

The Personalized Genome Report

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DOB: 06/10/62

City/State/Country of Birth on Record: Wilmington, Delaware. USA

Race/Ethnicity listed by patient: Black

Legal Disclaimer for the Patient

This Personalized Genome Report (PGR) was created from the DNA and cells you recently submitted for analysis. This PGR assumes you have read and understand the Patient Handout you were given earlier. With your DNA and cells, the testing company performed Whole Genome Sequencing and epigenome analysis of this data. This report can be used by you and your primary care physician to make healthcare decisions for you. This report has been designed to clearly explain your genome (your DNA) and epigenome and what risks you carry for many different diseases. The entire report explains information obtained from your Nuclear Genome with the exception of the Mitochondrial Genome section at the end. You can refer to the glossary located in the Patient Handout to better define unfamiliar concepts.

The information given in this report is based on analysis from thousands of research articles stretching over the last several decades. It is important to understand that the information and risk calculation in some parts of your PGR will change over time. This is due to the fact that the genetic understanding of mutations (DNA changes) is still quite limited for many disorders. Your absolute risks are based on current knowledge. As tens of thousands and millions of genomes and medical/family histories are added to the pool, the testing company will have a more complete understanding of what mutations cause what disorder. Some mutations can be detrimental (increase the risk of disease) while others can be beneficial (provide a protective measure against disease). With new information on different mutations constantly being updated, calculated risks for some disorders will change over time either up or down. The information presented in this PGR is a best guess for genetic knowledge available at the time of the report. Every new genome and history added to the pool makes the PGR more robust for the next patient. **For this reason it is recommended to get you genome re-analyzed every 6 to 12 months.**

Risk calculations may change with re-analysis, especially for some minorities. As more genomes and medical histories are added for each minority group the information in the database will be updated.

Because calculated risks can change over time, it is important that you and your doctor agree on a plan that makes sense for you today. Invasive or risky procedures to avoid disease in many cases are not recommended and should only be discussed with your doctor if your calculated risk versus the general public is very high.

Please be aware that all disease is a combination of your genetics and your environment. Your environment can have a very strong impact on your health. In some cases your environment can also

create genetic changes in certain tissues and cells of your body. Your environment can include many things such as smoking or the water you drink but can also include risk factors such as stress levels and obesity. See the Patient Handout for a more complete explanation of a person's environment. As scientific understanding of genetics improves it will be possible to incorporate risk calculations for different diseases based on your specific environmental factors. Note though that someone's environment can radically change (a patient loses fifty pounds and stops smoking), this again would impact the scores for some diseases.

Information on some diseases for which there is no meaningful intervention or treatment will **not** be part of the PGR. These include Huntington Disease and Amyotrophic Lateral Sclerosis (Lou Gehrig's Disease) and several other diseases. The full list is on the Patient Handout. This information was analyzed and is available from the testing company by special request only. For specific questions regarding this separate report contact the testing company directly.

Part 1 Broad View of Genome

46 XY No clinically significant deletions or duplications. 50X Coverage

Copy Number Variations of clinical significance: None observed

Part 2 Race

In the PGR, race is defined by your geographic ancestry, (genetically, what groups of people in the world you come from). It is common to have percentages equal to less than 100% due to centuries of relocation and mixture.

Your Race: Western Central African 62%
Eastern European Caucasian 21%
Native American (Seminole Tribe) 6%

Part 3 Carrier Status for Mendelian Disorders

"Mendelian Disorders" is a term used by geneticists for single gene disorders. There are several thousand characterized Mendelian disorders. Adults and children can be carriers for these diseases and live normal, healthy lives. As a carrier for a disease there is the potential to have offspring with that disease. It is recommended that you compare this part of the PGR with the person with which you plan to have biological children. If there is a match for a disease you should discuss this with your doctors.

The diseases listed in this part of the PGR are based on specific disease causing mutations documented in the medical literature going back several decades. Many of them are rare and may be unfamiliar to you. **A positive result in this section of the PGR is NOT expected to change and should be taken as fact.** As new documented mutations in specific families are found there is a possibility new mutations/diseases previously undocumented could be discovered and reported in future PGRs.

Autosomal Recessive traits you possess in Alphabetical Order:

If a trait listed immediately below matches your partner you have a 25% chance with each new offspring of that child having the disease. Some autosomal recessive disorders lead to early childhood death and have no cure. Most people are carriers for more than ten autosomal recessive disorders.

