

Supplementary Appendix

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Figure S1. Haplotype sequence of the proband around the region including Var1 and Var2

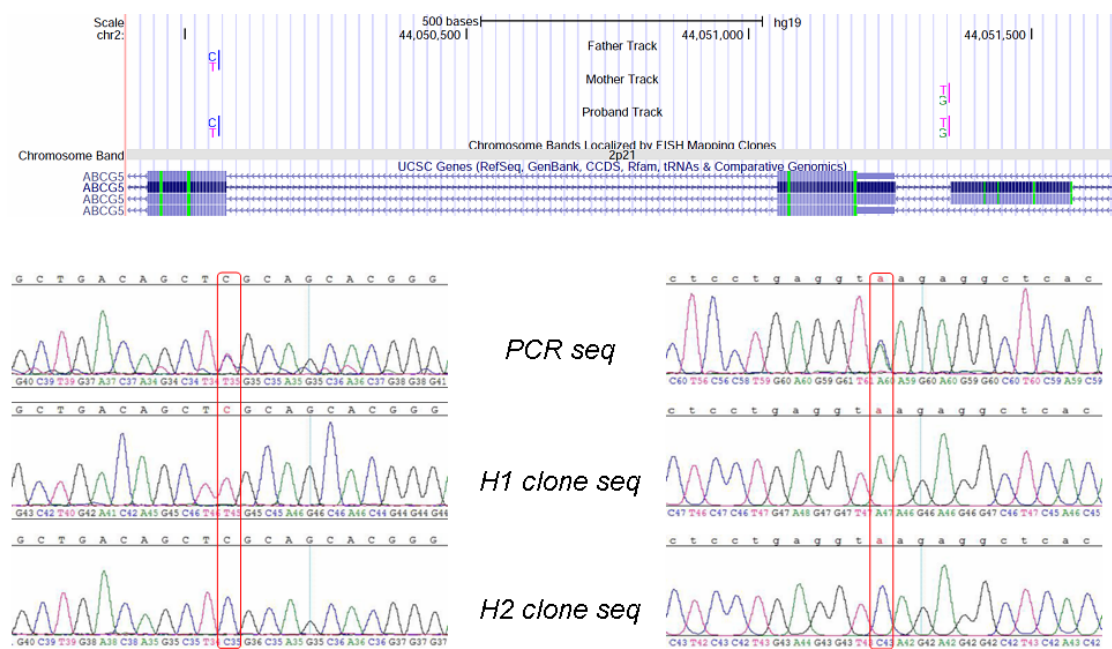


Figure S2. The size distribution of platelets (PLT) and the blood smear for the I-3, I-4, II-2 and II-3. The dash lines demonstrate the volume for the platelets, and the double dash lines show the 30 fL.

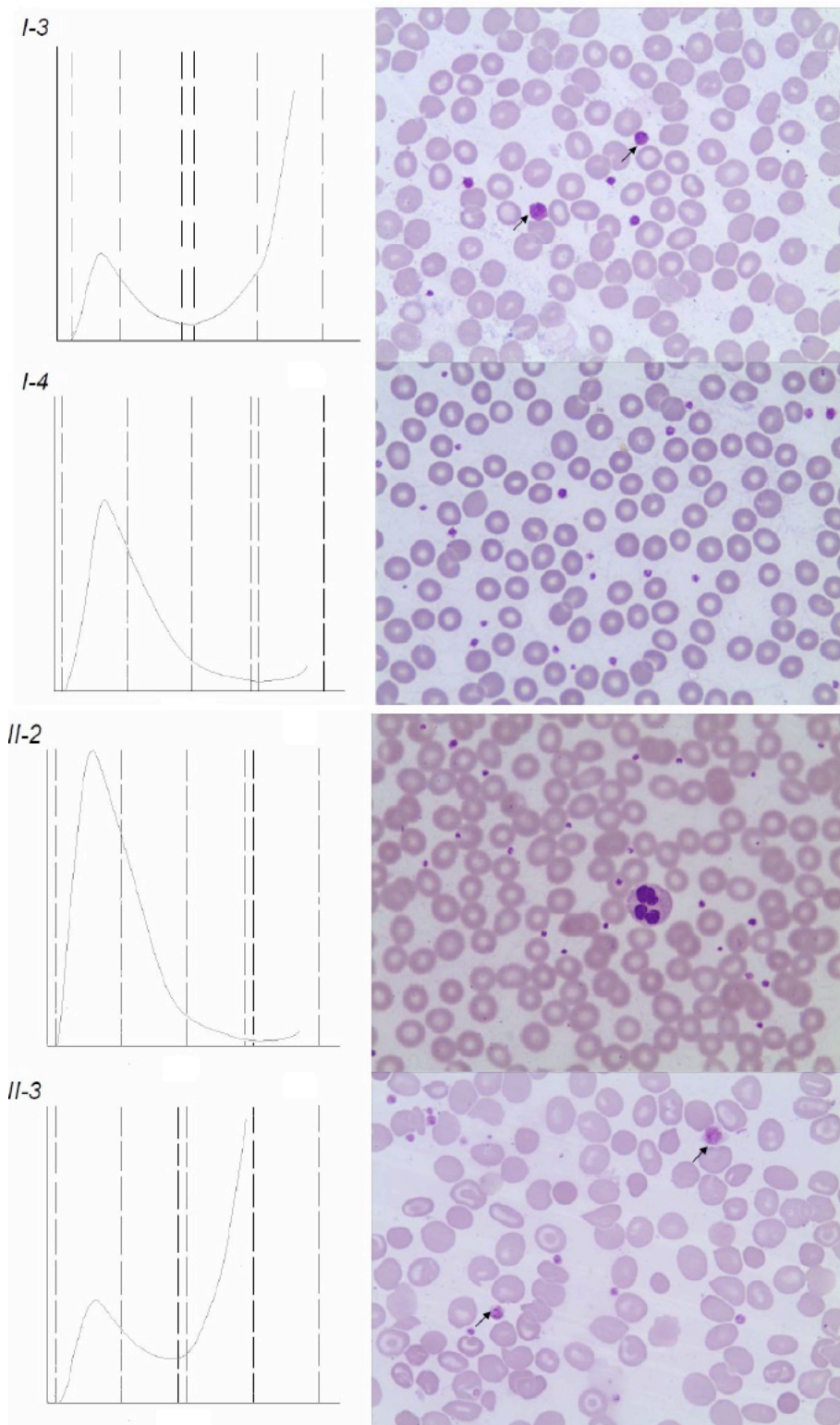


Figure S3. Monitoring of the platelets for the II-4. (A) Comparison of platelet size distribution before and after treatment. (B) Disappearance of giant platelets in blood smear for the II-4 after the treatments.

