



## Maximized Wellness

Genetic Report for

**Testy McTesterson**

Generated by GeneSavvy Report Pipeline v1.0  
Jun 30, 2021

FULL NAME

**Testy  
McTesterson**

PHONE

**N/A**

EMAIL

**N/A**

DATE OF BIRTH

**11/11/2011**

DOCTOR

**N/A**

This report is for research, informational, and educational use only. It is not intended for the diagnosis, prevention, or treatment of disease. GeneSavvy does not provide medical advice.

# Contents

---

Using this report

Suggested first steps for clinical actions

**Overview of all your health network results**

**Potentially significant variants impacting gene function**

**Summary of researched SNPs and variants**

**Functional health networks** (in order of priority)

[Musculoskeletal Stability](#)

[Methylation](#)

[Iron](#)

[Oxidative Stress](#)

[Detoxification: Phase 2](#)

[Lipid Metabolism](#)

[Detoxification: Phase 1](#)

[Inflammation](#)

[SARS-CoV-2 Severity](#)

**Reference library of genes and variants** (sorted alphabetically)

Technical data

Thank you

Practitioner notes

# Using This Report

Note that the source databases from which our data is derived are being updated as more research is published. This report summarizes research data available when the report was generated.

## Objectives

This report is designed to help visualize genetic patterns of disruption associated to functional health pathways and biological processes. These polygenic patterns and predictions of pathway disruption can also have compounded effects due to environmental exposures and infections. Further exploration of gene networks with predicted disruption via additional diagnostic laboratory testing is subsequently recommended. Networks with predicted genetic disruptions may also benefit from generalized nutrition and/or lifestyle support to slow symptom progression and move towards more vibrant health!

## Gene impact vs health impact

This report presents data about the impact of genetic variants on gene function. Be aware that while gene impact predicts health, they are not the same thing. Measure of gene disruption considers the amount of change from the expected reference nucleotides, with respect to where inside the gene the change is found (i.e. stop/start/regulatory areas have greater impact), the type of mutation (i.e. frameshift may have a much greater impact than a missense mutation), and the amount of research available about the impact of that allele on protein function. It is important to note that in some cases, variants found may have a large impact on the function of the gene with no negative health impact, or even with positive health impacts.

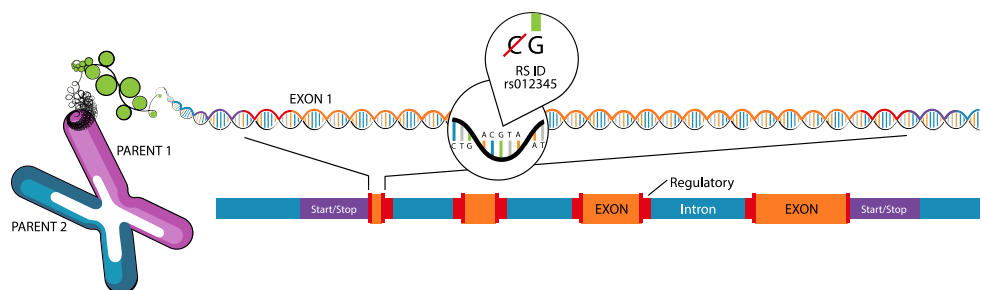
*Humans have 23 pairs of chromosomes, one set inherited from each parent. They vary in size and the largest (chr 1) contains over 2000 genes. A gene has the instructions to make a protein, encoded by a series of exons, introns, and regulatory sequences. Exons are like words; they directly produce amino acids. Introns space out those words to make them readable, and regulatory sequences act like punctuation.*

## Note on REF nucleotides

Some of the data in your report may include REF variants. During sequencing, we identify thousands of REF SNPs (locations where your DNA matches the human reference genome) for each of the health networks — but most of them are not included in this report as we are highlighting disrupted genetic function. It is important to note that being equal to the reference is not inherently good or bad — it is simply the most common nucleotide found in that location. Because of this, some REF nucleotides actually have pertinent health information and so those REF results will be included in this report.

## Didn't find the smoking gun? YOU OWN THE GOLDMINE for your diagnostic odyssey!

If your genetic testing was performed through GeneSavvy, congratulations! The data in this report is just a drop in the bucket of what can be discovered in your DNA. We have provided you with data about every protein-coding nucleotide (exons), many of the non-coding nucleotides (introns), and all your mitochondrial DNA in the companion Variant Exploration Report spreadsheet that came with this report. THE WHOLE GOLDMINE. With that spreadsheet, your DNA is an ocean of data ready for you to explore. If you need help, please reach out to us at [info@genesavvy.com](mailto:info@genesavvy.com).



## Suggested First Steps for Quick Clinical Actions

1. Review **HIGH** and **MEDIUM** priority health networks and utilize clinical tools to explore these networks as a differential diagnosis or potential underlying cause of current diagnosis that could be used for more personalized treatment protocols.

2. Review **SEVERE** and **HIGH** impact gene variants that are most likely to affect gene activity.

Prioritize variants that are **VERY RARE**, **RARE**, or **UNCOMMON** to identify potential genetic factors that are more unique to the individual.

**Variants with "VARIANT" link associations** have been referenced in at least one study of a genetic disorder, trait, or phenotype that is contained in the OMIM database. Click the link to learn about the related research.

**Remember to focus on the functional role of the GENE more than the specific variant.** Most variants increase or decrease gene expression and activity, so consider the upstream or downstream effects that might happen within related biological pathways if genes activity is altered due to these variants.

3. Review **STANDARD** priority health networks with highest relation to clinical presentation. Use these networks to further help identify genetic or environmental patterns that can be explored as a differential diagnosis or potential underlying cause of current diagnosis that could be used for more personalized treatment protocols.

4. **SNP results that contain a "\*" are PREDICTED results.** Genetic raw data files from non-GeneSavvy genetic testing companies do not contain the enhanced genetic data that is needed for our reports. When we detect this missing data, our GeneSavvy Bioinformatic Technology gathers information surrounding the missing data and makes a prediction of your most likely result. These predicted results will contain a " \* " and should be confirmed by additional testing as needed.

*\*For more information about this report and how to use it, please check out our [Report Information Page](#) or email us at [info@genesavvy.com](mailto:info@genesavvy.com) so we can help turn this genetic information into clinical action!*

# Overview of All Health Network Results

The pathway and gene network overview below gives a quick view of the systems reviewed in this report. Networks susceptible to disruption due to the genetic variants present should be addressed with priority to optimize health.

Functional Health Network	Priority
Musculoskeletal Stability	HIGH
Methylation	HIGH
Iron	MEDIUM
Oxidative Stress	STANDARD
Detoxification: Phase 2	LOW
Lipid Metabolism	LOW
Detoxification: Phase 1	LOW
Inflammation	LOW
SARS-CoV-2 Severity	LOW

# Potentially Significant Variants Impacting Gene Function

(ranked by predicted impact from high to low)

This significant variant overview highlights the variants found with the highest predicted potential to alter the transcription of the gene and/or the function of resulting enzyme. By identifying these high impact variants and potentially altered enzymes within the associated networks, targeted treatment options that account for these specific gene/enzyme alterations may be developed for more personalized and more effective health and lifestyle recommendations.

- SEVERE predicted impact on gene / VERY RARE variant / HOMOZYGOUS genotype
- HIGH predicted impact on gene / RARE variant
- MEDIUM predicted impact on gene / UNCOMMON variant / HETEROZYGOUS genotype
- LOW predicted impact on gene / COMMON variant / REFERENCE genotype

GENE SYMBOL	GENE NAME	VARIANT LOCATION	VARIANT	DEPTH	OMIM LINK	EFFECT	RESULT	IMPACT	FREQUENCY
TF	Transferrin	133756968	<a href="#">rs1799899</a>	175	<a href="#">VARIANT</a>	Missense	HET	Severe	Rare 4.5%
MTHFR	Methylenetetrahydrofolate Reductase	11796321	<a href="#">rs1801133</a>	169	<a href="#">VARIANT</a>	Missense	HET	Severe	Common 46.1%
VDR	Vitamin D Receptor	47879112	<a href="#">rs2228570</a>	209	<a href="#">GENE</a>	StartLoss	HOM	Severe	Common 66.4%
CILP	Cartilage Intermediate Layer Protein	65201874	<a href="#">rs2073711</a>	113	<a href="#">VARIANT</a>	Missense SpliceSite	HOM	Severe	Common 61.2%
NAT2	N-Acetyltransferase 2	18400593	<a href="#">rs1799930</a>	72	<a href="#">VARIANT</a>	Missense	HET	Severe	Uncommon 27.3%
COL2A1	Collagen Type II Alpha 1 Chain	47974193	<a href="#">rs2070739</a>	131	<a href="#">GENE</a>	Missense	HET	Severe	Uncommon 10.0%
MTHFR	Methylenetetrahydrofolate Reductase	11794419	<a href="#">rs1801131</a>	113	<a href="#">VARIANT</a>	Missense	HET	High	Uncommon 29.7%
BMP2	Bone Morphogenetic Protein 2	6778468	<a href="#">rs235768</a>	125	<a href="#">GENE</a>	Missense	HOM	High	Common 72.6%
NAT2	N-Acetyltransferase 2	18400860	<a href="#">rs1799931</a>	81	<a href="#">VARIANT</a>	Missense	HET	High	Rare 3.9%
GPX1	Glutathione Peroxidase 1	49357401	<a href="#">rs1050450</a>	90	<a href="#">VARIANT</a>	Missense	HET	High	Uncommon 30.2%
NOS2	Nitric Oxide Synthase 2	27760172	<a href="#">rs542585205</a>	105	<a href="#">GENE</a>	Missense	HET	High	Very Rare 0.0%
COL3A1	Collagen Type III Alpha 1 Chain	188999354	<a href="#">rs1800255</a>	159	<a href="#">VARIANT</a>	Missense	HET	High	Uncommon 30.0%
CETP	Cholesteryl Ester Transfer Protein	56982180	<a href="#">rs5882</a>	165	<a href="#">VARIANT</a>	Missense	HET	High	Common 58.8%
CYP1A1	Cytochrome P450 Family 1 Subfamily A Member 1	74720710	<a href="#">rs145198866</a>	117	<a href="#">GENE</a>	Missense	HET	High	Very Rare 0.4%
ICAM1	Intercellular Adhesion Molecule 1	10284532	<a href="#">rs1801714</a>	168	<a href="#">GENE</a>	Missense	HET	High	Rare 2.2%
NAT2	N-Acetyltransferase 2	18400806	<a href="#">rs1208</a>	74	<a href="#">VARIANT</a>	Missense	HOM	High	Common 59.7%
COL2A1	Collagen Type II Alpha 1 Chain	47974758	<a href="#">rs12721427</a>	175	<a href="#">GENE</a>	Missense	HET	High	Rare 5.3%
CYP2A6	Cytochrome P450 Family 2 Subfamily A Member 6	40843854	<a href="#">rs6413474</a>	479	<a href="#">GENE</a>	Missense	HET	High	Very Rare 1.1%

GENE SYMBOL	GENE NAME	VARIANT LOCATION	VARIANT	DEPTH	OMIM LINK	EFFECT	RESULT	IMPACT	FREQUENCY
COL10A1	Collagen Type X Alpha 1 Chain	116120483	rs2228547	99	GENE	Missense	HET	High	Uncommon 16.5%
TF	Transferrin	133775510	rs1049296	218	VARIANT	Missense	HET	High	Uncommon 13.5%
SOD2	Superoxide Dismutase 2	159692840	rs4880	113	VARIANT	Missense	HET	High	Common 47.3%
MTHFR	Methylenetetrahydrofolate Reductase	11790870	rs2274976	211	GENE	Missense	HET	High	Very Rare 1.8%
CYP2D7	.	42127941	rs16947	249	VARIANT	Missense	HET	Medium	Uncommon 39.0%
COL10A1	Collagen Type X Alpha 1 Chain	116125413	rs1064583	160	GENE	Missense	HET	Medium	Common 47.4%
COL3A1	Collagen Type Iii Alpha 1 Chain	189010695	rs1516446	171	GENE	Missense	HOM	Medium	Common 99.7%
COL4A1	Collagen Type Iv Alpha 1 Chain	110166251	rs3742207	95	GENE	Missense	HET	Medium	Uncommon 31.1%
COL2A1	Collagen Type Ii Alpha 1 Chain	48004297	rs3803183	181	GENE	Missense	HET	Medium	Common 75.8%
APOE	Apolipoprotein E	44905910	rs440446	142	GENE	Missense	HET	Medium	Common 69.7%
CILP	Cartilage Intermediate Layer Protein	65196790	rs938952	206	GENE	Missense	HOM	Medium	Common 75.0%
CILP	Cartilage Intermediate Layer Protein	65198563	rs2679118	84	GENE	Missense	HOM	Medium	Common 98.8%
CYP2C19	Cytochrome P450 Family 2 Subfamily C Member 19	94842866	rs3758581	229	GENE	Missense	HOM	Medium	Common 95.4%
TF	Transferrin	133766289	rs2692696	88	GENE	Missense	HOM	Medium	Common 99.6%
TFRC	Transferrin Receptor	196073940	rs3817672	144	GENE	Missense	HOM	Medium	Common 42.6%
GSTP1	Glutathione S-Transferase Pi 1	67585218	rs1695	204	GENE	Missense	HET	Medium	Uncommon 36.4%
CILP	Cartilage Intermediate Layer Protein	65197350	rs2679117	126	GENE	Missense	HOM	Medium	Common 98.8%
SLC11A2	Solute Carrier Family 11 Member 2	51009122	rs445520	96	GENE	Missense	HET	Medium	Common 90.8%
UGT1A8	Udp Glucuronosyltransferase Family 1 Member A8	233618225	rs1042597	193	GENE	Missense	HOM	Medium	Uncommon 25.4%
ICAM1	Intercellular Adhesion Molecule 1	10285007	rs5498	135	GENE	Missense	HET	Medium	Uncommon 36.7%
GSTM1	Glutathione S-Transferase Mu 1	109690516	rs1065411	6	GENE	Missense	HOM	Medium	Uncommon 38.8%
SOD3	Superoxide Dismutase 3	24799693	rs2536512	52	GENE	Missense	HET	Medium	Common 50.0%
APOE	Apolipoprotein E	Multiallelic	APOE_GenoType	N/A	.	GenoType	E3	GenoType	Common GenoType



# Summary of Researched SNPs & Variants

(grouped by Health Network)

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

## Musculoskeletal Stability: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Type 2 Collagen	Monitor & Support	COL2A1	rs2070739	HET	COL2A1 enzyme activity, type 2 collagen function and exercise induced tissue damage recovery.	Possible slightly reduced COL2A1 function and possible slightly reduced recovery time after exercise induced tissue damage.
Bone Health	Monitor & Support	VDR	rs2228570	HOM	VDR enzyme activity, Vitamin D metabolism, and bone health.	Possible reduced vitamin D metabolism function and increased risk for bone health issues due to chronic low vitamin D levels.
Type 1 Collagen	Monitor & Support	COL1A1	rs2734272	HOM	COL1A1 enzyme activity and type 1 collagen function.	Possible reduced COL1A1 function and possible reduced type 1 collagen stability.



# Methylation: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Vitamin B12	Supports Homeostasis	MTRR	rs1801394	REF	MTRR enzyme activity and B12 (methylcobalamin) levels.	Typical MTRR enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Vitamin B12	Supports Homeostasis	MTRR	rs1532268	REF	MTRR enzyme activity and B12 (methylcobalamin) levels.	Typical MTRR enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Homocysteine	Supports Homeostasis	MTR	rs1805087	REF	MTR enzyme activity and homocysteine levels.	Typical MTR enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Neurotransmitters	Optimize & Enhance	COMT	rs4680	REF	COMT enzyme activity and neurotransmitter metabolism. Altered dopamine, norepinephrine, pain tolerance, and stress resiliency.	Possible increased COMT enzyme activity. Possibly lower levels of dopamine and norepinephrine with higher pain tolerance.
Neurotransmitters	Optimize & Enhance	COMT	rs4633	REF	COMT enzyme activity and neurotransmitter metabolism. Altered dopamine, norepinephrine, and pain tolerance.	Possible increased COMT enzyme activity. Possibly lower levels of dopamine and norepinephrine with higher pain tolerance.
Neurotransmitters	Supports Homeostasis	COMT	rs6267	REF	COMT enzyme activity and neurotransmitter metabolism. Altered dopamine, norepinephrine, and pain tolerance.	Typical COMT enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Folate	Monitor & Support	MTHFR	rs1801133	HET	MTHFR enzyme activity and folate metabolism within the methylation cycle.	MTHFR enzyme activity possibly reduced by about 40%
Folate	Monitor & Support	MTHFR	rs1801131	HET	MTHFR enzyme activity and folate metabolism within the methylation cycle.	MTHFR enzyme activity possibly slightly reduced.
Neurotransmitters	Supports Homeostasis	COMT	rs6269	HET	COMT enzyme activity and neurotransmitter metabolism. Altered dopamine, norepinephrine, and pain tolerance.	Typical COMT enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.



