



## 23andMe Genetic Health Overview

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### What this overview includes

This overview includes brief summaries of your 23andMe results for:

- diseases for which you are at greater than average genetic risk,
- heritable diseases for which you carry one or more genetic variants (carrier status),
- and drugs to which you are likely to have an atypical response based on genetics.

These results are based on your genetic data and any sex and ancestry information you have provided along with population-level risk data for specified age ranges. They do not take into account non-genetic factors, family history, or additional genetic factors that may influence these conditions. Only results for genetic associations that are scientifically well established are included. This overview does not provide details regarding diseases for which you are at typical or lower than average genetic risk, heritable diseases for which you aren't known to carry a variant, or drugs to which you are likely to have a typical response. If you would like more information on any of your 23andMe results, please go to that topic's individual report page on our website at <https://www.23andme.com/you/health/>.

## Overview of Genetic Health






Petar Ivanov  
Year of Birth: 1989  
Eastern European

Disease risk results are included in this overview only if your risk based on genetics is greater than 1%. Note that certain conditions may have genetic information applicable only to specific populations.

*Components of this test were performed in a clinical laboratory regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) to perform high-complexity testing. The data provided are intended for informational and educational use and are not for diagnostic use.*

*\*All conditions tested are listed at the end of the report. You may not have data for every report.*

Disease risk	Your risk	Average risk
Restless Legs Syndrome	  2.5%	2.0%
29 conditions*	  Typical or decreased risk	

Carrier status	Status
Cystic Fibrosis	Variant Present
47 heritable conditions*	Variant Absent

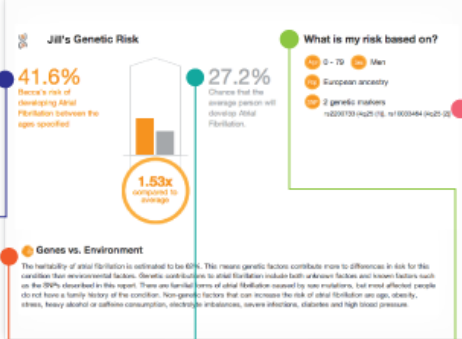
Drug response	Response
8 other drugs*	Typical Response

## How to read your reports

## Disease Risk reports

1 \_\_\_\_\_

2



**Average risk:** The probability of a person with the listed ancestry from the general population developing the disease between the ages specified.

Genetic variants (and the genes they're located in or near) used to determine your results for this condition.

Your genetic risk is based on the sex and ancestry settings in your profile and your genotype(s) for a specific set of genetic markers. Your risk does not take into account non-genetic factors, family history, or additional genetic factors that may contribute to risk for this disease.

## Carrier Status reports

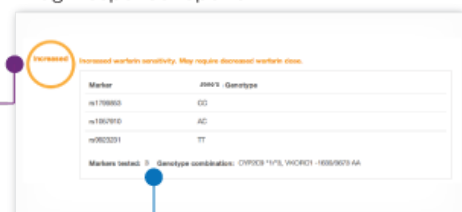
6



Details of the specific genetic marker(s) for which you carry a mutation linked to the condition. “Coverage” indicates the percentage of mutations for this disease that are covered by this test in specific populations. Other mutations not covered by this test may also be linked to the condition.

## Drug Response reports

8



Details of the specific genetic markers tested, the genes in which those markers are located, your genotypes at those markers and other names for your combination of genotypes if any.

## Restless Legs Syndrome

Imagine what it would be like to crawl into bed every night, ready to catch some much-needed Zs, only to be struck by an irrepressible urge to move your legs as soon as you began to relax. No matter how tired you were, instead of drifting off peacefully, you would be compelled to get up and move around. It may sound crazy, but this is exactly the situation people with restless legs syndrome (RLS) experience. Though the symptoms in many people are milder, it is estimated that about 4% of the U.S. population suffers from this puzzling disorder.