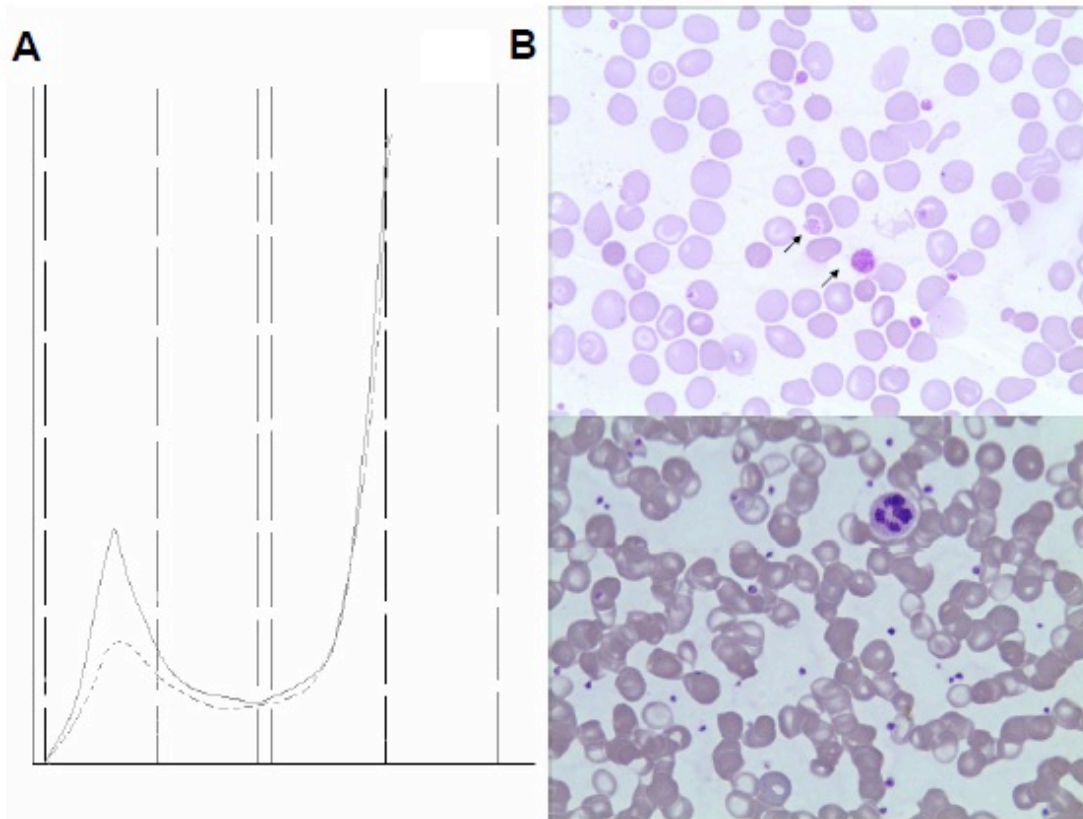
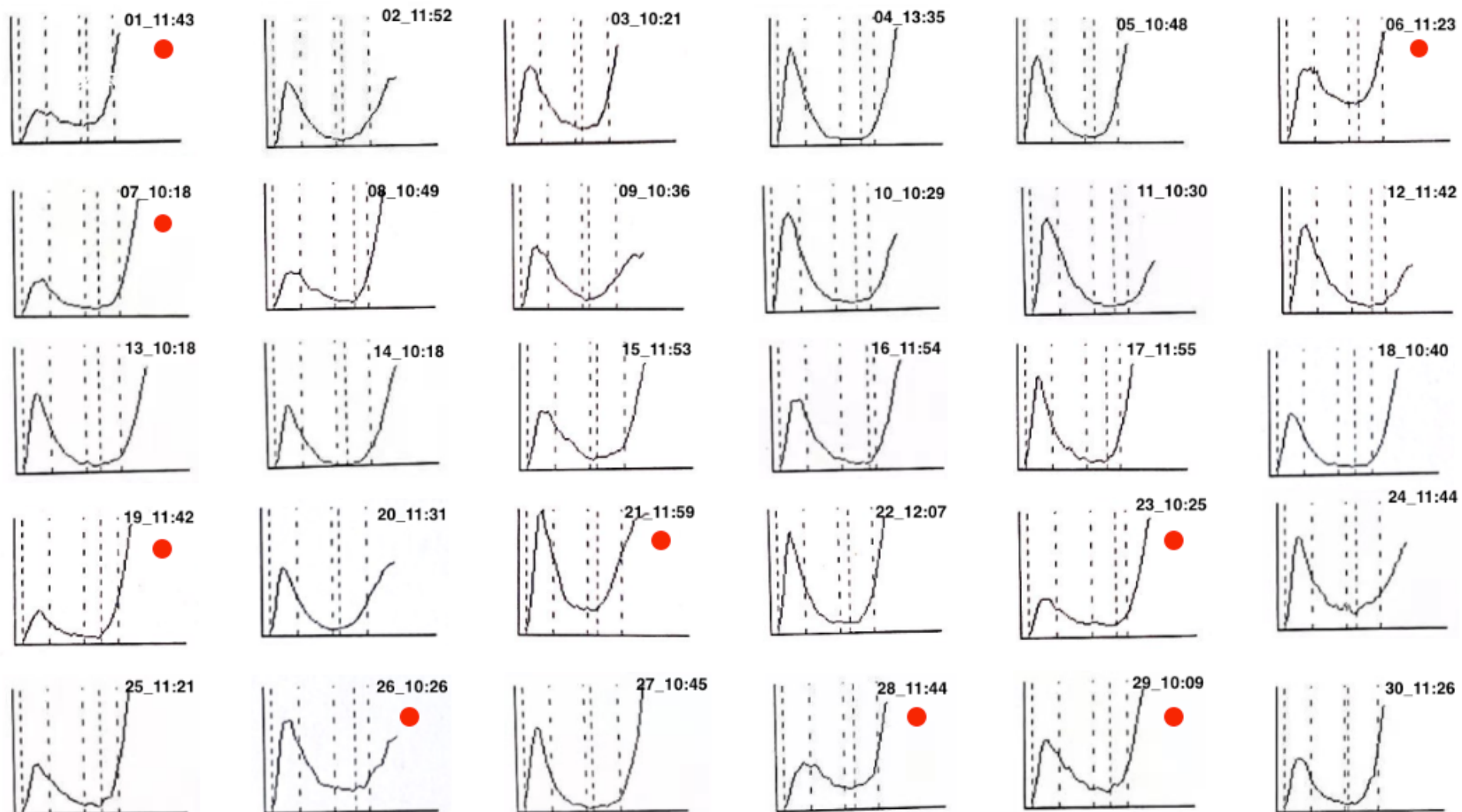


Figure S4 The size distribution of platelets (PLT) for 30 individuals with giant platelets.



Red dots indicate the cases for which instrument could not export values for all parameters of the platelets

II. Supplementary Tables

Supplementary Table 1. Clinical Characteristics of the Family Members

	ID									Normal Value of Rang
	I-1	I-2	I-3	I-4	II-1	II-2	II-3	II-4	III-1	
Gender	Male	Female	Male	Female	Female	Male	Female	Male	Female	
Age	69	66	58	52	45	40	34	32	9	
TC	169.76	213.46	155.07	200.31	212.69	258.7	134.96	97.84	638.06	<200 mg/dl
LDL-C	99	129.93	96.29	123.74	124.9	163.19	71.54	50.66	527.46	≤120 mg/dl
HDL-C	49.88	61.49	43.7	51.43	68.83	67.29	53.75	39.06	45.63	50-60 mg/dl
β-Sitosterol	1.06	1.44	1.58	1.14	1.3	1.23	1.52	1.49	3.26	0.31~0.80 mg/dl
PLT	275	324	172	298	363	240	250	217	289	85-303 10 ⁹ /L
PDW	10.5	10.9	NaN	15.4	11.4	12.1	NaN	NaN	15.7	11~26.5 fl
MPV	10.2	10.3	NaN	12	10.6	10.8	NaN	NaN	12.1	7.6~13.2 fl
P-LCR	25	26.1	NaN	39.2	28.5	30.1	NaN	NaN	41	13~43 %
PT			11(9-13)				9.4(9-13)	10(9-13)	10(9-13)	
APTT			29(20-40)				26.8(20-40)	28(20-40)	27.3(20-40)	
TT			17.1(14-24)				15(14-24)	16.7(14-24)	15(14-24)	
FIB			3.1(2-4)				3.84(2-4)	3.62(2-4)	3.6(2-4)	
PTA			120(70-150)				106.8(70-150)	117(70-150)	116.5(70-150)	
PAgT			61(35-75)				65.7(35-75)	63(35-75)	64.3(35-75)	

Total cholesterol, TC; Low density lipoprotein cholesterol, LDL-C; High density lipoprotein cholesterol, HDL-C; Platelet count, PLT; Platelet distribution width, PDW; Mean platelet volume, MPV; Platelet-large cell ratio, P-LCR; prothrombin time, PT; activated partial thromboplastin time, APTT ; thrombin time, TT; fibrinogen coagulative time, FIB;p rothrombin time activity, PTA; platelet agglutination test, PAgT.

