QuantumLife WGS Report

Genetic Risk Carriers Screening and Analysis

PATIENT NAME	GENDER
John Doe	Male
DATE OF BIRTH	**SAMPLE TYPE**
01/11/1990	Whole Blood
COLLECTION DATE	**SAMPLE ID**
01/01/2023	A123456
ACCESSION #	**REPORT DATE**
4316546	07/08/2023

Overview

At QuantumLife Inc, we specialize in Genetic Risk Carriers Screening and Analysis. Our mission is to provide comprehensive, personalized genomic information that can help you understand your genetic predisposition to various hereditary diseases. To do this, we use Whole Genome Sequencing (WGS), a cutting-edge technology that reads each of the roughly three billion base pairs of an individual's DNA sequence. WGS offers an in-depth view of the entire genome, allowing us to detect genetic variants that may influence disease risk. By understanding these risks, you're empowered to make informed decisions about your health, lifestyle, and preventive care. Our services are designed to provide clarity and guidance in the complex world of genomics, bridging the gap between genetic information and actionable health insights.

Section 1: Finding Summary

This is a brief summary of the WGS screening and analysis.

Category	Test Results	Your Risk
Genome sequenced	3.2 Billion base pairs analyzed	nan
Variants detected	5.3 millions	nan
Total genes assessed	1519	Carrying 5 disease-causing variants
Total hereditary diseases assessed	1652	1 hereditary cancer, 2 metabolic disorders, 1 endocrine disorder, and 1 respiratory disorder
Total pharmacogenetic genes assessed	1234	nan
Total drugs assessed	120	51 drugs potentially impacted
Total age-related phenotypes/traits assessed	25	High risk of developing muscle degeneration condition, one neurological condition
Total physical fitness traits assessed	12	Physically active, risk in developing cardiovascular conditions

Section 2: Hereditary Disease Risk

This section includes variants that, when found by themselves, are associated with high and moderate genetic risk of developing a disease. Depending on a variant zygosity and disease inheritance, the disease can affect you or represent a risk for the next generation (carrier variants).

Subsection 2.1: Variant Summary

HEALTH CATEGORY	GENE	VARIANT	ZYGOSITY	CLASSIFICATION	DISEASE ASSOCIATED	INHERITANCE
Hereditary Cancer	ATM	rsxxxxxx c.1564_1565delGA (p.Glu522fs) NM_000051.3	Heterozygous	Pathogenic	Breast cancer	Autosomal dominant
xxxxxx	xxxxx	xxxxx	xxxxxx	xxxxxx	xxxxxx	xxxxx
xxxxxx	xxxxx	xxxxx	xxxxxx	xxxxxx	xxxxxx	xxxxx

xxxxxx	xxxxx	xxxxx	xxxxxx	xxxxxx	xxxxxx	xxxxx
xxxxxx	xxxxx	xxxxx	xxxxxx	xxxxxx	xxxxxx	xxxxx

Subsection 2.2: Variant Details

Variants Associated with Hereditary Cancer

Patient was found to have a pathogenic variant associated with an increased risk for Hereditary Cancer

Variant ID	Gene	VARIANT	Effect allele	Effect allele frequency
rsxxxxx	ATM	c.1564_1565delGA (p.Glu522fs)	C>delGA	nan

The c.1564_1565delGA frameshift variant is predicted to truncate the ATM protein, resulting in a loss or deficient function. The variant has been identified in a homozygous or compound heterozygous state in individuals affected with ataxia-telangiectasia (PMID 9463314, PMID 10817650, PMID 9000145, PMID 12497634, PMID 21965147), as well as in an individual diagnosed with breast cancer (PMID 27083775). The ATM gene codes for an enzyme called serine-protein kinase. This enzyme plays a key role in DNA damage response by transducing checkpoint signaling, cell cycle regulation, and tumor suppression. Pathogenic ATM variants result in impaired response to DNA damage, cell death, and formation of cancerous tumors and are associated with autosomal dominant susceptibility to breast cancer (MIM 114480) and autosomal recessive ataxia-telangiectasia (AT, MIM 208900). Women with a pathogenic ATM variant have a 5-to 9-fold increased risk of breast cancer (PMID 1961222, PMID 15928302). There is also some evidence supporting an association with autosomal dominant pancreatic, colorectal, prostate, and possibly other cancers (PMID 26483394, PMID 15928302, PMID 26662178). Ataxia-telangiectasia a childhood-onset disease characterized by progressive cerebellar ataxia, neurologic degeneration, conjunctival telangiectasias, immunodeficiency and increased risk of leukemia and lymphoma as well as other cancers (PMID 20301790). The worldwide prevalence of ataxiatelangiectasia is unknown, however, the estimated prevalence in the US ranges from 1 in 40,000 to 1 in 100,000 live births (PMID 20301790).

