Bruce Wayne - Heart Health

June 21, 2018

PowerXomeGS + MitoPowerGS

GeneSavvy

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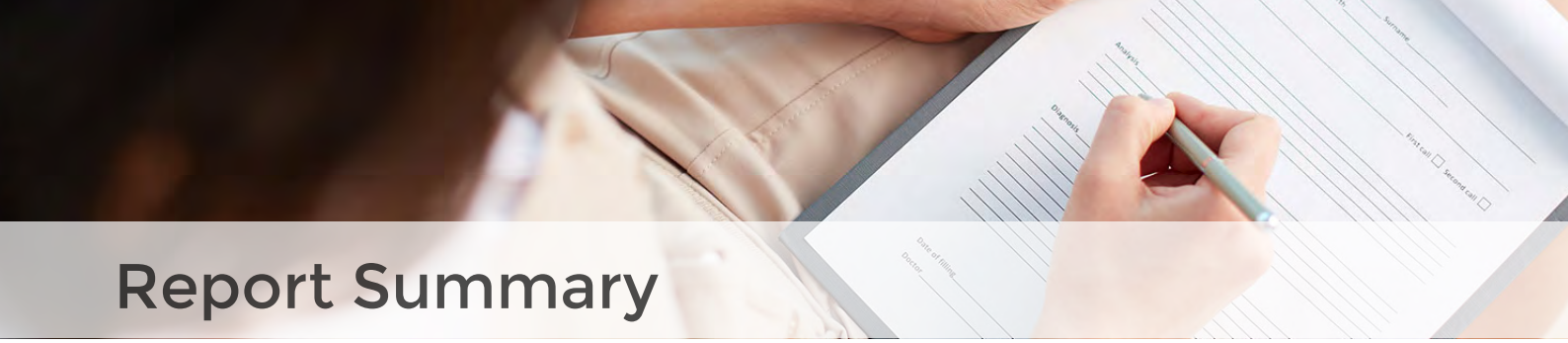
Patient Information

Full Name:	Bruce Wayne - Heart Health
Gender:	Male
Address:	1007 Mountain Drive, Gotham City, Metropolis 12345 United States
Phone:	123-456-7891
Email:	info@genesavvy.com
Date Of Birth:	1963-02-19
Marital Status:	Single
Accesession#:	BATMAN
Collected:	2018-04-01
Recieved:	2018-06-06
Reported:	2018-06-06
Tech:	ROBIN
Doctor:	Dr. Hugo Strange



Report Summary

After reviewing current concerns it seems that one of the biggest concerns is heart health so we started looking first for correlative patterns between the genetic data and heart health. Some **interesting patterns here popped up for us between heart health and pancreatic function. Variants in genes (CFTR, PRSS1, PRSS3, NPHP3) all contribute to possible pancreatic disruption.**On top of several potentially disruptive genetic variants for pancreatic function, there were also **several hits in Autophagy (ATG3, ATG9B, LAMP2, ULK2, ULK1), with LAMP2 having specific mention of stress induced Autophagy and the ULK genes having specific mention of Autophagy induced by starvation.**All of these factors could make a highly susceptible environment for **something like Takotsubo Myopathy (Broken Heart Disease) or a stress induced pancreatitis that is contributing to a cardiomyopathy.**

An LMNA variant also popped up with a **direct correlation to dilated cardiomyopathy** that I thought might be interesting to explore, as well as relations between **ATPase activity and cardiac issues.**

There were also genetic variants with possible disruption in **lipid metabolism, transport, and utilization (NPC's, APO's, LPL),**as well as **Glycogen/Glycogen Storage/G-6-S (GPI, PYGL, SLC's, LAMP2).**

Some potential **immune system/thyroid concern was also found with disruption in genes (HLA-DRB1, TPO, SLC25A16, TSHR)**

Mitochondrial Variantswith high potential for mitochondrial disruption were found with the following genes seeming to be **the most affected (MT-CYB, MT-ND5).**Mitochondrial genes **(MT-ATP6, MT-CO1, MT-ND1, MT-ND4) also carried some potentially disruptive variants.**

https://www.ncbi.nlm.nih.gov/pubmed/26634508 LMNA gene to dilated cardiomyopathy

https://academic.oup.com/cardiovascres/article/53/4/782/412514 NA/CA exchanger and cardiac hypertrophy (ATPase)

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4290869/ Stress, Autophagy, Pancreatitis

Potential Next Steps

Testing pancreatic functionto see if it has any contribution to heart concerns, also **checking for possible heavy environmental stressors contributing to concerns.**

Testing other potential disruptionsfound such as **lipid metabolism, transport and utilization** as well as **glycogen, glucose, g6p utilization and function**and **immune/thyroid markers**to see how they might contribute to current concerns would be good as well. **Removing unnecessary environmental load in these areas would also be good.**

Exploring treatments to support disrupted and possibly inefficient genes for better longterm health.Blood viscosity testingmight also be of interest. **Exploring mitochondrial gene variants**and possibly discussing variants found with **mitochondrial disease expert to get treatment options for potentially disrupted mitochondrial genes.**

Using this Report

This report was designed to visualize genetics in a polymorphic, gene network arrangement. Here at GeneSavvy we strongly believe that genetic susceptibilities are created by compounded genetic and environmental influences. The goal is to find genetic and environmental patterns that can show us possible biological processes that are more susceptible to environmental hits. If we find these susceptible processes we can adjust our environment to reduce toxic effects and increase biological efficiency.