Supplementary Table 2. Primers of customized Gene Panel Test for 4 common FH causal genes

Amplicon_ID	Ion_AmpliSeq_Fwd_Primer*	Ion_AmpliSeq_Rev_Primer*	Symbol	Chr	Amplicon_Start	Amplicon_Stop
AMPL3677489788	GGTGGTTACATCCAGCTTTAGGAA	CCTGAATGCTAACACTAAGAACCAGA	APOB	chr2	21228658	21228877
AMPL4702519983	CTTGGTTTTCTTCAGCAAGGCTTT	CCCTTTGGTGCTCTGATTAGAGATTAAG	APOB	chr2	21263842	21264053
AMPL4702563407	CAAGGGCAGACAGTGGCTAT	AGTCCTTCACAGGCAGATATTAACAAAA	APOB	chr2	21251109	21251326
AMPL4702569339	AGGGTATCCACCAAGGCTCT	GAGGCTGAATTGTTTTGGGAACATAA	APOB	chr2	21241887	21242066
AMPL4702572764	GCTGATCCCTCGCCATGTT	CTTTCTTCTCAGCCCCATCACTTT	APOB	chr2	21255269	21255465
AMPL587079551	GCAAATGATGAAGTTCTCAGCTTCTTAT	CATTATTCCTGGGCTGAAACTAAATGA	APOB	chr2	21228401	21228550
AMPL587080662	TGGTGAATGGAGACACTTCAACATT	GCTGTGCTTTGTGAGTTTATCAGTCA	APOB	chr2	21229947	21230169
AMPL587110603	TGCATCAATGACAGATGAAGATGAAGAA	CCAGTCCTTCATGTCCCTAGAAATC	APOB	chr2	21229612	21229827
AMPL587118187	CATAGAGAGAAATCTTTCAGTGGAGGAG	TCTGCTGGAAACAACGAGAACAT	APOB	chr2	21230282	21230433
AMPL587123574	AGTCAGTGCTTGAAAGGAAGGAA	GATAACCGTGCCTGAATCTCAGT	APOB	chr2	21228088	21228304
AMPL587126935	GGATGCAGTACTACTTCCACTTTTGTTA	CAACTGCAGACATATATGATACAATTTGATCAG	APOB	chr2	21232999	21233223
AMPL587127039	TGATTTTGGCATCATCTAATGCAATTTGT	CCCACAGCAAGCTAATGATTATCTGAA	APOB	chr2	21233244	21233386
AMPL4702574822	GCATAGCTCACCTTGACATTGTA	CCACGAATGTCTACAGCAACTTG	APOB	chr2	21236021	21236172
AMPL4702588225	TGAGTAGATTTTCCAGCAACTATGTGG	CAGGGAAAACCTTCAACATGGAAAG	APOB	chr2	21257647	21257848
AMPL4702597728	GGAAC TTGACCATTAACCCAGTACAAA	CTGATGCCATTTTCTTGTCTGATTTTCT	APOB	chr2	21249724	21249871
AMPL4702603581	CACATTGCCCTTCCTCGTCTT	GATTCCTTCTGCTTTTCTTTACGAT	APOB	chr2	21260068	21260231
AMPL4702622694	GAGGTCATGGGAGACTGGCAAA	TCCCACATTGGGAAGTAAAGG	APOB	chr2	21247865	21248068
AMPL4702622904	GGCTGGTGTCTAACAGTCTCT	AATATTGTTGTGAAGGTCAGAGGCA	APOB	chr2	21236288	21236489
AMPL4702631687	GTGCAGTAATTCAGGCCAGGAA	GCACCCTGAGAATTTGTGATGTC	APOB	chr2	21242664	21242851
AMPL4702639864	CAGTGTGTACTGACAGGACTGG	TCCTGCTCCCTTCTACCTTGAA	APOB	chr2	21258527	21258667
AMPL4702643442	CAGACTTGGCTGAAAGAATTACCC	GAGTTTGTGACAAATATGGGCATCAT	APOB	chr2	21245681	21245875
AMPL4702734718	TCTGTGATCCAGGAGTCTATTAGCA	CCAAAGTAATGCAGGTGTTGAGAC	APOB	chr2	21237996	21238211
AMPL4702739643	GCATCCTCCATACCTTGACAGTTG	CCCCTCTCTGCCCAGAATTG	APOB	chr2	21265220	21265425
AMPL587139369	CCTCTCAAAATATTCTTGCAAGGTCTCA	GACCTAACTCTACTAGACTCCCCAAT	APOB	chr2	21233491	21233694
AMPL4233140585	CTGTAAC TCTTGAGAAGCTTCTGAA	AAGATAACATTAACAGCTGAAAGAGATGAAAT	APOB	chr2	21225169	21225362
AMPL587144133	AAAAGCACTCTAACATCAATAGCCTCAA	GCATCCTGGATTCAAAATGTGGATACTA	APOB	chr2	21232847	21233004
AMPL587156123	GCTAAGCTTGTGGTCAACTGCT	AGTACTGTGAGCTTAACCACGAAAAAT	APOB	chr2	21229294	21229506

AMPL587182477	TGTTCACTGTTTGTGTGGTCAA	GCTTCAGGGAACACAATGCAAA	APOB	chr2	21234465	21234674
AMPL587201708	CTTCATATCTTCCTGTTATCTGTTGGT	GCTTCAGTTCATTTGGACTCCAAAA	APOB	chr2	21235125	21235341
AMPL587214973	CTGGGTCAAGTGATGGAAGAGA	GGGTGCAAGAGTCAAAGGATAAT	APOB	chr2	21250816	21251001
AMPL587228279	CCAATCCTTAGTGTCGCCTTGT	AATGGGAAGTATAAGAACTTTGCCACTT	APOB	chr2	21234797	21235017
AMPL587239776	ACCTCTGTGGTCTTGAGAGACT	CATAAGGGGAGGTAGGGACGGT	APOB	chr2	21225410	21225634
AMPL587243150	ATTCTTGAGTAACTCGTACCAAGCC	GGGCCATTAGGCAAATTGATGAT	APOB	chr2	21225750	21225941
AMPL587244175	CTTTGGTGCAGGTCCAGTTCATA	AACCACAGTCATGAACCCCTAC	APOB	chr2	21224438	21224662
AMPL587244614	TGTGCAGAAATTGCTCAACTTGAC	GGCTGGACAGTGAAATATTATGAACTTGA	APOB	chr2	21224958	21225111
AMPL587253207	TCTAGAGTCTCATCCCCCTAGTACCT	GTCCAAAGTACAAAGCCATCACTG	APOB	chr2	21252431	21252588
AMPL4702760901	TGAATGTCCAGGGTGAGTCTGTAA	CAAAACATAGCTTCTTACCACACATCTC	APOB	chr2	21239353	21239574
AMPL587253845	CCGTGATGTATATCAGAAATGTGTGGTA	CTTTCTGCCACTGCTCAGGAA	APOB	chr2	21224685	21224889
AMPL587271935	CCAGGAAAAGTGCTTCTGAAATGATGTA	GGGTTACCAAGTCCTGACTCT	APOB	chr2	21252718	21252925
AMPL587273518	CTTGGGCACGTTGTCTTTCAG	TTTTCTCTCTGTTTAGTCTCTCCAGAT	APOB	chr2	21226066	21226222
AMPL637590428	AAGTTGCTTACCGCCTGTCT	GGATATGGATGAAGATGACGACTTTTCTA	APOB	chr2	21227015	21227195
AMPL4653086575	GACAGACCAGGCTGACATTTTC	CCTCCCTCCTGAAAGGTGAAAC	APOB	chr2	21266388	21266529
AMPL637979996	TGTGGCGTAGAGACCCATCATAT	GCACAATTGGCCCAGAAAAATGG	APOB	chr2	21235455	21235634
AMPL1012820289	GGCAAAGAACTGTTCTACTTAGTAGCA	GAGTCTGCCAGTCAATAAATACATGGAT	APOB	chr2	21237715	21237936
AMPL1012976280	CCACTACTGGAGCTAGTGAGGTA	CAAGGCTGGAGTAAAAGTGAAGT	APOB	chr2	21246205	21246427
AMPL1226567828	TGACACTAGATTTTCTACAGTTTGGTTT	CTCAACCGTACAGTTCCTAGAATATGA	APOB	chr2	21227770	21227986
AMPL1498067538	GATTGATATCCAAATGGTCCCTGAGA	AGTCACATCTCTTTGCCACAG	APOB	chr2	21256066	21256209
AMPL1500364378	GGAAGTGCCTGGTGGTTCTTA	CATGCTGAAACTACCCTCACTG	APOB	chr2	21250587	21250754
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AMPL3676958730	GCTTTGCTTGATGTTCTCCGTT	AGTGAGGCCAACACTTACTTGAATT	APOB	chr2	21228980	21229197
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AMPL3677066071	GCTATCCAGGGTAAGCTGATTGTTTA	GAGTCAGTGAAGTTCTCCAGCA	APOB	chr2	21231085	21231264
AMPL3677151632	AGAACTTCTAATTTGGAATCTCCTTTGG	CTTCCCCACATCTCACACACAA	APOB	chr2	21231329	21231513
AMPL3677154866	CCCTGTAAACCTTAATGAAAAACGAAT	CTAAAGGCATGGCACTGTTTGG	APOB	chr2	21230542	21230729
AMPL3677216852	AAGCAGGGCACTGACTTTGT	CTGTAATGGCCCCGTTTACCAT	APOB	chr2	21233824	21234047
AMPL3677248777	AGGGTGATTCTCGCTAGAGGAAT	TCCACACCAGAATTTACCATCCTTAAC	APOB	chr2	21231617	21231804