Alport Syndrome
Aspartoacylase Deficiency/Canavan Disease
Chediak-Higashi Syndrome
Cystic Fibrosis
Fibrodysplasia Ossificans Progressiva
Glucose-6-Phosphate Dehydrogenase Deficiency
Glycogen Storage Disease Type 2/Pompe Disease
Hemorrhagic Thrombocytopenic Purpura/Bernard-Soulier Syndrome
Litterer-Siwe Syndrome
Long Chain Hydroxyl Acyl CoA Dehydrogenase Deficiency (LCHAD Deficiency)
Mucopolysaccharidosis Type 2/ Hunter Syndrome
Phenylketonuria
Phytanic Acid Oxidase Deficiency/Refsum disease
Sickle Cell trait
Seckel Syndrome
Usher Syndrome

Autosomal Dominant traits you possess in Alphabetical Order:

These traits are dominant in the genome. If you possess a Dominant mutation you are positive for this disease and may show signs and symptoms of this disease and you have a 50% chance of passing this disease on to your biological children.

NONE

X-Linked Recessive Disorders:

Females have two X chromosomes and males have one X and one Y chromosome. Females who are carriers of X-Linked Recessive Disorders (genes with mutations on the X-chromosome) are usually not symptomatic but will have a 50% chance of each new male offspring being positive for the disease. Males with an X-linked recessive disorder are positive for the disease and may show signs and symptoms of the disease. Males with X-linked recessive disorders will also pass the mutation on to their children.

NONE

X-Linked Dominant Disorders:

These are rare disorders carried on the X chromosome. Both males and females can have these disorders as the gene involved has a dominant role. Since males only have one X chromosome they are typically more severely affected.

NONE

Y-Linked Disorders:

These are disorders only on the Y chromosome.

NONE

Part 4 ICD-9 Codes/Documented Drug Allergies

Here are the ICD-9 codes (diagnosis codes) that your physician included on the intake form when you submitted your DNA. These codes are medical conditions that were diagnosed by your physician independently of this DNA test. Purely environmental diseases (ex: affliction with a virus) have been omitted by the testing company. The ICD-9 codes which follow have been analyzed in conjunction with your DNA in efforts to obtain a medical history.

This information will remain anonymous and will help scientists better understand what medical conditions are caused by which DNA changes.

272 Hypercholesterolemia

340 Multiple Sclerosis

477 Allergic Rhinitis

701.4 Keloid

724.2 Low Back Pain

Documented Drug Allergies: Morphine causes a hives-like skin reaction

Part 5 Complex Gene Disorders

Complex Gene Disorders are disorders that occur due to mutations/changes in several genes or in some cases, hundreds of genes. Changes in genes can also interact with one's environment to cause disease. Absolute risk is the percent chance of someone with your genome acquiring a disease. The easiest way to understand your risk for a disease is to ask what your **absolute risk** of the disease is in comparison to the absolute risk of the average person. If you would like to find out which genes/mutations were included to calculate these risks, we recommend you contact the testing laboratory. Also, different ethnic backgrounds, and their genetic effects on risk for different disorders can come into play. Absolute risk is different than relative risk or attributed risk.

All of us have risks for most diseases that are equal or close to risks of the general population. Hence, the decision was made to list specifically which diseases your genome indicates you would be 50% more likely or less likely to have versus the general population. The information in this section **is expected to change significantly in the future** as our understanding of the contribution of genetic determinants of complex disorders matures. This PGR provides you with the best available evidence using information as of the date of the report. Percentages may go up or down for any disease as our understanding of harmful or beneficial mutations and how they relate to disease increases. It is therefore recommended

that you obtain a new issue of the PGR at least once per year to ensure the health and wellness program established with your primary care physician is current and up to date. As more genomes have been sequenced and clinical histories added to the information database, a clearer picture of risks for each person will emerge.

It is important to understand that a very high risk of disease (90% lifetime risk) does not mean that you will get the disease. As well, a very low risk of disease (5% lifetime risk) does not mean that you will not get the disease. The easiest way to think about this is to say "if one-hundred people with my exact genome were in a room, ninety of them would get the disease I am at very high risk for and five of them would get the disease I am at very low risk for".

Diseases which you have a 50% chance or more of having in your lifetime versus the general public:

The first number is the risk for the general population and the second number is your risk. The third number is how many times more likely you are to get the disease versus the general population. In the third column 1.0 would equal the risk for the general population.