## Iron: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Iron	Supports Homeostasis	HFE	rs1800562	REF	HFE enzyme activity and hereditary hemochromatosis (iron overload)	Typical HFE enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Iron	Supports Homeostasis	HFE	rs1799945	REF	HFE enzyme activity and hereditary hemochromatosis (iron overload)	Typical HFE enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Iron	Supports Homeostasis	HFE	rs1800730	REF	HFE enzyme activity and hereditary hemochromatosis (iron overload)	Typical HFE enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.

## Oxidative Stress: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Nitric Oxide	Supports Homeostasis	NOS3	rs1799983	REF	NOS3 enzyme activity and related eNOS activity.	Typical NOS3 enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Antioxidant	Monitor & Support	GPX1	rs1050450	HET	GPX1 enzyme activity and defense against oxidative stress and DNA damage.	Possibly some reduced GPX1 enzyme activity and some reduced ability to manage oxidative stress and DNA damage. Some research suggests GPX activity may be increased in response to increased selenium intake for this result.
Antioxidant	Supports Homeostasis	SOD2	rs4880	HET	MnSOD or SOD2 enzyme activity and management of oxidative stress	Possible altered SOD2 enzyme activity with potentially increased ability to manage oxidative stress.
Nitric Oxide	Monitor & Support	NOS1	rs2682826	HET	NOS1 enzyme expression and related nNOS activity.	Possible reduced NOS1 enzyme expression with potentially decreased ability to manage oxidative stress but also possible reduced risk for neurotoxic nitric oxide levels.
Nitric Oxide	Monitor & Support	NOS2	rs1060826	HOM	NOS2 enzyme activity and related iNOS activity.	Possible reduced NOS2 enzyme expression with potentially decreased ability to manage oxidative stress.

## Detoxification: Phase 2: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Carcinogens	Supports Homeostasis	UGT1A6	rs17863783	REF	UGT1A6 enzyme activity, drug toxicity, and carcinogen removal.	Typical UGT1A6 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Bilirubin	Supports Homeostasis	UGT1A1	rs35003977	REF	UGT1A1 enzyme-substrate binding activity and bilirubin metabolism.	Typical UGT1A1 to bilirubin binding activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Bilirubin	Supports Homeostasis	UGT1A1	rs4148323	REF	UGT1A1 enzyme transcription and expression activity, and bilirubin metabolism.	Typical UGT1A1 transcription and expression activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Xenobiotics	Supports Homeostasis	NAT1	rs4987076	REF	NAT1 enzyme transcription and expression activity, and xenobiotic detoxification.	Typical NAT1 transcription and expression activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Acetylation	Monitor & Support	NAT2	rs1208	HOM	NAT2 enzyme activity, and acetylation rate.	Possibly reduced NAT2 enzyme activity. Possible slow acetylator phenotype.

# Lipid Metabolism: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
APOE	Supports Homeostasis	APOE	APOE_GenoType	E3	early onset Alzheimer's disease symptoms	This is the most common APOE Type, with no association to either increase or reduce Alzheimer's or CAD risk.
APOE	Monitor & Support	APOE	rs7412	REF	APOE genotype classification. Also associated with cognitive stability and cardio health.	Typical APOE enzyme function. Your GenoType result is associated with normal risk for early cognitive decline. Focus on maintaining cognitive function by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
APOE	Supports Homeostasis	APOE	rs429358	REF	APOE genotype classification. Also associated with cognitive stability and cardio health.	Typical APOE enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Cognition	Supports Homeostasis	APOE	rs121918393	REF	resistance to cognitive decline and APOE subtype result modification.	This Genotype has less effect on APOE subtypes or cognitive decline. Other genotypes at this SNP might have more resistance to early cognitive decline. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Heart	Supports Homeostasis	PON1	rs662	REF	altered PON1 activity, heart health, and risk for coronary artery disease.	Typical PON1 enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Lipids	Supports Homeostasis	LPL	rs328	REF	altered LPL activity, and altered triglycerides, cholesterol, HDL, and LDL levels. Possible response to dietary intake.	Typical LPL enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Lipids	Supports Homeostasis	LPL	rs1801177	REF	altered LPL activity, risk of combined hyperlipidemia, altered triglycerides, cholesterol, HDL, and apolipoproteins (apoB) levels..	Typical LPL enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Lipids	Optimize & Enhance	CETP	rs5882	HET	altered serum CETP levels, HDL, LDL, and aging.	Possible some reduced serum CETP levels. Possibly some increased HDL levels, increased HDL & LDL particle size, better cognitive function and healthy aging.



# Detoxification: Phase 1: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Estrogen	Supports Homeostasis	CYP1A1	rs1048943	REF	CYP1A1 enzyme activity and estrogen metabolism	Typical CYP1A1 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Nicotine	Monitor & Support	CYP2A6	rs5031017	REF	CYP2A6 enzyme activity and nicotine addiction risk.	Typical CYP2A6 enzyme activity. Higher risk for nicotine addiction
Nicotine	Monitor & Support	CYP2A6	rs1801272	REF	CYP2A6 enzyme activity and nicotine addiction risk.	Typical CYP2A6 enzyme activity. Higher risk for nicotine addiction
Warfarin	Supports Homeostasis	CYP2C9	rs1799853	REF	CYP2C9 enzyme activity and Warfarin metabolism.	Typical CYP2C9 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Warfarin	Supports Homeostasis	CYP2C9	rs1057910	REF	CYP2C9 enzyme activity and Warfarin metabolism.	Typical CYP2C9 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Pharma	Supports Homeostasis	CYP2C9	rs2256871	REF	CYP2C9 enzyme activity and drug metabolism activity.	Typical CYP2C9 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Pharma	Supports Homeostasis	CYP2D6	rs28371706	REF	CYP2D6 enzyme activity and drug metabolism activity.	Typical CYP2D6 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Coffee	Optimize & Enhance	CYP1A2	rs2472304	HET	CYP1A2 enzyme activity and coffee consumption.	Slightly increased CYP1A2 enzyme activity. Possible slight increase in coffee consumption.
Pharma	Monitor & Support	CYP2D6	rs16947	HET	CYP2D6 enzyme activity and drug metabolism activity.	Reduced CYP2D6 enzyme activity. Possible slight reduction in drug metabolism activity.



# Inflammation: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Inflammation	Supports Homeostasis	IL1A	rs17561	REF	IL1A enzyme activity and inflammatory conditions.	Typical IL1A expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	IL1B	rs1143634	REF	IL1B enzyme activity and inflammatory conditions.	Typical IL1B expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	IL1RN	rs878972	REF	IL1RN enzyme activity and inflammatory conditions.	Typical IL1RN expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	TNF	rs1800610	REF	TNF enzyme activity and inflammatory conditions.	Typical TNF expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	TNF	rs3093662	REF	TNF enzyme activity and inflammatory conditions.	Typical TNF expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	IL6	rs1524107	REF	IL6 enzyme activity and inflammatory conditions.	Typical IL6 expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	IL6	rs1800795	REF	IL6 enzyme activity and inflammatory conditions.	Typical IL6 expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.

# SARS-CoV-2 Severity: Researched SNPs & Variants

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Blood Type	Supports Homeostasis	ABO	rs8176719	REF	ABO blood types and increased risk for SARS-CoV-2 severity.	Most likely "O" blood type. Possibly lower COVID-19 related risk.
SARS-CoV-2	Monitor & Support	CXCR6	rs2234355	REF	chemokine interactions between pathogens and CD4 cells and associated increased risk for SARS-CoV-2 severity.	Typical CXCR6 enzyme activity which has been associated with increased depletion of CD4 counts under viral load and increased viral load severity.
SARS-CoV-2	Supports Homeostasis	F3	rs3917643	REF	blood coagulation rate and associated risks for SARS-CoV-2 severity.	Typical F3 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
SARS-CoV-2	Supports Homeostasis	C4BPA	rs45574833	REF	C4BPA binding activity and associated increased risk for SARS-CoV-2 severity.	Typical C4BPA enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
ARDS	Monitor & Support	IL17A	rs2275913	REF	IL-17 expression, activity risk for Acute Respiratory Distress Syndrome (ARDS).	Typical IL-17 expression and activity. This result may carry more risk for Acute Respiratory Distress than HET or HOM results.
ARDS	Monitor & Support	TNF	rs1800610	REF	TNF expression, activity risk for Acute Respiratory Distress Syndrome (ARDS).	Typical TNF expression and activity. This result may carry more risk for Acute Respiratory Distress than HET or HOM results but also possible increased pathogen clearance activity and protection from persistent infection.



## HIGH PRIORITY NETWORK

*Based on your genetic data, this network has a high probability of disruption. It should be prioritized for further exploration with your clinical team. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

# Musculoskeletal Stability

The musculoskeletal system consists of ligaments, bones, muscles, tendons, and cartilage, thus forming the body's framework. It provides the human body with stability, shape, movement, and support. Musculoskeletal pain refers to the pain in the bones, muscles, nerves, tendons, and ligaments. This pain could be in just one area of your body, such as your back, or you can feel it throughout your body in case you have a widespread condition like fibromyalgia. Some of the commonly linked genes to musculoskeletal stability are COL genes, BMP2, VDR, and CILP. An injury commonly causes musculoskeletal pain to the muscles, joints, bones, ligaments, or tendons. Car accidents, falls, and sports injuries are some examples of accidents that can lead to pain.

## Action Items for Maximized Wellness (Musculoskeletal Stability)



### ACTIVELY SEEK

- Focus on proper warm-up and stretching prior to exercise or physical activity
- Include Calcium-rich foods in your daily diet
- Monitor and support healthy Vitamin D levels
- Utilize proper form during exercise and physical activities



### ACTIVELY AVOID

- Reduce physical activities involving high impact and stress on joints
- Avoid bad posture habits
- Limit caffeine, alcohol, and salt consumption.

## Clinical Exploration & Diagnostic Tools (Musculoskeletal Stability)

**Bone Density Scan:** Bone density scans measures bone mineral density to assess if a person is at risk of osteoporosis or fracture

**Vitamin D3 (Cholecalciferol):** This test can be used to monitor Vitamin D levels and identify possible Vitamin D deficiencies that can contribute to low bone mineral densities.

**Beighton Score Assessment:** The Beighton Scoring System is used to assess joint and tissue hypermobility.

## Nutritional Support & Supplements (Musculoskeletal Stability)

**Xymogen SynovX® Formulas:** Xymogen's SynovX® formulas have been specifically designed to support joint and tissue repair, health, and longevity.

**Xymogen K2-D3 5000:** A highly-bioavailable Vitamin D formula, enhanced with Vitamin K2.

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## Musculoskeletal Stability: Genes Analyzed

The genes presented below are associated with **Musculoskeletal Stability**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

- HIGH priority for clinical exploration / MOST LIKELY altered gene function
- MEDIUM priority for clinical exploration / LIKELY altered gene function
- STANDARD priority for clinical exploration / POSSIBLE altered gene function
- LOW priority for clinical exploration / LESS LIKELY altered gene function

### COL2A1

**Collagen Type 2 Alpha 1 Chain:** Type 2 Collagen is most specific to joint cartilage, skeletal development, linear growth. Essential for tissue stability and the ability to withstand compressive forces.

### CILP

**Cartilage Intermediate Layer Protein:** Plays a role in cartilage scaffolding and the formation and function of the cartilage extracellular matrix. May antagonize and interact with TGF-Beta and IGF-1. Associated with Joint Disease and Osteoarthritis.

### VDR

**Vitamin D Receptor:** Vitamin D3 Receptor that activates and mediates Vitamin D3 responsive cellular activities. Essential for bone/calcium homeostasis and mineral metabolism.

### COL3A1

**Collagen Type 3 Alpha 1 Chain:** Type 3 Collagen is mostly found in extensible connective tissues such as skin, lungs, uterus, intestines, and the vascular system. Essential for tissue strength and elasticity, similar to Type 1 Collagen.

### BMP2

**Bone Morphogenetic Protein 2:** Ligand for TGF-Beta that binds TGF-Beta receptors to induce cartilage and bone formation. Also plays roles in regulating myogenesis.

### COL10A1

**Collagen Type 10 Alpha 1 Chain:** Type 10 Collagen is essential for new bone and articulate joint space and cartilage formation. Essential for healthy joint function and resistance to normal joint wear and tear.

### COL4A1

**Collagen Type 4 Alpha 1 Chain:** Type 4 Collagen is essential for the creation of sheet-type tissues and structures in our body like basement membranes. Important for wound healing, digestion, respiration, and filtration.

### COL1A1

**Collagen Type I Alpha 1 Chain:** Type 1 Collagen is the most abundant collagen in the body. Helps make tendons, ligaments, bone, skin, hair, and teeth. Essential for tissue strength and elasticity.

### COL5A1

**Collagen Type 5 Alpha 1 Chain:** Type 5 Collagen is mostly found in skin, hair, eyes, and placental tissues. Type 5 collagen also plays a role in the formation of type 1 and 3 collagens.

## Musculoskeletal Stability: Researched SNPS & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Type 2 Collagen	Monitor & Support	COL2A1	rs2070739	HET	COL2A1 enzyme activity, type 2 collagen function and exercise induced tissue damage recovery.	Possible slightly reduced COL2A1 function and possible slightly reduced recovery time after exercise induced tissue damage.
Bone Health	Monitor & Support	VDR	rs2228570	HOM	VDR enzyme activity, Vitamin D metabolism, and bone health.	Possible reduced vitamin D metabolism function and increased risk for bone health issues due to chronic low vitamin D levels.
Type 1 Collagen	Monitor & Support	COL1A1	rs2734272	HOM	COL1A1 enzyme activity and type 1 collagen function.	Possible reduced COL1A1 function and possible reduced type 1 collagen stability.

## HIGH PRIORITY NETWORK

*Based on your genetic data, this network has a high probability of disruption. It should be prioritized for further exploration with your clinical team. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

# Methylation

B vitamins provide building blocks for growing cells, which are constantly being renewed, and play an important role in many physiological processes. B vitamins also supply some of the chemicals necessary for protecting our genes, so that DNA doesn't accumulate damage from the wear and tear in the daily lives of our cells. These vitamins – including folate, vitamins B6 and B12 – help make new DNA for cells that are constantly growing and renewing themselves. B vitamins are also involved in turning many genes on and off, and also help repair DNA. The process of DNA repair is called methylation. Methylation uses the process of donating 'methyl groups' to a substrate. A methyl group consists of one carbon bound to three hydrogen atoms (CH<sub>3</sub>). Although B vitamins are only required in small amounts, they are crucial for methylation and in producing new DNA.

## Action Items for Maximized Wellness (Methylation)



### ACTIVELY SEEK

- Increase intake of folate rich foods like leafy greens, lentils, asparagus, liver, and broccoli.
- Work with your functional medicine practitioner to monitor homocysteine levels along with B12 and Zinc.
- Increase intake of choline rich foods like egg yolks, beef liver, and wheat germ.
- Increase intake of betaine rich foods like beets, quinoa, and spinach.



### ACTIVELY AVOID

- Avoid synthetic forms of folate such as "folic acid"
- Reduce exposure to environmental toxins such as cigarette smoke, chemicals, air pollution, pesticides, and toxic plastics

## Clinical Exploration & Diagnostic Tools (Methylation)

**Doctor's Data Methylation Profile:** This test can help identify methionine metabolism disruptions related to methylation.

**ZRT Labs NeuroAdvanced Profile:** Testing to help detect possible neurotransmitter dysregulations commonly associated to methylation gene mutations.

**Genova NutrEval- PLASMA:** Nutritional analysis to help identify areas needing addition nutrient support related to methylation function.