AMPL3677250584	TTGAGGGAGCCAGATTCATAAACC	TGCCACATAGCATGGACTTC	APOB	chr2	21230795	21230992
AMPL3677272201	AAGCCTCCTGTAACCAATTGATGATTAA	ATTACCACCAGTTTGTAGATGAAACCAA	APOB	chr2	21232242	21232448
AMPL3677303433	GACAGCATCATCAATAAATCCAACCAATT	TCAGAGCCAAAGTCCATGAGTTAATC	APOB	chr2	21232537	21232730
AMPL4703594094	GAGCCATAAGCTGTAGCAGATGA	CAAATGGAACCTTCTGTTTAATCCTGCAT	APOB	chr2	21238274	21238498
AMPL4703615584	CTGGATATTTTGCTCAGAGATGGTTAGT	CAATTTCCAGCTTCCAGTAGACAAAGATA	APOB	chr2	21256266	21256469
AMPL4703636088	CCTTGCTGCTTTCTTCTTTTACCTTTTT	CATCTGGGAGTCTTCCTTATACCC	APOB	chr2	21237298	21237440
AMPL4703648744	AGAAATGATGCCCCCTCTTGATGTTC	CTTCAGTGCCACCCAGCTTA	APOB	chr2	21260881	21261061
AMPL4703666536	TGTGTTGAGCCGATGGCTAAA	ACCTGAAATACAATGCTCTGGATCTC	APOB	chr2	21234130	21234341
AMPL3677036261	CCATTTCCATGACCCTTTTCCAGAA	GAGCTTAGTAATGGAGTGATTGTCAAGA	APOB	chr2	21230947	21231138
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AMPL3677111451	CATGCTCCGTCTCAGGTACT	GGTATCGCAGCTTCCATCACTG	APOB	chr2	21231222	21231378
AMPL3677168109	AGCAAGAGTCCACCAATCAGAAA	AGCCTACAGGACACCAAAATAACC	APOB	chr2	21232069	21232293
AMPL3677196226	CAGTAACTCTGCCTTCCCTTCT	GCAAACACCTAAGAGTAAACCAAACTT	APOB	chr2	21230685	21230846
AMPL3677218487	AGCTTGCCAAAAGTAGGTACTTCAA	AGGGATCTGAAGGTGGAGGAC	APOB	chr2	21231467	21231660
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AMPL3677499003	CCGGACTTCATTTTTCCATCTGATCT	AGCTAGAGGGCCTCTTTTTTAC	APOB	chr2	21228826	21229025
AMPL3766439257	GGGTTCACTTTTAATACAGAGATGCAC	AGCAGCTGCGAGAGATCTTC	APOB	chr2	21255143	21255307
AMPL587069627	CATTTATTCCTACATGGGCCTCCATA	CATCCACAAACAATGAAGGGAATTTGAA	APOB	chr2	21230385	21230597
AMPL587074088	GGAGGCAGGATATTCTTACCATTTAGT	CTCTGTACCTGCTGGAATTGTCA	APOB	chr2	21227932	21228133
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AMPL587096945	TGCCTGTCAAAGGATTTGATGCT	ATGCGTCTACCTTACACAATAATCACAA	APOB	chr2	21230120	21230337
AMPL587113071	CGTAGGCATGACAAGAACTGAATTTAGA	CAGTCTATGACAAGAGCTTATGGGAT	APOB	chr2	21228496	21228707
AMPL587129104	CCTTGAAATCTGGAAGAGAAAGCTTGA	CCTGGATACACTGTTCCAGTTGT	APOB	chr2	21229776	21229995
AMPL587132430	TGAAACTTGCTCTCCCCAATTGAATGA	TGTTCACTCCATTAACCTCCCATTITTT	APOB	chr2	21233332	21233545
AMPL587161788	GCGTGAACTGGGACACAGTTA	GTGCAAACCTTGACTTCAGAGAAATACAA	APOB	chr2	21228261	21228460
AMPL4702558855	AAGGAAGATTATCTGCTAGAAAGCCA	TCTGTGGTTGACCTGCTTTCC	APOB	chr2	21235921	21236065
AMPL4702567381	CTGTATCACTTGTTAGTCAGCAGACAT	CTTCTTTGGTGAAGGTAAGAGTTTCTGT	APOB	chr2	21257577	21257701
AMPL4702591921	CCCACTGAACCAAGGCTTGT	TGATTAAGGGCTCTCCTGGCA	APOB	chr2	21255421	21255549

