SUPPLEMENTAL MATERIAL

Methods

Data sources and study population

The individual level data will not be made available to other researchers without IRB approval from the VA.

Participants of multiple ethnicities were recruited from approximately 50 VA healthcare facilities across the United States.¹ Individuals consented to a blood draw and to have their DNA extracted for genomic profiling and linked to their full electronic health record within the VA. Both MVP biobank and this analysis were approved by the VA institutional review boards. Phenotypic measures

We obtained demographic variables such as age and sex from the MVP enrollment questionnaire and participants' electronic health record (EHR) data. We secured all measures of LDL-C performed in VA laboratories using standardized assays for up to 15 years. The MVP participants had a median of 11 measurements (1st quartile of 6 and 3rd quartile of 19) of LDL-C, with 4.4% having only one measurement of LDL-C. When more than one measure was available, we extracted the maximal level of LDL-C to approximate the most likely level of untreated LDL-C.² We also analyzed the single outpatient LDL-C value obtained closest to the time of enrollment into MVP during 2011-2016 and statin prescriptions going back to 2002. Evidence of statin use was obtained from prescribing and pharmacy records in the EHR. Genotypic measures and identification of FH variants

Blood samples drawn from consenting MVP participants were shipped to a central biorepository in Boston, Massachusetts, where DNA was extracted and shipped to two external genotyping centers for genotyping on an Affymetrix biobank array designed specifically for the MVP. Genotyping was performed at two sites using the same Affymetrix best practices pipeline including several quality control (QC) procedures targeting genotyping sites and batches. Genotype calling was performed on all samples together in batches grouped by site and sample processing date. Standard Axiom genotyping quality matrix (dish quality control (DQC) >0.95, QC call rate >0.97) were comparable across the batches and sites. Furthermore, an advanced marker and sample QC procedure was used to clean and harmonize genotype calls. Probe sets were analyzed for any inconsistencies across all batches and removed. QC metrics like the final sample call rates, non-missingness, minor allele frequencies, Ratio A Allele frequencies were all determined to be consistent across sites and batches. The MVP genomics working group applied genotype calling algorithms to the data in batches using the Affymetrix Power Tools Suite (v1.18). Standard quality control pipelines were used to exclude duplicate samples, samples with more heterozygosity than expected, or discordance between sex inferred by genotyping versus self-report. We also excluded related individuals (halfway between 2nd and 3rd degree relatives or closer) as measured by the KING ³ software.

We queried the ClinVar database⁴ (archive of June 2017) for "pathogenic" or "likely pathogenic" variants linked to familial hypercholesterolemia. We considered variants with conflicting interpretations of pathogenicity but at least one "pathogenic" or "likely pathogenic" assertion. These ClinVar variants were matched by unique identifiers (*i.e.*, rs ID) and chromosomal position to variants genotyped on the MVP biobank array. Additional annotation was obtained from the Human Gene Mutation Database⁵ to further understand the molecular and functional effects of selected FH variants.

Statistical analysis

We used ADMIXTURE ⁶ analysis with the 1000 Genome phase 3 samples and known

continental ancestries to assign genotyped individuals to European (>80% European Ancestry) and admixed African Americans (>50% African Ancestry). Global and ethnicity-specific principal component analyses were performed using flashPCA software.⁷ We then calculated carrier frequencies within each of two main ancestry groups.

We used linear regression models to estimate the effects of FH variants on untransformed LDL-C, adjusted for age, age² and sex for FH variants with a carrier count of 30 or greater (a carrier frequency of ~0.009%). For these variants, we considered associations to be significant if the p was < 0.05/the number of tested variants. FH variants with fewer than 30 carriers were combined into total burden and gene-specific scores (i.e., *LDLR, APOB* and *PCSK9*) to assess associations with maxLDL but they were not considered individually. All association analyses were performed in the R statistical environment version 3.2.5 (http://www.r-project.org/). Phenome-wide association analysis of FH variants with clinical outcomes

Genotyped MVP participants were included in the phenome-wide association study (PheWAS) if the electronic health record (EHR) reflected two or more separate encounters in the VA Healthcare System in each of the two years prior to enrollment in the MVP. We identified 277,531 veterans and 21,209,658 prevalent ICD-9 diagnosis codes were available for analysis. ICD-9 diagnosis codes were collapsed to clinical disease groups and corresponding controls.⁸ Diseases were required to have a prevalence of more than 400 cases to be included in the PheWAS analysis. Among participants with European ancestry, carriers of FH variants were compared to the other participants using logistic regression adjusting for age, sex, and ten principal components using the PheWAS R package.⁹ A total of 1,171 disease phenotypes were available for analysis. Additionally, we identified individuals with clinical CHD or peripheral artery disease (PAD) using inpatient and outpatient ICD-9 and Current Procedural Terminology codes available in EHR data from up to 15 years prior to enrollment in the MVP.

References

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Supplemental Table 1: Distribution of P/LP FH Variants (obtained from ClinVar database) among multi-ethnic MVP participants.

