

livewello

Gene Variance report

Elissa Washuta / F

ALLERGY

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|---|------|-----------|--------------|----------|-----------|
| 1 | HLA | rs2155219 | G | GT | +/- |
| 2 | HLA | rs7775228 | C | TT | -/- |

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CLOTTING FACTORS

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|----------|------------|--------------|----------|-----------|
| 1 | CETP | rs1800775 | C | AC | +/- |
| 2 | CYP4V2 | rs13146272 | C | AA | -/- |
| 3 | F10 | rs3211719 | G | AG | +/- |
| 4 | F11 | rs2036914 | T | CT | +/- |
| 5 | F11 | rs2289252 | T | CT | +/- |
| 6 | F12 | rs1801020 | A | GG | -/- |
| 7 | F3 | rs1324214 | A | AG | +/- |
| 8 | F5 | rs6025 | T | CC | -/- |
| 10 | F9 | rs6048 | G | AG | +/- |
| 11 | GP6 | rs1613662 | G | AA | -/- |
| 12 | HRG | rs9898 | T | CT | +/- |
| 13 | ITGB3 | rs5918 | C | TT | -/- |
| 14 | KNGI598T | rs2731672 | T | CC | -/- |
| 15 | NR1I2 | rs1523127 | C | AC | +/- |
| 16 | SERPINC1 | rs2227589 | T | CC | -/- |

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DETOX

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|------------------|------------|--------------|----------|-----------|
| 1 | CTH | rs1021737 | T | GT | +/- |
| 3 | CYP1A1 | rs1799814 | T | GG | -/- |
| 4 | CYP1A1 | rs4986883 | C | TT | -/- |
| 5 | CYP1A2 | rs762551 | C | AA | -/- |
| 7 | CYP1B1 N453S | rs1800440 | C | CT | +/- |
| 11 | CYP2C9 | rs1057910 | C | AA | -/- |
| 12 | CYP2C9*2 C430T | rs1799853 | T | CC | -/- |
| 13 | CYP2D6 | rs1065852 | A | GG | -/- |
| 15 | CYP2D6 T2850C | rs16947 | A | AG | +/- |
| 17 | CYP2E1*1B G9896C | rs2070676 | G | CG | +/- |
| 18 | CYP2E1*4 A4768G | rs6413419 | A | GG | -/- |
| 20 | CYP3A4 | rs2740574 | C | TT | -/- |
| 21 | CYP3A4 | rs4986910 | G | AA | -/- |
| 22 | CYP3A4*2 S222P | rs55785340 | G | AA | -/- |
| 23 | GPX3 | rs8177412 | C | TT | -/- |
| 24 | GSTM1 | rs1056806 | T | CC | -/- |
| 25 | GSTM1 | rs2239892 | G | AA | -/- |
| 26 | GSTM1 | rs4147565 | A | GG | -/- |
| 27 | GSTM1 | rs4147567 | G | AA | -/- |
| 28 | GSTP1 | rs1138272 | T | CC | -/- |
| 29 | GSTP1 | rs1695 | G | AA | -/- |
| 30 | NAT1 | rs4986782 | A | GG | -/- |
| 31 | NAT2 | rs1208 | G | AA | -/- |
| 32 | NAT2 | rs1799930 | A | GG | -/- |
| 33 | NAT2 | rs1799931 | A | AG | +/- |
| 34 | NAT2 | rs1801279 | A | GG | -/- |
| 35 | NAT2 | rs1801280 | C | TT | -/- |

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GLUTEN INTOLERANCE

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|---|----------|-----------|--------------|----------|-----------|
| 1 | HLA | rs2858331 | G | GG | +/+ |
| 2 | HLA-DQA1 | rs2187668 | T | CT | +/- |

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IGA

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|---------------------|-----------|--------------|----------|-----------|
| 1 | CFH | rs6677604 | A | GG | -/- |
| 2 | HLA | rs9271366 | G | AA | -/- |
| 3 | HLA-DPB2 / COL11A2P | rs1883414 | A | GG | -/- |
| 4 | HLA-DQA2 | rs9275224 | A | GG | -/- |
| 5 | HORMAD2 | rs2412971 | A | AG | +/- |
| 6 | IFIH1 | rs1990760 | T | TT | +/+ |
| 7 | IGF1R | rs2229765 | A | AG | +/- |
| 8 | IRF5 | rs4728142 | A | AG | +/- |
| 9 | MTC03P1 | rs2856717 | A | GG | -/- |
| 10 | MTC03P1 | rs9275596 | C | TT | -/- |
| 11 | PSMB8 | rs9357155 | A | AG | +/- |
| 12 | TRAF1 | rs3761847 | G | AG | +/- |

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IGE

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|------------------|------------|--------------|----------|-----------|
| 1 | C3 | rs10402876 | C | GG | -/- |
| 3 | CD14 | rs2569191 | C | -- | NC |
| 4 | DARC | rs2814778 | C | TT | -/- |
| 5 | FCER1A | rs2251746 | C | CC | +/+ |
| 6 | FCER1A | rs2427824 | T | CC | -/- |
| 7 | FCER1A | rs2427827 | T | CC | -/- |
| 8 | FCER1A | rs2427837 | A | AA | +/+ |
| 9 | FCER1A | rs2494262 | A | AA | +/+ |
| 10 | FCER1A / OR10J2P | rs10489854 | T | CC | -/- |
| 11 | IL13 | rs1295685 | A | AG | +/- |
| 12 | IL13 | rs1800925 | T | CC | -/- |
| 13 | IL5 | rs2069812 | G | GG | +/+ |
| 14 | RAD50 | rs17772565 | T | CC | -/- |
| 15 | RAD50 | rs17772583 | G | AA | -/- |
| 16 | RAD50 | rs2040704 | G | AA | -/- |
| 17 | RAD50 | rs2240032 | T | CC | -/- |
| 18 | RAD50 | rs6884762 | T | CC | -/- |
| 19 | RAG1 | rs3740955 | A | AA | +/+ |
| 20 | SOCS1 | rs33977706 | A | CC | -/- |

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IGG

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|---|------------|-----------|--------------|----------|-----------|
| 1 | FCGR2A | rs1801274 | G | GG | +/+ |
| 2 | GSTM3 | rs7483 | T | CT | +/- |
| 3 | Intergenic | rs2013111 | C | CT | +/- |
| 4 | MUC21 | rs1634731 | G | AA | -/- |
| 5 | TBC1D27 | rs3751987 | A | AG | +/- |
| 6 | TNFRSF13B | rs4792800 | G | AA | -/- |

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METHYLATION

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|-------------------|------------|--------------|----------|-----------|
| 1 | ACAT1 | rs3741049 | A | AG | +/- |
| 2 | ACE Del16 | rs4343 | G | AG | +/- |
| 3 | AGT M235T/C4072T | rs699 | A | AG | +/- |
| 4 | AHCY-01 | rs819147 | C | TT | -/- |
| 5 | AHCY-02 | rs819134 | G | AA | -/- |
| 6 | AHCY-19 | rs819171 | C | TT | -/- |
| 7 | ATG9B (NOS3 G10T) | rs7830 | T | GG | -/- |
| 8 | BHMT R239Q | rs3733890 | A | GG | -/- |
| 9 | BHMT-02 | rs567754 | T | CC | -/- |
| 10 | BHMT-04 | rs617219 | C | AA | -/- |
| 11 | BHMT-08 | rs651852 | T | CT | +/- |
| 12 | C18orf56 (TYMS) | rs502396 | T | TT | +/+ |
| 13 | CBS A13637G | rs2851391 | T | CT | +/- |
| 14 | CBS A360A | rs1801181 | A | AG | +/- |
| 15 | CBS C19150T | rs4920037 | A | AG | +/- |
| 16 | CBS C699T | rs234706 | A | AG | +/- |
| 17 | CBS N212N | rs2298758 | A | GG | -/- |
| 18 | CLCN6 | rs13306560 | T | CC | -/- |
| 19 | CLCN6 | rs13306561 | G | AA | -/- |
| 21 | DAO | rs2070586 | A | GG | -/- |
| 22 | DAO | rs2111902 | G | TT | -/- |
| 23 | DAO | rs3741775 | C | AC | +/- |
| 24 | DHFR | rs1643649 | C | CT | +/- |
| 25 | FOLR1 | rs2071010 | A | GG | -/- |
| 26 | FOLR2 | rs651933 | G | AG | +/- |
| 27 | FOLR3 | rs7925545 | G | AA | -/- |
| 28 | FOLR3 | rs7926875 | A | CC | -/- |
| 29 | FUT2 | rs492602 | G | AA | -/- |
| 30 | FUT2 | rs601338 | A | GG | -/- |
| 31 | FUT2 | rs602662 | A | GG | -/- |
| 32 | G6PD | rs1050828 | T | CC | -/- |
| 33 | G6PD | rs1050829 | C | TT | -/- |
| 34 | GAD1 | rs10432420 | A | AG | +/- |