## Nutritional Support & Supplements (Methylation)

---

**Xymogen Methyl Protect™:** Comprehensive formula to support optimal methylation and healthy homocysteine levels. \*\*Helps stabilize methylation functions in toxic environments.

**Xymogen SAM-e & TMG:** Provides highly bioavailable methyl donors. \*\*Methylation activity without sufficient methyl donors is like having a new car with no gas.

**Xymogen 5-MTHF + B12:** Staple nutrients for MTHFR and methylation support. \*\*Higher indication with reduced MTHFR gene activity

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## Methylation: Genes Analyzed

The genes presented below are associated with **Methylation**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

- HIGH priority for clinical exploration / MOST LIKELY altered gene function
- MEDIUM priority for clinical exploration / LIKELY altered gene function
- STANDARD priority for clinical exploration / POSSIBLE altered gene function
- LOW priority for clinical exploration / LESS LIKELY altered gene function

### MTHFR

**Methylenetetrahydrofolate Reductase:** Responsible for the conversion of folic acid to methylfolate which is a cofactor needed for serotonin, norepinephrine, and dopamine synthesis.

### COMT

**Catechol-O-Methyltransferase:** Introduces a methyl group to the catecholamines. Responsible for breakdown of dopamine in the frontal cortex of the brain.

### CBS

**Cystathionine Beta-Synthase:** Catalyze the conversion of homocysteine to cystathionine, the first step in the transsulfuration pathway.

### MTRR

**5-Methyltetrahydrofolate-Homocysteine Methyltransferase Reductase:** Catalyses methylcobalamin and maintains homocysteine concentrations at non-toxic levels.

### MTR

**5-Methyltetrahydrofolate-Homocysteine Methyltransferase:** Catalyses the re-methylation of homocysteine to methionine.

## Methylation: Researched SNPS & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Vitamin B12	Supports Homeostasis	MTRR	rs1801394	REF	MTRR enzyme activity and B12 (methylcobalamin) levels.	Typical MTRR enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Vitamin B12	Supports Homeostasis	MTRR	rs1532268	REF	MTRR enzyme activity and B12 (methylcobalamin) levels.	Typical MTRR enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Homocysteine	Supports Homeostasis	MTR	rs1805087	REF	MTR enzyme activity and homocysteine levels.	Typical MTR enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Neurotransmitters	Optimize & Enhance	COMT	rs4680	REF	COMT enzyme activity and neurotransmitter metabolism. Altered dopamine, norepinephrine, pain tolerance, and stress resiliency.	Possible increased COMT enzyme activity. Possibly lower levels of dopamine and norepinephrine with higher pain tolerance.
Neurotransmitters	Optimize & Enhance	COMT	rs4633	REF	COMT enzyme activity and neurotransmitter metabolism. Altered dopamine, norepinephrine, and pain tolerance.	Possible increased COMT enzyme activity. Possibly lower levels of dopamine and norepinephrine with higher pain tolerance.
Neurotransmitters	Supports Homeostasis	COMT	rs6267	REF	COMT enzyme activity and neurotransmitter metabolism. Altered dopamine, norepinephrine, and pain tolerance.	Typical COMT enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Folate	Monitor & Support	MTHFR	rs1801133	HET	MTHFR enzyme activity and folate metabolism within the methylation cycle.	MTHFR enzyme activity possibly reduced by about 40%
Folate	Monitor & Support	MTHFR	rs1801131	HET	MTHFR enzyme activity and folate metabolism within the methylation cycle.	MTHFR enzyme activity possibly slightly reduced.



HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Neurotransmitters	Supports Homeostasis	COMT	rs6269	HET	COMT enzyme activity and neurotransmitter metabolism. Altered dopamine, norepinephrine, and pain tolerance.	Typical COMT enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.



MEDIUM  
PRIORITY  
NETWORK

*Based on your genetic data, this network has a medium probability of disruption. We recommend exploring this network further with your clinical team, but is not as urgent as any high priority networks you may have. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

## Iron

Iron is a chemical element found in the body. Essentially, iron is needed by the body to make both myoglobin and hemoglobin, which are responsible for the transport of oxygen throughout the whole body. Some of the processes that are most commonly associated with iron include oxygen transport, electron transport, and the synthesis of DNA. TF (Transferrin), SLC (Solute Carrier Family), HFE (Hemochromatosis), and TFR (Transferrin Receptor) are some genes linked with iron.

### Action Items for Maximized Wellness (Iron)



#### ACTIVELY SEEK

- Donate blood regularly.
- Routine iron level testing and liver health checkups with your functional medicine practitioner.



#### ACTIVELY AVOID

- Reduce alcohol consumption
- Avoid iron fortified foods if excess iron levels indicated.

### Clinical Exploration & Diagnostic Tools (Iron)

**Iron, TIBC and Ferritin Panel:** Quest Lab ID# 5616 - This lab test measures Total Iron, Iron Binding Capacity, % Saturation (calculated), and Ferritin which could be useful for monitoring iron uptake, transport, and utilization activity.

### Nutritional Support & Supplements (Iron)

**Xymogen Iron Glycinate:** High bioavailability, low toxicity, Iron supplement.

**Xymogen Chelex™:** Helps clear oxidative elements and support the body during toxic burden from environmental toxins like heavy metals.

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## Iron: Genes Analyzed

The genes presented below are associated with **Iron**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

- HIGH priority for clinical exploration / MOST LIKELY altered gene function
- MEDIUM priority for clinical exploration / LIKELY altered gene function
- STANDARD priority for clinical exploration / POSSIBLE altered gene function
- LOW priority for clinical exploration / LESS LIKELY altered gene function

<b>TF</b>	<b>Transferrin:</b> Responsible for the transport of iron from sites of absorption to those of storage and utilization
<b>TFRC</b>	<b>Transferrin Receptor:</b> Transferrin receptor involved in the cellular uptake of iron.
<b>SLC11A2</b>	<b>Solute Carrier Family 11 Member 2:</b> Transports Iron and other metals throughout the body, as well as into the mitochondria.
<b>HFE</b>	<b>Homeostatic Iron Regulator:</b> Regulates iron absorption by altering the interaction of the transferrin receptor with transferrin
<b>SLC40A1</b>	<b>Solute Carrier Family 40 Member 1:</b> Transports Iron and other metals throughout the body, as well as into the mitochondria. Also transfers iron between maternal and fetal circulation.
<b>HAMP</b>	<b>Hepcidin Antimicrobial Peptide:</b> Essential for iron homeostasis and is a major regulator of dietary iron absorption, iron distribution across tissues, and iron recycling.
<b>HJV</b>	<b>Hemojuvelin:</b> Activates, modulates expression, and acts as a cellular receptor for Hepcidin. Essential for iron homeostasis via Hepcidin activity regulation.

## Iron: Researched SNPs & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Iron	Supports Homeostasis	HFE	rs1800562	REF	HFE enzyme activity and hereditary hemochromatosis (iron overload)	Typical HFE enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Iron	Supports Homeostasis	HFE	rs1799945	REF	HFE enzyme activity and hereditary hemochromatosis (iron overload)	Typical HFE enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Iron	Supports Homeostasis	HFE	rs1800730	REF	HFE enzyme activity and hereditary hemochromatosis (iron overload)	Typical HFE enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.

STANDARD  
PRIORITY  
NETWORK

*Based on your genetic data, this network has a mild probability of disruption. You may wish to explore this network further with your clinical team to optimize your health. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

## Oxidative Stress

Oxidative stress results from an imbalance between the production and collection of reactive oxygen species (ROS) in tissues and cells and the capability of a biological system to detoxify these reactive products. Oxidative stress is a natural process and plays a vital role in the aging process. Many factors contribute to oxidative stress. These include diet, smoking and alcohol consumption, environmental factors such as radiation and pollution, and exposure to pesticides and industrial chemicals. Oxidative stress is a major contributor to developing a range of chronic conditions such as diabetes, cancer, and heart disease. SOD1, CAT, NOS3, and GPX1 are some genes that are closely related to oxidative stress. Making changes to your diet and lifestyle can help reduce oxidative stress. These include exercising regularly, maintaining a healthy body weight, and eating a balanced diet rich in vegetables and fruits.

### Action Items for Maximized Wellness (Oxidative Stress)



#### ACTIVELY SEEK

- Focus on staying hydrated.
- Increase consumption of vegetables high in nitrates
- Consume lean protein to help maintain optimum glutathione levels.
- Increase physical activity and exercise.



#### ACTIVELY AVOID

- Reduce exposure to environmental toxins such as cigarette smoke, chemicals, air pollution, pesticides, and toxic plastics.
- Reduce mental and emotional stressors.
- Reduce overall toxic burden.

### Clinical Exploration & Diagnostic Tools (Oxidative Stress)

**Organix® Comprehensive Profile:** The Genova Diagnostics Organix® Comprehensive Profile is a nutritional test providing insights into organic acids and a view into the body's cellular metabolic processes.

**GDX Oxidative Stress 2.0 (Blood):** The Oxidative Stress Analysis 2.0 nutritional test utilizes a blood sample in order to evaluate the body's oxidative stress status and antioxidant reserve.

**DUTCH Complete Urine Panel:** Comprehensive assessment of hormones and their metabolites. Also includes daily&free cortisol pattern, organic acids, melatonin (6-OHMS), and oxidative stress (8-OHdG).

### Nutritional Support & Supplements (Oxidative Stress)

**Xymogen Nrf2 Activator™:** Enhances enzyme activity and expression within detoxification pathways while also downregulating damaging inflammatory cytokine release.

**Xymogen S-Acetyl Glutathione™:** Enhances enzyme activity and expression within detoxification pathways while also downregulating damaging inflammatory cytokine release.

**Xymogen AngiNOX™:** Based on the latest Nobel Prize-winning research on nitric oxide, this formula offers therapeutic levels of L-arginine, L-citrulline, and other nutrients to promote optimal nitric oxide activity.

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## Oxidative Stress: Genes Analyzed

The genes presented below are associated with **Oxidative Stress**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

- HIGH priority for clinical exploration / MOST LIKELY altered gene function
- MEDIUM priority for clinical exploration / LIKELY altered gene function
- STANDARD priority for clinical exploration / POSSIBLE altered gene function
- LOW priority for clinical exploration / LESS LIKELY altered gene function

### GPX1

**Glutathione Peroxidase 1:** Catalyzes the reduction of hydroperoxides and hydrogen peroxide to water and oxygen with glutathione as a substrate which helps maintain redox balance. Also involved in arachidonic acid metabolism.

### SOD2

**Superoxide Dismutase 2:** Potent mitochondrial antioxidant that binds iron/manganese and converts free superoxide radicals/reactive oxygen species (ROS) to oxygen and hydrogen peroxide.

### NOS2

**Nitric Oxide Synthase 2:** Inducible Nitric Oxide Synthase (iNOS). Produces Nitric Oxide (NO) and L-Citrulline from L-Arginine and Oxygen. iNOS activity is commonly associated with endotoxin, cytokine (IL-6/IL-8), and inflammatory activity.

### SOD3

**Superoxide Dismutase 3:** Potent extracellular antioxidant that binds copper/zinc/heparin and converts free superoxide radicals/reactive oxygen species (ROS) to oxygen and hydrogen peroxide.

### CAT

**Catalase:** Essential antioxidant enzyme responsible for rapidly converting hydrogen peroxide to water and oxygen to protect cells against oxidative stress and toxic effects of hydrogen peroxide.

### NOS3

**Nitric Oxide Synthase 3:** Endothelial Nitric Oxide Synthase (eNOS). Produces Nitric Oxide (NO) and L-Citrulline from L-Arginine and Oxygen. eNOS activity is commonly associated with angiogenesis, vascular functions, cardio health, and blood coagulation

### NOS1

**Nitric Oxide Synthase 1:** Neuronal Nitric Oxide Synthase (nNOS). Produces Nitric Oxide (NO) and L-Citrulline from L-Arginine and Oxygen. nNOS activity is commonly associated with neurotoxicity and neurodegenerative disorders.

### SOD1

**Superoxide Dismutase 1:** Potent intracellular antioxidant that binds copper/zinc and converts free superoxide radicals/reactive oxygen species (ROS) to oxygen and hydrogen peroxide.

## Oxidative Stress: Researched SNPS & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Nitric Oxide	Supports Homeostasis	NOS3	rs1799983	REF	NOS3 enzyme activity and related eNOS activity.	Typical NOS3 enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Antioxidant	Monitor & Support	GPX1	rs1050450	HET	GPX1 enzyme activity and defense against oxidative stress and DNA damage.	Possibly some reduced GPX1 enzyme activity and some reduced ability to manage oxidative stress and DNA damage. Some research suggests GPX activity may be increased in response to increased selenium intake for this result.
Antioxidant	Supports Homeostasis	SOD2	rs4880	HET	MnSOD or SOD2 enzyme activity and management of oxidative stress	Possible altered SOD2 enzyme activity with potentially increased ability to manage oxidative stress.
Nitric Oxide	Monitor & Support	NOS1	rs2682826	HET	NOS1 enzyme expression and related nNOS activity.	Possible reduced NOS1 enzyme expression with potentially decreased ability to manage oxidative stress but also possible reduced risk for neurotoxic nitric oxide levels.
Nitric Oxide	Monitor & Support	NOS2	rs1060826	HOM	NOS2 enzyme activity and related iNOS activity.	Possible reduced NOS2 enzyme expression with potentially decreased ability to manage oxidative stress.



## LOW PRIORITY NETWORK

*Based on your genetic data, this network has a low probability of disruption. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

# Detoxification: Phase 2

Phase 2 detoxification is governed primarily by the GST family of enzymes. Glutathione S-Transferases (GST's) are responsible for attaching Glutathione to the various drugs, chemicals, and other environmental toxins produced by phase I detoxification. This process makes them more water-soluble so they can be more easily removed from the body through sweat, urine, and feces.

## Action Items for Maximized Wellness (Detoxification: Phase 2)



### ACTIVELY SEEK

- Focus on staying hydrated.
- Increase consumption of cruciferous veggies such as cabbage, cauliflower, and broccoli.
- Consume lean protein to help maintain optimum glutathione levels.
- Support healthy liver function.



### ACTIVELY AVOID

- Reduce exposure to environmental toxins such as cigarette smoke, chemicals, air pollution, pesticides, and toxic plastics.
- Reduce mental and emotional stressors.
- Reduce overall toxic burden.

## Clinical Exploration & Diagnostic Tools (Detoxification: Phase 2)

**HEPATIC FUNCTION PANEL:** Make sure the liver is functioning properly and healthy enough to support detoxification efforts

**Oxidative Stress Testing:** Oxidative stress markers might help indicate excessive toxic burdens that need to be addressed.

**GPL-TOX Profile:** The Great Plains Labs GPL-TOX Panel screens for 173 different environmental pollutants using 18 different metabolites, all from a single urine sample.

**Organix® Comprehensive Profile:** The Genova Diagnostics Organix® Comprehensive Profile is a nutritional test providing insights into organic acids and a view into the body's cellular metabolic processes.

## Nutritional Support & Supplements (Detoxification: Phase 2)

---

**Xymogen 6 Day Detox Kit:** Pharmaceutical grade, detox rejuvenation kit. Designed to quickly enhance the body's detox capabilities.

**Xymogen Liver Protect™:** Powerful liver support formula that promotes antioxidant and immune function.

**Xymogen S-Acetyl Glutathione™:** Acetylated glutathione that's enhanced to be stable in the gut. Essential for optimal detoxification functions as well as protection against oxidative stress.

**Xymogen Nrf2 Activator™:** Enhances enzyme activity and expression within detoxification pathways while also downregulating damaging inflammatory cytokine release.

**Xymogen Green Tea 600™:** A powerful and potent green tea extract that is rich in antioxidant polyphenols that support optimal detoxification function as well as healthy gut flora.

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## Detoxification: Phase 2: Genes Analyzed

The genes presented below are associated with **Detoxification: Phase 2**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

- HIGH priority for clinical exploration / MOST LIKELY altered gene function
- MEDIUM priority for clinical exploration / LIKELY altered gene function
- STANDARD priority for clinical exploration / POSSIBLE altered gene function
- LOW priority for clinical exploration / LESS LIKELY altered gene function

### NAT2

**N-Acetyltransferase 2:** Attaches Acetyl-CoA to Various Drugs, Chemicals, and Toxins for Detoxification. Helps Identify Slow, Medium, Fast Acetylators Phenotypes

### GSTM1

**Glutathione S-Transferase Mu 1:** Conjugates environmental toxins & plays a role in the detoxification of oxidative stress byproducts and carcinogens.