Gene Networks: Our GeneSavvy gene networks are built using a Boolean model to find genes related to the functional health terms used in your report. After collecting the relational genes, we use a practical scoring function-based algorithm to calculate relevance and extract the top relevant genes for your report.

The Colors: We use colors to help you scan quickly for patterns within this report. Gene symbols in GREY mean there were no exome variants found in that gene. Gene symbols in GREEN mean there were variants found with only LOW predicted impact. Gene symbols in YELLOW mean there were variants found with MODERATE predicted impact. Gene symbols in RED mean there were variants found with HIGH predicted impact. Genes listed in RED should be the first genes to research as potentially causative to the health concern.

Low Predicted Impact: Low predicted impact variants are usually synonymous variants that don't cause any amino acid changes or variants in areas that don't usually lead to impact on gene efficiency.

Moderate Predicted Impact: Moderate predicted impact variants are usually non-synonymous variants that do change the amino acid. This category of impact also contains insertions or deletions in multiples of 3 that don't cause a disruptive frameshift.

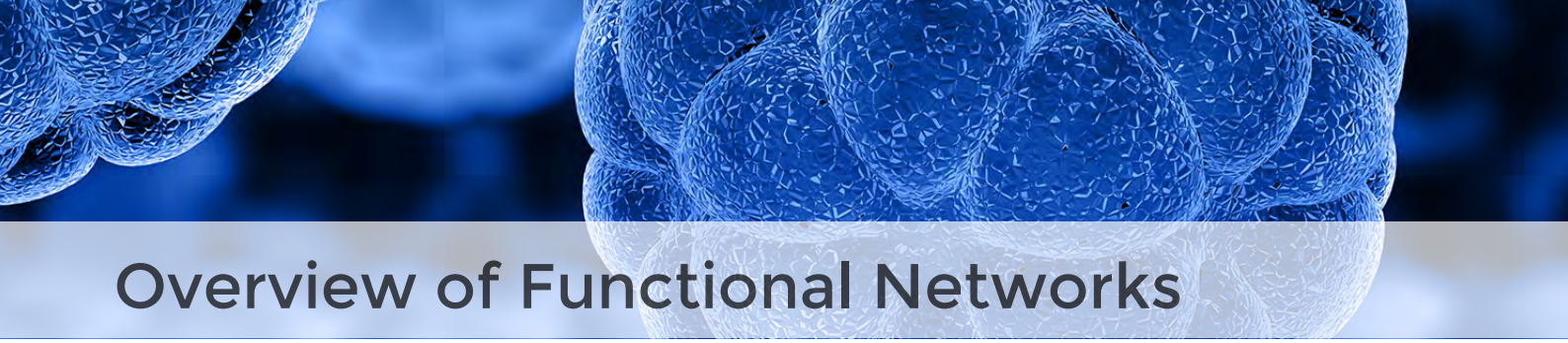
High Predicted Impact: High predicted impact variants are usually start or stop loss variants as well as disruptive frameshifts, major deletions or insertions or variants in splice site donors and splice site acceptors. These variants have high potential to impact gene functionality and efficiency.

Rarity: Rare genetic variants are usually given higher predicted impact compared to common variants. If the variant is found in 99% of our population then it has less chance to be a variant that directly leads to a health condition.

- **Very Rare:** Less than .01% of Population
- **Rare:** Less than 1% of Population
- **Uncommon:** Less than 5% of Population
- **Common:** Greater than 5% of Population
- **Unknown:** No Population Frequency Data

Significant Variants: Significant variants have high predicted impact. These variants should be first to research.

Common SNPs: Technically SNPs (Single Nucleotide Polymorphisms) are only classified as SNPs as they become commonly found in the population. These tend to have less impact on specific health conditions but they can still be key to finding patterns.



Overview of Functional Networks

The pathway and gene network overview below gives a quick view of the systems reviewed in this report. In general, networks with likely susceptibility should be addressed with priority for optimal health.

Antioxidant Defense	SUSCEPTABLE
ATPase	SUSCEPTABLE
Cholesterol	SUSCEPTABLE
Creatine Kinase	LIKELY LESS STABLE
Fat Soluble Vitamin Utilization	SUSCEPTABLE
G6P	SUSCEPTABLE
Glucose	LIKELY LESS STABLE
Glycogen	SUSCEPTABLE
Graves' disease	SUSCEPTABLE

Lipid Metabolism	SUSCEPTABLE
mTOR	LIKELY LESS STABLE
Pancreatitis	SUSCEPTABLE
T-lymphocytes T Cell	LIKELY LESS STABLE
Trypsinogen	SUSCEPTABLE
Wilson's Disease	SUSCEPTABLE

Potentially Significant Variants

This significant variant overview will show you the variants found with the highest predicted impact. In general, these variants will have high potential of affecting the output product of the gene its contained in. This data can be used to potentially super fine-tune treatment protocols by allowing you to increase or decrease enzyme activity to balance the impact of these variants.