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|--------------------------|------------|--------------|----------|-----------|
| 35 | GAD1 | rs12185692 | A | AA | +/+ |
| 36 | GAD1 | rs2058725 | C | TT | -/- |
| 37 | GAD1 | rs2241165 | C | TT | -/- |
| 38 | GAD1 | rs3749034 | A | GG | -/- |
| 39 | GAD1 | rs3791850 | A | GG | -/- |
| 40 | GAD1 | rs3791851 | C | TT | -/- |
| 41 | GAD1 | rs3791878 | T | GG | -/- |
| 42 | GAD1 | rs3828275 | T | TT | +/+ |
| 43 | GAD1 | rs701492 | T | CC | -/- |
| 44 | GAD1 | rs769395 | G | AA | -/- |
| 45 | GAD1 | rs769407 | C | GG | -/- |
| 46 | GAD2 | rs1805398 | T | GG | -/- |
| 47 | GAMT | rs17851582 | A | GG | -/- |
| 48 | GAMT | rs55776826 | T | CC | -/- |
| 49 | GIF | rs558660 | A | AG | +/- |
| 50 | MAOA R297R | rs6323 | G | TT | -/- |
| 51 | MAOB | rs1799836 | C | CT | +/- |
| 52 | MIR4761 (COMT -61 P199P) | rs769224 | A | GG | -/- |
| 53 | MIR4761 (COMT H62H) | rs4633 | T | CT | +/- |
| 54 | MIR4761 (COMT V158M) | rs4680 | A | AG | +/- |
| 55 | MIR4761 (COMT) | rs6269 | G | AG | +/- |
| 56 | MTHFD1 C105T | rs1076991 | T | CC | -/- |
| 57 | MTHFD1 G1958A | rs2236225 | A | AG | +/- |
| 58 | MTHFD1L | rs11754661 | A | GG | -/- |
| 59 | MTHFD1L | rs17349743 | C | CT | +/- |
| 60 | MTHFD1L | rs6922269 | A | AG | +/- |
| 61 | MTHFD1L | rs803422 | A | GG | -/- |
| 62 | MTHFR | rs12121543 | A | CC | -/- |
| 63 | MTHFR | rs1476413 | T | CC | -/- |
| 64 | MTHFR | rs17037390 | A | GG | -/- |
| 65 | MTHFR | rs17037396 | T | CC | -/- |
| 66 | MTHFR | rs4846049 | T | GG | -/- |
| 67 | MTHFR (LOC100506310) | rs4846048 | G | AA | -/- |
| 68 | MTHFR 03 P39P | rs2066470 | A | GG | -/- |
| 69 | MTHFR A1298C | rs1801131 | G | TT | -/- |
| 70 | MTHFR A1572G | rs17367504 | G | AA | -/- |
| 71 | MTHFR C677T | rs1801133 | A | AG | +/- |

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|----------------------|-----------|--------------|----------|-----------|
| 72 | MTHFR G1793A (R594Q) | rs2274976 | T | CC | -/- |
| 73 | MTHFS | rs6495446 | T | CT | +/- |
| 74 | MTR A2756G | rs1805087 | G | AA | -/- |
| 75 | MTRR A66G | rs1801394 | G | GG | +/+ |
| 76 | MTRR H595Y | rs10380 | T | CC | -/- |
| 77 | MTRR K350A | rs162036 | G | AA | -/- |
| 78 | MTRR R415T | rs2287780 | T | CC | -/- |
| 79 | MTRR-11 A664A | rs1802059 | A | AG | +/- |
| 80 | NOS1 | rs3782206 | T | CT | +/- |
| 81 | NOS2 | rs2248814 | A | AG | +/- |
| 82 | NOS2 | rs2274894 | T | GT | +/- |
| 83 | NOS2 | rs2297518 | A | AG | +/- |
| 84 | NOS3 | rs1800779 | G | GG | +/+ |
| 85 | NOS3 | rs1800783 | A | AA | +/+ |
| 87 | NOS3 T786C | rs2070744 | C | -- | NC |
| 88 | PEMT | rs4244593 | T | GT | +/- |
| 90 | SHMT1 C1420T | rs1979277 | A | GG | -/- |
| 91 | SLC19A1 | rs1888530 | C | -- | NC |
| 92 | SLC19A1 | rs3788200 | A | AA | +/+ |
| 93 | SOD2 | rs2758331 | A | AA | +/+ |
| 94 | SOD2 | rs4880 | G | GG | +/+ |
| 95 | SOD3 | rs2855262 | C | CC | +/+ |
| 96 | TCN1 | rs526934 | G | AA | -/- |
| 97 | TCN2 C766G | rs1801198 | G | CG | +/- |
| 98 | VDR Bsm | rs1544410 | T | CT | +/- |

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MITOCHONDRIAL FUNCTION

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|-----------------|------------|--------------|----------|-----------|
| 1 | ATP5C1 | rs1244414 | T | CC | -/- |
| 2 | ATP5C1 | rs1244422 | T | CT | +/- |
| 3 | ATP5C1 | rs12770829 | T | CC | -/- |
| 4 | ATP5C1 | rs2778475 | A | AG | +/- |
| 5 | ATP5C1 | rs4655 | C | TT | -/- |
| 6 | ATP5G3 | rs185584 | G | AA | -/- |
| 7 | ATP5G3 | rs36089250 | C | TT | -/- |
| 8 | CCL2 | rs1024611 | G | GG | +/+ |
| 9 | COX5A | rs8042694 | G | AA | -/- |
| 10 | COX6C | rs1135382 | A | GG | -/- |
| 11 | COX6C | rs12544943 | G | -- | NC |
| 12 | COX6C | rs4510829 | A | GG | -/- |
| 13 | COX6C | rs4626565 | C | TT | -/- |
| 14 | COX6C | rs7828241 | C | AA | -/- |
| 15 | COX6C | rs7844439 | A | CC | -/- |
| 16 | NDUFS3 | rs4147730 | A | GG | -/- |
| 17 | NDUFS3 | rs4147731 | A | GG | -/- |
| 18 | NDUFS3 (KBTBD4) | rs2233354 | C | TT | -/- |
| 19 | NDUFS7 | rs1142530 | T | CT | +/- |
| 20 | NDUFS7 | rs11666067 | A | AC | +/- |
| 21 | NDUFS7 | rs2074895 | A | AC | +/- |
| 22 | NDUFS7 | rs2332496 | A | AG | +/- |
| 23 | NDUFS7 | rs7254913 | G | AG | +/- |
| 24 | NDUFS7 | rs7258846 | T | GT | +/- |
| 25 | NDUFS7 | rs809359 | G | AA | -/- |
| 27 | NDUFS8 | rs1104739 | C | AA | -/- |
| 28 | NDUFS8 | rs1122731 | A | GG | -/- |
| 29 | NDUFS8 | rs2075626 | C | TT | -/- |
| 30 | NDUFS8 | rs3115546 | G | TT | -/- |
| 31 | NDUFS8 | rs4147776 | C | AA | -/- |
| 32 | NDUFS8 | rs999571 | A | GG | -/- |
| 33 | SLC19A1 | rs1051266 | C | TT | -/- |
| 34 | UQCRC2 | rs11648723 | T | GT | +/- |

