

Supplemental Table 1. High throughput sequencing, mapping and variant calling statistics

| | Patient 2-III-12 | Patient 2-III-13 | Healthy brother | Healthy sister | Father | Mother |
|--|-------------------------|-------------------------|------------------------|-----------------------|---------------|---------------|
| Number of reads | 31762456 | 41166167 | 35706401 | 35448194 | 24878776 | 34803835 |
| pairs | | | pairs | pairs | pairs | pairs |
| 63524912 | | 82332334 | 71412802 | 70896388 | 49757552 | 69607670 |
| reads | | reads | reads | reads | reads | reads |
| Number of mapped reads | 62807584 | 81380112 | 70507082 | 69804206 | 49133972 | 68797164 |
| properly mapped reads | 61383980 | 80785716 | 68750984 | 68016494 | 48045159 | 67689846 |
| Total variants | 408767 | 537638 | 437499 | 417501 | 312259 | 497203 |
| Variants not common polymorphisms | 124007 | 160764 | 131919 | 121868 | 87616 | 177446 |
| Coding changing non common variants | 990 | 1058 | 988 | 1026 | 1010 | 1061 |
| Recessive model variants | 7619 | 8170 | | | | |
| Recessive coding changing non common variants | 17 | 22 | | | | |
| Recessive coding changing non common variants in both patients | 5 | | | | | |

Supplemental Table 2. Final list of 5 coding changing, non-common recessive variants detected in both affected patients in Family 2

| Chromosome | Position | Gene | Gene description | Mutation | Type |
|-------------------|-----------------|-------------|---|-----------------|--|
| 1 | 47280785 | CYP4B1 | Cytochrome P450, family 4, subfamily B, polypeptide 1 | G/A | NM_001099772:c.922G>A; p.(Ala308Thr) |
| 1 | 45276025 | BTBD19 | BTB (POZ) domain containing 19 | C/G | NM_001136537:c.227C>G; p.(Thr76Ser) |
| 8 | 145738349 | RECQL4 | RecQ protein-like 4 | G/T | NM_004260: c.2636C>A; p.(Pro879His) |
| 8 | 145541756 | DGAT1 | Diacylglycerol O-acyltransferase 1 | A/G | NG_034192.1:g.13827T>C |
| 8 | 144789247 | CCDC166 | Coiled-coil domain containing 166 | C/T | NM_001162914: c.937G>A; p.(Val313Met) |

Supplemental Table 3. List of DGAT1 variants

| Chromosome | Position | RSID | Variant* | PolyPhen2 | SIFT | Allele Frequency | Annotation | Allele Count | Allele Number | Number of Homozygotes |
|------------|-----------|-------------|--|-------------------|-----------|------------------|-----------------|--------------|---------------|-----------------------|
| 8 | 145550151 | . | NM_012079.5:c.149C>T; p.(Ala50Val) | Benign | Tolerated | 0.001456 | missense | 2 | 1374 | 0 |
| 8 | 145545073 | rs145303862 | NG_034192.1(DGAT1):c. 201-2A>G | | | 0.000008284 | splice acceptor | 1 | 120716 | 0 |
| 8 | 145545068 | . | NM_012079.5:c.204C>A; p.(Cys68Ter) | | | 0.000008282 | stop gained | 1 | 120748 | 0 |
| 8 | 145545064 | . | NM_012079.5:c.208C>T; p.(Arg70Cys) | Probably damaging | Damaging | 0.00000828 | missense | 1 | 120780 | 0 |
| 8 | 145545063 | . | NM_012079.5:c.209G>A; p.(Arg70His) | Benign | Damaging | 0.000008277 | missense | 1 | 120814 | 0 |
| 8 | 145545024 | . | NM_012079.5:c.248G>A; p.(Ser83Asn) | Benign | Tolerated | 0.00001652 | missense | 2 | 121052 | 0 |
| 8 | 145545017 | . | NM_012079.5:c.255C>G; p.(Tyr85Ter) | | | 0.000008258 | stop gained | 1 | 121094 | 0 |
| 8 | 145545010 | rs189862978 | NM_012079.5:c.262A>G; p.(Ile88Val) | Benign | Tolerated | 0.00004954 | missense | 6 | 121108 | 0 |
| 8 | 145544994 | . | NM_012079.5:c.278T>C; p.(Val93Ala) | Probably damaging | Damaging | 0.000008255 | missense | 1 | 121144 | 0 |
| 8 | 145544982 | . | NG_034192.1(DGAT1):c. 288+1delG | | | 0.000008256 | splice donor | 1 | 121126 | 0 |
| 8 | 145542719 | . | NM_012079.5:c.301G>A; p.(Ala101Thr) | Possibly damaging | Tolerated | 0.00004498 | missense | 1 | 22234 | 0 |
| 8 | 145542716 | . | NM_012079.5:c.304C>T; p.(Arg102Trp) | Probably damaging | Damaging | 0.00004464 | missense | 1 | 22400 | 0 |
| 8 | 145542706 | . | NM_012079.5:c.314T>C; p.(Leu105Pro) | Probably damaging | Damaging | 0.00004342 | missense | 1 | 23030 | 0 |
| 8 | 145542581 | . | NM_012079.5:c.332A>G; p.(Tyr111Cys) | Probably damaging | Damaging | 0.0000216 | missense | 2 | 92614 | 0 |
| 8 | 145542532 | . | NM_012079.5:c.381T>G; p.(Asp127Glu) | Probably damaging | Tolerated | 0.0000191 | missense | 2 | 104718 | 0 |
| 8 | 145542530 | . | NM_012079.5:c.383C>T; p.(Pro128Leu) | Probably damaging | Tolerated | 0.000009574 | missense | 1 | 104450 | 0 |
| 8 | 145542530 | . | NM_012079.5:c.383C>A; p.(Pro128His) | Probably damaging | Tolerated | 0.000009574 | missense | 1 | 104450 | 0 |
| 8 | 145542525 | . | NM_012079.5:c.388A>G; p.(Ser130Gly) | Possibly damaging | Tolerated | 0.000009589 | missense | 1 | 104282 | 0 |
| 8 | 145542516 | . | NM_012079.5:c.397G>A; p.(Ala133Thr) | Possibly damaging | Tolerated | 0.00002929 | missense | 3 | 102436 | 0 |
| 8 | 145542500 | . | NM_012079.5:c.413T>C; p.(Ile138Thr) | Possibly damaging | Tolerated | 0.00002021 | missense | 2 | 98962 | 0 |