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Automation 11790000 1790 M00077 4100112 1700 1700 170 1700007 1700 1700007 1700 1700007 170000	AX-98075214 rs2	267607213	18780 NM 000527.4(LDLR):c.131G>A (p.Trp44Ter)	LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016)	19	11210962 19	11100286	3741	1
0.48607711 0.15607 0.6107 0.0007 0.	AX-98033604 rs:	121908041	18781 NM 000527.4(LDLR):c.137G>C (p.Cys46Ser)		19	11210968 19	11100292	3742	1
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A-6672771 print/2016 10112 10112144 10112744	AX-86712213 rs	749038326	245449 NM_000527.4(LDLR):c.268G>T (p.Asp90Tyr)	LDLR Likely pathogenic(Last reviewed: Nov 5, 2016)	19	11213417 19	11102741	251106	7
AR-2827070 r144172274 [11129] M 00027 4LUDIA: S0100-A (0) 1021111 DLB Partagenc/Likey partagencin, not providely transversed. Dec 15, 2010) 121 1112935 131 11129355 131 1112935 <td>AX-86574463 rs</td> <td>5750474121</td> <td>245462 NM_000527.4(LDLR):c.292G>C (p.Gly98Arg)</td> <td>LDLR Conflicting interpretations of pathogenicity(Last reviewed: Mar 25, 2016)</td> <td>19</td> <td>11213441 19</td> <td>11102765</td> <td>251119</td> <td>19</td>	AX-86574463 rs	5750474121	245462 NM_000527.4(LDLR):c.292G>C (p.Gly98Arg)	LDLR Conflicting interpretations of pathogenicity(Last reviewed: Mar 25, 2016)	19	11213441 19	11102765	251119	19
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Acc.239021 201103410 22740 ML 000572 ML00572	AX-82987078 rs	5144172724	171199 NM_000527.4(LDLR):c.301G>A (p.Glu101Lys)	LDLR Pathogenic/Likely pathogenic, not provided(Last reviewed: Dec 16, 2016)	19	11213450 19	11102774	161266	12
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AR-833338 11208020 245738 NM_00027_4(LUC)L(::283-C (6 L), C+221Tr) DLB Likely pathogenic(Lust reviewed: No 25, 2016) 19 1122726 19 1110558 25342 34 AR-863335 CFS65300 AS55 MM_00057_4(LUC)L(::275-CT) LAR Conflicting interpretations of pathogenic(Lust reviewed: Aug 31, 2016) 19 1122786 19 1110568 153500 44 AR-830357 ST200 NM_000572 4(LUD)L(::278-CT) (L) AL Conflicting interpretations of pathogenic(Lust reviewed: CE 15, 2016) 19 1122784 19 11106568 15267 55 AR-8642145 ST201 NM_000572 4(LUD)L(::2867-AL (L) (LA) 28507 LD Conflicting interpretations of pathogenic(Lust reviewed: CE 2, 2016) 19 1122817 19 110748 61228 40 AR-861245 ST202 NM_000572 4(LUD)L(::2867-AL (L) (L) 4280417) LDR Conflicting interpretations of pathogenic(Lust reviewed: De 15, 2016) 19 1122815 19 110748 61228 40 AR-8612745 ST202 NM_000572 4(LUD)L(: 20057A (L) AL Conflicting interpretations of pathogeninc(Lust reviewed: De 15, 2016) 19 <	AX-98067814 rs:	121908044	18786 NM_000527.4(LDLR):c.621C>T (p.Gly207=)	LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016)	19	11216203 19	11105527	3747	3
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Ax-866387 1512198040 92052 MM. 20027-4(LDR.R): ZP3CA-(Ap.Cy26TiY) UDA Ubit participant (Last reviewed: Dec 16, 2016) 19 1122728 19 11106652 275800 4 Ax8436457 175122098 340088 MM. 20057-4(LDR.R): ZD3A464(p) 1010284 Longituding interpretations of pathogenic/Lock perturbations of pathogenic/Lock perturbation perturbations of pathogenic/Lock perturbations of path	AX-86663356 rs	768563000	245758 NM_000527.4(LDLR):c.718G>T (p.Glu240Ter)	LDLR Pathogenic(Last reviewed: Nov 26, 2016)	19	11217264 19	11106588	251422	34
NA5846457 1312001 MM, 000027 4(LDUR): 29764 (p.Ap2A66(i), LDR. Conflicting interpretations of pathogenicity, not provide/(Last reviewed: Dct. 6, 2016) 19 1127744 19 1107866 16.1287 NA5802166 TS12208 354088 Notes 19 1127744 19 1107846 161286 20 NA5802166 TS1207122 TS1207122 TS12086716 19 11218112 19 1107486 151286 20 NA5812726 TS120868716 Notes SS120172 TS12086716 19 11218112 19 1110746 151286 20 NA5812726 TS1208680 11206 Notes SS120172 1212156 19 11110748 15128 20 NA5807268 TS1208680 11200 Notes SS1201777 1D Notes 1110748 15128 20 1110748 15128 20 1110748 15128 20 1110748 1110748 1110748 1110748 1110748 1110748 1110748 1110748 111111748 1110748<	AX-83503525 rs:	150673992	171200 NM_000527.4(LDLR):c.757C>T (p.Arg253Trp)	LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Aug 31, 2016)	19	11217303 19	11106627	161261	235
Ax86212466 n,75112298 34088 NM_000527.41LDR]; 82504-D6 (s10288; v) 110778	AX-86638837 rs:	121908040	362692 NM_000527.4(LDLR):c.782G>A (p.Cys261Tyr)	LDLR Likely pathogenic(Last reviewed: Dec 16, 2016)	19	11217328 19	11106652	375800	4