Potentially Significant Variants Were Found in The Following Genes

Gene	Variant Location	RSID	Depth	OMIM ID	Effect	Result	Severity	Frequency
PON1	14101493	rs5789820 rs869278364	26	604989	Frameshift	HOM	High	Common (100%)
ATP6V0A2	124229429	rs7135542	18	611716	SpliceSite, Silent	HET	High	Common (93.8%)
ATP7B	52511499	N/A	19	606882	Missense	HET	High	Uncertain
DNAH8	38854646	rs142328376	19	603337	Missense	HET	High	Very Rare (0.65%)
NPC1	21119777	rs1140458	57	607623	SpliceSite, Silent	HET	High	Common (62.5%)
NPC2	74947404	rs140130028	50	601015	Intron, SpliceDonor	HET	High	Very Rare (0.37%)
DYSF	71738977	rs34997054	147	603009	Missense	HET	High	Very Rare (0.72%)
LMNA	156107534	rs4641	57	150330	SpliceSite, Silent	HOM	High	Common (26.34%)
GPI	34868642	rs1801015	31	172400	SpliceSite, Silent	HET	High	Common (20.7%)
PYGL	51378590	rs11356035	17	613741	Intron, SpliceAcceptor	HET	High	Common (43.94%)
SLC37A4	118898435	rs56966114 rs547203028	49	602671	Intron, SpliceAcceptor, SpliceDonor	HOM	High	Common (100%)
LAMP2	119576455	rs73219144	38	309060	SpliceSite, Silent	HET	High	Uncommon (2.71%)
HLA-DRB1	32551955	rs778205073	2	142857	Frameshift	HOM	High	Uncommon (14.29%)

Gene	Variant Location	RSID	Depth	OMIM ID	Effect	Result	Severity	Frequency
HLA-DRB1	32551960	rs796436531	2	142857	Frameshift	HOM	High	Uncommon (11.88%)
SLC25A16	70243284	rs778512677	24	139080	Missense	HET	High	Uncertain
RICTOR	39074478	N/A	16	609022	Frameshift, StartLoss	HET	High	Uncertain
CFTR	117149147	rs1800076	39	602421	Missense	HET	High	Uncommon (3.08%)
NPHP3	132403418	rs34391943	27	608002	Missense	HET	High	Rare (1.3%)
PRSS1	142457375	rs747228052	40	276000	Missense, SpliceSite	HET	High	Uncertain
BPI	36939052	rs5743507	28	109195	SpliceSite, Silent	HET	High	Uncommon (9.4%)
PRSS3	33797928	rs779080843	77	613578	Frameshift	HET	High	Uncertain
PRSS3	33794797	rs773515866	41	613578	Frameshift	HET	High	Very Rare (0.16%)
PRSS3	33795613	rs756027884	51	613578	Intron, SpliceDonor	HET	High	Uncommon (2.03%)
PRSS3	33797930	rs797012348	80	613578	Frameshift	HET	High	Uncertain
F2	48549791	rs2305998	23	610465	SpliceSite, Silent	HET	High	Common (25.59%)

Mitochondrial Report

Mitochondrial Report

Gene	Result	AA Change	rsID	Clinvar	OMIM	MitoMap Disease	Conser-vation	Depth	Ref Allele	Alt Allele	Population Frequency	Disease Score	Hetero Fraction
MT-ATP6	8620T	P32S	NA	NA	NA	NA	-1.28141	440	C	T	NA	High	Homoplasmy (1.40%)
MT-ATP6	8728A	Stop-gain	NA	NA	NA	NA	0.824732	540	T	A	NA	NA	Homoplasmy (0.90%)
MT-ATP6	8641T	N39Y	NA	NA	NA	NA	1.00024	486	A	T	NA	High	Homoplasmy (1.00%)
MT-ATP6	9130T	syn	NA	NA	NA	NA	-0.0528268	698	C	T	NA	NA	Homoplasmy (0.70%)
MT-ATP6	8865A	syn	NA	NA	NA	NA	-0.316094	407	G	A	NA	NA	Homoplasmy (2.20%)
MT-ATP8	8435T	T24S	NA	NA	NA	NA	0.0349291	516	A	T	NA	Low	Homoplasmy (2.10%)
MT-CO1	6459A	Stop-gain	NA	NA	NA	NA	0.889087	681	T	A	NA	NA	Homoplasmy (0.90%)
MT-CO1	6708A	Stop-gain	NA	NA	NA	MM & Rhabdomyolysis	0.889087	409	G	A	NA	NA	Homoplasmy (1.20%)
MT-CO1	6493G	L197R	NA	NA	NA	NA	0.889087	613	T	G	NA	High	Homoplasmy (1.10%)
MT-CO1	7152T	M417L	NA	NA	NA	NA	0.988543	380	A	T	NA	Low	Homoplasmy (1.30%)
MT-CO1	6177C	M92L	NA	NA	NA	NA	0.0934331	428	A	C	NA	Medium	Homoplasmy (1.40%)
MT-CO1	6059G	H52Q	NA	NA	NA	NA	-7.80469	624	C	G	NA	Low	Homoplasmy (1.40%)
MT-CO1	6125G	syn	NA	NA	NA	NA	-0.287157	420	A	G	NA	NA	Homoplasmy (1.20%)
MT-CO1	6989T	syn	NA	NA	NA	NA	-1.29896	510	A	T	NA	NA	Homoplasmy (1.20%)
MT-CO2	7615C	Q10H	NA	NA	NA	NA	0.130654	397	A	C	NA	High	Homoplasmy (1.30%)
MT-CO2	7878G	Stop-gain	NA	NA	NA	NA	1.088	594	A	G	NA	NA	Homoplasmy (1.20%)
MT-CO2	7649T	T22S	NA	NA	NA	NA	-5.78749	541	A	T	NA	Low	Homoplasmy (1.30%)
MT-CO2	7657A	H24Q	NA	NA	NA	NA	-5.35233	555	T	A	NA	High	Homoplasmy (1.10%)
MT-CO3	9867T	R221C	NA	NA	NA	NA	-0.0270551	549	C	T	0.04%	Low	Homoplasmy (1.30%)
MT-CO3	9893T	syn	NA	NA	NA	NA	-4.07143	554	C	T	NA	NA	Homoplasmy (1.10%)
MT-CO3	9583T	P126L	NA	NA	NA	NA	0.764236	465	C	T	NA	High	Homoplasmy (1.90%)
MT-CO3	9570T	T122S	NA	NA	NA	NA	-2.22509	426	A	T	NA	Low	Homoplasmy (1.60%)