### GSTP1

**Glutathione S-Transferase Pi 1:** Conjugates environmental toxins & involved in estrogen metabolism. Also known to increase IL-6 inflammation with increased Vitamin E intake and protect against neurodegeneration.

### UGT1A8

**UDP Glucuronosyltransferase Family 1 Member A8:** Attaches glucuronic acid to bilirubin, phenols, opioids, mycophenolic acid, various drugs, chemicals, and toxins for detoxification.

### NFE2L2

**Nuclear Factor, Erythroid 2 Like 2:** Regulates the Nrf2 signaling pathways and activates antioxidant defense mechanisms.

### UGT1A1

**UDP Glucuronosyltransferase Family 1 Member A1:** Attaches glucuronic acid to bilirubin, estrogen, carcinogens, various drugs, chemicals, and toxins for detoxification.

### UGT1A6

**UDP Glucuronosyltransferase Family 1 Member A6:** Attaches glucuronic acid to bilirubin, hormones, drugs (aspirin, acetaminophen), chemicals, and toxins for detoxification.

### UGT1A9

**UDP Glucuronosyltransferase Family 1 Member A9:** Attaches glucuronic acid to bilirubin, steroids, hormones, drugs, chemicals, and toxins for detoxification.

### NQO1

**NAD(P)H Quinone Dehydrogenase 1:** Uses NAD(P)H to reduce quinones to hydroquinones. Protects against oxidative stress and metabolizes CoQ10 and Vitamin K. Binds FAD as cofactor.

### GSTA1

**Glutathione S-Transferase Alpha 1:** Conjugates Environmental Toxins & Protects Cells from Reactive Oxygen Species (ROS) and Products of Peroxidation.

**GSTO1**

**Glutathione S-Transferase Omega 1:** Conjugates environmental toxins & pro-inflammatory to endotoxins and bacterial infection. Also helps neutralize oxidative stress.

**NAT1**

**N-Acetyltransferase 1:** Attaches Acetyl-CoA to Various Drugs, Chemicals, and Toxins for Detoxification. Helps Identify Slow, Medium, Fast Acetylator Phenotypes

## Detoxification: Phase 2: Researched SNPS & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Carcinogens	Supports Homeostasis	UGT1A6	rs17863783	REF	UGT1A6 enzyme activity, drug toxicity, and carcinogen removal.	Typical UGT1A6 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Bilirubin	Supports Homeostasis	UGT1A1	rs35003977	REF	UGT1A1 enzyme-substrate binding activity and bilirubin metabolism.	Typical UGT1A1 to bilirubin binding activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Bilirubin	Supports Homeostasis	UGT1A1	rs4148323	REF	UGT1A1 enzyme transcription and expression activity, and bilirubin metabolism.	Typical UGT1A1 transcription and expression activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Xenobiotics	Supports Homeostasis	NAT1	rs4987076	REF	NAT1 enzyme transcription and expression activity, and xenobiotic detoxification.	Typical NAT1 transcription and expression activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Acetylation	Monitor & Support	NAT2	rs1208	HOM	NAT2 enzyme activity, and acetylation rate.	Possibly reduced NAT2 enzyme activity. Possible slow acetylator phenotype.

## LOW PRIORITY NETWORK

*Based on your genetic data, this network has a low probability of disruption. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

# Lipid Metabolism

Lipid metabolism refers to both the synthesis as well as the breakdown of fats consumed through food inside the body cells to release energy for bodily processes or to be stored for later use. Optimal health depends on a complex balance of environmental, dietary, and genetic factors that can influence lipid metabolism. Genes listed in this section can influence LDL ('bad' cholesterol) levels, HDL ('good' cholesterol) levels, and other lipid metabolism markers.

## Action Items for Maximized Wellness (Lipid Metabolism)



### ACTIVELY SEEK

- Increase intake of phytonutrient rich foods
- Integrate physical activity into everyday routines.
- Routine heart, liver and lipid metabolism health checkups with your functional medicine practitioner.



### ACTIVELY AVOID

- Focus on avoiding environmental pro-carcinogens
- Avoid high cholesterol foods
- Reduce alcohol consumption.

## Clinical Exploration & Diagnostic Tools (Lipid Metabolism)

**Spectracell CardioMetabolic Risk:** SpectraCell's CardioMetabolic Risk panel covers biomarkers related to metabolic dysfunction, lipids & lipoproteins, cardio health risk, and inflammation.

**LIPID PANEL:** Quest Lab ID# 7600 - Lipid Panel, Standard: This Lipid Panel measures serum cholesterol and triglyceride (TG) levels; it includes evaluation of the cholesterol/HDL-C ratio (calculated), HDL-C, LDL-C (calculated), non-HDL-C (calculated), total cholesterol, and TG.

## Nutritional Support & Supplements (Lipid Metabolism)

**Xymogen LipotropiX™:** Provides fat metabolism, bile flow, and healthy liver function support. \*\*Higher indication with reduced CETP gene activity\*\*











**Xymogen ALAmax™ CR:** Provides Fat-Soluble and Water-Soluble Antioxidant Activity. \*\*Higher indication with reduced PON1 gene activity\*\*

**Xymogen CholeRex™:** Prevents peroxidation of (LDL) and protects from oxidized LDL damage. \*\*Higher indication with reduced PON1 gene activity\*\*

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## Lipid Metabolism: Genes Analyzed

The genes presented below are associated with **Lipid Metabolism**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

	HIGH priority for clinical exploration / MOST LIKELY altered gene function
	MEDIUM priority for clinical exploration / LIKELY altered gene function
	STANDARD priority for clinical exploration / POSSIBLE altered gene function
	LOW priority for clinical exploration / LESS LIKELY altered gene function
	<b>CETP</b> <b>Cholesteryl Ester Transfer Protein:</b> Regulates Reverse Cholesterol Transport. Strong and Independent Risk Factor for CAD.
	<b>APOE</b> <b>Apolipoprotein E:</b> Lipid Transporter Essential for Lipid Metabolism Homeostasis. Famously Associated to Risk of Alzheimer's Disease.
	<b>LPL</b> <b>Lipoprotein Lipase:</b> Key Player in Triglyceride Metabolism. Essential for Lipid Clearance, Utilization, and Storage
	<b>APOC3</b> <b>Apolipoprotein C3:</b> Essential for Triglyceride Homeostasis. Inhibits LPL & LIPC. Altered Function Associated to Low Triglyceride and High HDL Levels
	<b>PON1</b> <b>Paraoxonase 1:</b> Protects LDL from Peroxidation. Responsible for HDL Antioxidant Function. Major Anti-Atherosclerotic Component of HDL
	<b>APOA2</b> <b>Apolipoprotein A2:</b> 2nd most abundant protein in HDL particles, helps to stabilize HDL structure and metabolism activity.



## Lipid Metabolism: Researched SNPs & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
APOE	Supports Homeostasis	APOE	APOE_GenoType	E3	early onset Alzheimer's disease symptoms	This is the most common APOE Type, with no association to either increase or reduce Alzheimer's or CAD risk.
APOE	Monitor & Support	APOE	rs7412	REF	APOE genotype classification. Also associated with cognitive stability and cardio health.	Typical APOE enzyme function. Your GenoType result is associated with normal risk for early cognitive decline. Focus on maintaining cognitive function by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
APOE	Supports Homeostasis	APOE	rs429358	REF	APOE genotype classification. Also associated with cognitive stability and cardio health.	Typical APOE enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Cognition	Supports Homeostasis	APOE	rs121918393	REF	resistance to cognitive decline and APOE subtype result modification.	This Genotype has less effect on APOE subtypes or cognitive decline. Other genotypes at this SNP might have more resistance to early cognitive decline. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Heart	Supports Homeostasis	PON1	rs662	REF	altered PON1 activity, heart health, and risk for coronary artery disease.	Typical PON1 enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Lipids	Supports Homeostasis	LPL	rs328	REF	altered LPL activity, and altered triglycerides, cholesterol, HDL, and LDL levels. Possible response to dietary intake.	Typical LPL enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Lipids	Supports Homeostasis	LPL	rs1801177	REF	altered LPL activity, risk of combined hyperlipidemia, altered triglycerides, cholesterol, HDL, and apolipoproteins (apoB) levels..	Typical LPL enzyme function. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Lipids	Optimize & Enhance	CETP	rs5882	HET	altered serum CETP levels, HDL, LDL, and aging.	Possible some reduced serum CETP levels. Possibly some increased HDL levels, increased HDL & LDL particle size, better cognitive function and healthy aging.

## LOW PRIORITY NETWORK

*Based on your genetic data, this network has a low probability of disruption. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

# Detoxification: Phase 1

Phase 1 detoxification is primarily responsible for processing and breaking down environmental exposures like toxins, pollutants, substances, chemicals, medications, etc... into metabolites that our bodies can utilize or remove. Toxic metabolites formed during phase 1 detoxification are then passed through phase 2 detoxification where they get further modified to promote removal from the body.

## Action Items for Maximized Wellness (Detoxification: Phase 1)



### ACTIVELY SEEK

- Focus on staying hydrated.
- Increase consumption of polyphenol rich, high-antioxidant foods and teas.
- Increase consumption of cruciferous veggies such as cabbage, cauliflower, and broccoli.
- Support healthy liver function.



### ACTIVELY AVOID

- Reduce exposure to environmental toxins such as cigarette smoke, chemicals, air pollution, pesticides, and toxic plastics.
- Reduce alcohol consumption
- Avoid frying or using high heat cooking methods that can char or burn food.
- Reduce overall toxic burden.

## Clinical Exploration & Diagnostic Tools (Detoxification: Phase 1)

**HEPATIC FUNCTION PANEL:** Make sure the liver is functioning properly and healthy enough to support detoxification efforts

**Oxidative Stress Testing:** Oxidative stress markers might help indicate excessive toxic burdens that need to be addressed.

**GPL-TOX Profile:** The Great Plains Labs GPL-TOX Panel screens for 173 different environmental pollutants using 18 different metabolites, all from a single urine sample.

**Organix® Comprehensive Profile:** The Genova Diagnostics Organix® Comprehensive Profile is a nutritional test providing insights into organic acids and a view into the body's cellular metabolic processes.

## Nutritional Support & Supplements (Detoxification: Phase 1)

---

**Xymogen 6 Day Detox Kit:** Pharmaceutical grade, detox rejuvenation kit. Designed to quickly enhance the bodies detox capabilities.

**Xymogen Liver Protect™:** Powerful liver support formula that promotes antioxidant and immune function.

**Xymogen S-Acetyl Glutathione™:** Acetylated glutathione that is enhanced to be stable in the gut. Essential for optimal detoxification functions as well as protection against oxidative stress.





**Xymogen Nrf2 Activator™:** Enhances enzyme activity and expression within detoxification pathways while also downregulating damaging inflammatory cytokine release.

**Xymogen Green Tea 600™:** A powerful and potent green tea extract that is rich in antioxidant polyphenols that support optimal detoxification function as well as health gut flora.

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## Detoxification: Phase 1: Genes Analyzed

The genes presented below are associated with **Detoxification: Phase 1**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

	HIGH priority for clinical exploration / MOST LIKELY altered gene function
	MEDIUM priority for clinical exploration / LIKELY altered gene function
	STANDARD priority for clinical exploration / POSSIBLE altered gene function
	LOW priority for clinical exploration / LESS LIKELY altered gene function
<b>CYP1A1</b>	<b>Cytochrome P450-1A1:</b> Metabolizes Estrogen (17B-estradiol), Aromatic Hydrocarbons, Arachidonic Acid, DHA, Drugs, Chemicals, and more...
<b>CYP2A6</b>	<b>Cytochrome P450-2A6:</b> Metabolizes Nicotine, Retinoid Acid, Steroid Compounds, Drugs, Chemicals, and more...
<b>CYP2C19</b>	<b>Cytochrome P450 2C19:</b> Metabolizes Proton Pump Inhibitors, Progesterone, Estradiol, Melatonin, SSRIs, Drugs, Chemicals, and more...
<b>CYP1A2</b>	<b>Cytochrome P450-1A2:</b> Metabolizes Estrogen, Caffeine, Aflatoxin B1(Mold), Acetaminophen, Drugs, Chemicals, and more...
<b>CYP2C9</b>	<b>Cytochrome P450 2C9:</b> Metabolizes serotonin, THC, NSAIDs, fatty acids, cholesterol, linoleic acid, arachidonic acid, drugs, chemicals, and more...
<b>CYP2D6</b>	<b>Cytochrome P450-2D6:</b> Metabolizes Serotonin, Pregnenolone, Cholesterol, Androsterone, Eicosanoids, Opioids, Cough Syrup, Drugs, Chemicals, and more...
<b>CYP3A4</b>	<b>Cytochrome P450-3A4:</b> Plays a role in metabolizing about 50% of the drugs commonly prescribed. Can easily become overwhelmed trying to metabolize too many drugs.
<b>CYP2E1</b>	<b>Cytochrome P450-2E1:</b> Metabolizes fatty acids, alcohol, acrylamide, acetaminophen, chlorzoxazone, drugs, chemicals, and more... Expressed and active in the brain.
<b>CYP3A5</b>	<b>Cytochrome P450-3A5:</b> Plays a role in metabolizing about 50% of the drugs commonly prescribed. Can easily become overwhelmed trying to metabolize too many drugs.

## Detoxification: Phase 1: Researched SNPS & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Estrogen	Supports Homeostasis	CYP1A1	rs1048943	REF	CYP1A1 enzyme activity and estrogen metabolism	Typical CYP1A1 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Nicotine	Monitor & Support	CYP2A6	rs5031017	REF	CYP2A6 enzyme activity and nicotine addiction risk.	Typical CYP2A6 enzyme activity. Higher risk for nicotine addiction
Nicotine	Monitor & Support	CYP2A6	rs1801272	REF	CYP2A6 enzyme activity and nicotine addiction risk.	Typical CYP2A6 enzyme activity. Higher risk for nicotine addiction
Warfarin	Supports Homeostasis	CYP2C9	rs1799853	REF	CYP2C9 enzyme activity and Warfarin metabolism.	Typical CYP2C9 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Warfarin	Supports Homeostasis	CYP2C9	rs1057910	REF	CYP2C9 enzyme activity and Warfarin metabolism.	Typical CYP2C9 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Pharma	Supports Homeostasis	CYP2C9	rs2256871	REF	CYP2C9 enzyme activity and drug metabolism activity.	Typical CYP2C9 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Pharma	Supports Homeostasis	CYP2D6	rs28371706	REF	CYP2D6 enzyme activity and drug metabolism activity.	Typical CYP2D6 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
Coffee	Optimize & Enhance	CYP1A2	rs2472304	HET	CYP1A2 enzyme activity and coffee consumption.	Slightly increased CYP1A2 enzyme activity. Possible slight increase in coffee consumption.
Pharma	Monitor & Support	CYP2D6	rs16947	HET	CYP2D6 enzyme activity and drug metabolism activity.	Reduced CYP2D6 enzyme activity. Possible slight reduction in drug metabolism activity.

## LOW PRIORITY NETWORK

*Based on your genetic data, this network has a low probability of disruption. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

# Inflammation

Inflammation is a normal activity of the human body and is necessary to begin the healing process resulting from injury. The keyword here is "injury"... When the human body continuously battles against "injury," it is in a constant state of inflammation. We often think of inflammation as red, hot to the touch, swelling, and pain. Most people don't realize that the inflammatory process can occur without those obvious warning signs, potentially creating constant damage to our healthy cells and body tissues. Inflammation is a valuable warning sign, but when inflammation becomes out of balance, it can be disastrous.

## Action Items for Maximized Wellness (Inflammation)



### ACTIVELY SEEK

- Increase anti-inflammatory food intake.
- Reduce emotional, physical, and psychological stress.
- Maintain good oral health and hygiene.
- Focus on developing positive emotions like gratitude, inspiration, happiness, excitement, and pride.



### ACTIVELY AVOID

- Avoid inflammatory meals that contain things like alcohol, refined sugars and fast food.
- Avoid excessive blue light exposures from things like phone, iPad, TV, and laptop screens.
- Be mindful of negative emotions like hate, anger, jealousy and sadness.