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|----------|------------------|-------------|--|------------------------------|-----------------|--------------------|----------------------|-----------|---------------|----------|
| 8 | 145542414 | rs140443241 | NM_012079.5:c.416C>T; p.(Ala139Val) | Benign | Tolerated | 0.00004594 | missense | 4 | 87066 | 0 |
| 8 | 145542411 | . | NM_012079.5:c.419C>T; p.(Ala140Val) | Benign | Tolerated | 0.00005686 | missense | 5 | 87940 | 0 |
| 8 | 145542408 | rs150538509 | NM_012079.5:c.422A>G; p.(Asn141Ser) | Probably damaging | Tolerated | 0.0000567 | missense | 5 | 88178 | 0 |
| 8 | 145542387 | . | NM_012079.5:c.443T>G; p.(Phe148Cys) | Probably damaging | Damaging | 0.0001117 | missense | 10 | 89532 | 0 |
| 8 | 145542375 | rs55907012 | NM_012079.5:c.455A>G; p.(Lys152Arg) | Benign | Tolerated | 0.0164 | missense | 1482 | 90390 | 20 |
| 8 | 145542372 | . | NM_012079.5:c.458G>T; p.(Arg153Leu) | Benign | Tolerated | 0.00001108 | missense | 1 | 90246 | 0 |
| 8 | 145542371 | . | NM_012079.5:c.458delG; p.(Arg153ProfsTer7) | | | 0.00001104 | frameshift | 1 | 90576 | 0 |
| 8 | 145542366 | . | NM_012079.5:c.464C>T; p.(Ala155Val) | Possibly damaging | Tolerated | 0.00006645 | missense | 6 | 90294 | 0 |
| 8 | 145542364 | . | NM_012079.5:c.466G>A; p.(Val156Met) | Benign | Tolerated | 0.000022 | missense | 2 | 90908 | 0 |
| 8 | 145542219 | . | NM_012079.5:c.479C>T; p.(Thr160Met) | Probably damaging | Damaging | 0.0000937 | missense | 10 | 106720 | 0 |
| 8 | 145542210 | . | NM_012079.5:c.488C>T; p.(Ala163Val) | Benign | Tolerated | 0.00002772 | missense | 3 | 108228 | 0 |
| 8 | 145542204 | . | NM_012079.5:c.493delC; p.(Leu165CysfsTer34) | | | 0.000009161 | frameshift | 1 | 109156 | 0 |
| 8 | 145542196 | . | NM_012079.5:c.502C>T; p.(His168Tyr) | Benign | Tolerated | 0.000009071 | missense | 1 | 110238 | 0 |
| 8 | 145542193 | rs144065666 | NM_012079.5:c.505G>A; p.(Val169Met) | Benign | Tolerated | 0.0002085 | missense | 23 | 110330 | 0 |
| 8 | 145542184 | . | NM_012079.5:c.514C>G; p.(Leu172Val) | Benign | Tolerated | 0.000008996 | missense | 1 | 111156 | 0 |
| 8 | 145542166 | . | NM_012079.5:c.530_531del e1GT; p.(Cys177PhefsTer10) | | | 0.000008957 | frameshift | 1 | 111648 | 0 |
| 8 | 145542160 | . | NM_012079.5:c.538G>T; p.(Ala180Ser) | Benign | Tolerated | 0.00001798 | missense | 2 | 111246 | 0 |
| 8 | 145542159 | rs139015645 | NM_012079.5:c.539C>T; p.(Ala180Val) | Benign | Tolerated | 0.00002701 | missense | 3 | 111086 | 0 |
| 8 | 145542155 | . | NM_012079.5:c.540_542dupGGC; p.(Ala181dup) | | | 0.00007227 | inframe insertion | 8 | 110692 | 0 |
| 8 | 145542144 | . | NM_012079.5:c.554T>C; p.(Leu185Pro) | Possibly damaging | Tolerated | 0.000009138 | missense | 1 | 109438 | 0 |
| 8 | 145542133 | . | NM_012079.5:c.565A>G; p.(Ile189Val) | Benign | Tolerated | 0.00001856 | missense | 2 | 107744 | 0 |
| 8 | 145542129 | . | NM_012079.5:c.569C>T; p.(Thr190Ile) | Probably damaging | Tolerated | 0.00002805 | missense | 3 | 106934 | 0 |