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|--------|-----------|--------------|----------|-----------|
| 35 | UQCRC2 | rs4850 | A | GG | -/- |
| 36 | UQCRC2 | rs6497563 | C | TT | -/- |

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OTHER IMMUNE FACTORS

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|-------------|------------|--------------|----------|-----------|
| 1 | 4q27 Region | rs6822844 | T | GT | +/- |
| 2 | ADD1 G460W | rs4961 | T | GG | -/- |
| 3 | APOE | rs429358 | C | TT | -/- |
| 4 | ATG16L1 | rs10210302 | T | CT | +/- |
| 5 | HLA-DRB1 | rs660895 | G | AA | -/- |
| 6 | IL13 | rs20541 | A | AG | +/- |
| 7 | IL4R | rs1801275 | G | AG | +/- |
| 8 | MEFV | rs11466023 | A | GG | -/- |
| 10 | STAT4 | rs10181656 | G | CC | -/- |
| 11 | TNF | rs1800629 | A | GG | -/- |
| 12 | TNF | rs361525 | A | GG | -/- |

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SULFOTRANSFERASE

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|----|---------|------------|--------------|----------|-----------|
| 2 | SULT1A1 | rs1801030 | C | TT | -/- |
| 3 | SULT1A1 | rs4149381 | G | TT | -/- |
| 4 | SULT1A1 | rs6498090 | A | GG | -/- |
| 5 | SULT1A1 | rs7192559 | T | CC | -/- |
| 6 | SULT1A1 | rs7193599 | C | AA | -/- |
| 7 | SULT1A1 | rs8057055 | A | CC | -/- |
| 8 | SULT1A1 | rs9282862 | C | TT | -/- |
| 9 | SULT2A1 | rs11083907 | A | GG | -/- |
| 10 | SULT2A1 | rs11569679 | T | CC | -/- |
| 11 | SULT2A1 | rs2547231 | C | CC | +/+ |
| 12 | SULT2A1 | rs2547242 | C | TT | -/- |
| 13 | SULT2A1 | rs2910393 | T | TT | +/+ |
| 15 | SULT2A1 | rs296366 | T | TT | +/+ |
| 16 | SULT2A1 | rs4149448 | G | AA | -/- |
| 17 | SULT2A1 | rs4149449 | T | CC | -/- |
| 18 | SULT2A1 | rs4149452 | T | CC | -/- |
| 19 | SULT2A1 | rs8113396 | G | AA | -/- |

A gene variance report is a graphical representation of your genetic raw data, displayed as a color coded chart. Phenotypes are determined based on the presence of known variant alleles in your genotype. Alleles are considered variant if they are the minor allele i.e they occur with less frequency (MAF) in the default global population. In very few instances, the minor allele will not represent a mutation. In such cases, homozygous and/or heterozygous phenotypes may actually be normal. As such, what is normal or abnormal should not be determined solely based on phenotypes displayed in this report. All alleles are reported in reference to the forward strand. rsIDs and genotype information are obtained from the genetic raw data prepared by your personal genomic service. Minor allele frequency (MAF), RefSNP and gene variation/SNP names are obtained directly from dbSNP which is a free public archive for genetic variation maintained by the NCBI <http://www.ncbi.nlm.nih.gov/snp/>.

Report generated by Livewello Gene Variance Software. <http://livewello.com/genetics> v2.0

Do not make any decisions about your health solely based on the information contained in this report. Always consult with a licensed and experienced health practitioner when you receive your report

Elissa Washuta / F

THYROID

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|---|-------|------------|--------------|----------|-----------|
| 1 | CTLA4 | rs231775 | G | AA | -/- |
| 2 | FOXE1 | rs10984009 | A | AG | +/- |
| 3 | FOXE1 | rs1867277 | A | AA | +/+ |
| 4 | FOXE1 | rs7043516 | C | AC | +/- |

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Elissa Washuta / F

TONGUE TIE

| | Gene | rsID | Minor Allele | Genotype | Phenotype |
|---|-------|------------|--------------|----------|-----------|
| 1 | IRF6 | rs861020 | A | GG | -/- |
| 2 | IRF6 | rs987525 | A | AA | +/+ |
| 3 | RARA | rs7217852 | G | AA | -/- |
| 4 | RARA | rs9904270 | T | CC | -/- |
| 5 | TBX22 | rs41307258 | A | AT | +/- |

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Gene Definitions

HLA [+/-]

SS2; BTN7; BTL-II; HSBLMHC1

CETP [+/-]

The protein encoded by this gene is found in plasma, where it is involved in the transfer of cholesteryl ester from high density lipoprotein (HDL) to other lipoproteins. Defects in this gene are a cause of hyperalphalipoproteinemia 1 (HALP1). Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Oct 2013]

F10 [+/-]

Normally, if you get hurt, your body forms a blood clot to stop the bleeding. For blood to clot, your body needs cells called platelets and proteins known as clotting factors. If you have a bleeding disorder, you either do not have enough platelets or clotting factors or they don't work the way they should. Bleeding disorders can be the result of other diseases, such as severe liver disease. They can also be inherited. Hemophilia is an inherited bleeding disorder. Bleeding disorders can also be a side effect of medicines.

F11 [+/-]

This gene encodes coagulation factor XI of the blood coagulation cascade. This protein is present in plasma as a zymogen, which is a unique plasma coagulation enzyme because it exists as a homodimer consisting of two identical polypeptide chains linked by disulfide bonds. During activation of the plasma factor XI, an internal peptide bond is cleaved by factor XIIa (or XII) in each of the two chains, resulting in activated factor XIa, a serine protease composed of two heavy and two light chains held together by disulfide bonds. This activated plasma factor XI triggers the middle phase of the intrinsic pathway of blood coagulation by activating factor IX. Defects in this factor lead to Rosenthal syndrome, a blood coagulation abnormality. [provided by RefSeq, Jul 2008]

F3 [+/-]

This gene encodes coagulation factor III which is a cell surface glycoprotein. This factor enables cells to initiate the blood coagulation cascades, and it functions as the high-affinity receptor for the coagulation factor VII. The resulting complex provides a catalytic event that is responsible for initiation of the coagulation protease cascades by specific limited proteolysis.

F9 [+/-]

Mutations in the F9 gene cause a type of hemophilia called hemophilia B. More than 900 alterations in this gene have been identified. The most common mutations change single DNA building blocks (base pairs) in the gene. A small percentage of mutations delete or insert multiple base pairs or rearrange segments of DNA within the gene. Mutations in the F9 gene lead to the production of an abnormal version of coagulation factor IX or reduce the amount of this protein. The altered or missing protein cannot participate effectively in the blood clotting process. As a result, blood clots cannot form properly in response to injury. These problems with blood clotting lead to excessive bleeding that can be difficult to control. Mutations that completely eliminate the activity of coagulation factor IX result in severe

hemophilia. Mutations that reduce but do not eliminate the protein's activity usually cause mild or moderate hemophilia. Several mutations near the beginning of the F9 gene sequence cause an unusual form of hemophilia known as hemophilia B Leyden. People with these mutations are born with very low levels of functional coagulation factor IX, but hormonal changes cause the levels of this protein to increase gradually during puberty. As a result, adults with hemophilia B Leyden rarely experience episodes of abnormal bleeding.

HRG [+/-]

This histidine-rich glycoprotein contains two cystatin-like domains and is located in plasma and platelets. The physiological function has not been determined but it is known that the protein binds heme, dyes and divalent metal ions. It can inhibit rosette formation and interacts with heparin, thrombospondin and plasminogen. Two of the protein's effects, the inhibition of fibrinolysis and the reduction of inhibition of coagulation, indicate a potential prothrombotic effect. Mutations in this gene lead to thrombophilia due to abnormal histidine-rich glycoprotein levels. [provided by RefSeq, Jul 2008]

NR1I2 [+/-]

This gene product belongs to the nuclear receptor superfamily, members of which are transcription factors characterized by a ligand-binding domain and a DNA-binding domain. The encoded protein is a transcriptional regulator of the cytochrome P450 gene CYP3A4, binding to the response element of the CYP3A4 promoter as a heterodimer with the 9-cis retinoic acid receptor RXR.