Gene	Result	AA Change	rsID	Clinvar	OMIM	MitoMap Disease	Conser-vation	Depth	Ref Allele	Alt Allele	Population Frequency	Disease Score	Hetero Fraction
MT-CO3	9254G	syn	NA	NA	NA	NA	-0.818346	299	A	G	0.74%	NA	Homoplasmy (97.00%)
MT-CYB	15590A	Stop-gain	NA	NA	NA	NA	-0.0987244	702	C	A	NA	NA	Homoplasmy (0.70%)
MT-CYB	15041T	G99W	NA	NA	NA	NA	0.700866	547	G	T	NA	High	Homoplasmy (1.10%)
MT-CYB	14882T	T46S	NA	NA	NA	NA	-0.0876693	520	A	T	NA	Low	Homoplasmy (1.70%)
MT-CYB	15551T	Stop-gain	NA	NA	NA	NA	0.807039	658	A	T	NA	NA	Homoplasmy (0.90%)
MT-CYB	14806T	syn	NA	NA	NA	NA	-1.66474	465	C	T	NA	NA	Homoplasmy (1.30%)
MT-CYB	15755A	Stop-gain	NA	NA	NA	NA	0.731559	527	T	A	NA	NA	Homoplasmy (0.90%)
MT-CYB	15206T	P154S	NA	NA	NA	NA	0.700866	482	C	T	NA	High	Homoplasmy (1.50%)
MT-CYB	15356A	Stop-gain	NA	NA	NA	NA	0.700866	542	G	A	NA	NA	Homoplasmy (0.90%)
MT-CYB	15037G	H97Q	NA	NA	NA	NA	-3.43894	561	C	G	NA	High	Homoplasmy (1.80%)
MT-CYB	14878G	syn	NA	NA	NA	NA	-2.15757	507	A	G	NA	NA	Homoplasmy (1.20%)
MT-CYB	14950G	H68Q	NA	NA	NA	NA	-1.86187	614	C	G	NA	High	Homoplasmy (1.00%)
MT-CYB	15380T	T212S	NA	NA	NA	NA	-4.62754	583	A	T	NA	Low	Homoplasmy (1.20%)
MT-DLOOP	506T	NA	NA	NA	NA	NA	-0.807252	438	C	T	NA	NA	Homoplasmy (1.60%)
MT-DLOOP	545C	NA	NA	NA	NA	NA	0.051874	427	G	C	NA	NA	Homoplasmy (1.60%)
MT-DLOOP	41T	NA	NA	NA	NA	NA	-1.5342	82	C	T	NA	NA	Homoplasmy (9.80%)
MT-DLOOP	16077T	NA	NA	NA	NA	NA	-3.19342	487	A	T	NA	NA	Homoplasmy (1.00%)
MT-DLOOP	114T	NA	NA	NA	NA	BD-associated	-1.00551	207	C	T	0.25%	NA	Homoplasmy (98.10%)
MT-DLOOP	499C	NA	NA	NA	NA	NA	-3.18637	394	G	C	NA	NA	Homoplasmy (1.30%)
MT-DLOOP	16390A	NA	rs41378955	NA	NA	NA	-2.2728	384	G	A	9.88%	NA	Homoplasmy (1.30%)
MT-DLOOP	310C	NA	NA	NA	NA	NA	0.31622	216	T	TC C	26.63%	NA	Multi-Het TC (9.70%) C (77.90%)
MT-DLOOP	16172C	NA	rs2853817	NA	NA	NA	-1.45737	444	T	C	7.45%	NA	Homoplasmy (97.30%)
MT-DLOOP	16304C	NA	NA	NA	NA	NA	-0.174205	517	T	C	6.26%	NA	Homoplasmy (97.30%)
MT-ND1	3692A	L129Q	rs193303027	NA	NA	NA	0.873961	571	T	A	NA	High	Homoplasmy (2.10%)
MT-ND1	3500G	Stop-gain	NA	NA	NA	NA	0.753441	551	C	G	NA	NA	Homoplasmy (1.50%)
MT-ND1	3533T	T76I	NA	NA	NA	NA	-0.572276	559	C	T	NA	Low	Homoplasmy (1.30%)
MT-ND1	3663T	syn	NA	NA	NA	NA	-0.331236	549	A	T	NA	NA	Homoplasmy (1.10%)
MT-ND1	3511T	T69S	NA	NA	NA	NA	-0.813315	540	A	T	0.04%	Low	Homoplasmy (1.50%)
MT-ND2	5376T	T303S	NA	NA	NA	NA	0.0795512	617	A	T	NA	Medium	Homoplasmy (0.80%)