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nx-292930 rsi120722 17.208 MM 000527 4(LDR): 2007-T (p.ArgB03Trp) LDR Conflicting interpretations of pathogenic/(Last reviewed: Dec 16, 2016) 19 11218150 19 11107481 4251517 AK-8313728 rsi37868746 171206 MM 000527 4(LDR): 2005-T (p.ArgB03Trp) LDR Pathogenic/(Last reviewed: Dec 16, 2016) 19 1122154 19 111107681 451522 5 AK-84707238 rsi37866746 171200 MM 000527 4(LDR): 2005-T (p.ArgB03Trp) LDR Conflicting interpretations of pathogenic/(Last reviewed: Dec 16, 2016) 19 1122151 19 11110768 161282 5 AK-93020794 rsi39326652 S721278 M000527 4(LDR): 2005-CA (p.ArgB03Trp) LDR Conflicting interpretations of pathogenic/(Last reviewed: Dec 16, 2016) 19 1122141 19 11110766 36450 6 AK-9602676 S52422789 S30850205 171212 MM 000527.4(LDR): 12226-A (p.ArgB0450H) LDR Conflicting interpretations of pathogenic/(Last reviewed: Dec 16, 2016) 19 1122384 19 11113305 251603 78 AK-8602067 S73785807 X40DK12NL LDR Conflicting interpretations of pathogenic/(Last reviewed: Dec 16, 2016)	AX-86721456 rs	751122998	354088 NM_000527.4(LDLR):c.820delA (p.T274Hfs*95)	LDLR Pathogenic/Likely pathogenic(Last reviewed: Oct 9, 2016)	19	11218070 19	11107394	369863	4
AX-8312722 r121908030 245800 NM. 000527.4(LDR): e30676.A (p. Ghy2325er) LDIR Conflicting interpretations of pathogenic/Llast reviewed: Dec 16, 2016) 19 11221354 19 11110678 161282 5 X-8470104 r72658460 171200 NM. 000527.4(LDR): e3066.A (p. Ghy2325er) LDIR Conflicting interpretations of pathogenic/Llast reviewed: Dac 16, 2016) 19 11221351 19 11110678 161282 5 X-84370164 rri2058460 171207 NM. 000527.4(LDR): L0266.A (p. Ghy2325er) LDIR Conflicting interpretations of pathogenic/Llast reviewed: Per 12, 2016) 19 11221431 19 11110756 254603 798 X-886072667 r552422789 230981 NM. 000527.4(LDR): L1276-A (p.Ag0406Gin) LDIR Conflicting interpretations of pathogenic/Llast reviewed: Dec 16, 2016) 19 1122394 19 11113308 228798 9 X-86672687 r552422789 230081 NM. 000527.4(LDR): L1286-A (p.Gu/40847b) LDIR Conflicting interpretations of pathogenic/Llast reviewed: Dec 16, 2016) 19 1122301 11113308 122767 19 11224005 19 11113332 161276 9 Ak66723647 r58/r58/r58/r58/r58/r5	AX-86619878 rs	368657165	171204 NM_000527.4(LDLR):c.862G>A (p.Glu288Lys)	LDLR Conflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016)	19	11218112 19	11107436	161268	20
N 4507238 n 2738074 171205 NM 000574 (LDBR): c9763-A (p. 6); 2325er) LDB. Conflicting interpretations of pathogenicity, lost reviewed: Dec 16, 2010) 19 11221357 19 11110678 16681 16526 AK-54781084 rs72858780 127207 NM_0005274 (LDBR): c9763-A (p. 6); 23248er) LDB. Comflicting interpretations of pathogenicity, last reviewed: Nev 5, 2016) 19 11221414 19 1111075 521603 798 AK-9806794 ri139302564 54113 NM_0005274 (LDBR): c12256A (p. ApgdeGin) LDR Pathogenicity, last reviewed: Dec 16, 2016) 19 11223484 19 1111078 254607 AK-8662667 ri13943601 45116 NM_0005274 (LDBR): c12276-A (p. ApgdeGin) LDR Comflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016) 19 11223498 19 1111333 25452 AK-8670240 ri237865002 240651 NM_0005274 (LDBR): c1276-A (p. ApgdaGin) LDR Comflicting interpretations of pathogenicity(Last reviewed: Dec 13, 2016) 19 11224041 1111333 251747 1 AK-8670304 ri778505037 264066 NM_	AX-82929130 rs:	151207122	171205 NM_000527.4(LDLR):c.907C>T (p.Arg303Trp)	LDLR Conflicting interpretations of pathogenicity(Last reviewed: Mar 25, 2016)	19	11218157 19	11107481	161281	49
N:S-34781064 r72658800 171207 NM 000527.4 (LDLR): c.10246>+ (D.6)3245er) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Ian 24, 2017) 19 11221411 19 11110781 161263 17829 Ax 83216162 rs139361633 245927 NM. 000527.4 (LDLR): c.10246>+ (D.6)43237/r) LDLR Conflicting interpretations of pathogenic/Llast reviewed: De 12, 2016) 19 11221442 19 11110763 251663 6518 Ax 8662676 r55242278 230981 NM. 000527.4 (LDLR): c.1026>- (D.6)408410 LDLR Conflicting interpretations of pathogenic/Lust reviewed: De 16, 2016) 19 11223984 19 11113303 226738 33 Ax 86621429 r36855602 71212 NM. 000527.4 (LDLR): L3276>- (D.7)47413441 LDLR Conflicting interpretations of pathogenic/Lust reviewed: De 16, 2016) 19 11224005 19 1113333 251747 11 Ax 86503801 r373658937 246061 NM. 000527.4 (LDLR): L376>- (D.A)74156(ILL pathogenic/Lust reviewed: De 13, 2016) 19 11224014 19 11113333 251747 14 Ax 86503081 r37375557	AX-83137728 rs:	121908030	245850 NM_000527.4(LDLR):c.910G>T (p.Asp304Tyr)	LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016)	19	11218160 19	11107484	251517	30