## Clinical Exploration & Diagnostic Tools (Inflammation)

**High Sensitivity C-Reactive Protein (HS-CRP):** This acute phase protein (CRP) increases when you have certain diseases or biological functions causing inflammation.

**Erythrocyte Sedimentation Rate (ESR):** ESR tests red blood cell stickiness. Sticky red blood cells or increased ESR can indicate that you have some inflammation, somewhere in the body.

**Blood/Plasma Viscosity:** Blood coagulation cascades are activated during many inflammatory conditions. Detecting high viscosity may indicate inflammation.

## Nutritional Support & Supplements (Inflammation)

---

**Xymogen ALAmax™ CR:** Alpha-lipoic acid is an antioxidant that can reduce inflammation and may improve the symptoms of certain diseases.

**Xymogen CurcuPlex CR™:** Curcumin is a potent anti-inflammatory supplement that reduces inflammation in a wide range of diseases.





**Xymogen Omega MonoPure® DHA EC:** Fish oil supplements containing omega-3 fatty acids can improve inflammation in several diseases and conditions.

**Photobiomodulation:** Also known as laser light therapy. Specific light wavelengths are known to reduce IL1B, IL6, and general inflammation levels.

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## Inflammation: Genes Analyzed

The genes presented below are associated with **Inflammation**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

-  HIGH priority for clinical exploration / MOST LIKELY altered gene function
-  MEDIUM priority for clinical exploration / LIKELY altered gene function
-  STANDARD priority for clinical exploration / POSSIBLE altered gene function
-  LOW priority for clinical exploration / LESS LIKELY altered gene function

### ICAM1

**Intercellular Adhesion Molecule 1:** Also known as CD54, this glycoprotein can be induced by IL-1 and TNF activity. Levels increase greatly upon cytokine stimulation. Binds to Integrins of CD11/CD18.

### IL1RN

**Interleukin 1 Receptor Antagonist:** Binds functional IL1A/IL1B receptors to inhibit IL1A/IL1B activity and regulate IL1 related immune and inflammatory responses.

### IL1B

**Interleukin 1 Beta:** This cytokine plays a central role in regulating acute immune, and inflammatory response. Contributes to inflammatory pain hypersensitivity.

### IL1A

**Interleukin 1 Alpha:** This cytokine plays a central role in regulating acute immune, and inflammatory response. Induces apoptosis in response to cell injury.

### TNF

**Tumor Necrosis Factor:** Essential inflammatory cytokine produced by our immune system during acute and chronic inflammatory conditions to signal apoptosis and more.

### IL6

**Interleukin 6:** A potent cytokine involved in a wide variety of acute and chronic inflammatory reactions. Capable of inducing fever during autoimmune disease and infection response. Required for the generation of TH17 cells.

### CRP

**C-Reactive Protein:** C-Reactive Protein levels increase greatly during acute phase response to tissue injury, infection, or other inflammatory stimuli to promote host defense mechanisms.



## Inflammation: Researched SNPS & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Inflammation	Supports Homeostasis	IL1A	rs17561	REF	IL1A enzyme activity and inflammatory conditions.	Typical IL1A expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	IL1B	rs1143634	REF	IL1B enzyme activity and inflammatory conditions.	Typical IL1B expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	IL1RN	rs878972	REF	IL1RN enzyme activity and inflammatory conditions.	Typical IL1RN expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	TNF	rs1800610	REF	TNF enzyme activity and inflammatory conditions.	Typical TNF expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	TNF	rs3093662	REF	TNF enzyme activity and inflammatory conditions.	Typical TNF expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	IL6	rs1524107	REF	IL6 enzyme activity and inflammatory conditions.	Typical IL6 expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.
Inflammation	Supports Homeostasis	IL6	rs1800795	REF	IL6 enzyme activity and inflammatory conditions.	Typical IL6 expression and activity. This result may carry more risk for more severe acute inflammatory response than HET or HOM results but also possible increased protection from chronic inflammatory conditions.

## LOW PRIORITY NETWORK

*Based on your genetic data, this network has a low probability of disruption. Lifestyle tips to support this network and lab testing options to further explore network activity are provided.*

# SARS-CoV-2 Severity

Severe acute respiratory syndrome coronavirus 2 is a highly pathogenic variant belonging to the family of coronaviruses. It was first detected in 2019 in China and has since spread all around the world, causing a pandemic. Some common symptoms of this virus include the common cold, fever, loss of smell and taste, cough, shortness of breath or difficulty in breathing, tiredness, etc. The clinical presentation of this virus ranges from asymptomatic and mild respiratory tract infections to influenza-like disease and severe illnesses that can result in multiorgan failure, lung injury, and even death. Although it is commonly believed that the SARS-CoV-2 replicates in the lungs, infected patients have reported symptoms that indicate the involvement of the heart, kidneys, cardiovascular systems, and other organs. TNF, CXCR6, C4BPA, F3, ABO, and IL17 are some of the genes closely linked to SARS-CoV-2 Severity.

## Action Items for Maximized Wellness (SARS-CoV-2 Severity)



### ACTIVELY SEEK

- Follow CDC guidelines for COVID safety.
- Rest and relaxation. Get plenty of sleep!
- Get out in the sun, enjoy the outdoors, and possibly do some "forest bathing" or "shinrin-yoku".
- Reduce stress.. Deep Breathe. Pray. Meditate. Exercise. Express gratitude.



### ACTIVELY AVOID

- Intermittent fasting which can increase cytokine storm intensity
- Areas or activities with increased risk of COVID-19 exposure.
- Touching your hands to your mouth, nose, or eyes
- Reactive or inflammatory meals that contain things like dairy, gluten, trans fats, sugar, and processed foods.

## Clinical Exploration & Diagnostic Tools (SARS-CoV-2 Severity)

**COVID-19 Viral Detection:** Helps detect an active viral infection

**COVID-19 Antibodies:** Helps detect a previous viral infection and proper immune response from a previous infection or vaccine.

## Nutritional Support & Supplements (SARS-CoV-2 Severity)

---

**Xymogen OlivDefense®:** Promotes antiviral & antioxidant activity, helps manage inflammatory response & cytokine release, and supports defense against immune challenges.

**Creating Health's Bo-D-Zyme™:** Proteolytic enzymes that help break down damaged tissue to prevent an ongoing inflammatory response. Helps the body recover, heal, and maintain joint and tissue integrity.

**Xymogen Viragraphis™:** This formula may help improve recovery from upper respiratory viral infection and support overall energy.

**Creating Health's OncoDetox™ ES:** Potent NRF-2 stimulator that promotes glutathione production and supports lung health as well as immune, nervous, and cardiovascular system health.

If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.

## SARS-CoV-2 Severity: Genes Analyzed

The genes presented below are associated with **SARS-CoV-2 Severity**. Click the gene symbols to see the variants we found in that gene. To explore additional data about your other genes, SNPs, and variants not included in this document, see your GeneSavvy Variant Exploration Report spreadsheet.

- HIGH priority for clinical exploration / MOST LIKELY altered gene function
- MEDIUM priority for clinical exploration / LIKELY altered gene function
- STANDARD priority for clinical exploration / POSSIBLE altered gene function
- LOW priority for clinical exploration / LESS LIKELY altered gene function

### ABO

**ABO Blood Group:** The basis of our A-B-O blood types. A, B and AB blood types express glycosyltransferase activity that converts the H antigen to A or B antigens. O Blood types lack this activity.

### CXCR6

**C-X-C Motif Chemokine Receptor 6:** Chemokine receptor which can work in conjunction with CD4 to act as an entry co-receptor which can be used by retroviruses (Lentivirus) and possibly other pathogens to enter target cells.

### F3

**Coagulation Factor 3, Tissue Factor:** Also known as "Tissue Factor", this potent cell surface glycoprotein enables cells to initiate the blood coagulation cascade through the "Extrinsic" coagulation cascade pathway.

### C4BPA

**Complement Component 4 Binding Protein Alpha:** Required for assembly of the C4B protein which controls the activation of the complement cascade through the "classical" complement pathway. Also interacts with the Protein S anticoagulant enzyme and Amyloid P.

### TNF

**Tumor Necrosis Factor:** Essential inflammatory cytokine produced by our immune system during acute and chronic inflammatory conditions to signal apoptosis and more.

### IL17A

**Interleukin 17 Alpha:** Pro-Inflammatory Cytokine produced by activated T-Cells that can stimulate IL-6/COX-2 expression and enhance NO production. Also known to regulate NF-KappaB and MAPK activity.

## SARS-CoV-2 Severity: Researched SNPS & Variants

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype
- Personalized Environmental Optimization

HEALTH CATEGORY	CLINICAL ACTION	GENE	VARIANT	GENOTYPE	VARIANT ASSOCIATIONS	NOTES FOR YOUR GENOTYPE
Blood Type	Supports Homeostasis	ABO	rs8176719	REF	ABO blood types and increased risk for SARS-CoV-2 severity.	Most likely "O" blood type. Possibly lower COVID-19 related risk.
SARS-CoV-2	Monitor & Support	CXCR6	rs2234355	REF	chemokine interactions between pathogens and CD4 cells and associated increased risk for SARS-CoV-2 severity.	Typical CXCR6 enzyme activity which has been associated with increased depletion of CD4 counts under viral load and increased viral load severity.
SARS-CoV-2	Supports Homeostasis	F3	rs3917643	REF	blood coagulation rate and associated risks for SARS-CoV-2 severity.	Typical F3 enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
SARS-CoV-2	Supports Homeostasis	C4BPA	rs45574833	REF	C4BPA binding activity and associated increased risk for SARS-CoV-2 severity.	Typical C4BPA enzyme activity. Your GenoType result supports pathway homeostasis. Focus on maintaining this stability by utilizing the "Action Items for Maximized Wellness" mentioned in this report.
ARDS	Monitor & Support	IL17A	rs2275913	REF	IL-17 expression, activity risk for Acute Respiratory Distress Syndrome (ARDS).	Typical IL-17 expression and activity. This result may carry more risk for Acute Respiratory Distress than HET or HOM results.
ARDS	Monitor & Support	TNF	rs1800610	REF	TNF expression, activity risk for Acute Respiratory Distress Syndrome (ARDS).	Typical TNF expression and activity. This result may carry more risk for Acute Respiratory Distress than HET or HOM results but also possible increased pathogen clearance activity and protection from persistent infection.

# Reference Library of Genes and Variants

(sorted alphabetically)



*If your data came from a GeneSavvy Starter Kit, congratulations! Our labs utilize next generation sequencing technology to scan every single protein-coding piece of DNA in your body (full exome), some non-coding regions (introns), and mitochondrial DNA as well. The breadth of this data gives us confidence that if something in your genes is affecting your health, we will find it.*

- HIGH priority for clinical exploration / HOMOZYGOUS genotype
- MEDIUM priority for clinical exploration
- STANDARD priority for clinical exploration / HETEROZYGOUS genotype
- LOW priority for clinical exploration / REFERENCE genotype

## **ABO** ABO Blood Group *Basepairs Analyzed By GeneSavvy: All Exon Locations*

This gene encodes for the ABO enzyme which is the basis of our A-B-O blood types system. A, B and AB blood types express glycosyltransferase activity that converts the H antigen to A or B antigens. O Blood types lack this activity.

### Client's Genetic Variants and SNPs Identified Within the **ABO** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>ABO</b>	rs8176707	<b>HET</b>	Uncommon 7.4%	Intron	Low

## **APOA2** Apolipoprotein A2 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

2nd most abundant protein in HDL particles, helps to stabilize HDL structure and metabolism activity.

### Client's Genetic Variants and SNPs Identified Within the **APOA2** Gene

No variants were found for this gene.

## APOC3 Apolipoprotein C3 Basepairs Analyzed By GeneSavvy: 3,163

APOC3 plays an important role in cholesterol metabolism. It inhibits lipoprotein lipase and hepatic lipase, delaying catabolism of triglyceride-rich particles. APOC3 plays a multifaceted role in triglyceride homeostasis. APOC3 intracellularly, promotes hepatic VLDL1 assembly and secretion; extracellularly, attenuates hydrolysis and clearance of triglyceride-rich lipoproteins. APOC3 impairs the lipolysis of TRLs by inhibiting lipoprotein lipase and the hepatic uptake of TRLs by remnant receptors.

### Client's Genetic Variants and SNPs Identified Within the APOC3 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
APOC3	rs2070669	HET	Common 53.0%	Intron	Low
APOC3	rs2070668	HET	Common 42.8%	Intron	Low
APOC3	rs4520	HOM	Common 70.4%	Silent	Low
APOC3	rs5128	HOM	Common 86.6%	UTR3	Low
APOC3	rs4225	HET	Common 45.0%	UTR3	Low

## APOE Apolipoprotein E Basepairs Analyzed By GeneSavvy: 3,877

Apolipoprotein E (APOE) transports fat-soluble vitamins and cholesterol into the lymph system and then into the blood. APOE has been shown to reduce cholesterol levels, reduce the risk of heart disease, and reduce inflammation. There are at least three slightly different versions (alleles) of the APOE gene. The major subtypes of APOE are called e2, e3, and e4 which are comprised of alleles at 2 different locations within the gene: rs429358, and rs7412. Other notable variants are rs429358, rs7412, and the rs121918393 "ChristChurch" allele.

### Client's Genetic Variants and SNPs Identified Within the APOE Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
APOE	rs440446	HET	Common 69.7%	Missense	Medium

## **BMP2** Bone Morphogenetic Protein 2 *Basepairs Analyzed By GeneSavvy: All Exon*

### *Locations*

The BMP2 gene encodes for a secreted ligand of the TGF-Beta superfamily of proteins. This ligand for TGF-Beta binds various TGF-Beta receptors to activate SMAD enzymes to induce cartilage and bone formation. Also plays a role in regulating myogenesis.

### Client's Genetic Variants and SNPs Identified Within the **BMP2** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>BMP2</b>	rs235768	HOM	Common 72.6%	Missense	High
<b>BMP2</b>	rs1049007	HOM	Common 70.6%	Silent	Low

## **C4BPA** Complement Component 4 Binding Protein Alpha *Basepairs*

### *Analyzed By GeneSavvy: All Exon Locations*

This gene encodes for the Complement Component 4 Binding Protein Alpha enzyme which is required for assembly of the C4B protein that controls the activation of the complement cascade through the "classical" complement pathway. C4BPA also interacts with the Protein S anticoagulant enzyme and Amyloid P.

### Client's Genetic Variants and SNPs Identified Within the **C4BPA** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>C4BPA</b>	rs1126618	HOM	Common 76.9%	Silent	Low

## **CAT** Catalase *Basepairs Analyzed By GeneSavvy: All Exon Locations*

The CAT gene encodes for the Catalase enzyme which plays essential for protecting our cells against oxidative stress. The CAT enzyme is responsible for rapidly converting hydrogen peroxide to water and oxygen to protect cells against oxidative stress and toxic effects of hydrogen peroxide. Can convert hydrogen peroxide to water and oxygen at a rate of 1 million hydrogen peroxide molecules per second!

### Client's Genetic Variants and SNPs Identified Within the **CAT** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>CAT</b>	rs1049982	HET	Common 60.7%	UTR5	Low



## CBS Cystathionine Beta-Synthase *Basepairs Analyzed By GeneSavvy: 23,171*

Cystathionine beta synthase catalyses the conversion of homocysteine to cystathione and is directly involved in the removal of homocysteine from the methionine cycle, thus any alterations in its activity could affect homocysteine levels.

### Client's Genetic Variants and SNPs Identified Within the CBS Gene

No variants were found for this gene.