Gene	Result	AA Change	rsID	Clinvar	OMIM	MitoMap Disease	Conser-vation	Depth	Ref Allele	Alt Allele	Population Frequency	Disease Score	Hetero Fraction
MT-ND2	4847T	syn	NA	NA	NA	NA	-8.17515	526	C	T	NA	NA	Homoplasmy (1.00%)
MT-ND2	5325T	T286S	NA	NA	NA	NA	-0.745543	598	A	T	NA	Low	Homoplasmy (1.30%)
MT-ND4	10835T	N26Y	rs386829113	NA	NA	NA	0.0791811	866	A	T	NA	High	Homoplasmy (0.60%)
MT-ND4	11208G	L150R	NA	NA	NA	NA	0.886236	631	T	G	NA	High	Homoplasmy (1.10%)
MT-ND4	11162A	Stop-gain	NA	NA	NA	NA	-0.52611	628	C	A	NA	NA	Homoplasmy (1.10%)
MT-ND4	10938T	P60L	NA	NA	NA	NA	-0.0217008	622	C	T	NA	Medium	Homoplasmy (1.10%)
MT-ND4	11921A	Stop-gain	NA	NA	NA	NA	0.858575	636	T	A	NA	NA	Homoplasmy (0.80%)
MT-ND4	10891G	syn	NA	NA	NA	NA	-0.0217008	812	A	G	NA	NA	Homoplasmy (0.90%)
MT-ND4	10991T	M78L	NA	NA	NA	NA	-0.425228	587	A	T	NA	Low	Homoplasmy (1.00%)
MT-ND4	11711T	A318S	NA	NA	NA	NA	-0.0703386	567	G	T	NA	High	Homoplasmy (0.90%)
MT-ND4	12085T	syn	NA	NA	NA	NA	-4.50848	551	C	T	NA	NA	Homoplasmy (1.30%)
MT-ND4	11029T	syn	NA	NA	NA	NA	-0.425228	661	A	T	NA	NA	Homoplasmy (0.80%)
MT-ND4	11014T	syn	NA	NA	NA	NA	-4.15786	654	C	T	NA	NA	Homoplasmy (0.90%)
MT-ND4	10936T	syn	NA	NA	NA	NA	-1.23228	639	C	T	NA	NA	Homoplasmy (1.10%)
MT-ND4L	10641C	I58L	NA	NA	NA	NA	-0.425228	691	A	C	NA	Low	Homoplasmy (0.90%)
MT-ND4L	10638G	N57D	NA	NA	NA	NA	-1.03052	684	A	G	NA	Medium	Homoplasmy (0.90%)
MT-ND5	13120T	R262C	NA	NA	NA	NA	0.602984	414	C	T	NA	High	Homoplasmy (1.20%)
MT-ND5	13783T	P483S	rs368889908	NA	NA	NA	-0.170291	557	C	T	NA	High	Homoplasmy (0.90%)
MT-ND5	13201A	A289T	NA	NA	NA	NA	0.675787	520	G	A	0.12%	High	Homoplasmy (1.20%)
MT-ND5	13156T	Stop-gain	NA	NA	NA	NA	-0.125047	469	C	T	NA	NA	Homoplasmy (1.10%)
MT-ND5	14143T	T603S	NA	NA	NA	NA	-1.37388	611	A	T	NA	Low	Homoplasmy (1.10%)
MT-ND5	12898A	Stop-gain	NA	NA	NA	NA	0.748591	509	T	A	NA	NA	Homoplasmy (1.00%)
MT-ND5	12652A	Stop-gain	rs386829161	NA	NA	NA	0.748591	589	T	A	NA	NA	Homoplasmy (1.40%)
MT-ND5	13004G	Stop-gain	NA	NA	NA	NA	0.821394	406	A	G	NA	NA	Homoplasmy (1.20%)
MT-ND5	12802T	S156C	NA	NA	NA	NA	-4.20202	558	A	T	NA	High	Homoplasmy (1.10%)
MT-ND5	13186T	T284S	NA	NA	NA	NA	0.821394	499	A	T	NA	High	Homoplasmy (1.40%)
MT-ND5	13518G	H394Q	NA	NA	NA	NA	-3.48017	576	C	G	NA	Low	Homoplasmy (0.90%)
MT-ND5	14065T	T577S	NA	NA	NA	NA	-0.471189	705	A	T	NA	Medium	Homoplasmy (1.00%)
MT-ND5	12907T	syn	rs28690070	NA	NA	NA	-2.01793	512	C	T	NA	NA	Homoplasmy (1.00%)

