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Genetic Insights Report

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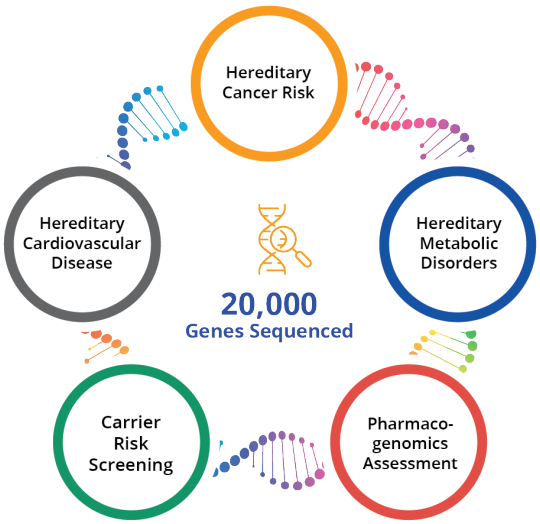
Sample ID :	Sample Type :	Sample Collected :
Sample Received :	Report Generated :	

Genomic Health Insights Overview

- The DNA of an individual is organised into known functional components called genes.
- There are over 20,000 genes in an individual.
- Genomic Health Insights identifies variants in these genes that may have an impact on one's health or on the health of one's progeny.
- Cutting edge sequencing and bioinformatics technology are used to identify relevant variants in these genes that pass a high enough bar of scientific evidence.
- Variants are then categorized based on their impact into individual sections on cancer risk, cardiac/cardiovascular risk, metabolic health risk, and carrier status.
- An additional section on how a person's genomic variants may modulate their response to drugs is also provided.



Disclaimer: Note that the above information is not intended for any action on your part for diagnosis, cure, mitigation, treatment or prevention of disease. Any action based on this information should be taken only on the recommendation of a medical specialist.

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Notes and Limitations

Note that this analysis does not cover Variants of Uncertain Significance - those genomic variants that have not yet been categorized by current science as harmful or benign. Only variants classified as Pathogenic or Likely Pathogenic per the American College of Medical Genetics guidelines for variant classification are included in this report.

Also, modern genome sequencing processes of the type used here, can identify genomic variants that involve one genomic character being changed to another with high confidence. Such variants are also called Single Nucleotide Variants (SNVs). These comprise a majority of all genomic variants.

However, there are also genomic variants, albeit fewer, where several consecutive genomic characters are either deleted or repeated multiple times. These are called Copy Number Variants (CNVs). Careful analysis of sequencing data can identify some CNVs. However, these may need to be confirmed using alternative methods before we can be confident of their presence. If any CNVs are included in this report, we will explicitly indicate whether you should consider confirmation through alternative methods, or if we have already done that for you.

For further information on limitations please refer to the detailed note at the end of the report.

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Hereditary Cancer Risk

What is Hereditary Cancer Risk?

Cancer is caused by the spontaneous accumulation of genomic changes over decades. This accumulation may proceed at different rates in different individuals. Some individuals are born with genomic variants that make them much more susceptible to this accumulation, leading to an increased risk for cancer. Variants in several genes have been discovered to increase cancer risk for 12 relatively common cancers. Together, ~10% of some common cancers occur on account of these variants.

Results

Your report describes 1 observation(s) of significance.

Summary of Observations of Significance

Gene	Zygosity	Inheritance	Variant	Variant Classifcation
MSH6	Heterozygous	Dominant	chr2:48032833dupT c.3633dupT p.Val1212CysfsTer3	Pathogenic

Schedule appointment with Genetic Counsellor for detailed explanation

Meaning of Terms Used

Gene: Genes carry recipes for the creation of molecules that keep the body running. There are ~20,000 genes in the human body.

Variant: This refers to a character in the gene where you are different from the "average human".

Zygosity: We inherit 3 billion genomic characters from each parent. A gene variant is Homozygous if it is found in the characters inherited from BOTH parents and Heterozygous if it is found in the characters inherited from only one parent.

Variant Classification: Not all variants have a significant impact on health. We evaluate current scientific evidence to classify variants accordingly as per ACMG guidelines. A 'Pathogenic' variant has conclusive scientific evidence, while a 'Likely Pathogenic' variant has enough but inconclusive evidence.

Organ: Organ is a collection of tissues that constitutes the a biological function.

Tissue: Tissue is a group of cells performing the same function.

Detailed Findings

Organ/ Tissue	Associated Gene(s)	Variant Detected	Gene(s) with Variants
Blood	*REDACTED*	No	-
Breast	*REDACTED*	No	-
Colorectal	*REDACTED*	Yes	*MSH6*
Endocrine	*REDACTED*	No	-
Endometrial/ Uterine	*REDACTED*	Yes	*MSH6*
Nervous system	*REDACTED*	Yes	*MSH6*
Ovarian	*REDACTED*	Yes	*MSH6*
Pancreatic	*REDACTED*	Yes	*MSH6*
Prostate	*REDACTED*	N/A	-
Kidney	*REDACTED*	No	-
Skin	*REDACTED*	No	-
Stomach	*REDACTED*	Yes	*MSH6*

Recommendations

To understand the implications of the above variants, if any, please schedule a report consultation appointment with a Strand counselor by writing to **hello@strandls.com**. Subsequently, you can also take this report to a medical specialist.