AX-83216162 n139361632 425927 NM. 000527.4(LDLR): L024G-T (p.Asp342Tyr) LDR Conflicting interpretations of pathogenic/Lust reviewed: Nov 5, 2016) 19 11221411 19 11110755 251603 798 AX-98067994 rs193922566 45113 NM. 000527.4(LDLR): C10556-A(p.Cys32Tyr) LDR Conflicting interpretations of pathogenic/Lust reviewed: Dec 16, 2016) 19 1122494 19 11110755 251603 66 AX-8660267 S52422789 230981 NM. 000527.4(LDLR): L322G-A (p.Glu408Lys) LDLR Conflicting interpretations of pathogenic/Lust reviewed: Dec 16, 2016) 19 1122398 19 11113333 26433 33 AX-866703204 r573856027 240661 NM. 000527.4(LDLR): L2417-6 (p.Lev414Arg) LDLR Conflicting interpretations of pathogenic/Lust reviewed: an 3, 2017) 19 11224005 19 11113332 251747 1 AX-86670861 r573858027 240661 NM. 000527.4(LDLR): L2476-A (p.Arg4166in) LDLR Rangenic/Likely pathogenic/Likely pathogenic/Likely reviewed: Mar 25, 2016) 19 11224005 19 11113451 369662 3 AX-86670861 r573075755 172125 NM. 000527.4(LDLR): L3467-C5 (p.P7952564	AX-86707238 rs	373869746	171206 NM_000527.4(LDLR):c.967G>A (p.Gly323Ser)	LDLR Conflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016)	19			161282	5
N:9807994 h:193922566 45113 NM2 00527.4(LDR):: L0376>A(p.cy3527/r) UDR Pathogenic/List reviewed: Dec 12, 2016) 19 11222442 19 11110766 36459 6 AX866266 r5552422789 23081 NM 000527.4(LDR):: L2176>A(p.Arg406GIN LDR Conflicting interpretations of pathogenic/ty(Last reviewed: Dec 16, 2016) 19 11222408 19 1113308 284533 3 AX8662288 r137943601 45116 NM_000527.4(LDR):: L2276>A(p.Glu408ky) LDR Conflicting interpretations of pathogenicity(Last reviewed: Jan 3, 2017) 19 11224005 19 1113332 161727 AX8672364 r736555037 246661 NM_000527.4(LDR):: L2470>A (p.Arg416Gin) LDR Pathogenic/Likely pathogenic(Last reviewed: Arz 5, 2016) 19 1122404 19 1113338 251747 1 AX-867364 r53092504 7300505 NM_000527.4(LDR):: L3767> LDR Likely pathogenic(Last reviewed: Arz 5, 2016) 19 1122402 19 1113338 251747 1 AX-8670667 r539925767 S5905 NM_0000527.4(LDR):: L3767 LDR	AX-54781084 rs	72658860	171207 NM_000527.4(LDLR):c.970G>A (p.Gly324Ser)	LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Jan 24, 2017)	19	11221357 19	11110681	161263	1629
AX 86262676 rs552422789 230981 NM 000527.4(LDLR): L217G>A (p.Arg406Gin) LDLR Conflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016) 19 1122388 19 AX 86623288 rs137943601 AS116 NM<000527.4(LDLR): L212G>A (p.Glu408Ly) LDR Conflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016) 19 11223089 19 11113312 161276 9 AX 86673204 rs748555492 246061 NM<00527.4(LDLR): L124T>G (p.Leu414Arg) LDR Conflicting interpretations of pathogenic(Last reviewed: Last 2, 2016) 19 11224005 19 1111332 251772 11 AX 86073026 rs73057307 246061 NM<00527.4(LDLR): L1247G>A (p.Arg406In) LDR Pathogenic/Liker pathogenic(Last reviewed: Dat 3, 2017) 19 11224127 19 1111332 251772 14 AX 86070284 rs73087307 S400557.4(LDLR): L1376C>T (p.Pro5265er) LDR Pathogenic/Liker pathogenic(Last reviewed: Dat 3, 2017) 19 11224127 19 11113451 369662 33 AX 86570284 rs73087307 S40058 MM<000527.4(LDLR): L1376C>T (p.Pro5265er) LDR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016	AX-83216162 rs:	139361635	245927 NM_000527.4(LDLR):c.1024G>T (p.Asp342Tyr)	LDLR Conflicting interpretations of pathogenicity(Last reviewed: Nov 5, 2016)	19	11221411 19	11110735	251603	798
AX-862289 rs13794301 45116 NM_000527.4(LDLR):c12226A (p.Glu408Lys) LDR Conflicting interpretations of pathogenic/k(Last reviewed: Jan 3, 2017) 19 1112329 19 11113323 26453 3 AX-86631429 rs368562025 171212 NM_000527.4(LDLR):c.1236C>T (p.Thr413Met) LDR Conflicting interpretations of pathogenic/k(Last reviewed: Jan 3, 2017) 19 11224008 19 11113332 251747 1 AX-8603204 rs773658037 246661 NM_000527.4(LDLR):c.12476-A (p.Arg41661) LDR Nucleopenic/Likely pathogenic(Last reviewed: Mar 25, 2016) 19 11224018 19 11113332 251752 144 AX-86593083 rs37077955 171215 NM_000527.4(LDLR):c1476C>G (p.Tyr489Ter) LDR Pathogenic/Likely pathogenic 10 11224319 19 111134543 161270 4 AX-8670584 rs37082026 181262 NM_000527.4(LDLR):c1376C>F (p.Pr52565er) LDR Conflicting interpretations of pathogenic/Likely tartogenic/Likely pathogenic 10 11224431 19 111134543 161270 4 AX-8672647 rs37037572 171215 NM_000527.4(LDLR):c.13765CF (p.Pr52565er) LDR Conflicting interpret	AX-98067994 rs:	193922566	45113 NM_000527.4(LDLR):c.1055G>A (p.Cys352Tyr)	LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 12, 2016)	19	11221442 19	11110766	36450	6