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|----------|------------------|--------------------|---|------------------------------|-----------------|--------------------|---------------------|----------|---------------|----------|
| 8 | 145542001 | . | NM_012079.5:c.599C>T; p.(Ala200Val) | Benign | Tolerated | 0.00005194 | missense | 6 | 115522 | 0 |
| 8 | 145541995 | . | NM_012079.5:c.605C>A; p.(Thr202Asn) | Probably damaging | Damaging | 0.00002573 | missense | 3 | 116602 | 0 |
| 8 | 145541993 | . | NM_012079.5:c.607A>G; p.(Ile203Val) | Benign | Tolerated | 0.000008567 | missense | 1 | 116722 | 0 |
| 8 | 145541990 | . | NM_012079.5:c.610C>T; p.(Leu204Phe) | Probably damaging | Tolerated | 0.00005135 | missense | 6 | 116852 | 0 |
| 8 | 145541974 | rs142970193 | NM_012079.5:c.626T>C; p.(Phe209Ser) | Benign | Tolerated | 0.00005954 | missense | 7 | 117566 | 0 |
| 8 | 145541971 | rs138248019 | NM_012079.5:c.629C>T; p.(Ser210Phe) | Probably damaging | Damaging | 0.000008502 | missense | 1 | 117624 | 0 |
| 8 | 145541968 | . | NM_012079.5:c.629_631de ICCT; p.(Ser210del) | | | 0.00007649 | inframe deletion | 9 | 117664 | 0 |
| 8 | 145541966 | rs150434452 | NM_012079.5:c.634C>G; p.(Arg212Gly) | Benign | Tolerated | 0.000008501 | missense | 1 | 117628 | 0 |
| 8 | 145541966 | rs150434452 | NM_012079.5:c.634C>T; p.(Arg212Cys) | Probably damaging | Tolerated | 0.000136 | missense | 16 | 117628 | 0 |
| 8 | 145541965 | . | NM_012079.5:c.635G>A; p.(Arg212His) | Possibly damaging | Tolerated | 0.000008501 | missense | 1 | 117634 | 0 |
| 8 | 145541963 | . | NM_012079.5:c.637G>A; p.(Asp213Asn) | Possibly damaging | Tolerated | 0.000017 | missense | 2 | 117664 | 0 |
| 8 | 145541960 | . | NM_012079.5:c.640G>A; p.(Val214Ile) | Probably damaging | Tolerated | 0.0001616 | missense | 19 | 117602 | 0 |
| 8 | 145541950 | . | NM_012079.5:c.650G>A; p.(Trp217Ter) | | | 0.000008506 | stop gained | 1 | 117558 | 0 |
| 8 | 145541947 | . | NM_012079.5:c.653G>T; p.(Cys218Phe) | Probably damaging | Damaging | 0.000008511 | missense | 1 | 117498 | 0 |
| 8 | 145541945 | . | NM_012079.5:c.655C>T; p.(Arg219Cys) | Probably damaging | Damaging | 0.00003407 | missense | 4 | 117394 | 0 |
| 8 | 145541944 | . | NM_012079.5:c.656G>A; p.(Arg219His) | Probably damaging | Damaging | 0.00001704 | missense | 2 | 117346 | 0 |
| 8 | 145541942 | . | NM_012079.5:c.658A>G; p.(Arg220Gly) | Benign | Tolerated | 0.00003409 | missense | 4 | 117320 | 0 |
| 8 | 145541930 | . | NM_012079.5:c.670A>G; p.(Lys224Glu) | Benign | Tolerated | 0.000008553 | missense | 1 | 116916 | 0 |
| 8 | 145541923 | . | NG_034192.1(DGAT1):c. 676+1G>A | | | 0.00001715 | splice donor | 2 | 116618 | 0 |
| 8 | 145541829 | . | NM_012079.5:c.680C>T; p.(Ser227Phe) | Possibly damaging | Damaging | 0.000008756 | missense | 1 | 114210 | 0 |
| 8 | 145541818 | . | NM_012079.5:c.691A>G; p.(Lys231Glu) | Benign | Tolerated | 0.00001774 | missense | 2 | 112768 | 0 |
| 8 | 145541817 | . | NM_012079.5:c.692A>C; p.(Lys231Thr) | Possibly damaging | Tolerated | 0.000008877 | missense | 1 | 112646 | 0 |