AMPL4702597715	TGTTCCCATGGTAGAATTTGGACAA	GCTAGCAGACAATGCCAGTGT	APOB	chr2	21251274	21251495
AMPL4702600052	AGGAGGCGGACCAGTTGTA	GCAAATCCTCCAGAGATCTAAAGATGTT	APOB	chr2	21236131	21236337
AMPL4702600811	TTTTCTTCCAAGCCAATCTGAGAA	TGGTATTCAAATACTTGTTTAGATAAATGGACA	APOB	chr2	21249819	21249945
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AMPL587200225	GCCGTAGTTTCCCATTTGTTGGT	TGATGGGCTCATATGCTGAAATGAAAT	APOB	chr2	21234294	21234515
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AMPL587207454	GTCAGCTGAAGCAAATCCAAAGG	CTCGGAACCTATCAACTCTACAAATCTGTT	APOB	chr2	21250709	21250866
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AMPL587237827	CGCAATGGCCTGGCTTTTAATTAT	CCTCTAACTTTACTTCCCAACTCTCAA	APOB	chr2	21224845	21225008
AMPL587238021	TGAAGATTGTGTTGATCTCATCTTGAT	AGCCCAAGAGGTATTTAAAGCCATT	APOB	chr2	21225282	21225457
AMPL587244227	GACTTTCGAATATACCTGGGACAGT	GGGACTCAAGGATAACGTGTTTGAT	APOB	chr2	21225589	21225799
AMPL587251003	GTTCTCCTGGAGCAAGCTTCAT	CAAAAGATTGATTGACCTGTCCATTCA	APOB	chr2	21224619	21224743
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AMPL637430943	AGAAGGCTAGTGACAGGGTCT	TGGTCAGTTTGCAAGCAAGTCT	APOB	chr2	21242483	21242707
AMPL4702608977	GTACAAGCATCTTTTCGGGCTT	TCTTGGGAGAGGAGCTTGGT	APOB	chr2	21247694	21247905
AMPL4702615695	TCCTCAGGACCTGAATGATCTCAA	CAGTCAAGCTGCTCAGTGGA	APOB	chr2	21245592	21245724
AMPL4702625136	GCCTGGAACAGAGCACTTGAG	TTGTCAACTCTGATCAGCAGCA	APOB	chr2	21258375	21258571
AMPL4702659253	CTCCTAGCGAAGTCCGGAATG	AAATCTTTGCCCGGAGGTGA	APOB	chr2	21245829	21246009
AMPL4702694445	ACTGAAGTCCTTCATATTTGCCATCTTC	GGTTAGGATAGAATTCTCCAGTTTTCA	APOB	chr2	21227431	21227638
AMPL4702714942	AACATGGCTTGGTGAGGTATGAA	TGAATCTACTGAGGGCAAAACGTC	APOB	chr2	21239217	21239400
AMPL637944367	GACCCTCAACTCAGTTTTGAATATGGT	CCATAAAACAAACATGAACCTGACAT	APOB	chr2	21226159	21226368
AMPL4233139900	AAATTTACACGGATATGATAGTGCTCATCAA	ACAAATTGCATTAGATGATGCCAAATCA	APOB	chr2	21233075	21233272
AMPL1012216917	GGCTGACTTACAAACAGAGGCA	GATTCAAGAAGTGCCACCAGGAT	APOB	chr2	21265056	21265265
AMPL1012880407	CTTCCTGACAGGGTTGGTGTT	CCACATCCCAGAAAACCTCTTCTT	APOB	chr2	21237128	21237349
AMPL1013060206	AGTATATTTTGAGCTGACACACCATGTT	GCTTCTGAAACAAACACTTGTCAAAT	APOB	chr2	21250951	21251109
AMPL4567386395	TCCATGTATTTATTGACTGGCAGACTC	CCGATTATCCTAAGAGCTTGCATATGTA	APOB	chr2	21237910	21238048
AMPL1498159057	CATTAGATACCTGGACACCTCAATCAG	TTTGAAGACTCTCCAGGAACTGAAAAA	APOB	chr2	21256161	21256320
AMPL4567917755	TGTCTCTTAAGCTGCTGCAGTT	AAAAACAATCCATGATCTACATTTGTTTATTGA	APOB	chr2	21232925	21233071
AMPL4652891859	GGTGCCCACTAGCTCAAAAGTT	CTGCTCCACTCACTTTACCGT	APOB	chr2	21259887	21260110

AMPL1637061913	CAGGTTCTTGATCAGACTGACTATCTTT	AACGAGCTTCAGGAAGCTTCT	APOB	chr2	21225052	21225201
AMPL4652934767	CTGGTGCAAACACACAAGTTCA	GGTGTATGGCTTCAACCCTGAG	APOB	chr2	21263737	21263890
AMPL4652943930	TTTTGAGGACTTCCATGCTTAGAAA	GATTTTTCCACAGACAGTGTCAACAAAG	APOB	chr2	21249574	21249778
AMPL4653069015	GCCATCTCAGCCCTGTAGAGT	CCTGTCTTACAGAAGAGGAAATGCTG	APOB	chr2	21266285	21266435
AMPL4703586227	TGTGAATGGTACTAGTTCAGCCTGTA	CCAAACTGCTTCTCCAAATGGAC	APOB	chr2	21238155	21238319
AMPL4703597808	GGATGGCAGCTTTCTGGATCAT	AGCACTTTTCTGGACACCTTTTA	APOB	chr2	21252543	21252731
AMPL4703623413	TCATGGAGCTGACTCAGTGATCT	CGGAGAAAGATGAACCTACTTACATCCT	APOB	chr2	21260726	21260933
AMPL4701683945	GCAAACAGAACTTTACGTTGGCT	GGTCTACAGATTGGAGAGGTCATC	APOB	chr2	21246381	21246573
AMPL3676976464	CATGTCTTCTCCTCATGAATTCTGAA	CCTATGAGCTCCAGAGAGAGGAC	APOB	chr2	21241791	21241929
AMPL3677020586	ACTGAAGACCGTGTGCTCTTG	AATGCTGTACTCTACCGCTAAAGG	APOB	chr2	21229151	21229339
AMPL4703642609	CTATTGAGGTGGTCTTGCAAAGTCT	GGTATTTTTGCCTAATGTTTATTGCTCT	APOB	chr2	21237392	21237543
AMPL4703654789	CCATTTGTATGTGCATCGATGGT	GCTAAGGTTTCAGGGTGTGGAG	APOB	chr2	21234002	21234171
AMPL413502387	CCCTGACTCCGCTTCTTCT	CGCAGAAACAAGGCGTGTG	LDLR	chr19	11231020	11231223
AMPL413542372	ATTCTTTAGTTGGCAGGAAATAGACACA	GGACTCACAGCACGTCTCC	LDLR	chr19	11210808	11211029
AMPL413576067	CGGCGAAGGGATGGGTA	GCACTCGTAGCCGATCTTAAGG	LDLR	chr19	11221268	11221401
AMPL413629258	CGAAGATGGCTCGGATGAGT	CATACCGCAGTTTTCTCGTC	LDLR	chr19	11216101	11216281
AMPL413640942	TGAGAAGTAGGTGGCCTCCA	TGCTTTGGTCTTCTCTGTCTTTGAATAA	LDLR	chr19	11241881	11242080
AMPL413738884	GCCTGAATGGTGTGGACATCT	GAAACCTTCAGGGAGCAGCTT	LDLR	chr19	11226820	11226996
AMPL3989014358	TCAGTGGGTCTTTCCTTTGAGTG	CGTTGTGCGCAGTCCACTTG	LDLR	chr19	11213291	11213441
AMPL3989324171	AAAGGCCCTGCTTCTTTTCTCT	CCAACCTTCATCGCTCATGTCCTT	LDLR	chr19	11217171	11217352
AMPL3989544606	GGTGGCCAGCAATAGAATCTACT	TGATGACGGTGTCATAGGAAGAGA	LDLR	chr19	11224072	11224260
AMPL3989689733	TTCCCGTTGGGAGGTCTTTTC	TGTA CTGCTCCGGTCCA	LDLR	chr19	11223802	11224025
AMPL3989797055	CGCAAGGCGATCTCTAAACAAA	GTCAAAGTTGATGCTGTTGATGTTCT	LDLR	chr19	11240052	11240271
AMPL3989824420	CGTCATTAGGCGCACACCTAT	GTGTCCTTACGGCTGTGGAG	LDLR	chr19	11233776	11233924
AMPL4055019071	CACAGACTTGGGAAGTTCTCCAA	CAGAGCCCTCACGCTACTG	LDLR	chr19	11238547	11238742
AMPL4137497464	AGCGTCCCCGGCTATAGAA	GGTCTGAGTCACAGACGAACTG	LDLR	chr19	11215817	11215976
AMPL4137530416	GGAATCAGAGCTTCACGGGTTA	AGTGTCCCGACCCGGAT	LDLR	chr19	11199980	11200204
AMPL4703442186	CTTCGAAGGTGTGGGTTTTGG	CCTTCCTCACACTGGCACTTG	LDLR	chr19	11222096	11222274
AMPL4703449287	GAGAGAGGGTGGCCTGTG	ACAGTAGGTTTTACGCCAACAAGTT	LDLR	chr19	11230699	11230864
AMPL4702062301	TCAGCACGTGACCTCTCCTTA	CTCGTACGTAAGCCACACCT	LDLR	chr19	11227497	11227692