CTH [+/-]

This gene encodes a cytoplasmic enzyme in the trans-sulfuration pathway that converts cystathione derived from methionine into cysteine. Glutathione synthesis in the liver is dependent upon the availability of cysteine. Mutations in this gene cause cystathioninuria. Alternative splicing of this gene results in three transcript variants encoding different isoforms. [provided by RefSeq, Jun 2010]

CYP1B1 N453S [+/-]

More than 140 CYP1B1 gene mutations have been identified to cause early-onset glaucoma. People with this condition experience increased pressure within the eye before the age of 40. This pressure causes damage to the optic nerves connecting the eyes and the brain. Between 20 percent and 40 percent of people with glaucoma before the age of 5 (primary congenital glaucoma) have mutations in the CYP1B1 gene. The mutations that cause early-onset glaucoma may result in an enzyme that is unstable or the wrong shape. It is not well understood how defects in the CYP1B1 enzyme cause signs and symptoms of glaucoma. Recent studies suggest that the defects may interfere with the early development of the trabecular meshwork, which is a network of mesh-like canals that helps drain excess fluid from the eye. If fluid cannot be drained, pressure inside the eye may increase, which is characteristic of glaucoma. The CYP1B1 enzyme may interact with another protein called myocilin, which is produced from the MYOC gene. Individuals with mutations in both the MYOC and CYP1B1 genes may develop glaucoma at an earlier age and have more severe symptoms than do those with mutations in only one of the genes.

CYP2D6 T2850C [+/-]

Caffeine is a bitter substance found in coffee, tea, soft drinks, chocolate, kola nuts, and certain medicines. It has many effects on the body's metabolism, including stimulating the central nervous system. This can make you more alert and give you a boost of energy. For most people, the amount of caffeine in two to four cups of coffee a day is not harmful. However, too much caffeine can cause problems. It can make you jittery and shaky. Make it hard to fall asleep or stay

asleepCause headaches or dizzinessMake your heart beat faster or cause abnormal heart rhythmsCause dehydrationMake you dependent on it so you need to take more of it. If you stop using caffeine, you could get withdrawal symptoms.Some people are more sensitive to the effects of caffeine than others. They should limit their use of caffeine. So should pregnant and nursing women. Certain drugs and supplements may interact with caffeine. If you have questions about whether caffeine is safe for you, talk with your health care provider.Food and Drug Administration

CYP2E1*1B G9896C [+/-]

This gene encodes a member of the cytochrome P450 superfamily of enzymes. The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids and other lipids. This protein localizes to the endoplasmic reticulum and is induced by ethanol, the diabetic state, and starvation. The enzyme metabolizes both endogenous substrates, such as ethanol, acetone, and acetal, as well as exogenous substrates including benzene, carbon tetrachloride, ethylene glycol, and nitrosamines which are premutagens found in cigarette smoke. Due to its many substrates, this enzyme may be involved in such varied processes as gluconeogenesis, hepatic cirrhosis, diabetes, and cancer. [provided by RefSeq, Jul 2008]

NAT2 [+/-]

Note: Loci in other organisms that are functionally homologous to this one are validly referred to as both NAT1 and NAT2; i.e., the functional homologs of NAT1 include mouse and rat Nat2, while the functional homologs of human NAT2 include mouse and rat Nat1. Name:sequence associations are consistent with current use in the field. [27 Apr 2009]

HLA-DQA1 [+/-]

HLA-DQA1 belongs to the HLA class II alpha chain paralogues. The class II molecule is a heterodimer consisting of an alpha (DQA) and a beta chain (DQB), both anchored in the membrane. It plays a central role in the immune system by presenting peptides derived from extracellular proteins. Class II molecules are expressed in antigen presenting cells (APC: B Lymphocytes, dendritic cells, macrophages). The alpha chain is approximately 33-35 kDa. It is encoded by 5 exons; exon 1 encodes the leader peptide, exons 2 and 3 encode the two extracellular domains, and exon 4 encodes the transmembrane domain and the cytoplasmic tail. Within the DQ molecule both the alpha chain and the beta chain contain the polymorphisms specifying the peptide binding specificities, resulting in up to four different molecules. Typing for these polymorphisms is routinely done for bone marrow transplantation. [provided by RefSeq, Jul 2008]

HORMAD2 [+/-]

CT46.2

IFIH1 [+/+]

DEAD box proteins, characterized by the conserved motif Asp-Glu-Ala-Asp (DEAD), are putative RNA helicases. They are implicated in a number of cellular processes involving alteration of RNA secondary structure such as translation initiation, nuclear and mitochondrial splicing, and ribosome and spliceosome assembly. Based on their distribution patterns, some members of this family are believed to be involved in embryogenesis, spermatogenesis, and cellular growth and division. This gene encodes a DEAD box protein that is upregulated in response to treatment with beta-interferon and a protein kinase C-activating compound, mezerein. Irreversible reprogramming of melanomas can be achieved by treatment with both these agents; treatment with either agent alone only achieves reversible differentiation. Genetic variation in this gene is associated with diabetes mellitus insulin-dependent type 19. [provided by RefSeq, Jul 2012]

IGF1R [+/-]

This receptor binds insulin-like growth factor with a high affinity. It has tyrosine kinase activity. The insulin-like growth factor I receptor plays a critical role in transformation events. Cleavage of the precursor generates alpha and beta subunits. It is highly overexpressed in most malignant tissues where it functions as an anti-apoptotic agent by enhancing cell survival. Alternatively spliced transcript variants encoding distinct isoforms have been found for this gene. [provided by RefSeq, May 2014]

IRF5 [+/-]

This gene encodes a member of the interferon regulatory factor (IRF) family, a group of transcription factors with diverse roles, including virus-mediated activation of interferon, and modulation of cell growth, differentiation, apoptosis, and immune system activity. Members of the IRF family are characterized by a conserved N-terminal DNA-binding domain containing tryptophan (W) repeats. Multiple transcript variants encoding different isoforms have been found for this gene, and a 30-nt indel polymorphism (SNP rs60344245) can result in loss of a 10-aa segment. [provided by RefSeq, Mar 2010]

PSMB8 [+/-]

At least one mutation in the PSMB8 gene has been found to cause Nakajo-Nishimura syndrome, a condition that has been described only in the Japanese population. The identified mutation changes a single protein building block (amino acid) in the protein produced from the PSMB8 gene, replacing the amino acid glycine with the amino acid valine at protein position 201 (written as Gly201Val or G201V). This mutation greatly reduces the production of this protein, which impairs the normal assembly of immunoproteasomes and causes the immune system to malfunction. For unknown reasons, the malfunctioning immune system triggers abnormal inflammation that can damage tissues throughout the body. Abnormal inflammation likely underlies many of the signs and symptoms of Nakajo-Nishimura syndrome, including the development of red, swollen lumps (nodular erythema) on the skin, recurrent fevers, joint problems, and an enlarged liver and spleen (hepatosplenomegaly). It is less clear how mutations in the PSMB8 gene lead to other characteristic features of the condition, including muscle weakness and wasting and a loss of fatty tissue (lipodystrophy), mainly in the upper body. Because the protein produced from the PSMB8 gene may be involved in the maturation of adipocytes, studies suggest that a shortage of this protein may interfere with the normal development and function of these cells.