Hereditary Cardiovascular Disease

What is Hereditary Cardiovascular Risk?

Cardio-vascular disease is a group of diseases that affect your heart and blood vessels. Hereditary cardiovascular conditions is a term used to describe a variety of inherited, familial diseases of the heart and blood vessels. They represent a small percentage of cardio-vascular diseases, but are very important to assess risk. A number of genes are known to carry variants that cause an increased risk of abnormal structure or function of the heart or the blood vessels. These represent a small but serious yet silent fraction of the overall burden of cardiovascular disease. Knowledge of these variants can help clinicians manage any risks more effectively.

Results

No observation(s) of significance were observed.

Detailed Findings

Broad Disease Category	Disorder Type	Associated Gene(s)	Variant Detected	Gene(s) with Variants
Cardiomyopathy	Arrhythmogenic right ventricular cardiomyopathy	*REDACTED*	No	-
	Dilated cardiomyopathy (non syndromic)	*REDACTED*	No	-
	Hypertrophic cardiomyopathy (non syndromic)	*REDACTED*	No	-

Detailed Findings

Broad Disease Category	Disorder Type	Associated Gene(s)	Variant Detected	Gene(s) with Variants
	Restrictive cardiomyopathy	*REDACTED*	No	-
Arrhythmia	Long QT syndrome	*REDACTED*	No	-
	Short QT syndrome	*REDACTED*	No	-
	Brugada syndrome	*REDACTED*	No	-
	Atrial fibrillation	*REDACTED*	No	-
	Catecholaminergic polymorphic ventricular tachycardia	*REDACTED*	No	-
Other	Familial hypercholesterolemia	*REDACTED*	No	-
	Aortopathies	*REDACTED*	No	-
	Ehlers–Danlos syndrome, vascular type	*REDACTED*	No	-

Recommendations

To understand the implications of the above variants, if any, please schedule a report consultation appointment with a Strand counselor by writing to **hello@strandls.com**. Subsequently, you can also take this report to a medical specialist.

Hereditary Metabolic Disorders

What is Hereditary Metabolic Disorders Risk?

Inherited metabolic disorders refer to medical conditions caused by inherited genetic defects that interfere with the body's metabolism. Very often, the variants that are seen in these genes manifest in early childhood and adolescence. Sometimes, however, it can take years before symptoms can be seen, e.g., as in Wilson's disease where copper accumulates in various organs over time. Such disorders manifest clinically as adults or under conditions of stress. An assessment for variants in the genes known to be associated with inherited metabolic disorders presents an opportunity for your clinician to prepare you for a mitigation solution where needed.

Results

No observation(s) of significance were observed.

Detailed Findings

Broad Disease Category	Disorder Type	Associated Gene(s)	Variant Detected
Amino acid, Peptide metabolism & Urea cycle disorder	Argininosuccinic aciduria	*REDACTED*	No
	Hyperoxaluria	*REDACTED*	No
	Homocystinuria	*REDACTED*	No
	Glutaric aciduria type 1	*REDACTED*	No
	L-2-hydroxyglutaric aciduria	*REDACTED*	No
	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	*REDACTED*	No
	Ornithine transcarbamylase deficiency	*REDACTED*	No

Detailed Findings

Broad Disease Category	Disorder Type	Associated Gene(s)	Variant Detected
	Phenylketonuria	*REDACTED*	No
	Cystinuria	*REDACTED*	No
Carbohydrate metabolism disorder	Fructosemia	*REDACTED*	No
	Galactosialidosis	*REDACTED*	No
	Retinitis pigmentosa type 59	*REDACTED*	No
	Pompe disease	*REDACTED*	No
	Krabbe disease	*REDACTED*	No
	Gaucher disease	*REDACTED*	No
	GBE1-related disorders (adult polyglucosan body disease)	*REDACTED*	No
	GM1 gangliosidosis (type III)	*REDACTED*	No
	Mucopolysaccharidoses type 1 late onset (Scheie syndrome)	*REDACTED*	No
	Maturity onset diabetes type 13	*REDACTED*	No
	Sialidosis type 1	*REDACTED*	No
	Phosphomannomutase 2 deficiency (PMM2-CDG-1A)	*REDACTED*	No
	Carbohydrate-deficient glycoprotein syndrome type Ia	*REDACTED*	No