### Petar's Genetic Risk

2.5%

Petar's risk of developing Restless Legs Syndrome between the ages specified



2.0%

Chance that the average person will develop Restless Legs Syndrome

### What is my risk based on?

- Age** 0 - 79
- Sex** Men
- Pop** European ancestry
- SNP** 1 genetic markers  
rs3923809 (BTBD9)

### Genes vs. Environment

The heritability of restless legs syndrome is estimated to be 54%. This means that genetic and environmental factors contribute nearly equally to differences in risk for this condition. Genetic factors that play a role in restless legs syndrome include both unknown factors and known factors such as the SNPs we describe here. Environmental factors include pregnancy. Low iron levels, dialysis for end-stage renal disease, and damage to the nerves of the hands and feet tend to worsen the condition.

### Additional Information

#### Other Medical Conditions

[Chronic diseases](#) such as kidney failure, diabetes, Parkinson's, and peripheral neuropathy can exacerbate symptoms of RLS. If you have RLS, your health care provider may work with you to manage these conditions to reduce your symptoms. [Pregnancy](#) can sometimes trigger symptoms of RLS. If this happens, the symptoms will usually disappear once the pregnancy is completed.

#### Lifestyle Factors

- **Limit caffeine, alcohol, and tobacco use:** Caffeine, alcohol, and tobacco intake can trigger or aggravate symptoms in predisposed individuals.
- **Get enough iron:** Insufficient iron levels can also trigger or aggravate symptoms.

#### Medications and Treatment

Taking certain drugs can sometimes cause symptoms of RLS. These symptoms usually disappear once the drug regimen is stopped. Your health care provider can work with you to manage drug regimens that may be triggering RLS.

View the full report online for links to resources, references, and more detailed genetic results and information.

## Carrier status: Cystic Fibrosis

Cystic fibrosis (CF) is a serious disease caused by mutations in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene that affect the ability of cells lining the inner surfaces of organs to properly transport salt and water. Most significantly, these mutations can cause mucus to build up in the lungs, which leads to infections and damage. There are also effects on the pancreas and reproductive system. A person must inherit a mutated copy of the gene from each parent to develop the disease. Approximately 30,000 children and adults in the United States (70,000 worldwide) are living with CF and an estimated eight million people carry one copy of a CFTR mutation.

### Petar's Genetic Results

**Variant  
Present**

**Has one copy of the DeltaF508 mutation in the CFTR gene. A person with one copy of this mutation typically does not have cystic fibrosis, but may pass the mutation to offspring.**

Gene	Variant	DNA change	Petar's genotype
CFTR	DeltaF508	CTT to —	—,CTT
<b>Markers tested:</b> 31 <b>Coverage:</b> 85-97% in individuals with European ancestry; 50-75% in individuals with African American, Hispanic, and Asian ancestry			

### What does this test cover?

More than 500 mutations in the CFTR gene causing cystic fibrosis have been documented. 23andMe reports data for 31 of the most common mutations in various populations, including the 23 mutations recommended for testing by the American College of Medical Genetics. Some mutations that have been identified in populations outside northern Europe are not included in this report.

### How is Cystic Fibrosis inherited?

Cystic fibrosis is inherited in a recessive manner, meaning that only a child who receives two mutated copies of the CFTR gene (one from each parent) will develop the disease.

### How common is this condition?

Approximately one in 3,200 babies of European descent is born with cystic fibrosis. Incidence is lower in non-European populations—for example, about one in 15,000 African American babies and one in 31,000 Asian American babies is born with cystic fibrosis. One out of 29 Caucasians, including Ashkenazi Jews, carries one copy of a CFTR mutation. In African Americans and Hispanic Americans the carrier rates are one in 65 and one in 46, respectively. CFTR mutations are rarer in people with Asian ancestry—the carrier rate is about one in 90.