Gene	Result	AA Change	rsID	Clinvar	OMIM	MitoMap Disease	Conser-vation	Depth	Ref Allele	Alt Allele	Population Frequency	Disease Score	Hetero Fraction
MT-ND5	12853T	syn	rs28689615	NA	NA	NA	-0.707472	531	C	T	NA	NA	Homoplasmy (1.10%)
MT-ND5	13227G	D297E	NA	NA	NA	NA	-3.91081	527	C	G	NA	High	Homoplasmy (1.10%)
MT-ND5	13764T	syn	NA	NA	NA	NA	-6.38884	544	C	T	NA	NA	Homoplasmy (0.90%)
MT-ND5	13062G	syn	rs28670513	NA	NA	NA	-7.55097	387	A	G	NA	NA	Homoplasmy (3.10%)
MT-ND5	12684A	syn	rs28410409	NA	NA	NA	-1.14429	585	G	A	0.25%	NA	Homoplasmy (1.00%)
MT-ND5	13095C	syn	rs28477492	NA	NA	NA	-5.22127	412	T	C	0.08%	NA	Homoplasmy (2.90%)
MT-ND5	13948T	P538S	NA	NA	NA	NA	0.632102	624	C	T	NA	High	Homoplasmy (0.80%)
MT-ND5	13542T	syn	NA	NA	NA	NA	-2.47717	579	A	T	NA	NA	Homoplasmy (0.90%)
MT-ND5	13966C	T544P	NA	NA	NA	NA	-0.671787	634	A	C	NA	Medium	Homoplasmy (1.10%)
MT-ND6	14258A	P139L	rs202227543	NA	NA	NA	0.531803	500	G	A	0.17%	Low	Homoplasmy (1.00%)
MT-RNR2	2985T	NA	NA	NA	NA	NA	0.762693	647	C	T	NA	NA	Homoplasmy (0.80%)
MT-RNR2	3062A	NA	NA	NA	NA	NA	0.0360787	688	T	A	NA	NA	Homoplasmy (1.30%)
MT-RNR2	3091A	NA	NA	NA	NA	NA	0.873961	634	G	A	NA	NA	Homoplasmy (3.20%)
MT-RNR2	3077T	NA	NA	NA	NA	NA	0.753441	667	C	T	NA	NA	Homoplasmy (1.80%)
MT-RNR2	2481G	NA	NA	NA	NA	NA	0.944346	462	A	G	NA	NA	Homoplasmy (1.30%)
MT-RNR2	3000T	NA	NA	NA	NA	NA	0.944346	653	A	T	NA	NA	Homoplasmy (0.80%)
MT-RNR2	2846T	NA	NA	NA	NA	NA	-0.963016	607	G	T	NA	NA	Homoplasmy (1.20%)
MT-RNR2	3009A	NA	NA	NA	NA	NA	-1.05384	654	C	A	NA	NA	Homoplasmy (1.40%)
MT-RNR2	3008T	NA	NA	NA	NA	NA	0.762693	652	C	T	NA	NA	Homoplasmy (0.80%)
MT-RNR2	2524T	NA	NA	NA	NA	NA	0.0360787	502	A	T	NA	NA	Homoplasmy (2.80%)
MT-RNR2	2281T	NA	NA	NA	NA	NA	-0.418055	500	A	T	NA	NA	Homoplasmy (1.80%)
MT-RNR2	2010A	NA	NA	NA	NA	NA	-0.0390394	431	T	A	NA	NA	Homoplasmy (1.20%)
MT-TH	12163T	NA	NA	NA	NA	NA	0.858575	564	A	T	NA	NA	Homoplasmy (0.90%)
MT-TL1	3240A	NA	NA	NA	NA	NA	-0.572276	546	C	A	NA	NA	Homoplasmy (0.90%)
MT-TR	10451A	NA	NA	NA	NA	NA	-0.0217008	518	T	A	NA	NA	Homoplasmy (1.00%)

Functional Networks

Antioxidant Defense

SUSCEPTABLE

ALB APOB APOE GGT1 GPX1 HMOX1 MPO MTTP PON1 PRDX5 SOD2 XDH

Functional Network Summary

ATPase

SUSCEPTABLE

ATP13A2 ATP1A3 ATP1B2 ATP2A1 ATP2A3 ATP6V0A2 ATP6V0A4 ATP6V1E1 ATP6V1H ATP7B ATP8A2 DNAH8 TCIRG1

Functional Network Summary

ATPase is an enzyme whose function is to hydrolyze ATP to form ADP and release energy when the bond is broken. It is found in the mitochondria of the cell. Underlying processes of ATPase include the movement of ions and the regulation of renal function. The primary genes related to ATPase include DNAH8 (Dynein Axonemal Heavy Chain 8), ATP2A (ATPase Sarcoplasmic/Endoplasmic Reticulum Ca2+), ATP6V0 (ATPase H+ Transporting V0), and ATP7B (ATPase Copper Transporting Bet). Cell signaling and the NA/K ATPase system, movement of ions through the cell membrane from the ATPase ion channels are some of the associated molecular pathways of ATPase.