Variants Associated with Endocrine Disorder

Patient is a carrier for a pathogenic variant associated with a Endocrine Disorder. Reproductive partner and/or first-degree relatives may benefit from screening

Variant ID	Gene	VARIANT	Effect allele	Effect allele frequency
rsxxxxx	ATM	c.1564_1565delGA (p.Glu522fs)	C>delGA	nan

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Section 3: Pharmacogenomic Findings

Unnamed: 0	REPORTING SCOPE	YOUR RISK
Total genes assessed	13	YOUR PHARMACOGENOMIC RESULTS
Total drugs assessed	88	51 Drugs Potentially Impacted

The way you respond to therapeutic drugs depends on your genes as well as other factors. This analysis found all your genetic alterations (genotypes) that may have a impact on how your body processing or metabolizing drugs (phenotypes). It helps your doctor better understand your personal situation, and your doctor will take these factors into account when deciding which drug or what drug dose is the best for you. Do not stop taking or alter the dosage of any prescribed medications without consulting with your doctor beforehand.

Subsection 3.1: Drugs Potentially Impacted

Symbol	Genotype	Phenotype	Drugs
ABCG2	rs2231142 reference (G)/rs2231142 variant (T)	Decreased Function	Rosuvastatin
CACNA1S	No CPIC variants found	Uncertain Susceptibility	Desflurane, Enflurane, Halothane, Isoflurane, Methoxyflurane, Sevoflurane, Succinylcholine
CFTR	No CPIC variants found	ivacaftor non-responsive in CF patients	Ivacaftor
CYP2B6	*9/*9	Poor Metabolizer	Efavirenz, Sertraline
CYP2C19	*38/*38	Normal Metabolizer	Amitriptyline, Citalopram, Clomipramine, Clopidogrel, Dexlansoprazole, Doxepin, Escitalopram, Imipramine, Lansoprazole, Omeprazole, Pantoprazole, Sertraline, Trimipramine, Voriconazole
CYP2C9	*3/*3	Poor Metabolizer	Celecoxib, Flurbiprofen, Fluvastatin, Ibuprofen, Lornoxicam, Meloxicam, Piroxicam, Tenoxicam, Warfarin
CYP2D6	*1/*1	Normal Metabolizer	Amitriptyline, Atomoxetine, Clomipramine, Codeine, Desipramine, Doxepin, Fluvoxamine, Hydrocodone, Imipramine, Nortriptyline, Ondansetron, Paroxetine, Tamoxifen, Tramadol, Trimipramine, Tropisetron, Venlafaxine, Vortioxetine
CYP3A5	*3/*3	Poor Metabolizer	Tacrolimus
CYP4F2	*1/*4	nan	Warfarin
DPYD	c.1627A>G (*5)/c.1627A>G (*5)	Normal Metabolizer	Capecitabine, Fluorouracil
G6PD	B (reference)/B (reference)	Normal	Aminosalicylic Acid, Aspirin, Chloramphenicol, Chloroquine, Ciprofloxacin, Dapsone, Dimercaprol, Doxorubicin, Furazolidone, Glyburide, Hydroxychloroquine, Mafenide, Methylene Blue, Nalidixic Acid, Nitrofurantoin, Norfloxacin, Ofloxacin, Pegloticase, Phenazopyridine, Primaquine, Quinine, Rasburicase, Sulfadiazine, Sulfadimidine, Sulfamethoxazole / Trimethoprim, Sulfanilamide, Sulfasalazine, Sulfisoxazole, Tafenoquine, Tolbutamide, Toluidine Blue, Vitamin C, Vitamin K
IFNL3	rs12979860 reference (C)/rs12979860 reference (C)	nan	Peginterferon Alfa-2a, Peginterferon Alfa-2b
NUDT15	*1/*1	Normal Metabolizer	Azathioprine, Mercaptopurine, Thioguanine
RYR1	No CPIC variants found	Uncertain Susceptibility	Desflurane, Enflurane, Halothane, Isoflurane, Methoxyflurane, Sevoflurane, Succinylcholine
SLCO1B1	*37/*37	Normal Function	Atorvastatin, Fluvastatin, Lovastatin, Pitavastatin, Pravastatin, Rosuvastatin, Simvastatin
TPMT	*1/*1	Normal Metabolizer	Azathioprine, Mercaptopurine, Thioguanine
UGT1A1	*1/*6	Intermediate Metabolizer	Atazanavir

VKORC1	rs9923231 variant (T)/rs9923231 variant (T)	nan	Warfarin

Subsection 3.2: Gene and Phenotype Findings

Symbol	Genotype	Phenotype	Drugs
ABCG2	rs2231142 reference (G)/rs2231142 variant (T)	Decreased Function	Rosuvastatin
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