TRAF1 [+/-]

The protein encoded by this gene is the fifth component of complement, which plays an important role in inflammatory and cell killing processes. This protein is comprised of alpha and beta polypeptide chains that are linked by a disulfide bridge. An activation peptide, C5a, which is an anaphylatoxin that possesses potent spasmogenic and chemotactic activity, is derived from the alpha polypeptide via cleavage with a convertase. The C5b macromolecular cleavage product can form a complex with the C6 complement component, and this complex is the basis for formation of the membrane attack complex, which includes additional complement components. Mutations in this gene cause complement component 5 deficiency, a disease where patients show a propensity for severe recurrent infections. Defects in this gene have also been linked to a susceptibility to liver fibrosis and to rheumatoid arthritis. [provided by RefSeq, Jul 2008]

FCER1A [+/+]

The immunoglobulin epsilon receptor (IgE receptor) is the initiator of the allergic response. When two or more high-affinity IgE receptors are brought together by allergen-bound IgE molecules, mediators such as histamine that are responsible for allergy symptoms are released. This receptor is comprised of an alpha subunit, a beta subunit, and two gamma subunits. The protein encoded by this gene represents the alpha subunit. [provided by RefSeq, Aug 2011]

IL13 [+/-]

This gene encodes an immunoregulatory cytokine produced primarily by activated Th2 cells. This cytokine is involved in several stages of B-cell maturation and differentiation. It up-regulates CD23 and MHC class II expression, and promotes IgE isotype switching of B cells. This cytokine down-regulates macrophage activity, thereby inhibits the production of pro-inflammatory cytokines and chemokines. This cytokine is found to be critical to the pathogenesis of allergen-induced asthma but operates through mechanisms independent of IgE and eosinophils. This gene, IL3, IL5, IL4, and CSF2 form a cytokine gene cluster on chromosome 5q, with this gene particularly close to IL4. [provided by RefSeq, Jul 2008]

IL5 [+/+]

This gene encodes a cytokine that acts as a growth and differentiation factor for both B cells and eosinophils. The encoded cytokine plays a major role in the regulation of eosinophil formation, maturation, recruitment and survival. The increased production of this cytokine may be related to pathogenesis of eosinophil-dependent inflammatory diseases. This cytokine functions by binding to its receptor, which is a heterodimer, whose beta subunit is shared with the receptors for interleukin 3 (IL3) and colony stimulating factor 2 (CSF2/GM-CSF). This gene is located on chromosome 5 within a cytokine gene cluster which includes interleukin 4 (IL4), interleukin 13 (IL13), and CSF2 . This gene, IL4, and IL13 may be regulated coordinately by long-range regulatory elements spread over 120 kilobases on chromosome 5q31. [provided by RefSeq, Jul 2013]

RAG1 [+/+]

Your immune system is a complex network of cells, tissues, and organs that work together to defend against germs. It helps your body to recognize these "foreign" invaders. Then its job is to keep them out, or if it can't, to find and destroy them. If your immune system cannot do its job, the results can be serious. Disorders of the immune system include Allergy and asthma - immune responses to substances that are usually not harmful Immune deficiency diseases - disorders in which the immune system is missing one or more of its parts Autoimmune diseases - diseases causing your immune system to attack your own body's cells and tissues by mistake NIH: National Institute of Allergy and Infectious Diseases

FCGR2A [+/+]

This gene encodes one member of a family of immunoglobulin Fc receptor genes found on the surface of many immune response cells. The protein encoded by this gene is a cell surface receptor found on phagocytic cells such as macrophages and neutrophils, and is involved in the process of phagocytosis and clearing of immune complexes. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Oct 2008]

GSTM3 [+/-]

Cytosolic and membrane-bound forms of glutathione S-transferase are encoded by two distinct supergene families. At present, eight distinct classes of the soluble cytoplasmic mammalian glutathione S-transferases have been identified: alpha, kappa, mu, omega, pi, sigma, theta and zeta. This gene encodes a glutathione S-transferase that belongs to the mu class. The mu class of enzymes functions in the detoxification of electrophilic compounds, including carcinogens,

therapeutic drugs, environmental toxins and products of oxidative stress, by conjugation with glutathione. The genes encoding the mu class of enzymes are organized in a gene cluster on chromosome 1p13.3 and are known to be highly polymorphic. These genetic variations can change an individual's susceptibility to carcinogens and toxins as well as affect the toxicity and efficacy of certain drugs. Mutations of this class mu gene have been linked with a slight increase in a number of cancers, likely due to exposure with environmental toxins. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Nov 2008]

TBC1D27 [+/-]

Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo

ACAT1 [+/-]

More than 40 mutations in the ACAT1 gene have been identified in people with beta-ketothiolase deficiency. Some of these genetic changes disrupt the normal function of the enzyme, while other mutations prevent cells from producing any functional enzyme. A shortage of the ACAT1 enzyme prevents the body from processing proteins and fats properly. As a result, chemical byproducts called organic acids can build up to toxic levels in the blood. These substances cause the blood to become too acidic (ketoacidosis), which can damage the body's tissues and organs, particularly in the nervous system. This damage leads to episodes of vomiting, dehydration, and other health problems associated with beta-ketothiolase deficiency.

ACE Del16 [+/-]

This gene encodes an enzyme involved in catalyzing the conversion of angiotensin I into a physiologically active peptide angiotensin II. Angiotensin II is a potent vasopressor and aldosterone-stimulating peptide that controls blood pressure and fluid-electrolyte balance. This enzyme plays a key role in the renin-angiotensin system. Many studies have associated the presence or absence of a 287 bp Alu repeat element in this gene with the levels of circulating enzyme or cardiovascular pathophysiologies. Multiple alternatively spliced transcript variants encoding different isoforms have been identified, and two most abundant spliced variants encode the somatic form and the testicular form, respectively, that are equally active. [provided by RefSeq, May 2010]

AGT M235T/C4072T [+/-]

At least six mutations in the AGT gene have been found to cause a severe kidney disorder called renal tubular dysgenesis. This condition is characterized by abnormal kidney development before birth, the inability to produce urine (anuria), and severe low blood pressure (hypotension). These problems result in a reduction of amniotic fluid (oligohydramnios), which leads to a set of birth defects known as the Potter sequence. Renal tubular dysgenesis can be caused by mutations in both copies of any of the genes involved in the renin-angiotensin system. Most of the mutations in the AGT gene that cause this disorder change single protein building blocks (amino acids) in the angiotensinogen protein. These changes occur in a region of the protein that is necessary for its conversion to angiotensin I. It is thought that the altered angiotensinogen cannot be converted, leading to a nonfunctional renin-angiotensin system. Without this system, the kidneys cannot control blood pressure. Because of low blood pressure, the flow of blood is reduced (hypoperfusion), and the body does not get enough oxygen during fetal development. As a result, kidney development is impaired, leading to the features of renal tubular dysgenesis.

CBS A13637G [+/-]

More than 150 mutations that cause homocystinuria have been identified in the CBS gene. Most of these mutations change single amino acids in cystathionine beta-synthase. The most common mutation substitutes the amino acid threonine for the amino acid isoleucine at position 278 in the enzyme (written as Ile278Thr or I278T). Another common mutation, which is the most frequent cause of homocystinuria in the Irish population, replaces the amino acid glycine with the amino acid serine at position 307 (written as Gly307Ser or G307S). These mutations disrupt the normal function of cystathionine beta-synthase. As a result, homocysteine and other potentially toxic compounds build up in the blood, and homocysteine is excreted in urine. Researchers have not determined how excess homocysteine leads to the signs and symptoms of homocystinuria.

CBS A360A [+/-]

More than 150 mutations that cause homocystinuria have been identified in the CBS gene. Most of these mutations change single amino acids in cystathionine beta-synthase. The most common mutation substitutes the amino acid threonine for the amino acid isoleucine at position 278 in the enzyme (written as Ile278Thr or I278T). Another common mutation, which is the most frequent cause of homocystinuria in the Irish population, replaces the amino acid glycine with the amino acid serine at position 307 (written as Gly307Ser or G307S). These mutations disrupt the normal function of cystathionine beta-synthase. As a result, homocysteine and other potentially toxic compounds build up in the blood, and homocysteine is excreted in urine. Researchers have not determined how excess homocysteine leads to the signs and symptoms of homocystinuria.

CBS C19150T [+/-]

More than 150 mutations that cause homocystinuria have been identified in the CBS gene. Most of these mutations change single amino acids in cystathionine beta-synthase. The most common mutation substitutes the amino acid threonine for the amino acid isoleucine at position 278 in the enzyme (written as Ile278Thr or I278T). Another common mutation, which is the most frequent cause of homocystinuria in the Irish population, replaces the amino acid glycine with the amino acid serine at position 307 (written as Gly307Ser or G307S). These mutations disrupt the normal function of cystathionine beta-synthase. As a result, homocysteine and other potentially toxic compounds build up in the blood, and homocysteine is excreted in urine. Researchers have not determined how excess homocysteine leads to the signs and symptoms of homocystinuria.