Detailed Findings

Broad Disease Category	Disorder Type	Associated Gene(s)	Variant Detected
Iron, porphyrin and heme-related disorders	Spastic paraplegia type 43	*REDACTED*	No
	Aceruloplasminemia	*REDACTED*	No
	Neuroferritinopathy	*REDACTED*	No
	Hemochromatosis	*REDACTED*	No
	Hypochromic microcytic anemia	*REDACTED*	No
	Iron-refractory iron deficient anaemia	*REDACTED*	No
Lipid and lipoprotein metabolism disorders	Very long chain acyl-CoA dehydrogenase deficiency	*REDACTED*	No
	Metachromatic leukodystrophy (late onset)	*REDACTED*	No
	Neuronal ceroid lipofuscinosis (Kufs disease, type A)	*REDACTED*	No
	Neuronal ceroid lipofuscinosis (Kufs disease, type B)	*REDACTED*	No
	Fabry disease	*REDACTED*	No
	GM1 gangliosidosis (Type III)	*REDACTED*	No
	Tay-Sachs disease	*REDACTED*	No
	Sandhoff disease	*REDACTED*	No

Detailed Findings

Broad Disease Category	Disorder Type	Associated Gene(s)	Variant Detected
	PLA2G6-related dystonia-parkinsonism (atypical)	*REDACTED*	No
	Niemann Pick disease type C	*REDACTED*	No
	Spinal muscular atrophy (type 1-3)	*REDACTED*	No
Lysosomal Storage Disease	Mucopolysaccharidoses type 1 late onset (Scheie sydrome)	*REDACTED*	No
	Gaucher disease	*REDACTED*	No
	Fabry disease	*REDACTED*	No
Neurotransmitter metabolism disorders	Dopa-responsive dystonia, classic Segawa	*REDACTED*	No
	Dopamine transporter deficiency syndrome (DTDS, atypical late onset type)	*REDACTED*	No
Peroxisomal Disorders	Zellweger spectrum disorders	*REDACTED*	No
Purine and pyramidine metabolism	Fanconi anemia	*REDACTED*	No
	SANDO syndrome (sensory ataxic neuropathy, dysarthria, opthalmoparesis)	*REDACTED*	No
Sterol Metabolism	X-linked adrenoleukodystrophy	*REDACTED*	No
	2-methyl acyl-CoA racemase deficiency (AMACR deficiency)	*REDACTED*	No
	Cerebrotendinous xanthomatosis	*REDACTED*	No

Detailed Findings

Broad Disease Category	Disorder Type	Associated Gene(s)	Variant Detected
	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (non-classic)	*REDACTED*	No
	Congenital adrenal hypoplasia	*REDACTED*	No
Trace element and mineral metabolism	Wilson disease	*REDACTED*	No
	Hypophosphatasia, adult	*REDACTED*	No
	Syndrome of liver cirrhosis, dystonia and hypermanganesemia	*REDACTED*	No
	Acrodermatitis enteropathica	*REDACTED*	No
Transport and channel metabolism	Spinocerebellar ataxia type 10	*REDACTED*	No
	Kufor Rakeb disease	*REDACTED*	No
	Retinitis pigmentosa type 74	*REDACTED*	No
	Myasthenic syndrome type 4A (adult)	*REDACTED*	No
	Dopamine transporter deficiency syndrome (DTDS, atypical late onset type)	*REDACTED*	No
	Basal ganglia disease, biotin-responsive	*REDACTED*	No
	Hermansky-Pudlak syndrome type 1	*REDACTED*	No
	Hermansky-Pudlak syndrome type 3	*REDACTED*	No

Detailed Findings

Broad Disease Category	Disorder Type	Associated Gene(s)	Variant Detected
Vitamin, Mineral and co-factor metabolism	Basal ganglia disease, biotin-responsive	*REDACTED*	**No**

Recommendations

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Carrier Risk Screening

What is Carrier Risk Screening?

Many people carry variants in their genes that are disease-causing ONLY when both gene copies are affected. When only one copy is affected by the gene variant, the person remains unaffected and is said to be a carrier. In the event that a person and their spouse are both carriers, each child has a quarter chance of becoming seriously affected. Screening for carrier status can help you control this risk.

Results

No observation(s) of significance were observed.