## CETP Cholesteryl Ester Transfer Protein *Basepairs Analyzed By GeneSavvy: 21,954*

CETP is a strong and independent risk factor for CAD. CETP plays a key role in the metabolism of HDL and mediates the exchange of lipids between lipoproteins, resulting in the eventual uptake of cholesterol by hepatocytes (reverse cholesterol transport). High plasma CETP concentration is associated with reduced HDL-C concentrations. CETP is involved in the transfer of neutral lipids, including cholesteryl ester and triglyceride, among lipoprotein particles. CETP allows the net movement of cholesteryl ester from high density lipoproteins/HDL to triglyceride-rich very low density lipoproteins/VLDL, as well as transport of triglyceride from VLDL to HDL. CETP also regulates the reverse cholesterol transport, by which excess cholesterol is removed from peripheral tissues and returned to the liver for elimination

### Client's Genetic Variants and SNPs Identified Within the CETP Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CETP	rs5882	HET	Common 58.8%	Missense	High
CETP	rs3816117	HET	Common 52.2%	Intron	Low
CETP	rs711752	HET	Uncommon 38.3%	Intron	Low
CETP	rs34620476	HET	Uncommon 36.0%	Intron	Low
CETP	rs891141	HOM	Common 96.0%	Intron	Low
CETP	rs891142	HOM	Common 97.2%	Intron	Low
CETP	rs891143	HOM	Common 97.4%	Intron	Low
CETP	rs7205804	HET	Uncommon 34.2%	Intron	Low
CETP	rs1532625	HET	Uncommon 34.1%	Intron SpliceSite	Low

## CILP Cartilage Intermediate Layer Protein Basepairs Analyzed By GeneSavvy: All

### Exon Locations

This gene encodes for the Cartilage Intermediate Layer Protein (CILP) which plays a role in cartilage scaffolding and the formation and function of the cartilage extracellular matrix. May antagonize and interact with TGF-Beta and IGF-1. Associated with Joint Disease and Osteoarthritis. Overexpression of this gene may lead to impaired chondrocyte growth and cartilage extracellular matrix repair. Overexpression of this gene may also indirectly promote inorganic pyrophosphate supersaturation in aging and osteoarthritic cartilage.

### Client's Genetic Variants and SNPs Identified Within the CILP Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CILP	rs2073711	HOM	Common 61.2%	Missense SpliceSite	Severe
CILP	rs938952	HOM	Common 75.0%	Missense	Medium
CILP	rs2679118	HOM	Common 98.8%	Missense	Medium
CILP	rs2679117	HOM	Common 98.8%	Missense	Medium

## COL10A1 Collagen Type 10 Alpha 1 Chain Basepairs Analyzed By GeneSavvy: All

### Exon Locations

The Collagen Type 10 Alpha 1 Chain (COL10A1) gene encodes for a protein that is an essential piece for the creation of Type 10 Collagen. Collagens are the main proteins used to create the body's various connective tissues, tendons, ligaments, bones, blood vessels, organs, and other structures. Type 10 Collagen is essential for new bone and articulate joint space and cartilage formation. Essential for healthy joint function and resistance to normal joint wear and tear.

### Client's Genetic Variants and SNPs Identified Within the COL10A1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
COL10A1	rs2228547	HET	Uncommon 16.5%	Missense	High
COL10A1	rs1064583	HET	Common 47.4%	Missense	Medium

## COL1A1 Collagen Type I Alpha 1 Chain *Basepairs Analyzed By GeneSavvy: All Exon*

### Locations

The Collagen Type I Alpha 1 Chain (COL1A1) gene encodes for a protein that is an essential piece for the creation of Type 1 Collagen. Collagens are the main proteins used to create the body's various connective tissues, tendons, ligaments, bones, blood vessels, organs, and other structures. Type 1 Collagen is the most abundant collagen of the family and helps make tendons, ligaments, bone, skin, hair, and teeth. Type 1 Collagen is also essential for tissue strength and elasticity.

### Client's Genetic Variants and SNPs Identified Within the COL1A1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
COL1A1	rs1061237	HET	Uncommon 30.9%	UTR3	Low
COL1A1	rs2249492	HET	Common 52.3%	Intron	Low
COL1A1	rs2734272	HOM	Common 100.0%	Silent	Low

## COL2A1 Collagen Type 2 Alpha 1 Chain *Basepairs Analyzed By GeneSavvy: All Exon*

### Locations

The Collagen Type 2 Alpha 1 Chain (COL2A1) gene encodes for a protein that is an essential piece for the creation of Type 2 Collagen. Collagens are the main proteins used to create the body's various connective tissues, tendons, ligaments, bones, blood vessels, organs, and other structures. Type 2 Collagen is most specific to joint cartilage, skeletal development, linear growth. Essential for tissue stability and the ability for cartilage to withstand compressive forces.

### Client's Genetic Variants and SNPs Identified Within the COL2A1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
COL2A1	rs2070739	HET	Uncommon 10.0%	Missense	Severe
COL2A1	rs12721427	HET	Rare 5.3%	Missense	High
COL2A1	rs3803183	HET	Common 75.8%	Missense	Medium
COL2A1	rs12721425	HET	Rare 5.3%	Intron	Low

## COL3A1 Collagen Type 3 Alpha 1 Chain

Basepairs Analyzed By GeneSavvy: All Exon

### Locations

The Collagen Type 3 Alpha 1 Chain (COL3A1) gene encodes for a protein that is an essential piece for the creation of Type 3 Collagen. Collagens are the main proteins used to create the body's various connective tissues, tendons, ligaments, bones, blood vessels, organs, and other structures. Type 3 Collagen is mostly found in extensible connective tissues such as skin, lungs, uterus, intestines, and the vascular system. Essential for tissue strength and elasticity, similar to Type 1 Collagen.

### Client's Genetic Variants and SNPs Identified Within the COL3A1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
COL3A1	rs1800255	HET	Uncommon 30.0%	Missense	High
COL3A1	rs1516446	HOM	Common 99.7%	Missense	Medium
COL3A1	rs150919697	HET	Very Rare 0.1%	Intron	Low
COL3A1	rs1801184	HET	Uncommon 34.4%	Silent	Low



## COL4A1 Collagen Type 4 Alpha 1 Chain *Basepairs Analyzed By GeneSavvy: All Exon*

### Locations

The Collagen Type 4 Alpha 1 Chain (COL4A1) gene encodes for a protein that is an essential piece for the creation of Type 4 Collagen. Collagens are the main proteins used to create the body's various connective tissues, tendons, ligaments, bones, blood vessels, organs, and other structures. Type 4 Collagen is essential for the creation of sheet-type tissues and structures in our body like basement membranes. Important for wound healing, digestion, respiration, and filtration.

### Client's Genetic Variants and SNPs Identified Within the COL4A1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
COL4A1	rs3742207	HET	Uncommon 31.1%	Missense	Medium
COL4A1	rs681884	HET	Common 76.7%	Intron	Low
COL4A1	rs1133219	HET	Uncommon 35.4%	Silent	Low
COL4A1	rs1213026	HET	Common 61.8%	Intron	Low
COL4A1	rs652572	HET	Common 58.2%	Intron	Low
COL4A1	rs1778817	HET	Common 61.7%	Intron	Low
COL4A1	rs589985	HET	Common 64.1%	Intron	Low
COL4A1	rs2289799	HET	Uncommon 34.4%	Intron	Low
COL4A1	rs874203	HET	Uncommon 33.9%	Silent	Low
COL4A1	rs874204	HET	Uncommon 33.9%	Silent	Low
COL4A1	rs16975492	HET	Uncommon 32.8%	Silent	Low
COL4A1	rs1373744	HOM	Common 93.0%	Silent	Low
COL4A1	rs995224	HET	Uncommon 25.9%	Silent	Low

## COL5A1 Collagen Type 5 Alpha 1 Chain *Basepairs Analyzed By GeneSavvy: All Exon*

### Locations

The Collagen Type 5 Alpha 1 Chain (COL5A1) gene encodes for a protein that is an essential piece for the creation of Type 5 Collagen. Collagens are the main proteins used to create the body's various connective tissues, tendons, ligaments, bones, blood vessels, organs, and other structures. Type 5 Collagen is mostly found in skin, hair, eyes, and placental tissues. Type 5 collagen also plays a role in the formation of type 1 and 3 collagens.

### Client's Genetic Variants and SNPs Identified Within the COL5A1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
COL5A1	rs3124299	HOM	Uncommon 35.0%	Silent	Low
COL5A1	rs3124302	HOM	Uncommon 39.5%	Intron	Low
COL5A1	rs3124308	HOM	Uncommon 39.4%	Intron	Low
COL5A1	rs3128598	HOM	Uncommon 39.3%	Intron	Low
COL5A1	rs3109675	HOM	Common 41.8%	Intron	Low
COL5A1	rs2228560	HET	Uncommon 39.7%	Silent	Low

## COMT Catechol-O-Methyltransferase *Basepairs Analyzed By GeneSavvy: 28,235*

Soluble catechol-O-methyltransferase (S-COMT) helps control the levels of certain hormones and is involved in the inactivation of the catecholamine neurotransmitters (dopamine, epinephrine, and norepinephrine). The enzyme introduces a methyl group to the catecholamine, which is donated by S-adenosyl methionine (SAM). Any compound having a catechol structure, like catecholestrogens and catechol-containing flavonoids, are substrates of COMT.

### Client's Genetic Variants and SNPs Identified Within the COMT Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
COMT	rs6269	HET	Uncommon 37.8%	UTR5	Low
COMT	rs4818	HET	Uncommon 32.1%	Silent	Low



## CRP C-Reactive Protein *Basepairs Analyzed By GeneSavvy: All Exon Locations*

C-Reactive Protein is involved in several host defense-related functions based on its ability to recognize foreign pathogens and damaged cells of the host and to initiate their elimination. C-Reactive Protein levels increase greatly during acute phase response to tissue injury, infection, or other inflammatory stimuli to promote host defense mechanisms such as agglutination, bacterial capsular swelling, phagocytosis, and complement fixation through its calcium-dependent binding to phosphorylcholine.

### Client's Genetic Variants and SNPs Identified Within the CRP Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CRP	rs1130864	HET	Uncommon 30.6%	UTR3	Low
CRP	rs1417938	HET	Uncommon 30.6%	Intron	Low

## CXCR6 C-X-C Motif Chemokine Receptor 6 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

This gene encodes for the C-X-C Motif Chemokine Receptor 6 enzyme which can work in conjunction with CD4 to act as an entry co-receptor that can be used by retroviruses (Lentivirus) and possibly other pathogens to enter target cells.

### Client's Genetic Variants and SNPs Identified Within the CXCR6 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CXCR6	rs2234358	HOM	Common 47.8%	UTR3	Low

## CYP1A1 Cytochrome P450-1A1 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

The CYP1A1 enzyme is part of the P450 superfamily and is heavily involved in drug metabolism and the synthesis of estrogen (17B-estradiol), cholesterol, steroids, arachidonic acid, DHA, and other lipids. CYP1A1 is also associated with the detoxification of environmental toxins and chemicals such as aromatic hydrocarbons commonly found in cigarette smoke.

### Client's Genetic Variants and SNPs Identified Within the CYP1A1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CYP1A1	rs145198866	HET	Very Rare 0.4%	Missense	High

## CYP1A2 Cytochrome P450-1A2 Basepairs Analyzed By GeneSavvy: All Exon Locations

The CYP1A2 enzyme is part of the P450 superfamily and is heavily involved in drug metabolism and the synthesis of estrogen, caffeine, aflatoxin B1(mold), acetaminophen, cholesterol, steroids, and other lipids. CYP1A1 is also associated with the detoxification of environmental toxins and chemicals such as aromatic hydrocarbons commonly found in cigarette smoke.

### Client's Genetic Variants and SNPs Identified Within the CYP1A2 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CYP1A2	rs762551	HET	Common 67.3%	Intron	Low
CYP1A2	rs2472304	HET	Common 43.3%	Intron	Low
CYP1A2	rs2470890	HET	Common 57.0%	Silent	Low

## CYP2A6 Cytochrome P450-2A6 Basepairs Analyzed By GeneSavvy: All Exon Locations

The CYP2A6 enzyme is part of the P450 superfamily and is heavily involved in drug metabolism and the synthesis of retinoic acid, steroids, and other lipids. CYP2A6 is also associated with the detoxification of environmental toxins and chemicals and is the main enzyme responsible for metabolizing nicotine. Because of CYP2A6's role in nicotine metabolism, it is often associated with nicotine addiction and tobacco consumption.

### Client's Genetic Variants and SNPs Identified Within the CYP2A6 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CYP2A6	rs6413474	HET	Very Rare 1.1%	Missense	High
CYP2A6	rs8192733	HET	Uncommon 37.0%	UTR3	Low
CYP2A6	rs56113850	HET	Common 51.9%	Intron	Low
CYP2A6	rs4986892	HET	Rare 2.1%	Silent	Low
CYP2A6	rs79584535	HET	Rare 2.3%	Silent	Low
CYP2A6	rs1137115	HET	Common 76.1%	Silent	Low





## CYP2C19 Cytochrome P450 2C19 Basepairs Analyzed By GeneSavvy: All Exon

### Locations

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 known to metabolize many xenobiotics, including the anticonvulsive drug mephenytoin, omeprazole, diazepam and some barbiturates. Polymorphism within this gene is associated with variable ability to metabolize mephenytoin, known as the poor metabolizer and extensive metabolizer phenotypes. The gene is located within a cluster of cytochrome P450 genes on chromosome 10q24.

### Client's Genetic Variants and SNPs Identified Within the CYP2C19 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CYP2C19	rs3758581	HOM	Common 95.4%	Missense	Medium
CYP2C19	rs17885098	HOM	Common 92.0%	Silent	Low

## CYP2C9 Cytochrome P450 2C9 Basepairs Analyzed By GeneSavvy: All Exon Locations

The CYP2C9 enzyme is part of the P450 superfamily and is heavily involved in drug metabolism and the synthesis of serotonin, THC, NSAIDs, fatty acids, cholesterol, linoleic acid, arachidonic acid, steroids, and other lipids. CYP2C9 is also associated with the detoxification of environmental toxins and chemicals and is the main enzyme responsible for metabolizing warfarin.

### Client's Genetic Variants and SNPs Identified Within the CYP2C9 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CYP2C9	rs9332104	HET	Uncommon 19.4%	Intron	Low
CYP2C9	rs9332120	HET	Uncommon 19.4%	Intron	Low
CYP2C9	rs28371675	HET	Uncommon 13.5%	Intron	Low

## CYP2D6 Cytochrome P450-2D6 Basepairs Analyzed By GeneSavvy: All Exon Locations

The CYP2D6 enzyme is part of the P450 superfamily and is heavily involved in metabolizing about 25% of commonly available pharmaceutical drugs such as SSRIs, opioids, tamoxifen, cough syrup, beta-blockers, antiarrhythmics, adrenoceptor antagonists, and tricyclic antidepressants. CYP2D6 also metabolizes serotonin, eicosanoids, and neurosteroids such as pregnenolone, cholesterol, androsterone.

### Client's Genetic Variants and SNPs Identified Within the CYP2D6 Gene

No variants were found for this gene.

## CYP2E1 Cytochrome P450-2E1 Basepairs Analyzed By GeneSavvy: All Exon Locations

The CYP2E1 enzyme is part of the P450 superfamily and is heavily involved in drug metabolism and the synthesis of fatty acids, alcohol, acrylamide, acetaminophen, chlorzoxazone. CYP2E1 is also associated with the detoxification of environmental toxins and chemicals and is commonly associated with metabolizing alcohol and other substances within the brain. CYP2E1 alcohol metabolism in the brain can be induced by nicotine which would explain why smokers who also drink can have a higher rate of alcohol metabolism.

### Client's Genetic Variants and SNPs Identified Within the CYP2E1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CYP2E1	rs943975	HOM	Common 79.2%	Intron	Low
CYP2E1	rs2070676	HOM	Common 72.8%	Intron	Low
CYP2E1	rs2249694	HOM	Common 64.0%	Intron	Low
CYP2E1	rs2249695	HOM	Common 64.0%	Intron	Low
CYP2E1	rs2480257	HOM	Common 60.3%	UTR3	Low
CYP2E1	rs2480256	HOM	Common 63.8%	UTR3	Low

## CYP3A4 Cytochrome P450-3A4 Basepairs Analyzed By GeneSavvy: All Exon Locations

Plays a role in metabolizing about 50% of the drugs commonly prescribed. Can easily become overwhelmed trying to metabolize too many drugs.

### Client's Genetic Variants and SNPs Identified Within the CYP3A4 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CYP3A4	rs3735451	HET	Uncommon 30.8%	Intron	Low
CYP3A4	rs12721620	HET	Uncommon 8.0%	Intron	Low
CYP3A4	rs2242480	HET	Uncommon 31.6%	Intron	Low
CYP3A4	rs10267228	HET	Rare 6.2%	Intron	Low
CYP3A4	rs4646437	HET	Uncommon 29.9%	Intron	Low
CYP3A4	rs2687116	HOM	Common 78.8%	Intron	Low



## CYP3A5 Cytochrome P450-3A5 Basepairs Analyzed By GeneSavvy: All Exon Locations

Plays a role in metabolizing about 50% of the drugs commonly prescribed. Can easily become overwhelmed trying to metabolize too many drugs.