CBS C699T [+/-]

More than 150 mutations that cause homocystinuria have been identified in the CBS gene. Most of these mutations change single amino acids in cystathionine beta-synthase. The most common mutation substitutes the amino acid threonine for the amino acid isoleucine at position 278 in the enzyme (written as Ile278Thr or I278T). Another common mutation, which is the most frequent cause of homocystinuria in the Irish population, replaces the amino acid glycine with the amino acid serine at position 307 (written as Gly307Ser or G307S). These mutations disrupt the normal function of cystathionine beta-synthase. As a result, homocysteine and other potentially toxic compounds build up in the blood, and homocysteine is excreted in urine. Researchers have not determined how excess homocysteine leads to the signs and symptoms of homocystinuria.

DAO [+/-]

Schizophrenia is a serious brain illness. People who have it may hear voices that aren't there. They may think other people are trying to hurt them. Sometimes they don't make sense when they talk. The disorder makes it hard for them to keep a job or take care of themselves. Symptoms of schizophrenia usually start between ages 16 and 30. Men often develop symptoms at a younger age than women. People usually do not get schizophrenia after age 45. There are three types of symptoms: Psychotic symptoms distort a person's thinking. These include hallucinations (hearing or seeing things that are not there), delusions (beliefs that are not true), trouble organizing thoughts, and strange movements. "Negative" symptoms make it difficult to show emotions and to function normally. A person may seem

depressed and withdrawn. Cognitive symptoms affect the thought process. These include trouble using information, making decisions, and paying attention. No one is sure what causes schizophrenia. Your genes, environment, and brain chemistry may play a role. There is no cure. Medicine can help control many of the symptoms. You may need to try different medicines to see which works best. You should stay on your medicine for as long as your doctor recommends. Additional treatments can help you deal with your illness from day to day. These include therapy, family education, rehabilitation, and skills training. NIH: National Institute of Mental Health

DHFR [+/-]

Dihydrofolate reductase converts dihydrofolate into tetrahydrofolate, a methyl group shuttle required for the de novo synthesis of purines, thymidylic acid, and certain amino acids. While the functional dihydrofolate reductase gene has been mapped to chromosome 5, multiple intronless processed pseudogenes or dihydrofolate reductase-like genes have been identified on separate chromosomes. Dihydrofolate reductase deficiency has been linked to megaloblastic anemia. Several transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Mar 2014]

FOLR2 [+/-]

The protein encoded by this gene is a member of the folate receptor (FOLR) family, and these genes exist in a cluster on chromosome 11. Members of this gene family have a high affinity for folic acid and for several reduced folic acid derivatives, and they mediate delivery of 5-methyltetrahydrofolate to the interior of cells. This protein has a 68% and 79% sequence homology with the FOLR1 and FOLR3 proteins, respectively. Although this protein was originally thought to be specific to placenta, it can also exist in other tissues, and it may play a role in the transport of methotrexate in synovial macrophages in rheumatoid arthritis patients. Multiple transcript variants that encode the same protein have been found for this gene. [provided by RefSeq, Jul 2008]

GAD1 [+/-]

Cerebral palsy is a group of disorders that affect a person's ability to move and to maintain balance and posture. The disorders appear in the first few years of life. Usually they do not get worse over time. People with cerebral palsy may have difficulty walking. They may also have trouble with tasks such as writing or using scissors. Some have other medical conditions, including seizure disorders or mental impairment. Cerebral palsy happens when the areas of the brain that control movement and posture do not develop correctly or get damaged. Early signs of cerebral palsy usually appear before 3 years of age. Babies with cerebral palsy are often slow to roll over, sit, crawl, smile, or walk. Some babies are born with cerebral palsy; others get it after they are born. There is no cure for cerebral palsy, but treatment can improve the lives of those who have it. Treatment includes medicines, braces, and physical, occupational and speech therapy. NIH: National Institute of Neurological Disorders and Stroke

GIF [+/-]

This gene encodes a lymphokine involved in cell-mediated immunity, immunoregulation, and inflammation. It plays a role in the regulation of macrophage function in host defense through the suppression of anti-inflammatory effects of glucocorticoids. This lymphokine and the JAB1 protein form a complex in the cytosol near the peripheral plasma membrane, which may indicate an additional role in integrin signaling pathways. [provided by RefSeq, Jul 2008]

MAOB [+/-]

The protein encoded by this gene belongs to the flavin monoamine oxidase family. It is an enzyme located in the mitochondrial outer membrane. It catalyzes the oxidative deamination of biogenic and xenobiotic amines and plays an

important role in the metabolism of neuroactive and vasoactive amines in the central nervous system and peripheral tissues. This protein preferentially degrades benzylamine and phenylethylamine. [provided by RefSeq, Jul 2008]

MIR4761 (COMT H62H) [+/-]

Catechol-O-methyltransferase catalyzes the transfer of a methyl group from S-adenosylmethionine to catecholamines, including the neurotransmitters dopamine, epinephrine, and norepinephrine. This O-methylation results in one of the major degradative pathways of the catecholamine transmitters. In addition to its role in the metabolism of endogenous substances, COMT is important in the metabolism of catechol drugs used in the treatment of hypertension, asthma, and Parkinson disease. COMT is found in two forms in tissues, a soluble form (S-COMT) and a membrane-bound form (MB-COMT). The differences between S-COMT and MB-COMT reside within the N-termini. Several transcript variants are formed through the use of alternative translation initiation sites and promoters. [provided by RefSeq, Sep 2008]

MIR4761 (COMT V158M) [+/-]

Catechol-O-methyltransferase catalyzes the transfer of a methyl group from S-adenosylmethionine to catecholamines, including the neurotransmitters dopamine, epinephrine, and norepinephrine. This O-methylation results in one of the major degradative pathways of the catecholamine transmitters. In addition to its role in the metabolism of endogenous substances, COMT is important in the metabolism of catechol drugs used in the treatment of hypertension, asthma, and Parkinson disease. COMT is found in two forms in tissues, a soluble form (S-COMT) and a membrane-bound form (MB-COMT). The differences between S-COMT and MB-COMT reside within the N-termini. Several transcript variants are formed through the use of alternative translation initiation sites and promoters. [provided by RefSeq, Sep 2008]

MIR4761 (COMT) [+/-]

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MTHFD1 G1958A [+/-]

Neural tube defects are birth defects of the brain, spine, or spinal cord. They happen in the first month of pregnancy, often before a woman even knows that she is pregnant. The two most common neural tube defects are spina bifida and anencephaly. In spina bifida, the fetal spinal column doesn't close completely. There is usually nerve damage that causes at least some paralysis of the legs. In anencephaly, most of the brain and skull do not develop. Babies with anencephaly are either stillborn or die shortly after birth. Another type of defect, Chiari malformation, causes the brain tissue to extend into the spinal canal. The exact causes of neural tube defects aren't known. You're at greater risk of having an infant with a neural tube defect if you are obese, have poorly controlled diabetes, or take certain antiseizure medicines. Getting enough folic acid, a type of B vitamin, before and during pregnancy prevents most neural tube defects. Neural tube defects are usually diagnosed before the infant is born, through lab or imaging tests. There is no cure for neural tube defects. The nerve damage and loss of function that are present at birth are usually permanent. However, a variety of treatments can sometimes prevent further damage and help with complications. NIH: National Institute of Child Health and Human Development

MTHFD1L [+/-]

The protein encoded by this gene is involved in the synthesis of tetrahydrofolate (THF) in the mitochondrion. THF is important in the de novo synthesis of purines and thymidylate and in the regeneration of methionine from homocysteine. Several transcript variants encoding different isoforms have been found for this gene.[provided by RefSeq, Jun 2011]

MTHFR C677T [+/-]

At least 40 mutations in the MTHFR gene have been identified in people with homocystinuria. Most of these mutations change single amino acids in methylenetetrahydrofolate reductase. These changes impair the function of the enzyme, and some cause the enzyme to be turned off (inactivated). Other mutations lead to the production of an abnormally small, nonfunctional version of the enzyme. Without functional methylenetetrahydrofolate reductase, homocysteine cannot be converted to methionine. As a result, homocysteine builds up in the bloodstream, and the amount of methionine is reduced. Some of the excess homocysteine is excreted in urine. Researchers have not determined how altered levels of homocysteine and methionine lead to the health problems associated with homocystinuria.