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|---|-----------|-------------|--|----------------------|-----------|-------------|---------------------|------|--------|-----|
| 8 | 145541815 | . | NM_012079.5:c.691_694de 1AAGGinsAAGA; p.(Ala232Thr) | Possibly damaging | Tolerated | 0.000107 | missense | 12 | 112170 | 0 |
| 8 | 145541815 | . | NM_012079.5:c.691_694de 1AAGGinsG; p.(Lys231del) | | | 0.000008915 | inframe deletion | 1 | 112170 | 0 |
| 8 | 145541812 | . | NM_012079.5:c.697A>G; p.(Ser233Gly) | Benign | Tolerated | 0.000008939 | missense | 1 | 111874 | 0 |
| 8 | 145541805 | . | NM_012079.5:c.704C>T; p.(Ala235Val) | Benign | Tolerated | 0.000009016 | missense | 1 | 110916 | 0 |
| 8 | 145541796 | . | NM_012079.5:c.713C>T; p.(Pro238Leu) | Benign | Tolerated | 0.0002813 | missense | 31 | 110204 | 0 |
| 8 | 145541788 | . | NM_012079.5:c.721G>A; p.(Val241Met) | Probably damaging | Damaging | 0.00001822 | missense | 2 | 109788 | 0 |
| 8 | 145541784 | . | NM_012079.5:c.725G>A; p.(Ser242Asn) | Possibly damaging | Tolerated | 0.000009117 | missense | 1 | 109682 | 0 |
| 8 | 145541782 | rs146322937 | NM_012079.5:c.727T>C; p.(Tyr243His) | Probably damaging | Damaging | 0.00006384 | missense | 7 | 109656 | 0 |
| 8 | 145541778 | . | NM_012079.5:c.731C>T; p.(Pro244Leu) | Probably damaging | Damaging | 0.000009124 | missense | 1 | 109600 | 0 |
| 8 | 145541772 | . | NM_012079.5:c.737A>G; p.(Asn246Ser) | Possibly damaging | Damaging | 0.000009129 | missense | 1 | 109546 | 0 |
| 8 | 145541766 | rs55962377 | NM_012079.5:c.743C>A; p.(Thr248Asn) | Benign | Tolerated | 0.06813 | missense | 7464 | 109560 | 347 |
| 8 | 145541761 | rs144473757 | NM_012079.5:c.748C>T; p.(Arg250Cys) | Benign | Damaging | 0.000192 | missense | 21 | 109400 | 0 |
| 8 | 145541760 | . | NM_012079.5:c.749G>T; p.(Arg250Leu) | Benign | Tolerated | 0.00002743 | missense | 3 | 109350 | 0 |
| 8 | 145541760 | . | NM_012079.5:c.749G>A; p.(Arg250His) | Probably damaging | Tolerated | 0.000009145 | missense | 1 | 109350 | 0 |
| 8 | 145541758 | . | NM_012079.5:c.751G>A; p.(Asp251Asn) | Probably damaging | Tolerated | 0.00003659 | missense | 4 | 109312 | 0 |
| 8 | 145541756 | rs148665132 | NG_034192.1(DGAT1):c. 751+2T>C; p.(Ala226_Arg250del) | | | 0.0001098 | splice donor | 12 | 109286 | 0 |
| 8 | 145541678 | . | NM_012079.5:c.754C>T; p.(Leu252Phe) | Probably damaging | Damaging | 0.000009213 | missense | 1 | 108544 | 0 |
| 8 | 145541669 | . | NM_012079.5:c.763T>C; p.(Phe255Leu) | Probably damaging | Damaging | 0.000009221 | missense | 1 | 108450 | 0 |
| 8 | 145541668 | . | NM_012079.5:c.764T>A; p.(Phe255Tyr) | Probably damaging | Damaging | 0.000009222 | missense | 1 | 108438 | 0 |
| 8 | 145541660 | rs144983092 | NM_012079.5:c.772G>A; p.(Ala258Thr) | Probably damaging | Damaging | 0.00007386 | missense | 8 | 108318 | 0 |
| 8 | 145541653 | . | NM_012079.5:c.779C>A; p.(Thr260Asn) | Probably damaging | Damaging | 0.000009235 | missense | 1 | 108278 | 0 |