### Client's Genetic Variants and SNPs Identified Within the CYP3A5 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
CYP3A5	rs15524	HET	Uncommon 24.8%	UTR3	Low
CYP3A5	rs28383472	HET	Very Rare 1.2%	Silent	Low

## F3 Coagulation Factor 3, Tissue Factor Basepairs Analyzed By GeneSavvy: All Exon Locations

This gene encodes for the Coagulation Factor 3 (F3) enzyme which is also commonly referred to as Tissue Factor. F3 is a cell surface glycoprotein that enables cells to initiate the blood coagulation cascade through the "Extrinsic" coagulation cascade.

### Client's Genetic Variants and SNPs Identified Within the F3 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
F3	rs33354	HOM	Uncommon 32.2%	UTR3	Low
F3	.	HOM	Subtype: -5000	Intron	Low
F3	rs2391424	HOM	Uncommon 33.1%	Intron	Low

## GPX1 Glutathione Peroxidase 1 Basepairs Analyzed By GeneSavvy: All Exon Locations

The GPX1 gene encodes for the Glutathione Peroxidase 1 enzyme which is crucial for protection against oxidative stress. GPX1 catalyzes the reduction of hydroperoxides and hydrogen peroxide to water and oxygen with glutathione as a substrate which helps maintain redox balance. GPX1 is also essential for arachidonic acid metabolism in platelets.

### Client's Genetic Variants and SNPs Identified Within the GPX1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
GPX1	rs1050450	HET	Uncommon 30.2%	Missense	High
GPX1	rs1800668	HET	Uncommon 28.4%	UTR5	Low

## **GSTA1** Glutathione S-Transferase Alpha 1 *Basepairs Analyzed By GeneSavvy: All*

### *Exon Locations*

Glutathione S-Transferase Alpha 1 is a biologically active member of the GST super-family that's involved in phase 2 detoxification and the glutathione-ascorbic acid (Vitamin C) antioxidant cycle. GSTA1 along with most other GST enzymes, attach Glutathione to various drugs, chemicals, and other environmental toxins which makes them more water-soluble so they can be more easily removed from the body through sweat, urine, and feces.

### Client's Genetic Variants and SNPs Identified Within the **GSTA1** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>GSTA1</b>	rs4715326	HET	Common 63.3%	Intron	Low
<b>GSTA1</b>	rs1051775	HET	Uncommon 30.3%	Silent	Low

## **GSTM1** Glutathione S-Transferase Mu 1 *Basepairs Analyzed By GeneSavvy: All*

### *Exon Locations*

Glutathione S-Transferase Mu 1 is a biologically active member of the GST super-family that's involved in phase 2 detoxification and the metabolism of oxidative stress byproducts and carcinogens. GSTM1 along with most other GST enzymes, attach Glutathione to various drugs, chemicals, and other environmental toxins which makes them more water-soluble so they can be more easily removed from the body through sweat, urine, and feces.

### Client's Genetic Variants and SNPs Identified Within the **GSTM1** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>GSTM1</b>	rs1065411	HOM	Uncommon 38.8%	Missense	Medium

## **GSTO1** Glutathione S-Transferase Omega 1 *Basepairs Analyzed By GeneSavvy:*

### *All Exon Locations*

Glutathione S-Transferase Omega 1 is a biologically active member of the GST super-family that's involved in phase 2 detoxification but also differs from most GST enzymes by playing a pro-inflammatory role in response to endotoxins and bacterial infection. GSTO1 along with most other GST enzymes, attach Glutathione to various drugs, chemicals, and other environmental toxins which makes them more water-soluble so they can be more easily removed from the body through sweat, urine, and feces. GSTO1 is also associated with neutralizing oxidative stress.

### Client's Genetic Variants and SNPs Identified Within the **GSTO1** Gene

No variants were found for this gene.

## **GSTP1** Glutathione S-Transferase Pi 1 *Basepairs Analyzed By GeneSavvy: All Exon*

### *Locations*

Glutathione S-Transferase Pi 1 is a biologically active member of the GST super-family that's involved in phase 2 detoxification, estrogen metabolism, pro-inflammatory (IL-6) responses associated with increase Vitamin E consumption, and protection against neurodegeneration. GSTP1 along with most other GST enzymes, attach Glutathione to various drugs, chemicals, and other environmental toxins which makes them more water- soluble so they can be more easily removed from the body through sweat, urine, and feces.

### Client's Genetic Variants and SNPs Identified Within the **GSTP1** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>GSTP1</b>	rs1695	<b>HET</b>	Uncommon 36.4%	Missense	Medium
<b>GSTP1</b>	rs1079719	<b>HOM</b>	Uncommon 38.1%	Intron	Low
<b>GSTP1</b>	rs1871041	<b>HOM</b>	Common 88.5%	Intron	Low
<b>GSTP1</b>	rs2370143	<b>HOM</b>	Uncommon 31.9%	Intron	Low
<b>GSTP1</b>	rs762803	<b>HOM</b>	Uncommon 38.2%	Intron	Low
<b>GSTP1</b>	rs1871042	<b>HET</b>	Uncommon 30.0%	Intron	Low
<b>GSTP1</b>	rs4891	<b>HET</b>	Uncommon 38.5%	Silent	Low

## **HAMP** Hepcidin Antimicrobial Peptide *Basepairs Analyzed By GeneSavvy: All Exon*

### *Locations*

This gene encodes for the liver-produced hormone called Hepcidin. Hepcidin is essential for iron homeostasis and is the main regulator dietary of iron absorption, iron distribution across tissues, and iron recycling. Hepcidin acts by promoting endocytosis and degradation of ferroportin, leading to iron retention in iron-exporting cells and decreased flow of iron into plasma. Hepcidin is also known to have antimicrobial activity against bacteria and fungi.

### Client's Genetic Variants and SNPs Identified Within the **HAMP** Gene

No variants were found for this gene.

## **HFE** Homeostatic Iron Regulator *Basepairs Analyzed By GeneSavvy: All Exon*

### *Locations*

It is thought that this protein functions to regulate iron absorption by regulating the interaction of the transferrin receptor with transferrin. The iron storage disorder, hereditary hemochromatosis, is a recessive genetic disorder that results from defects in this gene.

### Client's Genetic Variants and SNPs Identified Within the **HFE** Gene

No variants were found for this gene.



## HJV Hemojuvelin *Basepairs Analyzed By GeneSavvy: All Exon Locations*

Hemojuvelin (HJV) is known to activate and modulate the expression of Hepcidin (HAMP) as well as act as a cellular receptor for Hepcidin. HJV is essential for iron homeostasis via its ability to regulate Hepcidin activity. HJV also acts as a bone morphogenetic protein (BMP) and modulates Hepcidin activity in response to BMP signaling.

### Client's Genetic Variants and SNPs Identified Within the HJV Gene

No variants were found for this gene.

## ICAM1 Intercellular Adhesion Molecule 1 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

Intercellular Adhesion Molecule 1 (ICAM1), also known as CD54 is part of a large family of transmembrane proteins called cell adhesion molecules (CAMs). CAMs are heavily involved in the binding of a cell to another cell or to the extracellular matrix and they have roles in cell proliferation, differentiation, motility, trafficking, apoptosis, and tissue architecture. ICAM1 binds to Integrins of CD11/CD18, can be induced by IL-1/TNF activity, and shows levels increase greatly upon cytokine stimulation.

### Client's Genetic Variants and SNPs Identified Within the ICAM1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
ICAM1	rs1801714	HET	Rare 2.2%	Missense	High
ICAM1	rs5498	HET	Uncommon 36.7%	Missense	Medium
ICAM1	rs5030352	HOM	Common 47.1%	Intron	Low

## IL17A Interleukin 17 Alpha *Basepairs Analyzed By GeneSavvy: All Exon Locations*

Interleukin 17 Alpha (IL-17A) is a potent pro-Inflammatory Cytokine produced by activated T-Cells that can stimulate IL-6/COX-2 expression and enhance NO production. Also known to regulate NF-KappaB and MAPK activity.

### Client's Genetic Variants and SNPs Identified Within the IL17A Gene

No variants were found for this gene.

## IL1A Interleukin 1 Alpha *Basepairs Analyzed By GeneSavvy: All Exon Locations*

Interleukin 1 Alpha (IL1A) cytokines are produced by monocytes and macrophages as proproteins, which are proteolytically processed and released in response to cell injury, and thus induces apoptosis. IL1A stimulates thymocyte proliferation by inducing IL2A release, B-cell maturation and proliferation, and fibroblast growth factor activity. IL1A proteins are involved in the inflammatory response, being identified as endogenous pyrogens, and are reported to stimulate the release of prostaglandin and collagenase from synovial cells.

### Client's Genetic Variants and SNPs Identified Within the IL1A Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
IL1A	rs3783550	HET	Common 73.5%	Intron	Low
IL1A	rs2071376	HET	Common 73.4%	Intron	Low
IL1A	rs2071374	HET	Uncommon 27.9%	Intron	Low
IL1A	rs2071373	HET	Common 73.5%	Intron	Low
IL1A	rs1609682	HET	Common 73.4%	Intron	Low

## IL1B Interleukin 1 Beta *Basepairs Analyzed By GeneSavvy: All Exon Locations*

Interleukin 1 Beta (IL1B) cytokines are produced by monocytes and macrophages as proproteins, which are proteolytically processed and released in response to cell injury, and thus induces apoptosis. This potent proinflammatory cytokine induces COX2 in the CNS which is known to contribute to inflammatory pain hypersensitivity. IL1B proteins are involved in the inflammatory response, being identified as the major endogenous pyrogen, induces prostaglandin synthesis, neutrophil influx and activation, T-cell activation and cytokine production, B-cell activation and antibody production, and fibroblast proliferation and collagen production.

### Client's Genetic Variants and SNPs Identified Within the IL1B Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
IL1B	rs1143629	HET	Common 65.3%	Intron	Low

## IL1RN Interleukin 1 Receptor Antagonist *Basepairs Analyzed By GeneSavvy: All Exon Locations*

Interleukin 1 Receptor Antagonist (IL1RN) binds functional IL1A/IL1B receptors to inhibit IL1A/IL1B activity and regulate IL1 related immune and inflammatory responses.

### Client's Genetic Variants and SNPs Identified Within the IL1RN Gene

No variants were found for this gene.



## IL6 Interleukin 6 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

Interleukin 6 (IL-6) is a potent cytokine with well-defined pro- and anti-inflammatory properties that can regulate the immune system as well as play roles in cognitive function. IL6 can activate cells in two ways: One way is anti-inflammatory to assist in tissue regeneration while the other is pro-inflammatory, capable of inducing fever in people with autoimmune diseases or infections. IL6 levels are increased in nearly all disease states and is required for the generation of T(H)17 cells.

### Client's Genetic Variants and SNPs Identified Within the IL6 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
IL6	rs2069849	HET	Rare 6.2%	Silent	Low

## LPL Lipoprotein Lipase *Basepairs Analyzed By GeneSavvy: 28,188*

Lipoprotein lipase is anchored to the vascular endothelium and removes lipids from the circulation by hydrolysing triglycerides present in VLDL into free fatty acids. LPL is also heavily associated to body fat, fat distribution, plasma lipids and insulin concentrations.

### Client's Genetic Variants and SNPs Identified Within the LPL Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
LPL	rs316	HET	Uncommon 15.7%	Silent	Low





## **MTHFR** Methylenetetrahydrofolate Reductase *Basepairs Analyzed By*

*GeneSavvy: 20,373*

Methylenetetrahydrofolate Reductase is a key enzyme in the folate metabolism pathway – directing folate from the diet either to DNA synthesis or homocysteine re-methylation.

### Client's Genetic Variants and SNPs Identified Within the **MTHFR** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>MTHFR</b>	rs1801133	HET	Common 46.1%	Missense	Severe
<b>MTHFR</b>	rs1801131	HET	Uncommon 29.7%	Missense	High
<b>MTHFR</b>	rs2274976	HET	Very Rare 1.8%	Missense	High
<b>MTHFR</b>	rs1537516	HET	Rare 3.3%	UTR3	Low
<b>MTHFR</b>	rs1537515	HET	Rare 3.3%	UTR3	Low
<b>MTHFR</b>	rs4846051	HOM	Common 98.4%	Silent	Low
<b>MTHFR</b>	rs2066462	HET	Rare 3.3%	Silent	Low
<b>MTHFR</b>	rs2066470	HET	Rare 3.1%	Silent	Low

## **MTR** 5-Methyltetrahydrofolate-Homocysteine Methyltransferase

*Basepairs Analyzed By GeneSavvy: 108,700*

Methionine Synthase encodes the enzyme that catalyses the re-methylation of homocysteine to methionine.

### Client's Genetic Variants and SNPs Identified Within the **MTR** Gene

No variants were found for this gene.

## **MTRR** 5-Methyltetrahydrofolate-Homocysteine Methyltransferase Reductase *Basepairs Analyzed By GeneSavvy: 49,938*

Methionine Synthase Reductase catalyses methylcobalamin, an essential cofactor of methionine synthase (MTR), which is essential for maintaining adequate intracellular pools of methionine and is also responsible for maintaining homocysteine concentrations at non-toxic levels.

### Client's Genetic Variants and SNPs Identified Within the **MTRR** Gene

No variants were found for this gene.



## NAT1 N-Acetyltransferase 1 Basepairs Analyzed By GeneSavvy: All Exon Locations

The NAT1 gene encodes 1 of 2 Arylamine N-Acetyltransferase enzymes responsible for attaching Acetyl-CoA to various xenobiotics, drugs, chemicals, and toxins which is required for detoxification and removal from the body. NAT1 also plays a role in folate catabolism and genotypes within NAT1 help identify possible slow, medium, and fast acetylator phenotypes.

### Client's Genetic Variants and SNPs Identified Within the NAT1 Gene

No variants were found for this gene.

## NAT2 N-Acetyltransferase 2 Basepairs Analyzed By GeneSavvy: All Exon Locations

The NAT2 gene encodes 1 of 2 Arylamine N-Acetyltransferase enzymes responsible for attaching Acetyl-CoA to various xenobiotics, drugs, chemicals, and toxins which is required for detoxification and removal from the body. NAT2 genotypes within NAT2 help identify possible slow, medium, and fast acetylator phenotypes and have an association with higher incidences of drug toxicity.

### Client's Genetic Variants and SNPs Identified Within the NAT2 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
NAT2	rs1799930	HET	Uncommon 27.3%	Missense	Severe
NAT2	rs1799931	HET	Rare 3.9%	Missense	High
NAT2	rs1208	HOM	Common 59.7%	Missense	High
NAT2	rs1041983	HOM	Uncommon 36.0%	Silent	Low

## NFE2L2 Nuclear Factor, Erythroid 2 Like 2 Basepairs Analyzed By GeneSavvy: All Exon Locations

The NFE2L2 gene encodes for the enzyme commonly called "Nrf2" and is responsible for regulating the Nrf2 signaling pathway which activates the body's natural antioxidant defense systems to reduce oxidative stress in the cells. Since Nrf2 signaling can regulate the expression of other genes in this phase 2 detoxification network (GSTs, NQO1, UGTs, etc...) it is an important factor for overall detoxification, oxidative stress, and inflammation homeostasis.

### Client's Genetic Variants and SNPs Identified Within the NFE2L2 Gene

No variants were found for this gene.



## NOS1 Nitric Oxide Synthase 1 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

The NOS1 gene encodes for the Nitric Oxide Synthase 1 enzyme which is part of the nitric oxide synthase family. NOS1 is essential for producing Nitric Oxide (NO) and L-Citrulline from L-Arginine and Oxygen. NOS1 is also known as Neuronal Nitric Oxide Synthase (nNOS) and displays activity in the brain and peripheral nervous system similar to neurotransmitters with associations with neurotoxicity and neurodegenerative disorders as well.