NX-86631429 rs36852025 171212 NM.000527.4(LDR):: L238C+7 (p.Thr413Met) LDR Conflicting interpretations of pathogenic(Juctar teviewed: Jan 3, 2017) 19 11224005 19 1111332 151276 9 AX-86703204 rs73658037 246065 NM_000527.4(LDR):: L2347C-A (p.Arg416Gin) LDR Likely pathogenic(Last reviewed: Dec 13, 2016) 19 11224014 19 11113332 251752 14 AX-86070304 rs773658037 246066 NM_000527.4(LDR):: L1376-C (p.Tyr48Fer) LDR Pathogenic/Likely pathogenic(Last reviewed: Dec 13, 2016) 19 11224014 19 11113332 369862 3 AX-8607084 rs730882106 181262 NM_000527.4(LDR):: L1576C+ (p.Pro5265er) LDR Pathogenic/Likely pathogenic, not provided(Last reviewed: Dec 16, 2016) 19 11227402 19 1111352 18120 8 AX-86720647 rs730882106 181262 NM_000527.4(LDR):: L1576C+ (p.Pro5265er) LDR Conflicting interpretations of pathogenic/Likely pathogenic, not provided(Last reviewed: Dec 16, 2016) 19 11227612 19 11116328 161270 28 AX-86720647 rs73018271 NM_000527.4(LDR):: L13764C+7 (p.Arg9551rp) LDR <	AX-86626676 rs	552422789	230981 NM_000527.4(LDLR):c.1217G>A (p.Arg406GIn)	LDLR Conflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016)	19	11223984 19	11113308	228798	9
AX-86723634 rs7736554592 246061 NM_000527.4(LDR):: L12417>G (p.Leu414Arg) LDLR Likely pathogenic(Last reviewed: Mar 25, 2016) 19 11224014 19 11113332 251752 14 AX-86703204 rs773658037 246066 NM_000527.4(LDR):: L1358-TC-C LDLR Likely pathogenic(Last reviewed: Mar 25, 2016) 19 112242014 19 11113338 251752 14 AX-8670584 rs730582106 181262 NM_000527.4(LDR):: L1358-TC-C LDLR Pathogenic/Likely pathogenic(Last reviewed: Mar 25, 2016) 19 112242319 19 11113433 161270 4 AX-86670684 rs73082106 181262 NM_000527.4(LDR):: L1576C>T (p.Pro5256er) LDLR Pathogenic/Likely pathogenic, not provided(Last reviewed: Dec 16, 2016) 19 11224761 19 11116928 161271 277 AX-86723661 rs730382106 181262 NM_000527.4(LDR):: L78657 (p.Pro52557p) LDLR Conflicting interpretations of pathogenich/Last reviewed: Poc 16, 2016) 19 11227612 19 11116936 161290 8 AX-86720647 rs201102492 246329 NM_000527.4(LDR):: L78657 (p.Pre619Leu) LDLR Conflicting interpretations of pathogen	AX-86622889 rs:	137943601		LDLR Conflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016)	19			36453	3
AX-86703204 rs773658037 246066 NM_000527.4(LDLR):c.1247G-A (p.Arg416Gin) LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 13, 2016) 19 11224014 19 11113338 251752 14 AX-8607995 rs10707755 354095 NM_000527.4(LDLR):c.13876-C5 (p.Tyr489Ter) LDLR Pathogenic/Likely pathogenic(Last reviewed: Mar 25, 2016) 19 11224127 19 11113643 161270 4 AX-8667084 rs30882106 181262 NM_000527.4(LDLR):c.1576C-T (p.Pro526Ser) LDLR Conflicting interpretations of pathogenic/Ly not provided(Last reviewed: Dec 16, 2016) 19 11224428 19 11113543 161271 27 AX-86720647 rs30371272 111218 NM_000527.4(LDLR):c.1783-CF (p.Arg5957D) LDLR Conflicting interpretations of pathogenic/ty(Last reviewed: Feb 10, 2017) 19 11227613 19 11116938 161290 8 AX-86720647 rs201102492 246329 NM_000527.4(LDLR): c.1876C-A (p.Glu626Lys) LDLR Conflicting interpretations of pathogenicity(Last reviewed: Feb 10, 2017) 19 11220771 19 1122012 183127 167 AX-	AX-86631429 rs	368562025	171212 NM_000527.4(LDLR):c.1238C>T (p.Thr413Met)	LDLR Conflicting interpretations of pathogenicity(Last reviewed: Jan 3, 2017)	19				9
AX-98067995 rs139322567 354095 NM_000527.4(LDLR):::1358+27-C LDLR Likely pathogenic 19 11224127 19 11113451 369862 3 AX-86593083 rs37077955 171215 NM_000527.4(LDLR):::1376C>T (p.Pro5266er) LDLR Pathogenic/Likely pathogenic(Last reviewed: Mar 25, 2016) 19 11224129 19 11113451 369862 3 AX-86570684 rs370882106 181262 NM_000527.4(LDLR):::1775C>4 (p.ClyS5261u) LDLR Conflicting interpretations of pathogenic/Likely pathogenic, not provided(Last reviewed: Dec 16, 2016) 19 11224764 19 11116936 161270 A AX-86723961 rs3703371572 171217 NM_000527.4(LDLR):::1786-X (p.Arg5951rp) LDLR Conflicting interpretations of pathogenic/Likely pathogenic/Li			246061 NM_000527.4(LDLR):c.1241T>G (p.Leu414Arg)	LDLR Likely pathogenic(Last reviewed: Mar 25, 2016)	19				1