Prostate Cancer-Males	11%	72%	6.5
Sarcoidosis	1.3%	8%	6.2
Venous Thromboembolism	1.4%	8.2%	5.9
Keloids	4%	21%	5.3
Ankylosing Spondylitis	0.9%	4.6%	5.1
Alcohol Dependence	10%	48%	4.8
Osteoarthritis	14%	49%	3.5
Esophageal Cancer	0.4%	1.1%	2.8
Asthma	6%	15%	2.5
Kidney Disease	9%	17.9%	2.0
Gout	1.7%	2.6%	1.5

Diseases which you have a 50% chance or less of having in your lifetime versus the general public:

The first number is the risk for the general population and the second number is your risk. The third number is how many times less likely you are to get the disease versus the general population. In the third column 1.0 would equal the risk of the general population.

Melanoma	0.5%	0.06%	8.3
Basal Cell Carcinoma	1.2%	0.2%	6.0

Squamous Cell Carcinoma	1.1%	0.3%	3.7
Obesity	20%	6.8%	2.9
High Blood Pressure	22%	9%	2.4
Lung Cancer	2%	1%	2.0
Schizophrenia	4%	2.5%	1.6

List of Diseases in alphabetical order that were also correlated with your genome and found to have risks close to that of the general population.

Abdominal Aortic Aneurysm	Migraines
Age-related Macular Degeneration	Multiple Sclerosis
Alopecia Areata	Narcolepsy
Alzheimer's Disease	Nasopharyngeal Carcinoma
Anti-Social Personality Disorder	Neural Tube Defects
Arrhythmias	Neuroblastoma
Atopic Dermatitis	Nicotine Dependence
Atrial Fibrillation	Nonalcoholic Fatty Liver Disease
Attention-Deficit Hyperactivity Disorder	Obsessive-Compulsive Disorder
Behcet's Disease	Oral and Throat Cancer
Bipolar Disorder	Otosclerosis
Bladder Cancer	Page's Disease of the Bone
Brain Aneurysm	Pancreatic Cancer
Breast Cancer	Parkinson's Disease
Celiac Disease	Peripheral Arterial Disease
Chronic Lymphocytic Leukemia	Primary Biliary Cirrhosis
Chronic Obstructive Pulmonary Disease	Psoriasis
Cleft Lip and Cleft Palate	Renal Artery Stenosis
Cluster Headaches	Restless Legs Syndrome
Colorectal Cancer	Rheumatoid Arthritis
Coronary Heart Disease	Scleroderma
Creutzfeldt-Jakob Disease	Stomach Cancer
Crohn's Disease	Stroke
Degenerative Disc Disease	Systemic Lupus Erythematosus
Developmental Dyslexia	Tardive Dyskinesia
Essential Tremor	Thyroid Cancer
Follicular Lymphoma	Tourette's Syndrome
Gallstones	Type 1 Diabetes Mellitus
Generalized Vitiligo	Type 2 Diabetes Mellitus
Glaucoma	Ulcerative Colitis
Hashimoto's Thyroiditis	Whipple's Disease
Hip Dysplasia	Wolf-Parkinson-White Syndrome
Hodgkin's Lymphoma	xerosis
Hypertriglyceridemia	xerostomia
Kidney Cancer	

Part 6 Food Allergies

This list may change with re-analysis. Again, this is based on best and current information.

NONE

Part 7 Environmental Allergies

This list may change with re-analysis. Again, this is based on best and current information.

Cats, Dust Mites

Part 8 Drug Allergies/Sensitivities

This section is based on current evidence regarding your response to various drugs. By nature of the types of enzymes people possess, they can be fast metabolizers or slow metabolizers of certain drugs. This information can guide doctors to the correct dosage you may best response to certain drugs. For some drugs there also exists information on the potential for drug allergies. If present, these are also listed in bold type. Note, allergies for some drugs can be life threatening. This information will be amended in future PGR's as more genomes are analyzed.

Beta-Blocker Response: slow metabolizer, you would require less of this drug for the same effect

Warfarin Response: fast metabolizer, you would require more of this drug for the same effect

Penicillin Response: ****Allergy Suspected** You may experience nausea, vomiting and a hives-like skin reaction to this medication. NOTE: This allergy may exist for all "cillin" family of antibiotics.**

Genes leading to metabolism of other drugs were analyzed but were not found to be significantly different than the general population. These are mentioned below:

Antidepressant Response
Clopidogrel Response
Metformin Response
Naltrexone Response
Statin Response

Part 9 Your Mitochondrial Genome

All of us have a second genome outside of the nucleus called the Mitochondrial Genome. This is a very small genome and is used to help produce large amounts of energy or ATP in the cells. Mutations or errors in this genome can lead to significant or serious disease in some patients. Your mitochondrial genome was analyzed and is presented here. This information may change in future PGRs.

No clinically significant duplications or deletions. No clinically significant mutations.

END OF REPORT