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|---|-----------|-------------|--|----------------------|-----------|-------------|--------------------|-----|--------|---|
| 8 | 145541635 | rs146196839 | NM_012079.5:c.797A>G; p.(Asn266Ser) | Probably damaging | Tolerated | 0.001988 | missense | 215 | 108160 | 2 |
| 8 | 145541630 | rs141523495 | NM_012079.5:c.802C>T; p.(Pro268Ser) | Possibly damaging | Damaging | 0.0000185 | missense | 2 | 108104 | 0 |
| 8 | 145541627 | . | NM_012079.5:c.805C>T; p.(Arg269Cys) | Probably damaging | Damaging | 0.00002777 | missense | 3 | 108026 | 0 |
| 8 | 145541626 | rs139792712 | NM_012079.5:c.806G>A; p.(Arg269His) | Probably damaging | Damaging | 0.00008334 | missense | 9 | 107992 | 0 |
| 8 | 145541623 | . | NM_012079.5:c.809C>A; p.(Ser270Tyr) | Possibly damaging | Damaging | 0.00002779 | missense | 3 | 107946 | 0 |
| 8 | 145541620 | . | NM_012079.5:c.812C>T; p.(Pro271Leu) | Benign | Tolerated | 0.00000927 | missense | 1 | 107876 | 0 |
| 8 | 145541618 | . | NM_012079.5:c.814C>T; p.(Arg272Cys) | Probably damaging | Damaging | 0.00002783 | missense | 3 | 107814 | 0 |
| 8 | 145541614 | . | NM_012079.5:c.818T>C; p.(Ile273Thr) | Probably damaging | Damaging | 0.00001856 | missense | 2 | 107760 | 0 |
| 8 | 145541613 | rs146155230 | NM_012079.5:c.819C>G; p.(Ile273Met) | Probably damaging | Damaging | 0.00002784 | missense | 3 | 107742 | 0 |
| 8 | 145541612 | . | NM_012079.5:c.820C>T; p.(Arg274Trp) | Probably damaging | Damaging | 0.00001857 | missense | 2 | 107678 | 0 |
| 8 | 145541611 | . | NM_012079.5:c.821G>A; p.(Arg274Gln) | Probably damaging | Damaging | 0.000009285 | missense | 1 | 107704 | 0 |
| 8 | 145541607 | . | NM_012079.5:c.825G>C; p.(Lys275Asn) | Probably damaging | Tolerated | 0.000009295 | missense | 1 | 107590 | 0 |
| 8 | 145541606 | . | NM_012079.5:c.826C>T; p.(Arg276Cys) | Probably damaging | Damaging | 0.0000186 | missense | 2 | 107536 | 0 |
| 8 | 145541593 | . | NM_012079.5:c.839G>A; p.(Arg280Gln) | Probably damaging | Damaging | 0.000009339 | missense | 1 | 107080 | 0 |
| 8 | 145541591 | . | NM_012079.5:c.841C>T; p.(Arg281Trp) | Probably damaging | Damaging | 0.00000935 | missense | 1 | 106950 | 0 |
| 8 | 145541590 | . | NM_012079.5:c.842G>A; p.(Arg281Gln) | Probably damaging | Tolerated | 0.00003744 | missense | 4 | 106836 | 0 |
| 8 | 145541473 | . | NM_012079.5:c.868C>G; p.(Gln290Glu) | Probably damaging | Tolerated | 0.000008965 | missense | 1 | 111544 | 0 |
| 8 | 145541464 | . | NM_012079.5:c.877G>A; p.(Val293Met) | Possibly damaging | Tolerated | 0.000008871 | missense | 1 | 112724 | 0 |
| 8 | 145541457 | This study | NM_012079.5:c.884T>C; p.(Leu295Pro) | Probably damaging | Damaging | | | | | |
| 8 | 145541376 | . | NG_034192.1(DGAT1):c. 895-1G>A; p.? | | | 0.000008277 | splice acceptor | 1 | 120814 | 0 |
| 8 | 145541359 | . | NM_012079.5:c.911T>C; p.(Ile304Thr) | Possibly damaging | Damaging | 0.000008289 | missense | 1 | 120638 | 0 |
| 8 | 145541252 | . | NG_034192.1(DGAT1):c. 937-1G>A; p.? | | | 0.00002474 | splice acceptor | 3 | 121278 | 0 |