AX-86593083 rs370777955 171215 NM_000527.4(LDLR):c.1467C>G (p.Tyr489Ter) LDLR Pathogenic/Likely pathogenic(Last reviewed: Mar 25, 2016) 19 11224319 19 11113643 161270 4 AX-86570684 rs370382106 181262 NM_000527.4(LDLR)::c.1576C+T (p.Pro5265er) LDLR Conflicting interpretations of pathogenic/Likely pathogenic, not provided(Last reviewed: Dec 16, 2016) 19 111227604 19 11113752 183120 8 AX-86723961 rs373371572 171218 NM_000527.4(LDLR)::c.1786C>T (p.Arg595Trp) LDR Conflicting interpretations of pathogenic/Likely reviewed: Feb 10, 2017) 19 11227613 19 11116936 161290 8 AX-86720647 rs20102492 246329 NM_000527.4(LDR):: C.1786C>T (p.Phe619Leu) LDR Conflicting interpretations of pathogenic/Likely reviewed: Feb 10, 2017) 19 1112012 283027 AX-86034082 rs741314711 246378 NM_000527.4(LDR):: C.1876C>A (p.Glu62Giys) LDR Conflicting interpretations of pathogenic/Likely reviewed: Feb 3, 2017) 19 1112012 183127 127 AX-8664082 rs74131971122 184326 N	AX-86703204 rs	773658037	246066 NM_000527.4(LDLR):c.1247G>A (p.Arg416Gln)	LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 13, 2016)	19				
AX-86670684 rs730882106 181262 NM_000527.4(LDLR):c.1576C>T (p.Pro526Ser) LDLR Conflicting interpretations of pathogenic/t, not provided(Last reviewed: Dec 16, 2016) 19 11224428 19 11113752 183120 8 AX-8670684 rs73082106 1513729307 171217 NM_000527.4(LDLR):c.1778G>A (p.Gly592Glu) LDLR Pathogenic/Likely pathogenic, not provided(Last reviewed: Dec 16, 2016) 19 11227604 19 11116928 161271 277 AX-86720647 rs201102492 246329 NM_000527.4(LDLR):c.1786C>T (p.Arg595Te) LDLR Conflicting interpretations of pathogenicity(Last reviewed: Feb 10, 2017) 19 11120761 19 11227613 19 11116937 252029 9 AX-8634082 rs747134711 246378 NM_000527.4(LDLR):c.1876C>A (p.Glu626Lys) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Dec 16, 2016) 19 1122077 19 11120121 252083 2 AX-86047874 rs133991325 181269 NM_000527.4(LDLR):c.1978C>T (p.Glu626Lys) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 11230079 <t< td=""><td></td><td></td><td>354095 NM_000527.4(LDLR):c.1358+2T>C</td><td>,, , , , , , , , , , , , , , , , , , ,</td><td>19</td><td></td><td></td><td></td><td>3</td></t<>			354095 NM_000527.4(LDLR):c.1358+2T>C	,, , , , , , , , , , , , , , , , , , ,	19				3
AX-83471866 rs137929307 171217 NM_000527.4(LDLR):c.1775G>A (p.Gly592Glu) LDLR Pathogenic/Likely pathogenic, not provided(Last reviewed: Dec 16, 2016) 19 11227604 19 11116928 161271 27 AX-86723961 rs373371572 171218 NM_000527.4(LDLR):c.1783C>T (p.Arg595Trp) LDLR Conflicting interpretations of pathogenic/(Last reviewed: Feb 10, 2017) 19 11227612 19 11116936 161290 8 AX-86720647 rs20102492 246329 NM_000527.4(LDLR):c.1783C>T (p.Arg595Eu) LDLR Conflicting interpretations of pathogenicity(Last reviewed: Feb 10, 2017) 19 11220771 19 11120011 252029 9 AX-86720647 rs20102492 181269 NM_000527.4(LDLR):c.18756C>A (p.Glu62GLyS) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Feb 3, 2017) 19 11220721 183127 167 AX-80618749 rs139791325 181269 NM_000527.4(LDLR):c.1876C>A (p.Glu62GLYS) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 11220791 19 1122022 183127 167 AX-80618749 rs150021927 246458 NM_0000527.4(LDLR):c.1876C>A (p.Glu660Ter)									4
AX-86723961 rs373371572 171218 NM_000527.4(LDLR):c.1783C>T (p.Arg595Trp) LDLR Conflicting interpretations of pathogenic(Last reviewed: Feb 10, 2017) 19 11227612 19 11116936 161290 8 AX-86720647 rs201102492 246329 NM_000527.4(LDLR): c.1784G>T (p.Arg595Leu) LDLR Likely pathogenic(Last reviewed: Mar 25, 2016) 19 11227613 19 11110937 252029 9 AX-86720647 rs201102492 246329 NM_000527.4(LDLR): c.1784G>T (p.Arg595Leu) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 11220777 19 11120101 252083 2 AX-8034390 rs1393921325 181260 NM_000527.4(LDLR): c.1978C>T (p.Gln660Ter) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 11230700 19 112010224 36458 AX-9803790 rs193922569 45121 NM_000527.4(LDLR): c.2060CA> (p.Cys667Arg) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 1123050 19 1120303 252163 3 AX-86618749 rs150021927 246458 NM									-
AX-86720647 rs201102492 246329 NM_000527.4(LDLR):::1784G>T (p.Arg595Leu) LDLR Likely pathogenic(Last reviewed: Mar 25, 2016) 19 11227613 19 11116937 252029 9 AX-86634082 rs747134711 246378 NM_000527.4(LDLR):::1855T>C (p.Phe619Leu) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Feb 3, 2017) 19 11120012 252083 2 AX-83043349 rs139791325 181269 NM_000527.4(LDLR):::1876G>A (p.Glu62GLys) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Feb 3, 2017) 19 11120012 183127 167 AX-8033790 rs13991225 45121 NM_000527.4(LDLR):::0786C>A (p.Glu62GLys) LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11230078 19 1112022 36458 4 AX-86618749 rs150021927 246458 NM_000527.4(LDLR):::07967Tyr LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231058 19 11120382 252163 3 AX-86618749 rs774730452 246468 NM_000527.4(LDLR)::::0206C>A (p.Ala684Thr)									