### Client's Genetic Variants and SNPs Identified Within the NOS1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
NOS1	rs2682826	HET	Uncommon 25.0%	UTR3	Low
NOS1	rs2293054	HOM	Common 75.0%	Silent	Low

## NOS2 Nitric Oxide Synthase 2 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

The NOS2 gene encodes for the Nitric Oxide Synthase 2 enzyme which is part of the nitric oxide synthase family. NOS2 is essential for producing Nitric Oxide (NO) and L-Citrulline from L-Arginine and Oxygen. NOS2 is also known as Inducible Nitric Oxide Synthase (iNOS) which is known to be expressed in the liver and is commonly associated with endotoxin, cytokine (IL-6/IL-8), and inflammatory activity.

### Client's Genetic Variants and SNPs Identified Within the NOS2 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
NOS2	rs542585205	HET	Very Rare 0.0%	Missense	High
NOS2	rs2255929	HET	Common 49.3%	Intron	Low
NOS2	rs1060826	HOM	Common 67.4%	Silent	Low
NOS2	rs2297511	HOM	Common 66.6%	Intron	Low
NOS2	rs2297512	HOM	Common 67.4%	Intron SpliceSite	Low
NOS2	rs1060822	HOM	Common 67.6%	Silent	Low
NOS2	rs9282801	HET	Uncommon 32.0%	Intron	Low
NOS2	rs2274894	HOM	Common 68.0%	Intron	Low
NOS2	rs2248814	HOM	Common 67.8%	Intron	Low

## NOS3 Nitric Oxide Synthase 3 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

The NOS3 gene encodes for the Nitric Oxide Synthase 3 enzyme which is part of the nitric oxide synthase family. NOS3 is essential for producing Nitric Oxide (NO) and L-Citrulline from L-Arginine and Oxygen. NOS3 is also known as Endothelial Nitric Oxide Synthase (eNOS) and has activity commonly associated with angiogenesis, vascular functions, cardio health, and blood coagulation. Also known to be associated with neurotransmission and antimicrobial activities.

### Client's Genetic Variants and SNPs Identified Within the NOS3 Gene

No variants were found for this gene.

## NQO1 NAD(P)H Quinone Dehydrogenase 1 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

The NQO1 gene encodes the NAD(P)H: quinone oxidoreductase 1 enzyme which uses NADH or NADPH to reduce quinones to hydroquinones. Because quinones are highly reactive and toxic intermediate metabolites of many processes, it is important to be able to convert the quinones to hydroquinones quickly. Along with defense against oxidative stress, NQO1 is also involved in metabolizing CoQ10 and Vitamin K for the body to utilize. NQO1 uses FAD (flavin adenine-dinucleotide) as a cofactor for these activities so maintaining optimal levels of FAD/Riboflavin (B2) would be beneficial for NQO1 functionality.

### Client's Genetic Variants and SNPs Identified Within the NQO1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
NQO1	rs689452	HOM	Common 86.8%	Intron	Low

## PON1 Paraoxonase 1 *Basepairs Analyzed By GeneSavvy: 26,215*

PON1 encodes the glycoprotein enzyme paraoxonase. PON1 protects LDL and HDL from oxidation possibly by hydrolysing phospholipid or cholesteryl ester hydroperoxides, thus protecting against atherogenesis. Low serum PON activity has been associated with increased risk for coronary artery disease.

### Client's Genetic Variants and SNPs Identified Within the PON1 Gene

No variants were found for this gene.

## SLC11A2 Solute Carrier Family 11 Member 2 Basepairs Analyzed By GeneSavvy:

### All Exon Locations

SLC11A2 is important in iron transport but also important in transporting other metals such as manganese, cobalt, cadmium, nickel, vanadium, and lead. SLC11A2 may also play an important role in hepatic iron accumulation, tissue iron distribution, and importing iron into the mitochondria.

### Client's Genetic Variants and SNPs Identified Within the SLC11A2 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
SLC11A2	rs445520	HET	Common 90.8%	Missense	Medium
SLC11A2	rs161044	HET	Common 90.8%	Intron	Low
SLC11A2	rs1048230	HET	Uncommon 14.5%	Silent	Low
SLC11A2	rs11169655	HET	Uncommon 14.4%	Intron	Low

## SLC40A1 Solute Carrier Family 40 Member 1 Basepairs Analyzed By GeneSavvy:

### All Exon Locations

SLC40A1 is important in iron transport but also important in transporting other metals such as manganese, cobalt, cadmium, nickel, vanadium, and lead. SLC40A1 may also play an important role in hepatic iron accumulation, tissue iron distribution, and importing iron into the mitochondria. SLC40A1 is also involved in the transfer of iron between maternal and fetal circulation.

### Client's Genetic Variants and SNPs Identified Within the SLC40A1 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
SLC40A1	rs2304704	HOM	Common 71.6%	Silent	Low
SLC40A1	rs1439816	HOM	Common 81.9%	Intron	Low

## SOD1 Superoxide Dismutase 1 Basepairs Analyzed By GeneSavvy: All Exon Locations

The SOD1 gene provides instructions for making an enzyme called Superoxide Dismutase 1, which is abundant in cells throughout the body. SOD1 binds copper and zinc to convert intracellular free superoxide radicals and reactive oxygen species into oxygen and hydrogen peroxide. Balance is key with Superoxide Dismutase activity since pro-oxidants such as superoxide can also play important roles in cellular signaling, immune response, and pathogen defense.

### Client's Genetic Variants and SNPs Identified Within the SOD1 Gene

No variants were found for this gene.



## **SOD2** Superoxide Dismutase 2 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

The SOD2 gene provides instructions for making an enzyme called Superoxide Dismutase 2, which is abundant in mitochondria throughout the body. SOD2 binds iron/manganese to convert mitochondrial free superoxide radicals and reactive oxygen species into oxygen and hydrogen peroxide. Balance is key with Superoxide Dismutase activity since pro-oxidants such as superoxide can also play important roles in cellular signaling, immune response, and pathogen defense.

### Client's Genetic Variants and SNPs Identified Within the **SOD2** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>SOD2</b>	rs4880	HET	Common 47.3%	Missense	High
<b>SOD2</b>	rs2758332	HET	Common 49.4%	Intron	Low
<b>SOD2</b>	rs2855116	HET	Common 40.2%	Intron	Low
<b>SOD2</b>	rs2070994	HET	Common 40.2%	DownStream	Low

## **SOD3** Superoxide Dismutase 3 *Basepairs Analyzed By GeneSavvy: All Exon Locations*

The SOD3 gene provides instructions for making an enzyme called Superoxide Dismutase 3, which is abundant in extracellular spaces throughout the body. SOD3 binds copper, zinc, and heparin to convert extracellular free superoxide radicals and reactive oxygen species into oxygen and hydrogen peroxide. Balance is key with Superoxide Dismutase activity since pro-oxidants such as superoxide can also play important roles in cellular signaling, immune response, and pathogen defense.

### Client's Genetic Variants and SNPs Identified Within the **SOD3** Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
<b>SOD3</b>	rs2536512	HET	Common 50.0%	Missense	Medium

## TF Transferrin Basepairs Analyzed By GeneSavvy: All Exon Locations

Transferrins are iron binding transport proteins which can bind two Fe(3+) ions in association with the binding of an anion, usually bicarbonate. It is responsible for the transport of iron from sites of absorption and heme degradation to those of storage and utilization. Serum transferrin may also have a further role in stimulating cell proliferation.

### Client's Genetic Variants and SNPs Identified Within the TF Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
TF	rs1799899	HET	Rare 4.5%	Missense	Severe
TF	rs1049296	HET	Uncommon 13.5%	Missense	High
TF	rs2692696	HOM	Common 99.6%	Missense	Medium
TF	rs1130459	HET	Common 58.2%	UTR5	Low
TF	rs3811655	HET	Uncommon 23.2%	Intron	Low
TF	rs1799852	HET	Uncommon 10.5%	Silent	Low

## TFRC Transferrin Receptor Basepairs Analyzed By GeneSavvy: All Exon Locations

Transferrin receptor involved in the cellular uptake of iron.

### Client's Genetic Variants and SNPs Identified Within the TFRC Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
TFRC	rs3817672	HOM	Common 42.6%	Missense	Medium
TFRC	rs2239641	HOM	Common 48.3%	Intron	Low
TFRC	rs480760	HOM	Common 89.5%	Intron	Low

## TNF Tumor Necrosis Factor Basepairs Analyzed By GeneSavvy: All Exon Locations

Tumor Necrosis Factor is a multifunctional proinflammatory cytokine produced by our immune system during acute and chronic inflammatory conditions and is involved in the regulation of a wide spectrum of biological processes including cell proliferation, differentiation, apoptosis, lipid metabolism, and coagulation. Alzheimer's and Parkinson's diseases are both linked to higher TNF production in the brain.

### Client's Genetic Variants and SNPs Identified Within the TNF Gene

No variants were found for this gene.

## UGT1A1 UDP Glucuronosyltransferase Family 1 Member A1 Basepairs

Analyzed By GeneSavvy: All Exon Locations

The UGT1A1 gene encodes for a UDP-glucuronosyltransferase (abbreviated UGT) enzyme which facilitates glucuronidation reactions. Glucuronidation attaches glucuronic acid to things like bilirubin, retinoids, estrogen, testosterone, BPA, cortisol, fatty acids, aromatic hydrocarbons, drugs, chemicals, and various other environmental toxins which makes them more water-soluble so they can be more easily removed from the body through sweat, urine, and feces. UGT1A1 glucuronidation reactions are generally used to metabolize, inactivate, and eliminate bilirubin, estrogen, carcinogens, drugs, chemicals, and various other environmental toxins.

### Client's Genetic Variants and SNPs Identified Within the UGT1A1 Gene

No variants were found for this gene.

## UGT1A6 UDP Glucuronosyltransferase Family 1 Member A6 Basepairs

Analyzed By GeneSavvy: All Exon Locations

The UGT1A6 gene encodes for a UDP-glucuronosyltransferase (abbreviated UGT) enzyme which facilitates glucuronidation reactions. Glucuronidation attaches glucuronic acid to things like bilirubin, retinoids, estrogen, testosterone, BPA, cortisol, fatty acids, aromatic hydrocarbons, drugs, chemicals, and various other environmental toxins which makes them more water-soluble so they can be more easily removed from the body through sweat, urine, and feces. UGT1A6 glucuronidation reactions are generally used to metabolize, inactivate, and eliminate bilirubin, hormones, drugs (aspirin, acetaminophen), chemicals, and various other environmental toxins.

### Client's Genetic Variants and SNPs Identified Within the UGT1A6 Gene

No variants were found for this gene.

## UGT1A8 UDP Glucuronosyltransferase Family 1 Member A8 Basepairs

Analyzed By GeneSavvy: All Exon Locations

The UGT1A8 gene encodes for a UDP-glucuronosyltransferase (abbreviated UGT) enzyme which facilitates glucuronidation reactions. Glucuronidation attaches glucuronic acid to things like bilirubin, retinoids, estrogen, testosterone, BPA, cortisol, fatty acids, aromatic hydrocarbons, drugs, chemicals, and various other environmental toxins which makes them more water-soluble so they can be more easily removed from the body through sweat, urine, and feces. UGT1A8 glucuronidation reactions are generally used to metabolize, inactivate, and eliminate bilirubin, coumarins, phenols, anthraquinones, flavones, opioids, mycophenolic acid, drugs, chemicals, and various other environmental toxins

### Client's Genetic Variants and SNPs Identified Within the UGT1A8 Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
UGT1A8	rs1042597	HOM	Uncommon 25.4%	Missense	Medium



## UGT1A9 UDP Glucuronosyltransferase Family 1 Member A9 Basepairs

Analyzed By GeneSavvy: All Exon Locations

The UGT1A9 gene encodes for a UDP-glucuronosyltransferase (abbreviated UGT) enzyme which facilitates glucuronidation reactions. Glucuronidation attaches glucuronic acid to things like bilirubin, retinoids, estrogen, testosterone, BPA, cortisol, fatty acids, aromatic hydrocarbons, drugs, chemicals, and various other environmental toxins which makes them more water-soluble so they can be more easily removed from the body through sweat, urine, and feces. UGT1A9 glucuronidation reactions are generally used to metabolize, inactivate, and eliminate bilirubin, coumarins, phenols, anthraquinones, flavones, opioids, mycophenolic acid, drugs, chemicals, and various other environmental toxins

### Client's Genetic Variants and SNPs Identified Within the UGT1A9 Gene

No variants were found for this gene.

## VDR Vitamin D Receptor Basepairs Analyzed By GeneSavvy: All Exon Locations

This gene encodes for the Vitamin D Receptor enzyme which is the nuclear receptor for Calcitriol, the active form of Vitamin D3 which mediates the action of Vitamin D3 on cells. This Vitamin D3 Receptor activates and mediates Vitamin D3 responsive cellular activities and is essential for bone and calcium homeostasis as well as general mineral metabolism.

### Client's Genetic Variants and SNPs Identified Within the VDR Gene

GENE	VARIANT	GENOTYPE RESULT	FREQUENCY IN POPULATION	EFFECT	IMPACT ON GENE
VDR	rs2228570	HOM	Common 66.4%	StartLoss	Severe
VDR	rs731236	HET	Uncommon 33.9%	Silent	Low
VDR	rs7975232	HET	Common 55.3%	Intron	Low
VDR	rs1544410	HET	Uncommon 35.0%	Intron	Low



# Technical Information



*\*The performance, security, and data ownership information listed below does not apply to genetic information provided by non-GeneSavvy genetic testing companies\**

## GeneSavvy's Testing Performance

This test was designed to sequence the exons and canonical splice sites (+2NT) of the whole exome including extra coverage on genes associated with medical disorders and treatment options. GeneSavvy has developed highly advanced protocols to target and extract variants in exonic regions, intronic regions of interest, splice sites, and untranslated gene regions of interest.

**Mean Depth of Coverage: 40x**  
**Specificity: 99%**  
**Sensitivity: 96**

## Databases and Tools Used in OUR BIOPIPELINE v1.0

Our reports are built using our proprietary GeneSavvy bioinformatics pipeline to provide the most valuable and actionable results we can find. Below are some of the databases and tools we utilize to build actionable results for you!

<b>GENETIC DEFINITIONS</b> Reference Genome: HG38/GRCh38 RefSeq UCSC ENSEMBL GENCODE dbSNP dbSNV	<b>POPULATION FREQUENCY</b> 1000 Genome Project ExAC NHLBI-ESP gnomAD Complete Genomics	<b>DISEASE ASSOCIATIONS</b> ClinVar Cosmic ICGC NCI OMIM GWAS
<b>FRACTIONAL VARIANT PREDICTIONS</b> SIFT PolyPhen2 LRT MutationTaster MutationAssessor FATHMM InterVar CADD GERP++	<b>PIPELINE TOOLS</b> TriarkK GATK Galaxy snEff VEP Picard DRAGEN BWA	<b>ONTOLOGIES</b> Human Phenotype Ontology Gene Ontology MGI Jensen

## DATA SECURITY AND OWNERSHIP

GeneSavvy uses industry standards to keep your personal data safe. Your data will never be sold or distributed to any other party you do not choose to share it with; it is 100% owned by you! If you have further questions, please email us at [info@genesavvy.com](mailto:info@genesavvy.com)



# Thank you!

**Thank you for choosing GeneSavvy as your partner in functional genetics.**

Please let us know if we can do anything else to help you move toward more vibrant life! Remember that the genetic data in this report can be very complex, so give your functional genetic practitioner time to explore these findings in depth. Also, don't forget that data obtained from related laboratory testing and/or imaging can add to your practitioner's ability to best interpret and understand your genetic data. If you tested through GeneSavvy, you can also utilize the complete test results (full exome, partial introns, full mitochondrial DNA) contained in the accompanying GeneSavvy Variant Exploration Report spreadsheet to explore your other SNPs and data.

We're always looking for feedback to help us improve. Email us at [info@GeneSavvy.com](mailto:info@GeneSavvy.com) and let us know how you liked this report!

## Test, don't guess.

Generated by GeneSavvy Report Pipeline v1.0

PHONE	425-686-0000
EMAIL	<a href="mailto:info@GeneSavvy.com">info@GeneSavvy.com</a>
WEBSITE	<a href="http://www.GeneSavvy.com">www.GeneSavvy.com</a>

This report is for research, informational, and educational use only. It is not intended for the diagnosis, prevention, or treatment of disease. GeneSavvy does not provide medical advice.

[illegible]