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| 8 | 145541251 | . | NM_012079.5:c.937G>T; p.(Asp313Tyr) | Possibly damaging | Damaging | 0.000008246 | missense | 1 | 121278 | 0 |
| 8 | 145541236 | . | NM_012079.5:c.952C>T; p.(Arg318Cys) | Probably damaging | Tolerated | 0.00004947 | missense | 6 | 121288 | 1 |
| 8 | 145541227 | . | NM_012079.5:c.961G>A; p.(Glu321Lys) | Probably damaging | Damaging | 0.0001649 | missense | 20 | 121282 | 0 |
| 8 | 145541224 | . | NM_012079.5:c.964C>T; p.(Arg322Cys) | Probably damaging | Damaging | 0.00001649 | missense | 2 | 121276 | 0 |
| 8 | 145541211 | . | NM_012079.5:c.977T>A; p.(Leu326Gln) | Probably damaging | Damaging | 0.00001649 | missense | 2 | 121270 | 0 |
| 8 | 145541206 | . | NG_034192.1(DGAT1):c. 981+1G>T; p.? | | | 0.000008246 | splice donor | 1 | 121266 | 0 |
| 8 | 145541087 | . | NM_012079.5:c.1003C>T; p.(Leu335Phe) | Probably damaging | Damaging | 0.00000826 | missense | 1 | 121062 | 0 |
| 8 | 145541084 | . | NM_012079.5:c.1006A>G; p.(Ile336Val) | Benign | Tolerated | 0.00001652 | missense | 2 | 121060 | 0 |
| 8 | 145541081 | . | NM_012079.5:c.1009T>A; p.(Phe337Ile) | Probably damaging | Tolerated | 0.00003304 | missense | 4 | 121050 | 0 |
| 8 | 145541079 | rs149384210 | NM_012079.5:c.1011C>G; p.(Phe337Leu) | Possibly damaging | Tolerated | 0.00000826 | missense | 1 | 121064 | 0 |
| 8 | 145541074 | . | NM_012079.5:c.1013_101 6delTCTAinsTCTT; p.(Tyr339Phe) | Probably damaging | Tolerated | 0.000008261 | missense | 1 | 121046 | 0 |
| 8 | 145541074 | . | NM_012079.5:c.1013_101 6delTCTAinsA; p.(Phe338del) | | | 0.00002478 | inframe deletion | 3 | 121046 | 0 |
| 8 | 145541070 | . | NM_012079.5:c.1020G>A ; p.(Trp340Ter) | | | 0.00004957 | stop gained | 6 | 121036 | 0 |
| 8 | 145541045 | . | NM_012079.5:c.1045G>A; p.(Val349Met) | Possibly damaging | Tolerated | 0.000008265 | missense | 1 | 120996 | 0 |
| 8 | 145541029 | . | NM_012079.5:c.1061A>G; p.(Gln354Arg) | Benign | Tolerated | 0.00001653 | missense | 2 | 120968 | 0 |
| 8 | 145541017 | . | NM_012079.5:c.1073G>A ; p.(Arg358Gln) | Probably damaging | Damaging | 0.00003307 | missense | 4 | 120940 | 0 |
| 8 | 145541008 | . | NM_012079.5:c.1082A>G ; p.(Tyr361Cys) | Probably damaging | Damaging | 0.00000827 | missense | 1 | 120924 | 0 |
| 8 | 145541006 | . | NM_012079.5:c.1084C>T; p.(Arg362Trp) | Probably damaging | Damaging | 0.00002481 | missense | 3 | 120916 | 0 |
| 8 | 145541005 | rs138800445 | NM_012079.5:c.1085G>A; p.(Arg362Gln) | Possibly damaging | Tolerated | 0.00001654 | missense | 2 | 120900 | 0 |
| 8 | 145540902 | . | NM_012079.5:c.1108G>A; p.(Val370Ile) | Benign | Tolerated | 0.000008308 | missense | 1 | 120372 | 0 |
| 8 | 145540898 | . | NM_012079.5:c.1112C>T; p.(Thr371Ile) | Benign | Damaging | 0.000008303 | missense | 1 | 120438 | 0 |