AX-86634082 rs747134711 246378 NM_000527.4(LDLR):c.1855T>C (p.Phe619Leu) LDLR Conflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016) 19 11230777 19 11120101 252083 2 AX-86034082 rs747134711 246378 NM_000527.4(LDLR):c.1876G>A (p.Glu626Lys) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 11230778 19 11120122 183127 167 AX-8033790 rs139392569 45121 NM_000527.4(LDLR):: L1978C>T (p.Gln660Ter) LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 1123057 19 1112038 36458 4 AX-86618749 rs150021927 246458 NM_000527.4(LDLR):: C1999T>C (p.Cys667Arg) LDLR Iklely pathogenic(Last reviewed: Dec 16, 2016) 19 11231057 19 11120382 36458 4 AX-86618749 rs150021927 246458 NM_000527.4(LDLR): C.2000G>A (p.Cys667Arg) LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231058 19 11120432 256189 8 AX-8651791 rs774730452									8
AX-83048349 rs139791325 181269 NM_000527.4(LDLR): c.1876G>A (p.Glu626Lys) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Feb 3, 2017) 19 11230798 19 11120122 183127 167 AX-83048349 rs139322569 45121 NM_000527.4(LDLR): c.1978C>T (p.Gln660Ter) LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 1123057 19 11120324 36458 4 AX-86618749 rs150021927 246458 NM_000527.4(LDLR): c.1999T>C (p.Cys667Arg) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231057 19 11120382 252163 3 AX-86618749 rs150021927 246458 NM_000527.4(LDLR): c.2000G>A (p.Cys667Arg) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231058 19 11120382 252163 3 AX-8658179 rs774730452 246446 NM_000527.4(LDR): c.20060Ard (p.Gys67Tryr) LDLR Pathogenic(Last reviewed: Dec 16, 2016) 19 1123108 19 11120432 25219 0 AX-8668370 rs751228587 246496			246329 NM_000527.4(LDLR):c.1784G>T (p.Arg595Leu)	LDLR Likely pathogenic(Last reviewed: Mar 25, 2016)	19			252029	9
AX-98033790 rs193922569 45121 NM_000527.4(LDLR): c.1978C>T (p.Gln660Ter) LDLR Pathogenic/Likely pathogenic/List reviewed: Dec 16, 2016) 19 1123090 19 1120224 36458 4 AX-86618749 rs150021927 246458 NM_000527.4(LDLR): c.1978C>T (p.Gln660Ter) LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231057 19 11120224 36458 4 AX-86618749 rs150021927 246458 NM_000527.4(LDLR): c.1978C>T (p.Cgs667Arg) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231057 19 11120382 3689 8 AX-86581791 rs774730452 246486 NM_000527.4(LDLR): c.2050G>A (p.Ala684Thr) LDLR Pathogenic(Last reviewed: Dec 16, 2016) 19 11231108 19 11120432 252192 0 AX-86738370 rs751228587 246496 NM_000527.4(LDLR): c.2060dupT (p.Asn6880Infs) LDLR Pathogenic(Last reviewed: Mar 25, 2016) 19 11231154 19 11120442 252219 1 AX-86688504 rs368838866 181272 NM_000527.4(LDLR): c.21060C			246378 NM_000527.4(LDLR):c.1855T>C (p.Phe619Leu)	LDLR Conflicting interpretations of pathogenicity(Last reviewed: Dec 16, 2016)	19				2
AX-86618749 rs150021927 246458 NM_000527.4(LDLR):c.1999T>C (p.Cys667Arg) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231057 19 11120381 252163 3 AX-98067698 rs28942083 18728 NM_000527.4(LDLR):c.2000G>A (p.Cys667Tyr) LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231058 19 11120382 3689 8 AX-86581791 rs774730452 246486 NM_000527.4(LDLR): c.2050G>A (p.Ala684Thr) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231108 19 11120432 252192 0 AX-86738370 rs751228587 246496 NM_000527.4(LDLR): c.2060dupT (p.Asn688Glnfs) LDLR Pathogenic(Last reviewed: Mar 25, 2016) 19 1123118 19 11120442 252201 1 AX-86738370 rs751228587 246496 NM_000527.4(LDLR): c.2006C>T (p.Pro699Leu) LDLR Pathogenic(Last reviewed: Dec 16, 2016) 19 11231154 19 11120478 252219 51 AX-86688504 rs368838866 181272 NM_000527.4(LDLR): c.2100FORC)									
AX-98067698 rs28942083 18728 NM_000527.4(LDLR):c.2006>A (p.Cys667Tyr) LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231058 19 11120382 3689 8 AX-86581791 rs774730452 246486 NM_000527.4(LDLR):c.2050G>A (p.Ala684Thr) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 1123108 19 11120432 252192 0 AX-86738370 rs751228587 246496 NM_000527.4(LDLR):c.2060dupT (p.Asn688Ginfs) LDLR Pathogenic(Last reviewed: Mar 25, 2016) 19 1123118 19 11120442 252201 1 AX-86738370 rs751228587 246496 NM_000527.4(LDLR):c.2060dupT (p.Asn6886infs) LDLR Pathogenic(Last reviewed: Dec 16, 2016) 19 11231154 19 11120442 252201 1 AX-86688504 rs368838866 181272 NM_000527.4(LDLR):c.21006>A (p.Gly701Ser) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 11231159 19 11120483 31310 25 AX-83688866 181272 NM_000527.4(LDLR): c.2140+1G>A <									4
