDL-C, mmol/l	Mutation	Variant ID
G20	6	M	HC firstly detected in 4	HC	No	7.3	6.2	Missense Exon 26 c.9175C>T p.(Arg3059Cys)	rs146377316
G24	7	M	Hepatosplenomegaly, additionally diagnosed with epilepsy	HC	No	7.4	4.4	Missense Exon 26 c.10580G>A p.(Arg3527Gln)	rs5742904

C, Patient with mutation in the *ABCG8* gene

Patient ID	Age	Sex	Medical history if available	Family history	Xanthomas	TC, mmol/l	LDL-C, mmol/l	Mutation	Variant ID
G40	2	M		HC	No	6.4	Nd	Missense Exon 7 c.1083G>A p.(Trp361*)	rs137852987

D, Patient with mutation in the *LIPA* gene

Patient ID	Age	Sex	Medical history if available	Family history	Xanthomas	TC, mmol/l	LDL-C, mmol/l	Mutation	Variant ID
G48	11	F	Hepatomegaly	HC	No	7.9	Nd	spl Exon 8 c.894G>A (p.Q298=)	rs116928232

E, Patient with variant in the *PSCK9* gene (unknown significance)

Patient ID	Age	Sex	Medical history if available	Family history	Xanthomas	TC, mmol/l	LDL-C, mmol/l	Mutation	Variant ID
G34	8	F	Congenital heart defect, HC first detected in 4	HC	No	7.1	4.7	Missense c.1486C>T p.(Arg496Trp)	rs374603772

ASCVD, atherosclerotic cardiovascular disease; CAD, coronary artery disease; HC, hypercholesterolemia; LDL-C, low density lipoprotein cholesterol; LOVD, Leiden Open Variation Database; Nd, not determined; TC, total cholesterol; F, female; M, male.