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|---|-----------|-------------|---|----------------------|-----------|-------------|-------------|---|--------|---|
| 8 | 145540890 | . | NM_012079.5:c.1120T>A; p.(Trp374Arg) | Probably damaging | Damaging | 0.000008304 | missense | 1 | 120428 | 0 |
| 8 | 145540887 | . | NM_012079.5:c.1123C>T; p.(Gln375Ter) | | | 0.000008306 | stop gained | 1 | 120390 | 0 |
| 8 | 145540881 | . | NM_012079.5:c.1129T>C; p.(Trp377Arg) | Probably damaging | Damaging | 0.000008303 | missense | 1 | 120438 | 0 |
| 8 | 145540871 | rs146441969 | NM_012079.5:c.1139C>T; p.(Pro380Leu) | Probably damaging | Damaging | 0.000008312 | missense | 1 | 120314 | 0 |
| 8 | 145540868 | . | NM_012079.5:c.1142T>C; p.(Val381Ala) | Probably damaging | Damaging | 0.00000831 | missense | 1 | 120336 | 0 |
| 8 | 145540856 | . | NM_012079.5:c.1154G>A ; p.(Cys385Tyr) | Probably damaging | Damaging | 0.000008312 | missense | 1 | 120306 | 0 |
| 8 | 145540771 | . | NM_012079.5:c.1162C>T; p.(His388Tyr) | Probably damaging | Damaging | 0.00001657 | missense | 2 | 120684 | 0 |
| 8 | 145540765 | . | NM_012079.5:c.1168T>C; p.(Tyr390His) | Probably damaging | Damaging | 0.00002486 | missense | 3 | 120664 | 0 |
| 8 | 145540750 | . | NM_012079.5:c.1183C>T; p.(Arg395Ter) | | | 0.000008293 | stop gained | 1 | 120580 | 0 |
| 8 | 145540749 | . | NM_012079.5:c.1184G>A; p.(Arg395Gln) | Benign | Tolerated | 0.00002488 | missense | 3 | 120570 | 0 |
| 8 | 145540749 | . | NM_012079.5:c.1184G>C ; p.(Arg395Pro) | Probably damaging | Damaging | 0.00001659 | missense | 2 | 120570 | 0 |
| 8 | 145540747 | . | NM_012079.5:c.1186C>T; p.(Arg396Trp) | Probably damaging | Damaging | 0.00003318 | missense | 4 | 120558 | 0 |
| 8 | 145540746 | rs193169779 | NM_012079.5:c.1187G>A; p.(Arg396Gln) | Benign | Tolerated | 0.00007465 | missense | 9 | 120562 | 0 |
| 8 | 145540737 | . | NM_012079.5:c.1196G>C; p.(Ser399Thr) | Benign | Tolerated | 0.000008298 | missense | 1 | 120510 | 0 |
| 8 | 145540726 | . | NM_012079.5:c.1207G>T; p.(Ala403Ser) | Possibly damaging | Tolerated | 0.000008304 | missense | 1 | 120428 | 0 |
| 8 | 145540723 | . | NM_012079.5:c.1210A>G; p.(Arg404Gly) | Benign | Tolerated | 0.00001661 | missense | 2 | 120398 | 0 |
| 8 | 145540719 | . | NM_012079.5:c.1214C>T; p.(Thr405Ile) | Benign | Tolerated | 0.000008309 | missense | 1 | 120352 | 0 |
| 8 | 145540717 | . | NM_012079.5:c.1216G>A; p.(Gly406Arg) | Possibly damaging | Tolerated | 0.00001662 | missense | 2 | 120336 | 0 |
| 8 | 145540716 | . | NM_012079.5:c.1215_121 6delAG; p.(Phe408ProfsTer74) | | | 0.00001662 | frameshift | 2 | 120336 | 0 |
| 8 | 145540713 | . | NM_012079.5:c.1220T>C; p.(Val407Ala) | Probably damaging | Tolerated | 0.000008318 | missense | 1 | 120226 | 0 |
| 8 | 145540701 | . | NM_012079.5:c.1232C>T; p.(Ser411Leu) | Probably damaging | Damaging | 0.000008319 | missense | 1 | 120202 | 0 |
| 8 | 145540694 | . | NM_012079.5:c.1239C>A; p.(Phe413Leu) | Possibly damaging | Tolerated | 0.00001664 | missense | 2 | 120164 | 0 |