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
Amino acid, Peptide metabolism & Urea cycle disorders	Hyperoxaluria, primary type I	*AR*	*REDACTED*	**No**
	α-Methylacetoacetic aciduria	*AR*	*REDACTED*	**No**
	Aspartylglucosaminuria	*AR*	*REDACTED*	**No**
	Canavan disease	*AR*	*REDACTED*	**No**
	Argininosuccinate aciduria	*AR*	*REDACTED*	**No**
	Citrullinemia	*AR*	*REDACTED*	**No**
	Maple syrup urine disease	*AR*	*REDACTED*	**No**
	Homocystinuria, B6 responsive and non-responsive	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Maple syrup urine disease type II	*AR*	*REDACTED*	**No**
	Tyrosinemia type I	*AR*	*REDACTED*	**No**
	Trimethylaminuria	*AR*	*REDACTED*	**No**
	Glycine encephalopathy	*AR*	*REDACTED*	**No**
	Glutaricaciduria type I	*AR*	*REDACTED*	**No**
	Alkaptonuria	*AR*	*REDACTED*	**No**
	Methylmalonic aciduria, vitamin B12-responsive, cbl A and B type	*AR*	*REDACTED*	**No**
	3-methylcrotonyl CoA carboxylase 2 deficiency	*AR*	*REDACTED*	**No**
	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	*AR*	*REDACTED*	**No**
	Phenylketonuria	*AR*	*REDACTED*	**No**
	Propionicacidemia	*AR*	*REDACTED*	**No**
	Ornithine transcarbamylase deficiency	*X-Linked*	*REDACTED*	**No**
	Oculocutaneous albinism type 1A	*AR*	*REDACTED*	**No**
	Oculocutaneous albinism type 1B	*AR*	*REDACTED*	**No**
	Albinism, oculocutaneous, type III	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
Ataxia	Spinocerebellar ataxia type 10	*AD*	*REDACTED*	**No**
	Ataxia telangiectasia	*AR*	*REDACTED*	**No**
Carbohydrate metabolism disorders	Glycogen storage disease type 3	*AR*	*REDACTED*	**No**
	Hereditary fructosuria	*AR*	*REDACTED*	**No**
	Mucopolysaccharidosis type VI	*AR*	*REDACTED*	**No**
	Congenital disorder of glycosylation type 1	*AR*	*REDACTED*	**No**
	Dihydrolipoamide dehydrogenase deficiency	*AR*	*REDACTED*	**No**
	Walker–Warburg congenital muscular dystrophy	*AR*	*REDACTED*	**No**
	Glycogen storage disease type IA or von Gierke Disease	*AR*	*REDACTED*	**No**
	Glycogen storage disease type II (Pompe disease)	*AR*	*REDACTED*	**No**
	Galactosemia	*AR*	*REDACTED*	**No**
	Mucopolysaccharidosis IVA	*AR*	*REDACTED*	**No**
	GBE1-related disorders (adult polyglucosan body disease)	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Glycogen storage disease type IV	*AR*	*REDACTED*	**No**
	Mucopolysaccharidosis type IVB (Morquio)	*AR*	*REDACTED*	**No**
	Mucolipidosis type II alpha/beta	*AR*	*REDACTED*	**No**
	Mucolipidosis type III alpha/beta	*AR*	*REDACTED*	**No**
	Schindler disease type 1	*AR*	*REDACTED*	**No**
	Schindler disease type 3	*AR*	*REDACTED*	**No**
	Carbohydrate-deficient glycoprotein syndrome type Ia	*AR*	*REDACTED*	**No**
	Mucopolysaccharidosis, Ih (Hurler syndrome and Hurler-Scheie syndrome)	*AR*	*REDACTED*	**No**
	Glycogen storage disease Ib	*AR*	*REDACTED*	**No**
	Glycogen storage disease Ic	*AR*	*REDACTED*	**No**
Clotting and blood related disorders	Hemophilia A (HEMA)	*X-Linked*	*REDACTED*	**No**
	Hemophilia B (HEMB)	*X-Linked*	*REDACTED*	**No**
	Fanconi anemia, complementation group C	*AR*	*REDACTED*	**No**
Congineta l malformations	Opitz GBBB syndrome type I (GBBB1)	*X-Linked*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
Connective tissue disorders	Achondrogenesis type Ib	*AR*	*REDACTED*	**No**
	Epiphyseal dysplasia multiple 4	*AR*	*REDACTED*	**No**
	Ehlers–Danlos-like syndrome due to tenascin-X deficiency	*AR*	*REDACTED*	**No**
Cystic Fibrosis	Cystic fibrosis	*AR*	*REDACTED*	**No**
Dermatological disorders	Ichthyosis congenital AR 4A 4B (harlequin)	*AR*	*REDACTED*	**No**
	Bloom syndrome	*AR*	*REDACTED*	**No**
	Recessive dystrophic epidermolysis bullosa	*AR*	*REDACTED*	**No**
	Epidermolysis bullosa junctional 4 intermediate	*AR*	*REDACTED*	**No**
	Trichothiodystrophy 1 photosensitive	*AR*	*REDACTED*	**No**
	Fraser syndrome type 3	*AR*	*REDACTED*	**No**
	Epidermolysis bullosa junctional 1A, intermediate/epidermolysis bullosa, junctional 1B, severe	*AR*	*REDACTED*	**No**
	Epidermolysis bullosa junctional 3A, intermediate/epidermolysis bullosa, junctional 3B, severe	*AR*	*REDACTED*	**No**
	Hermansky-Pudlak syndrome type 1	*AR*	*REDACTED*	**No**
	Hermansky-Pudlak syndrome type 3	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Epidermolysis bullosa, junctional 5B, with pyloric atresia	*AR*	*REDACTED*	**No**