MTHFS [+/-]

The protein encoded by this gene is an enzyme that catalyzes the conversion of 5-formyltetrahydrofolate to 5,10-methenyltetrahydrofolate, a precursor of reduced folates involved in 1-carbon metabolism. An increased activity of the encoded protein can result in an increased folate turnover rate and folate depletion. Three transcript variants encoding two different isoforms have been found for this gene. [provided by RefSeq, Jun 2011]

MTRR A66G [+/+]

At least 20 mutations in the MTRR gene have been identified in people with homocystinuria. Some of these mutations change single amino acids in methionine synthase reductase. Other mutations lead to an abnormally small, nonfunctional version of the enzyme. All these mutations prevent the enzyme from functioning normally. Without methionine synthase reductase, methionine synthase cannot convert homocysteine to methionine. As a result, homocysteine builds up in the bloodstream, and the amount of methionine is reduced. Some of the excess homocysteine is excreted in urine. Researchers have not determined how altered levels of homocysteine and methionine lead to the health problems associated with homocystinuria.

MTRR-11 A664A [+/-]

The protein encoded by this gene catalyzes the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a co-substrate for homocysteine remethylation to methionine. Genetic variation in this gene influences susceptibility to occlusive vascular disease, neural tube defects, colon cancer and acute leukemia, and mutations in this gene are associated with methylenetetrahydrofolate reductase deficiency.[provided by RefSeq, Oct 2009]

NOS1 [+/-]

This gene encodes a cell surface receptor and transmembrane precursor protein that is cleaved by secretases to form a number of peptides. Some of these peptides are secreted and can bind to the acetyltransferase complex APBB1/TIP60 to promote transcriptional activation, while others form the protein basis of the amyloid plaques found in the brains of patients with Alzheimer disease. Mutations in this gene have been implicated in autosomal dominant Alzheimer disease and cerebroarterial amyloidosis (cerebral amyloid angiopathy). Multiple transcript variants encoding several different isoforms have been found for this gene. [provided by RefSeq, Jul 2008]

NOS2 [+/-]

Blood pressure is the force of your blood pushing against the walls of your arteries. Each time your heart beats, it pumps blood into the arteries. Your blood pressure is highest when your heart beats, pumping the blood. This is called systolic pressure. When your heart is at rest, between beats, your blood pressure falls. This is called diastolic pressure. Your blood pressure reading uses these two numbers. Usually the systolic number comes before or above the diastolic number. A reading of 119/79 or lower is normal blood pressure 140/90 or higher is high blood pressure Between 120 and 139 for the top number, or between 80 and 89 for the bottom number is called prehypertension. Prehypertension means you may end up with high blood pressure, unless you take steps to prevent it. High blood pressure usually has no symptoms, but it can cause serious problems such as stroke, heart failure, heart attack and kidney failure. You can control high blood pressure through healthy lifestyle habits and taking medicines, if needed. NIH: National Heart, Lung, and Blood Institute

NOS3 [+/+]

Alzheimer's disease (AD) is the most common form of dementia among older people. Dementia is a brain disorder that seriously affects a person's ability to carry out daily activities. AD begins slowly. It first involves the parts of the brain that control thought, memory and language. People with AD may have trouble remembering things that happened recently or names of people they know. A related problem, mild cognitive impairment (MCI), causes more memory problems than normal for people of the same age. Many, but not all, people with MCI will develop AD. In AD, over time, symptoms get worse. People may not recognize family members or have trouble speaking, reading or writing. They may forget how to brush their teeth or comb their hair. Later on, they may become anxious or aggressive, or wander away from home. Eventually, they need total care. This can cause great stress for family members who must care for them. AD usually begins after age 60. The risk goes up as you get older. Your risk is also higher if a family member has had the disease. No treatment can stop the disease. However, some drugs may help keep symptoms from getting worse for a limited time. NIH: National Institute on Aging

PEMT [+/-]

This gene encodes a membrane-bound protein that is a member of the mucin family. Mucins are O-glycosylated proteins that play an essential role in forming protective mucous barriers on epithelial surfaces. These proteins also play a role in intracellular signaling. This protein is expressed on the apical surface of epithelial cells that line the mucosal surfaces of many different tissues including lung, breast stomach and pancreas. This protein is proteolytically cleaved into alpha and beta subunits that form a heterodimeric complex. The N-terminal alpha subunit functions in cell-adhesion and the C-terminal beta subunit is involved in cell signaling. Overexpression, aberrant intracellular localization, and changes in glycosylation of this protein have been associated with carcinomas. This gene is known to contain a highly polymorphic variable number tandem repeats (VNTR) domain. Alternate splicing results in multiple transcript variants.[provided by RefSeq, Feb 2011]

SLC19A1 [+/+]

The membrane protein encoded by this gene is a transporter of folate and is involved in the regulation of intracellular concentrations of folate. Three transcript variants encoding different isoforms have been found for this gene.[provided by RefSeq, Mar 2011]

SOD2 [+/+]

This gene encodes a multifunctional proinflammatory cytokine that belongs to the tumor necrosis factor (TNF) superfamily. This cytokine is mainly secreted by macrophages. It can bind to, and thus functions through its receptors TNFRSF1A/TNFR1 and TNFRSF1B/TNFR. This cytokine is involved in the regulation of a wide spectrum of biological processes including cell proliferation, differentiation, apoptosis, lipid metabolism, and coagulation. This cytokine has been implicated in a variety of diseases, including autoimmune diseases, insulin resistance, and cancer. Knockout studies in mice also suggested the neuroprotective function of this cytokine. [provided by RefSeq, Jul 2008]

SOD3 [+/+]

This gene encodes a member of the superoxide dismutase (SOD) protein family. SODs are antioxidant enzymes that catalyze the dismutation of two superoxide radicals into hydrogen peroxide and oxygen. The product of this gene is thought to protect the brain, lungs, and other tissues from oxidative stress. The protein is secreted into the extracellular space and forms a glycosylated homotetramer that is anchored to the extracellular matrix (ECM) and cell surfaces through an interaction with heparan sulfate proteoglycan and collagen. A fraction of the protein is cleaved near the C-terminus before secretion to generate circulating tetramers that do not interact with the ECM. [provided by RefSeq, Jul 2008]

TCN2 C766G [+/-]

The protein encoded by this gene catalyzes the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a co-substrate for homocysteine remethylation to methionine. Genetic variation in this gene influences susceptibility to occlusive vascular disease, neural tube defects, colon cancer and acute leukemia, and mutations in this gene are associated with methylenetetrahydrofolate reductase deficiency.[provided by RefSeq, Oct 2009]

VDR Bsm [+/-]

Mutations in the VDR gene cause vitamin D-dependent rickets type 2 (VDDR2), also known as hereditary vitamin D-resistant rickets (HVDRR). This disorder of bone development is characterized by low levels of calcium (hypocalcemia) and phosphate (hypophosphatemia) in the blood, which lead to soft, weak bones (rickets) that are prone to fracture. A common feature of this condition is bowed legs. The VDR gene mutations that cause this condition prevent the VDR protein from functioning properly. Some changes in the VDR gene lead to an abnormally short version of the VDR protein; others result in the production of an abnormal receptor that cannot bind to calcitriol, to RXR, or to DNA. Despite plenty of calcitriol in the body, the altered VDR cannot stimulate gene activity important for mineral absorption. The lack of calcium and phosphate absorption in the intestines slows deposition of these minerals into developing bone (bone mineralization), which leads to soft, weak bones and other features of VDDR2. Hypocalcemia also causes muscle weakness and seizures in some affected individuals. Most VDR gene mutations impair hair growth, leading to alopecia; however, mutations that block VDR's ability to interact with calcitriol do not cause alopecia, indicating that calcitriol is not necessary for the receptor's role in hair development.