AMPL4703470320	GGACTGGATCCACAGCAACAT	TCCTGAAGCTCCTTCCTGCT	LDLR	chr19	11224295	11224500
AMPL4702067023	AATCCATTTGCATGCGTTCTTATGT	CAGTCTCTAGCCATGTTGCAGA	LDLR	chr19	11217957	11218155
AMPL413600486	CACGATGGGAAGTGCATCTCT	GGAACACGTAAAGACCCCTACA	LDLR	chr19	11215931	11216153
AMPL413602187	CTAGGACACAGCAGGTCGT	TCCCAGGGATGGAGTGATTATTTGTA	LDLR	chr19	11200168	11200392
AMPL413639801	GAATGCATCACCTGGACAAAG	CCTACAGCACTCATGTCTCAGT	LDLR	chr19	11218112	11218263
AMPL413643442	TGAACCTGGAGGGTGGCTA	GTGCAAAGTTTCAAGGATGAACTC	LDLR	chr19	11222235	11222401
AMPL413649256	CCCGACTGCAAGGACAAATCT	CGGCACCTAAATCACTGCATGT	LDLR	chr19	11216240	11216365
AMPL413662091	GGGTTCCCAGCAGGACTATTT	TGTTTTCACTACCAGCGAGT	LDLR	chr19	11226646	11226861
AMPL414667929	TCAGTTCTGGAGGTGCGATG	ACCCCGTAGAGACAAAGTCAGA	LDLR	chr19	11213401	11213536
AMPL4702074984	GCTGGCAGAGGAAATGAGAAGAAG	TGTCCAGGAGAAAAAGTGAACAGG	LDLR	chr19	11238698	11238861
AMPL4702081921	CAGTGTGACCGGAATATGACTG	CCCTCTGGCTTCACAAATCATTT	LDLR	chr19	11217306	11217441
AMPL4702091595	CCTTCTCCTTGGCCGTCTTTG	CCTCCTAGTCACAACCAAGTTTCTG	LDLR	chr19	11227652	11227777
AMPL4702110420	CACCGTCAGGCTAAAGGTCAG	CATCTCGTGACCAAAATGTTCTG	LDLR	chr19	11233884	11234102
AMPL3988950525	CCAGGATGGCTCTGATGAGT	GCCGCCATCATCAAAAAGGG	LDLR	chr19	11210987	11211210
AMPL3989538556	GCACGAGGTCAGGAAGATGAC	ATCATTCTCTGGGACAGGTCAGA	LDLR	chr19	11223985	11224119
AMPL3989604425	CATGAGGAGCTGCCTCACA	GTCAAGCCCGGTGCTGAT	LDLR	chr19	11231179	11231360
AMPL4054977229	TGTTCCCACGTCTGCAATGA	GTGGAATCTCATGAAACCTCCTA	LDLR	chr19	11221360	11221580
AMPL4055022853	CTCTCCAGGTGCTTTTCTGCTA	AGGAGATCTAGACACACAAGTGGA	LDLR	chr19	11227406	11227541
AMPL4137467001	CTATGGAAGAACTGGCGGCTTA	GTCCCTTGAGGATCATATGCCT	LDLR	chr19	11240224	11240421
AMPL4703455705	AATGATCTGCAGGTGAGCGT	AGGACAGAGTCGGTCCAGTAG	LDLR	chr19	11224114	11224336
AMPL4703467241	GCCTCACAGGTTCCGATGTC	ACCAGAAGATTCCAGAAATTTCCAGAAC	LDLR	chr19	11230820	11230997
AMPL4401981955	CTGGGATGGAGCTGCAGAA	CAGCCTGTGCCTGAAAACAG	LDLRAP1	chr1	25889004	25889144
AMPL871354742	GCTCTGCCTTCCAGGCTTG	CGCAGCAGGTGGCTTTG	LDLRAP1	chr1	25893325	25893546
AMPL871359545	GTGGAGTGGGAATAGCAGGTT	CAAACACCTTGTCGTGCATCT	LDLRAP1	chr1	25883531	25883687
AMPL1417508083	AGCCATTAGTCAGGTTCTCACTCT	ACTGTGAGGGATCCGGTCA	LDLRAP1	chr1	25889962	25890152
AMPL1417508102	GGTTGTGTGGCCAGCAGAT	TGGGAGCCAAGCCTGTTG	LDLRAP1	chr1	25890289	25890490
AMPL1500485340	TCCTAGGAAAGAAGGCTGGTG	ACTAGCGTCATGCCAGGTA	LDLRAP1	chr1	25880328	25880497
AMPL3676940843	TGGATGAAGCGTTTTTCAGGT	TCTGAGTGGCTTGTGTTCTTAATCAG	LDLRAP1	chr1	25891680	25891806
AMPL4702066737	GGGCTTAGGAACAGTGAAGTGT	GAGGTTGTCTGTCAGGATAATTCCC	LDLRAP1	chr1	25881266	25881428
AMPL4702080257	CTGAAACTGCCCTTGAGGT	GCTGGCTTTGTCCCTCTTCT	LDLRAP1	chr1	25889452	25889586

AMPL4702068190	GGGATGCTGTTCAGCCTCAAG	CAACTGGAGTAACAAGCTGGGA	LDLRAP1	chr1	25880457	25880647
AMPL871344379	GCTGATCTCCCACTGACAACC	GAGGTCAGGCTGAGGATCAC	LDLRAP1	chr1	25890113	25890329
AMPL871380385	GATCTCCTATTGCACAGCAGACA	GTAATGGTCACCTTTGGGTCCT	LDLRAP1	chr1	25883644	25883865
AMPL4401988444	GGAGGTGCTTTGATCTGAGGTTA	CTGAGGGTTTGTCCGAGACTG	LDLRAP1	chr1	25893180	25893366
AMPL4401996594	CCTCCCCATCCCCACTTC	CCACAATGTAGATGCAAAGTCTCAC	LDLRAP1	chr1	25889107	25889232
AMPL4402007081	GCCTTGGTCCTGCAGAGAAAG	GAAAAACCAAGAGGCTCTCCCA	LDLRAP1	chr1	25889546	25889748
AMPL4402050903	CAGAAGGTGACTCTGAAGGTGTC	CCCAAGTGGCAGAGTGGAT	LDLRAP1	chr1	25881375	25881506
AMPL1417508234	GCCCTGACACGTGATGGAC	AGCATAAGCCAGAAGTCACTGAATAAAT	LDLRAP1	chr1	25893511	25893720
AMPL3676917086	GCTTGTGTCTGAGTCCCTGTA	CCAGCACAGGAAGCTAGCATT	LDLRAP1	chr1	25891557	25891721
AMPL413508536	TTGCTGGGTTTCTTCCATGTCA	CTCATCTTACCAGGAAGCCA	PCSK9	chr1	55509468	55509688
AMPL3712526399	GCCAAGGTGCGGGTGTGA	TCCCCAAGATCGTGCCAAAG	PCSK9	chr1	55505712	55505864
AMPL413519077	GAAC TTCAGCTCCTGCACAGT	CCGTCCTCGTCCTCCTG	PCSK9	chr1	55505397	55505617
AMPL3712549622	GAGCGGATTACCCCTCCAC	GGAGGTGCTGAGTCCCAAA	PCSK9	chr1	55512271	55512440
AMPL3713799125	CTTAGAGGTGTGAGAGGAGGCT	AGACAGCATCATGGCTGCAAT	PCSK9	chr1	55523514	55523731
AMPL414595636	GCCTACGCCGTAGACAACAC	GACAAAGTCGGAACCATTTTAAAGCTC	PCSK9	chr1	55529117	55529331
AMPL1012467495	CCAGATGAGGAGCTGCTGA	GCTGGAGTCTGGAGGATGGA	PCSK9	chr1	55524252	55524446
AMPL1013168806	GCCTGATCAAGGAGCGAGAAAA	CCTCGATGTAGTCGACATGGG	PCSK9	chr1	55512010	55512229
AMPL1494547002	GCTCAACTGCCAAGGGAAGG	ACAGCATTCTTGGTTAGGAGACATTAG	PCSK9	chr1	55518421	55518570
AMPL4703355908	GTCTACGCCATTGCCAGGT	GGATCACACTTGTGAGGACCAA	PCSK9	chr1	55525213	55525428
AMPL4701788825	AGGCCTGAGTCTGCCTCT	GCTGGAGGCACCAATGATGT	PCSK9	chr1	55522933	55523126
AMPL4701791645	GTTGAGGCAGAGACTGATCCAC	GGGAGAGACTGTCAAGGTCACA	PCSK9	chr1	55523758	55523955
AMPL4701827905	CAGAGTGACCACCGGGAAATC	CCCAGGAGTACAGCTGCAA	PCSK9	chr1	55517995	55518185
AMPL4701829390	CTGTGCTGAGGCCACGA	GTGGCACAACTGACACAGAAAAG	PCSK9	chr1	55527096	55527308
AMPL3677194655	CAGGCCTGGAGTTATTTCGAAAA	ACCTACCTCGGGAGCTGA	PCSK9	chr1	55521663	55521868
AMPL413521968	CCTCATGGGCACCGTCA	CCCTACACCCGCACCTT	PCSK9	chr1	55505507	55505731
AMPL3677193311	GGGAGCAGGTCTCCCCAA	GTTGAGGACGCGGCTGTA	PCSK9	chr1	55521620	55521760
AMPL414611490	GACGATGCCTGCCTCTACTC	CGACTCCTTCCAAAGCCAGAA	PCSK9	chr1	55521824	55522014
AMPL414643158	ATGTCTTCCATGGCCTTCTTCC	ACTAAGCACAGTCCCCAGTGATAT	PCSK9	chr1	55509646	55509783
AMPL3712540308	GCTTCTGCAGGCCTTGAAGTT	GGTATT CATCCGCCCGGTA	PCSK9	chr1	55512186	55512310
AMPL422385088	CATCCCAGGATGGGTGTCTG	AGGCACCCAGAGTGAGTGA	PCSK9	chr1	55529264	55529454