	Ichthyosis, congenital, autosomal recessive 1	*AR*	*REDACTED*	**No**
	Xeroderma pigmentosum	*AR*	*REDACTED*	**No**
Endocrine disorders	Diabetes mellitus, permanent neonatal 3	*AR/AD*	*REDACTED*	**No**
	Adrenal insufficiency, congenital, with 46, XY sex reversal, partial or complete	*AR*	*REDACTED*	**No**
	Adrenal hypoplasia, congenital (AHC)	*X-Linked*	*REDACTED*	**No**
Epilepsy	Developmental and epileptic encephalopathy type 1 (DEE1)	*X-Linked*	*REDACTED*	**No**
Eye disorders	Bardet-Biedl syndrome type 1	*AR*	*REDACTED*	**No**
	Bardet-Biedl syndrome type 2	*AR*	*REDACTED*	**No**
	Retinitis pigmentosa type 74	*AR*	*REDACTED*	**No**
	Leber congenital amaurosis type 10	*AR*	*REDACTED*	**No**
	Usher syndrome type 3a	*AR*	*REDACTED*	**No**
	Achromatopsia type 3	*AR*	*REDACTED*	**No**
	Retinitis pigmentosa type 59	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Oculocutaneous albinism brown and type II	*AR*	*REDACTED*	**No**
	Usher syndrome type 1F	*AR*	*REDACTED*	**No**
	Macular degeneration, X-linked atrophic	*X-Linked*	*REDACTED*	**No**
	Retinitis pigmentosa type 3	*X-Linked*	*REDACTED*	**No**
	Retinitis pigmentosa, X-linked, and sinorespiratory Infections, with or without deafness	*X-Linked*	*REDACTED*	**No**
	Retinoschisis type 1, X-linked, juvenile (RS1)	*X-Linked*	*REDACTED*	**No**
	Usher syndrome type 2A	*AR*	*REDACTED*	**No**
Hearing disorders	Deafness AR type 12	*AR*	*REDACTED*	**No**
	Usher syndrome type 3a	*AR*	*REDACTED*	**No**
	Nonsyndromic hearing loss dominant type 3A	*AD*	*REDACTED*	**No**
	Nonsyndromic hearing loss recessive type 1A	*AR*	*REDACTED*	**No**
	Heimler syndrome type 1	*AR*	*REDACTED*	**No**
	Deafness, autosomal recessive type 23	*AR*	*REDACTED*	**No**
	Usher syndrome type 1F	*AR*	*REDACTED*	**No**
	Deafness autosomal recessive type 4	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Pendred syndrome	*AR*	*REDACTED*	**No**
	Deafness, autosomal recessive type 7	*AR*	*REDACTED*	**No**
	Deafness, autosomal recessive type 18A	*AR*	*REDACTED*	**No**
	Usher syndrome type 2A	*AR*	*REDACTED*	**No**
	Deafness, autosomal recessive type 8/10	*AR*	*REDACTED*	**No**
Heart disorders	Dialed cardiomyopathy type 1X	*AR*	*REDACTED*	**No**
Immune disorders	Severe combined immunodeficiency	*AR*	*REDACTED*	**No**
	Autoimmune polyendocrinopathy syndrome type I	*AR*	*REDACTED*	**No**
	Severe combined immunodeficiency, X-linked	*X-linked*	*REDACTED*	**No**
	Hemophagocytic lymphohistiocytosis familial type 2	*AR*	*REDACTED*	**No**
Inflammatory disorder	Aicardi Goutieres syndrome type 2	*AR*	*REDACTED*	**No**
Iron, porphyrin and heme-related disorders	Alpha thalassemia	*AR*	*REDACTED*	**No**
	Beta-thalassemia	*AR*	*REDACTED*	**No**
	Heme oxygenase-1 deficiency	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Atransferrinemia	*AR*	*REDACTED*	**No**
Kidney disorders	Finnish congenital nephrotic syndrome	*AR*	*REDACTED*	**No**
	Autosomal recessive polycystic kidney disease	*AR*	*REDACTED*	**No**
Lipid and lipoprotein metabolism disorders	Surfactant metabolism dysfunction, pulmonary 3	*AR*	*REDACTED*	**No**
	Adrenoleukodystrophy	*X-linked*	*REDACTED*	**No**
	Medium-chain acyl-coenzyme A dehydrogenase deficiency	*AR*	*REDACTED*	**No**
	Acyl-CoA dehydrogenase, short-chain, deficiency of	*AR*	*REDACTED*	**No**
	Very long chain acyl-CoA dehydrogenase deficiency	*AR*	*REDACTED*	**No**
	Metachromatic leukodystrophy	*AR*	*REDACTED*	**No**
	Carnitine palmitoyltransferase II deficiency, infantile	*AR*	*REDACTED*	**No**
	Krabbe disease	*AR*	*REDACTED*	**No**
	Gaucher disease type I & II	*AR*	*REDACTED*	**No**
	Tay-Sachs disease	*AR*	*REDACTED*	**No**
	Fabry disease	*X-Linked*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Mucolipidosis type IV (Berman disease/ ganglioside neurainidase type)	*AR*	*REDACTED*	**No**
	LDL cholesterol level QTL2	*AR*	*REDACTED*	**No**
	Niemann-Pick disease type C1/Niemann-Pick disease type D	*AR*	*REDACTED*	**No**
	Carnitine Deficiency, systemic primary	*AR*	*REDACTED*	**No**
	Niemann–Pick disease type A	*AR*	*REDACTED*	**No**
	Niemann–Pick disease type B	*AR*	*REDACTED*	**No**
Liver disorders	Progressive familial intrahepatic cholestasis type 3	*AR*	*REDACTED*	**No**