Cholesterol

SUSCEPTABLE

ABCA1 ABCG8 APOB APOC3 APOE CETP LCAT LDLR LIPC LPL MTTP NPC1 NPC2 PCSK9 STAR

Functional Network Summary

Cholesterol is a fat-like substance which is found in all the cells in the human body. The body needs cholesterol for making vitamin D, hormones, as well as enzymes that help to digest foods. The underlying molecular and biological processes that are associated with cholesterol include intestinal uptake of dietary cholesterol, synthesis of bile acid and steroid hormones, de novo cholesterol synthesis, and biliary secretion. The genes that are most often linked with cholesterol include LCAT (Lecithin-Cholesterol Acyltransferase), APO (Apolipoprotein), and ABC (ATP Binding Cassette).

Creatine Kinase

LIKELY LESS STABLE

CKB CKM CKMT1B DMD DYSF EGFR LAMA2 LMNA PIK3CA RAF1 TRAPPC11 TTN

Functional Network Summary

Creatine kinase is an enzyme expressed by various tissues and cell types. It is also known as creatine phosphokinase or phosphocreatine kinase. It is present in blood tests as a marker of damage to CK-rich tissue such as in myocardial infarction, rhabdomyolysis, muscular dystrophy, autoimmune myositides, and acute kidney injury. The biological processes and molecular pathways related to creatine kinase include providing site-specific high-energy phosphate and transferring phosphates between creatine and adenine nucleotides. Some of the associated genes include DMD (Dystrophin), CHKB (Choline Kinase Beta), and CKM (Creatine Kinase, M-Type). Statin-induced myopathy and beta-blocker treatment are ways to treat increased levels of creatine kinase. Creatine blood tests performed at various labs not only evaluate the levels of creatine in the blood, but also serve as an indicator for the underlying disorders that have still not become evident. R74.8 is a commonly associated ICD-10 code that can be used to indicate a diagnosis for abnormal levels of other serum enzymes.
https://healthyliving.azcentral.com/how-to-treat-high-creatine-kinase-levels-12228130.html

Fat Soluble Vitamin Utilization

SUSCEPTABLE

ALB APOB APOE CUBN CYP2R1 LCAT LDLR LPL MTHFR MTTP TJP2 TTPA

Functional Network Summary

Fat-soluble vitamin deficiency is a condition defined by subnormal levels of 25-hydroxy-vitamin D, vitamin E, and retinol, as well as prolonged prothrombin time and is common in people with bile and liver diseases. The underlying processes associated with the disease are inability to form mixed micelles, rapid passive absorption of unconjugated cholic acid, and reduced biliary secretion of conjugated bile acids. The genes linked to the deficiency include BAAT and SLC27A5.

G6P

SUSCEPTABLE

ACHE AGL G6PC3 GABRE GBE1 GCK GPI H6PD HK1 HK2 LPL PCK2 PGD PHKA2 PPARGC1A PYGL SLC37A4

Functional Network Summary

Glucose-6-phosphate dehydrogenase is a cytosolic enzyme. Its primary responsibility is to catalyze a chemical reaction. This enzyme participates in the pentose phosphate pathway, a metabolic pathway that supplies reducing energy to cells such as erythrocytes by maintaining the level of the co-enzyme nicotinamide adenine dinucleotide phosphate (NADPH). GP6C (Glucose-6-Phosphatase Catalytic Subunit), SLC37A4 (Solute Carrier Family 37 Member 4), and LPL (Lipoprotein Lipase) are some genes linked with G6P. The dietary treatment of G6P deficiency aims at avoiding hypoglycemia. The diagnosis is based on clinical presentation, on abnormal basal values, and absence of hyperglycemic response to glucagon. It can be confirmed by demonstrating a deficient activity of a G6P system component in a liver biopsy.
https://ghr.nlm.nih.gov/gene/G6PC

Glucose

LIKELY LESS STABLE

ABCC8 G6PC2 GCK GPI H6PD HNF1A INSR KCNJ11 SLC2A3 SLC2A4 SLC37A4 SLC5A1 SLC5A2

Functional Network Summary

Glucose can be best described as the simplest form of sugar. It is one of the preferred sources of the body for fuel. Glucose is also the key to keep all the mechanisms taking place in the body working in top order. Exocytosis is the primary process that is often linked to glucose. Generation of ATP and neurotransmitters and neurovascular couples are other processes related to the compound. Meanwhile, the genes that are often associated with glucose include SLC (Solute Carrier Family), INS (Insulin), GCK (Glucokinase), G6PC (Glucose-6-Phosphatase Catalytic Subunit), and GPI (Glucose-6-Phosphate Isomerase)