ATP5C1 [+/-]

The protein encoded by this gene is a transmembrane glycoprotein that is a member of the protein kinase superfamily. This protein is a receptor for members of the epidermal growth factor family. EGFR is a cell surface protein that binds to epidermal growth factor. Binding of the protein to a ligand induces receptor dimerization and tyrosine autophosphorylation and leads to cell proliferation. Mutations in this gene are associated with lung cancer. Multiple alternatively spliced transcript variants that encode different protein isoforms have been found for this gene. [provided by RefSeq, Jul 2010]

CCL2 [+/+]

This gene is one of several cytokine genes clustered on the q-arm of chromosome 17. Chemokines are a superfamily of secreted proteins involved in immunoregulatory and inflammatory processes. The superfamily is divided into four subfamilies based on the arrangement of N-terminal cysteine residues of the mature peptide. This chemokine is a member of the CC subfamily which is characterized by two adjacent cysteine residues. This cytokine displays chemotactic activity for monocytes and basophils but not for neutrophils or eosinophils. It has been implicated in the pathogenesis of diseases characterized by monocytic infiltrates, like psoriasis, rheumatoid arthritis and atherosclerosis. It binds to chemokine receptors CCR2 and CCR4. [provided by RefSeq, Jul 2013]

NDUFS7 [+/-]

A genetic brain disorder is caused by a variation or a mutation in a gene. A variation is a different form of a gene. A mutation is a change in a gene. Genetic brain disorders affect the development and function of the brain. Some genetic brain disorders are due to random gene mutations or mutations caused by environmental exposure, such as cigarette smoke. Other disorders are inherited, which means that a mutated gene or group of genes is passed down through a family. They can also be due to a combination of both genetic changes and other outside factors. Some examples of genetic brain disorders include Leukodystrophies, Phenylketonuria, Tay-Sachs disease, Wilson disease. Many people with genetic brain disorders fail to produce enough of certain proteins that influence brain development and function. These brain disorders can cause serious problems that affect the nervous system. Some have treatments to control symptoms. Some are life-threatening.

UQCRC2 [+/-]

Metabolism is the process your body uses to get or make energy from the food you eat. Food is made up of proteins, carbohydrates, and fats. Chemicals in your digestive system break the food parts down into sugars and acids, your body's fuel. Your body can use this fuel right away, or it can store the energy in your body tissues, such as your liver, muscles, and body fat. A metabolic disorder occurs when abnormal chemical reactions in your body disrupt this process. When this happens, you might have too much of some substances or too little of other ones that you need to stay healthy. You can develop a metabolic disorder when some organs, such as your liver or pancreas, become diseased or do not function normally. Diabetes is an example.

4q27 Region [+/-]

The protein encoded by this gene is a secreted cytokine that is important for the proliferation of T and B lymphocytes. The receptor of this cytokine is a heterotrimeric protein complex whose gamma chain is also shared by interleukin 4 (IL4) and interleukin 7 (IL7). The expression of this gene in mature thymocytes is monoallelic, which represents an unusual regulatory mode for controlling the precise expression of a single gene. The targeted disruption of a similar gene in mice leads to ulcerative colitis-like disease, which suggests an essential role of this gene in the immune response to antigenic stimuli. [provided by RefSeq, Jul 2008]

ATG16L1 [+/-]

At least one variation in the ATG16L1 gene is associated with an increased risk of Crohn disease, particularly a form of the disorder that affects the lower part of the small intestine (the ileum). This increased risk has been found primarily in Caucasian (white) populations. The identified ATG16L1 variation changes a single protein building block (amino acid) in a critical region of the autophagy related 16-like 1 protein. Specifically, it replaces the amino acid threonine with the amino acid alanine at protein position 300 (written as Thr300Ala or T300A). The effects of variations in the ATG16L1 gene on Crohn disease risk are unclear. Changes in this gene may affect the autophagy process, allowing worn-out cell parts and harmful bacteria to persist when they would otherwise be destroyed. These cell components and bacteria may trigger an inappropriate immune system response, leading to chronic inflammation in the intestinal walls and the

digestive problems characteristic of Crohn disease. Researchers continue to study the relationship between changes in the ATG16L1 gene and a person's risk of developing this disorder.

IL4R [+/-]

The protein encoded by this gene is a pleiotropic cytokine produced by activated T cells. This cytokine is a ligand for interleukin 4 receptor. The interleukin 4 receptor also binds to IL13, which may contribute to many overlapping functions of this cytokine and IL13. STAT6, a signal transducer and activator of transcription, has been shown to play a central role in mediating the immune regulatory signal of this cytokine. This gene, IL3, IL5, IL13, and CSF2 form a cytokine gene cluster on chromosome 5q, with this gene particularly close to IL13. This gene, IL13 and IL5 are found to be regulated coordinately by several long-range regulatory elements in an over 120 kilobase range on the chromosome. Two alternatively spliced transcript variants of this gene encoding distinct isoforms have been reported. [provided by RefSeq, Jul 2008]

SULT2A1 [+/+]

This gene encodes a member of the sulfotransferase family. Sulfotransferases aid in the metabolism of drugs and endogenous compounds by converting these substances into more hydrophilic water-soluble sulfate conjugates that can be easily excreted. This protein catalyzes the sulfation of steroids and bile acids in the liver and adrenal glands, and may have a role in the inherited adrenal androgen excess in women with polycystic ovary syndrome. [provided by RefSeq, Mar 2010]

FOXE1 [+/-]

Your thyroid is a butterfly-shaped gland in your neck, just above your collarbone. It is one of your endocrine glands, which make hormones. Thyroid hormones control the rate of many activities in your body. These include how fast you burn calories and how fast your heart beats. All of these activities are your body's metabolism. If your thyroid gland is not active enough, it does not make enough thyroid hormone to meet your body's needs. This condition is hypothyroidism. Hypothyroidism is more common in women, people with other thyroid problems, and those over 60 years old. Hashimoto's disease, an autoimmune disorder, is the most common cause. Other causes include thyroid nodules, thyroiditis, congenital hypothyroidism, surgical removal of part or all of the thyroid, radiation treatment of the thyroid, and some medicines. The symptoms can vary from person to person. They may include Fatigue, Weight gain, A puffy face, Cold intolerance, Joint and muscle pain, Constipation, Dry skin, Dry, thinning hair, Decreased sweating, Heavy or irregular menstrual periods and fertility problems, Depression, Slowed heart rate. To diagnose hypothyroidism, your doctor will look at your symptoms and blood tests. Treatment is with synthetic thyroid hormone, taken every day. NIH: National Institute of Diabetes and Digestive and Kidney Diseases

IRF6 [+/+]

Mutations in the IRF6 gene that cause popliteal pterygium syndrome may change the transcription factor's effects on the activity of certain genes. This affects the development and maturation of tissues in the face, skin, and genitals, resulting in the facial and genital abnormalities, skin webbing, and fusion of the fingers or toes (syndactyly) seen in popliteal pterygium syndrome.

TBX22 [+/-]

Many disorders can affect our ability to speak and communicate. They range from saying sounds incorrectly to being completely unable to speak or understand speech. Causes include Hearing disorders and deafness, Voice problems, such as dysphonia or those caused by cleft lip or palate, Speech problems like stuttering, Developmental

disabilities Learning disorders Autism Brain injury Stroke Some speech and communication problems may be genetic. Often, no one knows the causes. By first grade, about 5 percent of children have noticeable speech disorders. Speech and language therapy can help. NIH: National Institute on Deafness and Other Communication Disorders

A gene variance report is a graphical representation of your genetic raw data, displayed as a color coded chart. Phenotypes are determined based on the presence of known variant alleles in your genotype. Alleles are considered variant if they are the minor allele i.e they occur with less frequency (MAF) in the default global population. In very few instances, the minor allele will not represent a mutation. In such cases, homozygous and/or heterozygous phenotypes may actually be normal. As such, what is normal or abnormal should not be determined solely based on phenotypes displayed in this report. All alleles are reported in reference to the forward strand. rsIDs and genotype information are obtained from the genetic raw data prepared by your personal genomic service. Minor allele frequency (MAF), RefSNP and gene variation/SNP names are obtained directly from dbSNP which is a free public archive for genetic variation maintained by the NCBI <http://www.ncbi.nlm.nih.gov/snp/>.

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