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| 8 | 145540693 | . | NM_012079.5:c.1240T>C; p.(Phe414Leu) | Probably damaging | Tolerated | 0.000008323 | missense | 1 | 120156 | 0 |
| 8 | 145540691 | . | NM_012079.5:c.1242C>G; p.(Phe414Leu) | Probably damaging | Tolerated | 0.000008323 | missense | 1 | 120142 | 0 |
| 8 | 145540565 | . | NM_012079.5:c.1262T>A; p.(Val421Asp) | Probably damaging | Damaging | 0.00001883 | missense | 2 | 106222 | 0 |
| 8 | 145540557 | . | NM_012079.5:c.1270C>T; p.(Arg424Ter) | | | 0.000009569 | stop gained | 1 | 104504 | 0 |
| 8 | 145540547 | rs147655123 | NM_012079.5:c.1280G>A; p.(Arg427His) | Probably damaging | Tolerated | 0.000009797 | missense | 1 | 102072 | 0 |
| 8 | 145540545 | . | NM_012079.5:c.1282C>T; p.(Leu428Phe) | Benign | Tolerated | 0.000009822 | missense | 1 | 101814 | 0 |
| 8 | 145540532 | . | NM_012079.5:c.1295C>T; p.(Thr432Met) | Benign | Tolerated | 0.00004096 | missense | 4 | 97664 | 0 |
| 8 | 145540515 | . | NG_034192.1(DGAT1):c. 1311+1G>A; p.? | | | 0.00002157 | splice donor | 2 | 92716 | 0 |
| 8 | 145540514 | . | NG_034192.1(DGAT1):c. 1311+2T>C; p.? | | | 0.00003247 | splice donor | 3 | 92380 | 0 |
| 8 | 145540369 | . | NM_012079.5:c.1315C>T; p.(Pro439Ser) | Probably damaging | Damaging | 0.000009913 | missense | 1 | 100876 | 0 |
| 8 | 145540359 | . | NM_012079.5:c.1325G>A ; p.(Trp442Ter) | | | 0.00001976 | stop gained | 2 | 101192 | 0 |
| 8 | 145540354 | . | NM_012079.5:c.1330G>A; p.(Val444Met) | Probably damaging | Tolerated | 0.00005952 | missense | 6 | 100812 | 0 |
| 8 | 145540348 | . | NM_012079.5:c.1336C>T; p.(Arg446Cys) | Benign | Damaging | 0.00004983 | missense | 5 | 100342 | 0 |
| 8 | 145540347 | . | NM_012079.5:c.1337G>T; p.(Arg446Leu) | Benign | Damaging | 0.00003002 | missense | 3 | 99932 | 0 |
| 8 | 145540347 | . | NM_012079.5:c.1337G>A ; p.(Arg446His) | Probably damaging | Damaging | 0.00003002 | missense | 3 | 99932 | 0 |
| 8 | 145540340 | rs140874999 | NM_012079.5:c.1344C>G; p.(Phe448Leu) | Benign | Tolerated | 0.00001011 | missense | 1 | 98898 | 0 |
| 8 | 145540335 | . | NM_012079.5:c.1349G>T; p.(Gly450Val) | Probably damaging | Tolerated | 0.0000102 | missense | 1 | 98006 | 0 |
| 8 | 145540329 | . | NM_012079.5:c.1355A>G; p.(Tyr452Cys) | Probably damaging | Tolerated | 0.00001036 | missense | 1 | 96550 | 0 |
| 8 | 145540326 | . | NM_012079.5:c.1358G>A ; p.(Gly453Asp) | Probably damaging | Damaging | 0.00001052 | missense | 1 | 95072 | 0 |
| 8 | 145540310 | . | NM_012079.5:c.1374G>A ; p.(Trp458Ter) | | | 0.00002282 | stop gained | 2 | 87640 | 0 |
| 8 | 145540302 | . | NM_012079.5:c.1382T>C; p.(Leu461Pro) | Probably damaging | Damaging | 0.00001211 | missense | 1 | 82552 | 0 |
| 8 | 145540297 | . | NM_012079.5:c.1387A>G; p.(Ile463Val) | Benign | Tolerated | 0.00001245 | missense | 1 | 80294 | 0 |

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|---|-----------|---|---|----------------------|-----------|--------------------|------------|-----|-------|---|
| 8 | 145540294 | . | NM_012079.5:c.1390G>A ; p.(Gly464Arg) | Probably damaging | Damaging | 0.00002579 | missense | 2 | 77560 | 0 |
| 8 | 145540282 | . | NM_012079.5:c.1402G>A; p.(Ala468Thr) | Possibly damaging | Tolerated | 0.0000139 | missense | 1 | 71926 | 0 |
| 8 | 145540279 | . | NM_012079.5:c.1405G>A; p.(Val469Ile) | Possibly damaging | Tolerated | 0.00004196 | missense | 3 | 71504 | 0 |
| 8 | 145540267 | . | NM_012079.5:c.1417G>A; p.(Val473Ile) | Probably damaging | Tolerated | 0.00004565 | missense | 3 | 65722 | 0 |
| 8 | 145540257 | . | NM_012079.5:c.1427A>G ; p.(Tyr476Cys) | Probably damaging | Damaging | 0.00001629 | missense | 1 | 61396 | 0 |
| 8 | 145540249 | . | NM_012079.5:c.1435C>T; p.(Leu479Phe) | Probably damaging | Tolerated | 0.00003468 | missense | 2 | 57670 | 0 |
| 8 | 145540240 | . | NM_012079.5:c.1443dupT ; p.(Glu482Ter) | | | 0.00001836 | frameshift | 1 | 54452 | 0 |
| 8 | 145540230 | . | NM_012079.5:c.1454C>T; p.(Ala485Val) | Benign | Tolerated | 0.005766 | missense | 281 | 48736 | 7 |
| 8 | 145540224 | . | NM_012079.5:c.1460A>G; p.(Glu487Gly) | Benign | Tolerated | 0.00002207 | missense | 1 | 45310 | 0 |
| 8 | 145540222 | . | NM_012079.5:c.1462G>A; p.(Ala488Thr) | Benign | Damaging | 0.00002263 | missense | 1 | 44198 | 0 |
| TOTAL: | | | | | | | | | | |
| MAF of deleterious missense and null: | | | | | | 0.001860985 | | | | |
| Prevalence of deleterious missense and null: | | | | | | 288744.8552 | | | | |

*Intronic splice variants are based on genomic reference sequence (GRCh37/Hg19). MAF refers to mean allele frequency. Probably deleterious alleles were based on Polyphen2 (<http://genetics.bwh.harvard.edu/pph2/>) predictions, while the “damaging” alleles were based on SIFT (<http://sift.jcvi.org>) predictions.