Glycogen

SUSCEPTABLE

AGL ENO3 GBE1 GSK3A GYS1 GYS2 LAMP2 LDHA PFKM PHKA2 PHKG1 PYGB PYGL PYGM SLC37A4

Functional Network Summary

Graves' disease

SUSCEPTABLE

CTLA4 FAS GNAS HLA-DRB1 IFNG IGF1R IL2RA MPO PTPN22 SLC25A16 TPO TSHR

Functional Network Summary

Graves' disease, also called toxic diffuse goiter, is an immune system order that causes overproduction of thyroid hormones (hyperthyroidism). It causes an enlarged thyroid and is most common in women under 40 years of age. Underlying processes for Graves' disease include negative regulation of immune responses, immune system process, thyroid hormone generation, and regulation of NIK/NF-kappaB signaling. TSHR (Thyroid Stimulating Hormone Receptor), TG (Thyroglobulin), TPO (Thyroid Peroxidase), and LMOD1 (Leiomodin 1) genes are highly associated with Graves' disease.

Lipid Metabolism

SUSCEPTABLE

ABCA1 ACADM APOB APOE CETP CPT2 LCAT LDLR LIPC LPL MTTP NPC1 PLIN1 PNPLA2 SMPD1

Functional Network Summary

Lipid metabolism refers to either both the synthesis as well as breakdown of fats consumed through food inside the body cells to release energy for bodily processes or to be stored for later use. Underlying processes include lipoprotein particle remodeling, glucose metabolic process, and cholesterol efflux. Genes associated to lipid metabolism include APO (Apolipoprotein), INS (Insulin), and LPL (Lipoprotein Lipase). E78.9 is the ICD-10 code for lipid metabolism disorder.

mTOR

LIKELY LESS STABLE

DEPTOR MLST8 PIK3CA PIK3CB PIK3CG RHEB RICTOR RPS6KA1 RPS6KB1 RPTOR TSC2

Functional Network Summary

The mammalian target of rapamycin (mTOR), also known as the mechanistic target of rapamycin and FK506-binding protein 12-rapamycin-associated protein 1 (FRAP1), is a kinase that is encoded by the MTOR gene. It is a part of the phosphatidylinositol 3-kinase-related kinase family of protein kinases. mTOR kinase links extracellular signals with intracellular resources. It controls transcription, translation, protein degradation, and cytoskeleton. The ANGPTL7 gene is located in an intron of mTOR gene. mTOR gene is found in two complexes, mTORC1 and mTORC2. mTOR treatment in lymphangioleiomyomatosis, mTOR inhibitors in cancer therapy and combinatorial treatment with mTOR inhibitors, and streptozotocin leading to synergistic in vitro and in vivo antitumor effects in insulinoma cells are some of the treatments associated with mTOR. Dynamic Akt/mTOR signaling is used as a diagnostic tool. R48.2 is a specific ICD-10-CM code that can be used to indicate a diagnosis for reimbursement purposes.
https://ghr.nlm.nih.gov/gene/MTOR

Pancreatitis

SUSCEPTABLE

BRCA1 BRCA2 CASR CFTR GATA6 KRAS LPL MEN1 NPHP3 PALLD PNLIP PRSS1 PRSS2 REG3A TGFB1 TP53

Functional Network Summary

This is a condition which involves inflamed and damaged pancreas due to activated enzymes within the pancreas that begin attacking the pancreatic walls even before being released into the small intestine. Digestion, proteolysis, lipoprotein particle remodeling, and metabolic processes are the underlying processes of this disease. SPINK1 (Serine Peptidase Inhibitor, Kazal Type 1), PRSS1 (Protease, Serine 1), and REG3A (Regenerating Family Member 3 Alpha) are some of the associated genes. Hydrolase activity, peptidase activity, and triglyceride lipase activity are included in the molecular pathways. Fasting, use of IV fluids, and intravenous pain medication are used for the treatment of pancreatitis while blood tests and imaging studies are used for diagnoses. K85 is the ICD-10 code for the disease.

T-lymphocytes T Cell

LIKELY LESS STABLE

CASP8 CTLA4 GZMB IFNG IL2 IL2RA RAG1 STAT3 TP53

Functional Network Summary

T-lymphocytes, also known as, T-cells are a type of white blood cells that form an important part of the cell-mediated immunity. T-cells are produced in the thymus as compared to other lymphocytes that mature in the bone marrow. There are two types of T-lymphocytes that include helper T-cells and cytotoxic T-cell. Cytotoxic T-cells are responsible for eradicating all viral infections and possible tumors in the body. Helper T-cells on the other hand trigger an immune response and aid the immune system in fighting off diseases. The genes associated with T-cells are $CD4^{+}CD8^{-}$ or $CD4^{-}CD8^{+}$.

Trypsinogen

SUSCEPTABLE

BPI CFTR CPA6 EFL1 FCGR2A FLNB PNLIP PRSS1 PRSS2 PRSS3 REG3A SBDS TGFB1 TMPRSS15

Functional Network Summary

Wilson's Disease

SUSCEPTABLE

ABCA1 ALB APOE ATP7A ATP7B DRD2 EDNRB F2 NPC1 RET SLC17A5 TGFB1

Functional Network Summary

Thankyou!

Our team here at GeneSavvy thanks you for choosing GeneSavvy as your Genetic Testing!
We hope your experience with us was empowering.