	Progressive familial intrahepatic cholestasis type 2	*AR*	*REDACTED*	**No**
	Progressive familial intrahepatic cholestasis type 1	*AR*	*REDACTED*	**No**
	CPT deficiency hepatic type IA	*AR*	*REDACTED*	**No**
Mitochondrial disorders	Mitochondrial DNA depletion syndrome type 4A (Alpers- Huttenlocher Syndrome)	*AR*	*REDACTED*	**No**
	Mitochondrial DNA depletion syndrome type 4B (MNGIE form)	*AR*	*REDACTED*	**No**
	Pontocerebellar hypoplasia type 6	*AR*	*REDACTED*	**No**
	Mitochondrial complex IV deficiency, nuclear type 2	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
Motor Neuron diseases	Spastic paraplegia type 2, X-linked	*X-Linked*	*REDACTED*	**No**
	Spinal muscular atrophy type 2 (Dubowitz disease)	*AR*	*REDACTED*	**No**
	Spinal muscular atrophy type 3 (Kugelberg-Welander disease)	*AR*	*REDACTED*	**No**
	Spinal muscular atrophy type 4 (late onset)	*AR*	*REDACTED*	**No**
	Spinal muscular atrophy type 1 (Werdnig-Hoffmann disease)	*AR*	*REDACTED*	**No**
Muscular dystrophy	Becker muscular dystrophy	*X-Linked*	*REDACTED*	**No**
	Duchenne muscular dystrophy	*X-Linked*	*REDACTED*	**No**
	Limb girdle muscle dystrophy AR1/AD4	*AR/AD*	*REDACTED*	**No**
	Congenital myotonia, autosomal recessive form (Becker type)	*AR*	*REDACTED*	**No**
	Muscular dystrophy-dystroglycanopathy type A and B	*AR*	*REDACTED*	**No**
	Nemaline myopathy type 2	*AR*	*REDACTED*	**No**
Neurodevelopmental disorders	Adrenoleukodystrophy	*X-Linked*	*REDACTED*	**No**
	Joubert syndrome type 3	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Metachromatic leukodystrophy	*AR*	*REDACTED*	**No**
	Developmental and epileptic encephalopathy type 1	*X-Linked*	*REDACTED*	**No**
	Joubert syndrome type 9	*AR*	*REDACTED*	**No**
	Meckel syndrome type 6	*AR*	*REDACTED*	**No**
	Congenital hydrocephalus type 1	*AR*	*REDACTED*	**No**
	Joubert syndrom type 5	*AR*	*REDACTED*	**No**
	Cerebrooculofacioskeletal syndrome type 2	*AR*	*REDACTED*	**No**
	Krabbe disease	*AR*	*REDACTED*	**No**
	Gaucher disease type I & II	*AR*	*REDACTED*	**No**
	Tay-Sachs disease	*AR*	*REDACTED*	**No**
	Hydrocephalus due to congenital stenosis of aqueduct of Sylvius (HSAS)	*X-Linked*	*REDACTED*	**No**
	Donnai–Barrow syndrome	*AR*	*REDACTED*	**No**
	Primary microcephaly 1, recessive	*AR*	*REDACTED*	**No**
	Megalencephalic leukoencephalopathy with subcortical cysts type 2B (Van der Knapp disease)	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Basal ganglia disease, biotin-responsive	*AR*	*REDACTED*	**No**
	Cerebral creatine deficiency syndrome type 1	*X-Linked*	*REDACTED*	**No**
	Joubert syndrome type 2	*AR*	*REDACTED*	**No**
	Meckel syndrome type 2	*AR*	*REDACTED*	**No**
Neurodegenerative disorders	Ataxia telangiectasia	*AR*	*REDACTED*	**No**
	Neuronal ceroid lipofuscinosis type 3	*AR*	*REDACTED*	**No**
	Neuronal ceroid lipofuscinosis type 1	*AR*	*REDACTED*	**No**
	COACH syndrome type 1	*AR*	*REDACTED*	**No**
	Neuronal ceroid lipofuscinosis type 2	*AR*	*REDACTED*	**No**
Neuro-muscular disorders	Myasthenic syndrome congenital type 4A, slow-channel	*AD/AR*	*REDACTED*	**No**
	Myasthenic syndrome congenital type 4B, fast-channel	*AR*	*REDACTED*	**No**
	Familial dysautonomia (Riley-Day Syndrome)	*AR*	*REDACTED*	**No**
Peroxisomal disorders	Heimler syndrome type 1	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Zellweger spectrum disorders	*AR*	*REDACTED*	**No**
Pigmentation disorders	Oculocutaneous albinism brown and type II	*AR*	*REDACTED*	**No**
Pulmonary disorders	Surfactant metabolism dysfunction pulmonary type 3	*AR*	*REDACTED*	**No**
	Emphysema/Emphysema-cirrhosis due to AAT deficiency	*AR*	*REDACTED*	**No**
Sterol Metabolism disorders	Adrenal insufficiency, congenital, with 46, XY sex reversal, partial or complete	*AR*	*REDACTED*	**No**
	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	*AR*	*REDACTED*	**No**
	Cerebrotendinous xanthomatosis	*AR*	*REDACTED*	**No**
	Smith-Lemli-Opitz syndrome	*AR*	*REDACTED*	**No**
	Mevalonate Kinase Deficiency - Hyper-IgD syndrome + Mevalonic aciduria	*AR*	*REDACTED*	**No**
Skeletal disorders	Bardet-Biedl syndrome type 1	*AR*	*REDACTED*	**No**
	Bardet-Biedl syndrom type 2	*AR*	*REDACTED*	**No**
	Short-rib thoracic dysplasia 3 with or without polydactyly	*AR*	*REDACTED*	**No**