AX-86581791 rs774730452 246486 NM_000527.4(LDLR):c.2050G>A (p.Ala684Thr) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231108 19 11120432 252192 0 AX-86538370 rs751228587 246496 NM_000527.4(LDLR):c.2060dupT (p.Asn6886Infs) LDLR Pathogenic(Last reviewed: Mar 25, 2016) 19 11231118 19 11120442 252201 1 AX-86373870 rs751228587 246496 NM_000527.4(LDLR):c.2060dupT (p.Asn688GInfs) LDLR Pathogenic(Last reviewed: Mar 25, 2016) 19 11231154 19 11120478 252219 51 AX-8638840 rs36838866 181272 NM_000527.4(LDLR):c.2101G>A (p.Gly701Ser) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 11231159 19 11120478 252219 51 AX-86888264 rs36838866 181272 NM_000527.4(LDLR):c.2101G>A (p.Gly701Ser) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Nov 5, 2016) 19 11231159 19 11120478 3744 7 AX-86582518 rs72658867 45123 <td></td> <td></td> <td></td> <td></td> <td>-</td> <td></td> <td></td> <td></td> <td>3</td>					-				3
AX-86738370 rs751228587 246496 NM_000527.4(LDLR):c.2060dupT (p.Asn688Glnfs) LDLR Pathogenic(Last reviewed: Mar 25, 2016) 19 11231118 19 11120442 252201 1 AX-863738370 rs751228587 246496 NM_000527.4(LDLR):c.2060dupT (p.Asn688Glnfs) LDLR Pathogenic(Last reviewed: Mar 25, 2016) 19 11231118 19 11120442 252201 1 AX-86688504 rs36838866 181272 NM_000527.4(LDLR):c.2016>A (p.Gly7015er) LDLR Conflicting interpretations of pathogenic(Last reviewed: Nov 5, 2016) 19 11231159 19 11120438 183130 25 AX-8688504 rs36838866 181272 NM_000527.4(LDLR):c.2101G>A (p.Gly7015er) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 1123119 19 11120438 183130 25 AX-86885218 rs72658867 45123 NM_000527.4(LDLR): c.2140+16SA A LDLR Conflicting interpretations of pathogenic(ty(Last reviewed: Dec 25, 2016) 19 11231199 19 11120523 3744 7 AX-8682518 rs72658867 45123									-
AX-83379229 rs201573863 246513 NM_000527.4(LDLR):c.2096C>T (p.Pro699Leu) LDLR Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231154 19 11120478 252219 51 AX-8688504 rs368838866 181272 NM_000527.4(LDLR):c.2101G>A (p.Gly701Ser) LDLR Conflicting interpretations of pathogenic(Last reviewed: Nov 5, 2016) 19 11231159 19 11120478 252219 51 AX-86888504 rs368838866 181272 NM_000527.4(LDLR):c.2101G>A (p.Gly701Ser) LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 16, 2016) 19 11231159 19 11120478 252219 51 AX-83408984 rs145787161 18783 NM_000527.4(LDLR):c.2140+1G>A LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231199 19 11120523 3744 7 AX-86582518 rs72658867 45123 NM_000527.4(LDLR):c.2140+5G>A LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Dec 25, 2016) 19 11231203 19 11120527 36460 6501 AX-83148620 rs17853964									-
AX-86688504 rs36838866 181272 NM_000527.4(LDLR):c.2101G>A (p.Gly701Ser) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Nov 5, 2016) 19 11231159 19 11120483 183130 25 AX-83408984 rs145787161 18783 NM_000527.4(LDLR):c.2140+1G>A LDLR Pathogenic/Likely pathogenic(Last reviewed: Dec 16, 2016) 19 11231199 19 11120523 3744 7 AX-86582518 rs72658867 45123 NM_000527.4(LDLR):c.2140+5G>A LDLR Conflicting interpretations of pathogenic(Last reviewed: Dec 25, 2016) 19 1123103 19 11120527 36460 6501 AX-83148620 rs17853964 45125 NM_000527.4(LDLR):c.2479G>A (p.Val827Ile) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Jan 5, 2017) 19 1120207 36460 6501 AX-83148620 rs17853964 45125 NM_000527.4(LDLR):c.2479G>A (p.Val827Ile) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Jan 5, 2017) 19 11120207 36460 328									-
AX-83408984 rs145787161 18783 NM_000527.4(LDLR):c.2140+1G>A LDLR Pathogenic/Likely pathogenic/List reviewed: Dec 16, 2016) 19 11231199 19 11120523 3744 7 AX-86582518 rs72658867 45123 NM_000527.4(LDLR):c.2140+5G>A LDLR Pathogenic/Likely pathogenic/List reviewed: Dec 25, 2016) 19 11231203 19 11120527 36460 6501 AX-83148620 rs17853964 45125 NM_000527.4(LDLR):c.2479G>A (p.Val827Ile) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Jan 5, 2017) 19 1120202 36462 328					-				
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AX-83148620 rs137853964 45125 NM_000527.4(LDLR):c.2479G>A (p.Val827Ile) LDLR Conflicting interpretations of pathogenicity, not provided(Last reviewed: Jan 5, 2017) 19 11240278 19 11129602 36462 328									
AX-86643801 rs377437226 246635 NM_000527.4(LDLR):c.2546C>A (p.Ser849Ter) LDLR Pathogenic(Last reviewed: Mar 25, 2016) 19 11240345 19 11129669 252350 38									
	AX-86643801 rs	377437226	246635 NM_000527.4(LDLR):c.2546C>A (p.Ser849Ter)	LDLR Pathogenic(Last reviewed: Mar 25, 2016)	19	11240345 19	11129669	252350	38