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### Additional Information

#### *Other Medical Conditions*

In men, mutations in CFTR can cause a condition known as “congenital bilateral absence of the vas deferens” (CBAVD) leading to infertility. Different CFTR mutations are also associated with different types of symptoms and varying severity of disease. If you are concerned about CBAVD or cystic fibrosis, please consult your health care provider or a genetic counselor.

#### *Medications and Treatment*

In most cases, treatment for CF focuses on relieving symptoms. There is also a drug that can be used specifically to treat CF patients with a particular mutation called G551D.















































*View the full report online for links to resources, references, and more detailed genetic results and information.*















## Petar Ivanov's results for all conditions tested by 23andMe

Conditions and diseases tested by 23andMe: This list is continually expanding as new genetic associations are discovered and reported. Please visit our website at <https://www.23andme.com/health/all/> to view the most up-to-date list of conditions tested by 23andMe.

### About Risk Estimates:

23andMe reports results as genotype-specific incidence, which is an estimate of how many individuals in a population composed of people with a customer's genotype are expected to be diagnosed with a condition given a specified ancestry and age range. These estimates are based on well-established genetic associations reported in the biomedical literature and do not account for non-genetic factors, family history, or additional genetic factors that may modify a customer's risk. The genotype-specific incidence estimate combines the odds for a condition for a customer's genotypes at a set of SNPs with data about disease incidence. For more information on how 23andMe calculates these estimates, please see our technical papers available at <https://www.23andme.com/howitworks/>.

Disease risk (30)	Your risk	Average risk
Restless Legs Syndrome	  2.5%	2.0%
Esophageal Squamous Cell Carcinoma (ESCC)	  0.4%	0.4%
Stomach Cancer (Gastric Cardia Adenocarcinoma)	  0.3%	0.2%
Age-related Macular Degeneration	  Typical risk	
Atrial Fibrillation	  Typical risk	
Bipolar Disorder	  Typical risk	
Breast Cancer	  Typical risk	
Chronic Kidney Disease	  Typical risk	
Colorectal Cancer	  Typical risk	
Coronary Heart Disease	  Typical risk	
Gallstones	  Typical risk	
Lung Cancer	  Typical risk	
Lupus (Systemic Lupus Erythematosus)	  Typical risk	
Obesity	  Typical risk	
Scleroderma (Limited Cutaneous Type)	  Typical risk	
Type 2 Diabetes	  Typical risk	
Ulcerative Colitis	  Typical risk	
Venous Thromboembolism	  Typical risk	
Alzheimer's Disease	  Decreased risk	
Celiac Disease	  Decreased risk	
Crohn's Disease	  Decreased risk	
Exfoliation Glaucoma	  Decreased risk	
Melanoma	  Decreased risk	

Disease risk (30)	Your risk	Average risk
Multiple Sclerosis	  Decreased risk	
Parkinson's Disease	  Decreased risk	
Primary Biliary Cirrhosis	  Decreased risk	
Prostate Cancer	  Decreased risk	
Psoriasis	  Decreased risk	
Rheumatoid Arthritis	  Decreased risk	
Type 1 Diabetes	  Decreased risk	

**About Carrier Status:**

23andMe tests for specific genetic variants that are strongly linked to a number of inherited genetic conditions. These variants are typically the most common ones linked to the condition. Certain variants may be more common in certain populations than others. The absence of specific variants does not rule out the possibility that a customer may carry another variant linked to the condition.