AMPL3713058928	GTTGACTTTATGCTCATTCCCTCCT	TCGAAGTCGGTGACCATGAC	PCSK9	chr1	55517901	55518044
AMPL423279419	CCTCCTCTCTCCTACCATGAACTA	CCACTCCTGGAGAACTGGAG	PCSK9	chr1	55524085	55524295
AMPL3713751478	CGCTGTGTGGACCTCTTTG	CGGCACAGACCCTGACTG	PCSK9	chr1	55523076	55523257
AMPL4137386932	TTGAGTTGATCCTGTCTAGTCCCT	GTGTGGACGCTGCAGTTG	PCSK9	chr1	55525077	55525268
AMPL4137395318	CATCACCATCTTTCACCATTCACC	TCATTGATGACATCTTTGGCAGAGAA	PCSK9	chr1	55523675	55523805
AMPL1012192084	CCTGAATGGCACATTTGAAAGTGTATA	CCCATGCAAGGAGGAACATGA	PCSK9	chr1	55509290	55509510
AMPL1012506113	CACTTTGGCCTCACAGAAGGAT	CCGGCTCCTGACTACACAC	PCSK9	chr1	55528941	55529155
AMPL4701756358	CATCCAGCCACCTGCTGATT	CTATGAGGGTGCCGCTAACC	PCSK9	chr1	55518248	55518464
AMPL4701822259	GGTTTCCTAGCTCTTGCCTCAGA	GCACTGGTTGGGCTGAC	PCSK9	chr1	55526937	55527130

Supplementary Table 3. Primers and PCR protocols for amplification of ABCG5 exons and splice regions

Primers for amplicons ABCG5					PCR Protocols					
Primer ID	Sequences (5'to3')	Amplicon (bp)	Exon (bp)	Flanking region (bp)	94°C	94°C	Tm	72°C	cycles	72°C
ABCG5-E1 L:	CTCTCCACCCGATCCACTAA	580	283	297	5 min	30s	56°C	45s	30	5 min
ABCG5-E1 R:	TCACTCTGTTTCCTGGAGCA									
ABCG5-E2 L:	AACACGTTAGGAGCCTGTCC	395	122	273	5 min	30s	56°C	45s	30	5 min
ABCG5-E2 R:	TTGGTAGCAGTTCCATTACACA									
ABCG5-E3E4 L:	CACAGAGGGTCTCGGGAAG	499	315	184	5 min	30s	59°C	45s	30	5 min
ABCG5-E3E4 R:	GAGTGACGAGCAAAGGGAAG									
ABCG5-E5 L:	TGTGCTGCCTCTTTCATGTC	299	133	166	5 min	30s	59°C	30s	30	5 min
ABCG5-E5 R:	AAAGGGCCCAAAGTATCTGC									
ABCG5-E6 L:	CTCAGGCTGAAAGCCAGTTT	382	140	242	5 min	30s	59°C	30s	30	5 min
ABCG5-E6 R:	CCTGGCCACTGGTACAAATC									
ABCG5-E7 L:	TGCATCAGTGTTCCAGAGA	282	130	152	5 min	30s	59°C	30s	30	5 min
ABCG5-E7 R:	CATCCAGGCAGAAGTCTGAG									
ABCG5-E8E9 L:	AGAGAGGGAACCTGGAGAGG	788	520	268	5 min	30s	59°C	45s	30	5 min
ABCG5-E8E9 R:	AGGTGGGCATTAACCACAAA									
ABCG5-E10 L:	GATGGGCAAAGTGTAGATCCTC	400	139	261	5 min	30s	59°C	30s	30	5 min
ABCG5-E10 R:	CCTCCTTCTATGACTAGGGAACC									

ABCG5-E11 L:	CCCACTCACCACAAAGGATT	390	186	204	5 min	30s	59°C	30s	30	5 min
ABCG5-E11 R:	CCATAACCACTATCAGTTCTCTGG									
ABCG5-E12 L:	TTTCTAGGCAACGGTATTTCTTT	526	113	413	5 min	30s	56°C	45s	30	5 min
ABCG5-E12 R:	CCAAGTGTCCTTACCTGTTG									
ABCG5-E13 L:	AAGCGCTTGGTAAATACTTGTT	1000	838	162	5 min	30s	56°C	60s	30	5 min
ABCG5-E13 R:	TGCCTATAAGTAAACTCAGCTGAAAG									

Supplementary Table 4. Summary of all reported cases with *ABCG5* deficiency

Proband	Homozygous/Compound heterozygous	Mutation Position	Nucleotide alteration	PMID	TC (mg/dl)	β -sitosterol (mg/dl)	stomatocyte	Megaloplastocyte
1	Homozygous	exon 6	R243X	11452359	NaN	> 8	YES	YES
2	Homozygous	exon 9	R419H	11452359	NaN	> 8	NaN	NaN
3	Homozygous	exon 9	R389H	11452359	267	18.98	YES	YES
4	Homozygous	exon 3	del exon 3	11452359	NaN	> 8	NaN	NaN
5	Homozygous	exon 9	R389H	11452359	NaN	> 8	NaN	NaN
6	Compound heterozygous	exon 9/exon 11	R419H/R550S	11452359	NaN	> 8	NaN	NaN
7	Homozygous	exon 8	R408X	11452359	NaN	> 8	NaN	NaN
8	Homozygous	exon 9	R389H	11452359	NaN	> 8	NaN	NaN
9	Homozygous	exon 9	R419P	11452359	NaN	> 8	NaN	NaN
10	Compound heterozygous	exon 3/exon 9	E197X/N437K	11668628	432	NaN	NaN	NaN
11	Compound heterozygous	exon 9/intron 12	R419H/IVS12+1G>A	11855938, 12777658	707.66	NaN	NaN	NaN
12	Homozygous	exon 3	del388-428/ins388-408	15375183	290.03	52.2	NaN	NaN
13	Compound heterozygous	intron11/exon4	IVS11+3insT/E146X	16029460	193.35	57.67	YES	YES
14	Compound heterozygous	intron11/exon4	IVS11+3insT/E146X	16029460	255.22	60.77	YES	YES
15	Homozygous	exon 2	E77X	16029460, 17785700	197.22	38.04	YES	YES
16	Homozygous	exon 2	E77X	16029460, 17785700	143.08	25.08	YES	YES
17	Homozygous	exon 2	E77X	16029460, 17785700	NaN	16.16	YES	YES
18	Compound heterozygous	exon 10/exon 11	R446X/H510D	17228349	224.67	14.6	NaN	NaN