Detailed Findings

Broad Disease Category	Disorder Type	Inheritance	Associated Gene(s)	Variant Detected
	Chondroectodermal dysplasia (Ellis-van Creveld syndrome)	*AR*	*REDACTED*	**No**
	Osteopetrosis, autosomal recessive type 1	*AR*	*REDACTED*	**No**
Trace element and mineral metabolism and transport disorders	Wilson disease	*AR*	*REDACTED*	**No**
Vitamin, Mineral and co-factor metabolism disorders	Hypophosphatasia, adult	*AR/AD*	*REDACTED*	**No**
	Hypophosphatasia, childhood and infantile	*AR*	*REDACTED*	**No**
	Biotinidase deficiency	*AR*	*REDACTED*	**No**
	Vitamin D-dependent rickets type 1	*AR*	*REDACTED*	**No**
	Methylmalonic aciduria with homocystinuria cblC type	*AR*	*REDACTED*	**No**
	HARP syndrome	*AR*	*REDACTED*	**No**

Recommendations

To understand the implications of the above variants, if any, please schedule a report consultation appointment with a Strand counselor by writing to **hello@strandls.com**. Subsequently, you can also take this report to a medical specialist.

Pharmacogenomics Assessment

Pharmacogenomics Assessment

The liver is the site for drug breakdown in the human body. This activity is performed by many well-known liver enzymes. By analyzing variants in the genes for these liver enzymes, we can estimate how rapidly these enzymes process several known drugs and whether a standard dose would be an overdose or an underdose. In this section, we analyze 10 of these genes and their activity on over a hundred drugs. This will aid your clinician in prescribing the right drug and dosage for you.

Key terms in the results

- Ultra-Rapid Metabolizer: Increased enzyme activity compared to rapid metabolizers
- Rapid Metabolizer: Increased enzyme activity compared to normal metabolizers but less than ultrarapid metabolizers
- Normal Metabolizer: Fully functional enzyme activity
- Intermediate Metabolizer: Decreased enzyme activity (activity between normal and poor metabolizer)
- Poor Metabolizer: Little to no enzyme activity
- Indeterminate: Allele contains a variant that has not yet been characterized

Genes tested

CYP2D6	**1, *2, *3, *4, *6, *7, *8, *9, *10, *11, *12, *15, *17, *19, *29, *35, *41*
CYP2C19	**1, *2, *3, *4, *10*
CYP2C9	**1, *2, *3, *4, *5, *6, *8, *11*
CYP3A5	**3, *6, *7*

CYP4F2	*rs2108622*
DPYD	**1, *13*
F5	*rs6025*
NUDT15	*rs116855232, *1, *2, *4*
SLCO1B1	*rs4149056*
TPMT	**1, *2, *3A, *3C, *4*

Summary Results

Therapeautic Area	Drugs to be used with standard precaution	Drugs to be used with caution	Consider alternative drug
Dermatology Oncology	Fluorouracil	-	-
Oncology	Rucaparib Thioguanine Capecitabine Mercaptopurine	Tamoxifen Gefitinib Methotrexate -	- - - -
Rheumatology	Azathioprine Carisoprodol - -	Lesinurad Piroxicam Flurbiprofen Celecoxib	- - - -
Immunosuppression and transplantation	Sirolimus Azathioprine Tacrolimus	- - -	- - -