Carrier status (48)	Status
Cystic Fibrosis	Variant Present
ARSACS	Variant Absent
Agnesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)	Variant Absent
Alpha-1 Antitrypsin Deficiency	Variant Absent
Autosomal Recessive Polycystic Kidney Disease	Variant Absent
BRCA Cancer Mutations (Selected)	Variant Absent
Beta Thalassemia	Variant Absent
Bloom's Syndrome	Variant Absent
Canavan Disease	Variant Absent
Congenital Disorder of Glycosylation Type 1a (PMM2-CDG)	Variant Absent
Connexin 26-Related Sensorineural Hearing Loss	Variant Absent
D-Bifunctional Protein Deficiency	Variant Absent
DPD Deficiency	Variant Absent
Dihydrolipoamide Dehydrogenase Deficiency	Variant Absent
Factor XI Deficiency	Variant Absent
Familial Dysautonomia	Variant Absent
Familial Hypercholesterolemia Type B	Variant Absent
Familial Hyperinsulinism (ABCC8-related)	Variant Absent
Familial Mediterranean Fever	Variant Absent
Fanconi Anemia (FANCC-related)	Variant Absent
G6PD Deficiency	Variant Absent



Carrier status (48)	Status
GRACILE Syndrome	Variant Absent
Gaucher Disease	Variant Absent
Glycogen Storage Disease Type 1a	Variant Absent
Glycogen Storage Disease Type 1b	Variant Absent
Hemochromatosis (HFE-related)	Variant Absent
Hereditary Fructose Intolerance	Variant Absent
Hypertrophic Cardiomyopathy (MYBPC3 25bp-deletion)	Variant Absent
LAMB3-related Junctional Epidermolysis Bullosa	Variant Absent
Leigh Syndrome, French Canadian Type (LSFC)	Variant Absent
Limb-girdle Muscular Dystrophy	Variant Absent
Maple Syrup Urine Disease Type 1B	Variant Absent
Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency	Variant Absent
Mucopolidosis IV	Variant Absent
Neuronal Ceroid Lipofuscinosis (CLN5-related)	Variant Absent
Neuronal Ceroid Lipofuscinosis (PPT1-related)	Variant Absent
Niemann-Pick Disease Type A	Variant Absent
Nijmegen Breakage Syndrome	Variant Absent
Pendred Syndrome	Variant Absent
Phenylketonuria	Variant Absent
Primary Hyperoxaluria Type 2 (PH2)	Variant Absent
Rhizomelic Chondrodysplasia Punctata Type 1 (RCDP1)	Variant Absent
Salla Disease	Variant Absent
Sickle Cell Anemia & Malaria Resistance	Variant Absent
Tay-Sachs Disease	Variant Absent
Torsion Dystonia	Variant Absent
Tyrosinemia Type I	Variant Absent
Zellweger Syndrome Spectrum	Variant Absent

**About Drug Response:**

23andMe displays your likely response to a number of drugs based on genetic variants associated with differences in response. These may be differences in sensitivity, in the likelihood or severity of side

Drug response (8)	Response
Abacavir Hypersensitivity	Typical
Alcohol Consumption, Smoking and Risk of Esophageal Cancer	Typical

effects, or differences in disease risk tied to use of a drug. Only a medical professional can determine whether a drug is right for a particular patient. The information contained in this report should not be used to independently establish a drug regimen, or abolish or adjust an existing course of treatment.

Drug response (8)	Response
Clopidogrel (Plavix®) Efficacy	Typical
Fluorouracil Toxicity	Typical
Oral Contraceptives, Hormone Replacement Therapy and Risk of Venous Thromboembolism	Not Applicable
Pseudocholinesterase Deficiency	Typical
Response to Hepatitis C Treatment	Typical
Warfarin (Coumadin®) Sensitivity	Typical

## References

### Restless Legs Syndrome

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### About the 23andMe Personal Genome Service®

23andMe's Personal Genome Service provides customers with data on nearly 1,000,000 single nucleotide polymorphisms (SNPs) in their genome using a microarray-based genotyping assay. Customers provide saliva samples, which are analyzed by a CLIA-certified laboratory. Results are viewable on the 23andMe website at <https://www.23andme.com/you/> where reports are considered Established or Preliminary Research reports depending on the amount of evidence supporting the associations reported. We currently provide more than 60 Established Research reports on various disease risk, drug response, and carrier status topics, as well as Preliminary Research reports on more than 150 conditions and traits.