19	Homozygous	exon 10	R446X	17976197	382.06	15.76	NaN	NaN
20	Homozygous	exon 10	R446X	17976197	246.33	12.77	NaN	NaN
21	Homozygous	exon 10	R446X	17976197	200.08	13.93	NaN	NaN
22	Homozygous	exon 7	F283Ffs5X	19111681	391.11	27.5	NaN	NaN
23	Compound heterozygous	exon 7/exon 10	F283Ffs5X/R446X	19111681	416.24	21.4	NaN	NaN
24	Compound heterozygous	exon 8/exon 9	Y329X/N437K	20521169	429.31	NaN	NO	NO
25	Compound heterozygous	exon 9/exon 10	R389H/R446X	20521169	708.82	7.1	NO	NO
26	Compound heterozygous	exon 9/exon 10	R389H/R446X	20521169	404.18	9.17	NO	NO
27	Homozygous	exon 9	R389H	20521169	643.47	7.07	NO	NO
28	Compound heterozygous	exon 9/exon 7	R389H/G269R	20521169	344.86	6.14	NaN	NaN
29	Compound heterozygous	exon 1/exon10	Q16X/R446X	20719861	1028.54	8.4 ^a	NaN	NaN
30	Homozygous	exon 1	Q22X	24166850	205.72	27.6	YES	YES
31	Homozygous	exon 1	Q22X	24166850	220.03	48.52	YES	YES
32	Homozygous	exon 1	Q22X	24166850	134.57	35.98	YES	YES
33	Homozygous	exon 10	R446X	24166850	220.03	57.54	YES	YES
34	Compound heterozygous	Intron 9/exon10	IVS9 + 2A>G/R446X	24166850	331.79	48.9	YES	YES
35	Compound heterozygous	Intron 9/exon10	IVS9 + 2A>G/R446X	24166850	143.85	29.64	YES	YES
36	Compound heterozygous	Intron 9/exon10	IVS9 + 2A>G/R446X	24166850	343.78	23.64	YES	YES
37	Compound heterozygous	Intron 9/exon10	IVS9 + 2A>G/R446X	24166850	338.75	61.93	YES	YES

38	Homozygous	Exon 9	R419H	24166850	315.93	68.91	YES	YES
39	Homozygous	Exon 7	g.712G> A	24166850	227.77	90.23	YES	YES
40	Homozygous	intron10	IVS10-1 G>T	24623560	NaN	12.8	YES	YES
41	Compound heterozygous	Exon 12/exon 9	P558Efs14X/R389H	25665839	525.83	NaN	NaN	NaN
42	Compound heterozygous	Exon 12/exon 9	P558Efs14X/R389H	25665839	NaN	NaN	NaN	NaN
43	Compound heterozygous	exon 1/exon 9	S44A/R389H	25665839	874.72	NaN	NaN	NaN
44	Compound heterozygous	exon12/exon 10	P558Efs14X/R446X	25665839	760.10	NaN	NaN	NaN
45	Homozygous	exon 10	R446X	26813946	300.00	20.20	NaN	NaN
46	Heterozygote	intron 1/exon 11	IVS1-1G>A/L511X	26892138	295.00	68.10	NaN	YES
47	Compound heterozygous	exon 9/intron 12	R419H/IVS12-1G>A	27170062	681.00	182.00	NaN	NaN
48	Compound heterozygous	exon 9/exon 9	R419H/R389H	27401767, 28203044	866.00	24.60	NO	NO
49	Compound heterozygous	exon 6/exon 10	Q251X/R446X	28521186	770.00	78.80	NO	NO

^aSitosterolemia was initially ruled out because of the normal sitosterol:cholesterol ratio, but after one year sitosterol level increased to 8.4 mg/dL.

Supplementary Table 5. Phenotype survey of the proband

Variables	Pre-study	6 Weeks	9 Weeks	14 Weeks	18Weeks	22 Weeks	26Weeks	34 Weeks	68 Weeks	75 Weeks	127Weeks	131Weeks	136Weeks	140 Weeks	Normal Value of Rang
TC(mg/dl)															<200
Value	638.06	491.11	398.30	266.44	286.16	221.97	247.49	324.83	286.16	294.28	193.35	222.74	154.29	141.53	
Change from prestudy		-146.95	-239.75	-371.62	-351.90	-416.09	-390.57	-313.23	-351.90	-343.78	-444.71	-415.32	-483.76	-496.52	
LDL-C(mg/dl)															<120
Value	527.46	406.04	343.39	187.94	208.04	156.61	182.91	243.62	172.85	157.00	109.82	136.12	71.54	51.43	
Change from prestudy		-121.42	-184.07	-339.52	-319.41	-370.85	-344.55	-283.84	-354.60	-370.46	-417.64	-391.34	-455.92	-476.03	
LDH(Iu/l)	225.70	201.70	182.20	206.20	199.40	200.10	189.70	185.70	154.90	228.80	176.60	NaN	183.40	204.70	80~190
CK-MB(Iu/l)	21.00	20.40	22.70	33.00	21.00	26.74	24.90	17.60	20.80	26.70	22.30	NaN	18.70	22.30	0~25
GPT(u/l)	20.20	34.20	5.10	6.45	5.05	5.29	5.30	5.50	3.60	6.70	5.30	NaN	5.00	5.70	5~40
AST(u/l)	41.50	56.00	27.50	29.67	29.73	28.16	28.20	31.30	25.70	29.70	23.70	NaN	27.60	27.30	5~40
ALP(u/l)	240.90	243.80	215.70	216.80	217.80	222.70	216.60	200.50	201.10	NaN	270.50	NaN	257.50	256.10	34~104

Lactate dehydrogenase, LDH; Creatinine kinase, MB isoenzyme, CK-MB; Glutamic-pyruvic transaminase, GPT; Aspartate transaminase, AST; Alkaline phosphatase, ALP.

Supplementary Table 6. Nine variations identified in 30 individuals with abnormal platelet distribution

hg19	Variant_ID	REF	ALT	Consequence	alt allele count	all allele counts	alt frequency	#homozygotes	Xinjian Cohort (patient ID)
chr2:44039633	rs4148195	G	A	c.*622C>T 3'UTR	36488	151740	0.24	5308	2,5,8,9,16,18,27
chr2:44039644	v3	T	TTAATCCTCAGT AAAATCACTGGG		-	-	-	-	23
chr2:44039650	v4	C	CTCAGTAAAATC ACTGGGTGCTCTGTA		-	-	-	-	14
chr2:44039839	rs2278357	C	T	c.*416G>A 3'UTR	33312	152020	0.219	4266	2,4,18,27
chr2:44039875	rs2278356	A	C	c.*380T>G 3'UTR	66283	151930	0.436	16244	4,5,8,9,16,18,27
chr2:44040401	rs6720173	G	C	p.Gln604Glu missense	32178	152082	0.212	3874	18
chr2:44050063	rs199689137	G	A	p.Arg446Ter stop-gain	19	152042	0.000125	0	30
chr2:44065090	rs6756629	G	A	p.Arg50Cys missense	10274	152086	0.0676	425	2,4,10,11,16,27
chr2:44065755	rs781098379	G	A	p.Gln22Ter stop-gain	-	-	-	-	28

Alternative allele count, all allele counts, alternative frequency and number of homozygotes are all from gnomAD database.

IV. Supplementary References

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