Summary Results

Psychiatry	Doxepin	Iloperidone	-
	Trimipramine	Clozapine	-
	Fluvoxamine	aripiprazole lauroxil	-
	Risperidone	citalopram	-
	Escitalopram	Pimozide	-
	Amitriptyline	Venlafaxine	-
	Citalopram	Modafinil	-
	Clomipramine	Desipramine	-
	Imipramine	Protriptyline	-
	Sertraline	Duloxetine	-
	Paroxetine	Atomoxetine	-
	-	Trimipramine	-
	-	Thioridazine	-
	-	Brexpiprazole	-
	-	Escitalopram	-
	-	Amitriptyline	-
	-	Vortioxetine	-
	-	Imipramine	-
	-	Amphetamine	-
	-	Mirtazapine	-
	-	Doxepin	-
	-	Aripiprazole	-
	-	Pitolisant	-
	-	Amoxapine	-
	-	Clomipramine	-
	-	Perphenazine	-
	-	Nortriptyline	-
Neurology	Diazepam	Haloperidol	-
	Clobazam	Tetrabenazine	-
	Phenytoin	Donepezil	-
	Brivaracetam	meclizine	-
	-	Dextromethorphan/ Quinidine	-
	-	Phenytoin	-
	-	Deutetrabenazine	-
	-	Valbenazine	-
	-	Siponimod	-

Summary Results

Cardiology	Clopidogrel - - - - - -	Atorvastatin Simvistatin Carvedilol Propafenone Nebivolol Propranolol Metoprolol	- - - - - - -
Gastroenterology	Esomeprazole Omeprazole Lansoprazole Rabeprazole Pantoprazole Ondansetron Dexlansoprazole	Metoclopramide Dronabinol - - - - -	- - - - - - -
Gyneacology	Hormonal Contraceptives Flibanserin	Elagolix Flibanserin	- -
Infectious Disease	Voriconazole	Quinine Sulfate	-
Pain Management	- -	Oxycodone Ibuprofen	- -
Anesthesia/ Pain	- - -	Lofexidine Tramadol Codeine	Meloxicam - -
Cardiovascular	- - -	Pravastatin Acenocoumarol Flecainide	- - -
Dental	Cevimeline	-	-
Inborn Errors of Metabolism	Eliglustat	-	-

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Summary Results

Urology	Hydrocodone - - - -	Tamsulosin Mirabegron darifenacin Tolterodine Fesoterodine	- - - - -
Endocrinology	-	Rosuvastatin	-
Hematology	Avatrombopag Lusutrombopag	Warfarin -	- -

✓ The genotype is unlikely to affect the metabolism or cause any adverse effect. The medication can be prescribed to the patient in accordance with the standard guidelines and recommendations.

✗ The genotype might significantly affect the metabolism of the drug or can have an adverse effect. The patient is might not benefit from the medication. An alternate medication is recommended..

⚠ The genotype is likely to have a some effect on the metabolism of the drug or there are evidence suggesting likely adverse effect or loss of efficacy. Appropriate dosing adjustment is required while prescribing the medication. Possibility of adverse reaction may exist for few medication. Increased vigilance is required for drugs where a possibility of adverse reaction exists.

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Recommendations

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Limitation Details

As with any laboratory test, there is a small chance that this result may be inaccurate for a procedural reason, such as an error during specimen collection and labeling (incorrect patient identification), an error in processing, data collection, or interpretation. Currently available data indicates that technical error rate for analysis involving DNA tests is anywhere between 1-3%. Variants that have not been confirmed by an independent analysis could represent technical artifacts. However, our validation study (Document No. VR-017v1) showed 100% concordance between the results obtained by NGS data and Sanger sequencing (confirmation of the variant), when the supporting read fraction of the variant with at least 20 reads was >30%.

Large insertions, deletions, duplications, inversions, repeat expansions and complex rearrangements cannot be characterized accurately by NGS as it uses short-read sequencing data. Such structural variants have a much higher false-positive and false-negative rate than seen for SNVs (single nucleotide variant). It is possible that the genomic region where a disease causing variant exists in the person being tested (which may impact the phenotype) was not captured using the current technologies and therefore was not detected.

For an autosomal dominant condition, if the variant does not seem to be inherited from parents, it could be due to a de novo (new) event or due to a germline mosaicism in an unaffected parent. In case of germline mosaicism, there is a risk of the disease recurrence in the family. However, due to technical limitations of this test, germline mosaicism cannot be determined by this test. Additionally, it is possible that a particular genetic abnormality may not be recognized as the underlying cause of the genetic disorder due to incomplete scientific knowledge about the function of all genes in the human genome and the impact of variants on those genes.

Not all variants detected may be listed in the report. Inclusion of variants is dependent upon our assessment of their significance. The quality of sequencing and coverage varies between regions. Specific genomic regions, such as homopolymers, tandem repeat sequence, GCrich regions, high sequence homology, etc. influence the quality of sequencing and coverage. This may result in an occasional error in sequence reads or lack of detection of a particular genetic lesion. Accurate interpretation of this report is dependent on detailed clinical history of the patient. In the event of unavailability of detailed clinical history, the lab cannot guarantee the accuracy of the interpretation. This report is strictly not a medical diagnostic report and shall not be construed as the medical certificate or medical laboratory report.

Signature

REDACTED

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