# Parkinson's Disease

Parkinson's disease is characterized by tremor, muscle stiffness, and problems with movement. Many factors, including genetics, can influence a person's chances of developing Parkinson's disease. This test includes two genetic variants associated with increased risk of developing the condition.

Overview Scientific Details Frequently Asked Questions

## Jamie, you have **one** of the two genetic variants we tested.

People with this variant have an increased risk of developing Parkinson's disease. Your risk may also be influenced by other factors.


## How To Use This Test

**This test does not diagnose Parkinson's disease or any other health conditions.**

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Genetic Health Risk tutorial
See Scientific Details
See Frequently Asked Questions

### Intended Uses

- Tests for the **G2019S** variant in the LRRK2 gene and the **N370S** variant in the GBA gene associated with an increased risk of developing Parkinson's disease.

### Limitations

- Does **not** test for all possible variants or genes associated with Parkinson's disease.
- Does **not** test for any variants or genes linked to early-onset or young-onset Parkinson's disease.

### Ethnicity Considerations

- The variants included in this test are most common and best studied in people of **European**, **Ashkenazi Jewish**, and **North African Berber** descent.

## You may have an **increased risk** of developing Parkinson's disease based on your genetic result.

However, many people with this variant do not develop Parkinson's disease. Consider discussing your risk with a healthcare professional, especially if you have a family history or other risk factors for this condition.

We detected the G2019S variant in the LRRK2 gene.

See Scientific Details

### Although your risk may be increased, many people with this variant do not develop Parkinson's disease.

Studies in people of **European** and **Ashkenazi Jewish** descent have shown that about 25% of individuals with this variant develop Parkinson's disease during their lifetime. This variant has only been studied in a few ethnicities.

See Scientific Details

### Other factors may also influence your risk of developing Parkinson's disease.

It's important to know your family history. Your risk may be higher if Parkinson's disease runs in your family.

Even though nothing has been proven to prevent Parkinson's disease, research is ongoing to understand what causes this condition and the potential benefits of lifestyle factors, such as physical activity. See Resources for more information.

## Environment and other factors can also influence the chances of developing Parkinson's disease.

Consult with a healthcare professional before making any major lifestyle changes.

**Age**

The risk of developing Parkinson's disease increases as a person ages. This condition is most often diagnosed in people over the age of 55.

See Scientific Details for more information


## About Parkinson's Disease

### When it develops

Parkinson's disease typically develops in adulthood, after 55 years of age.

### Typical signs and symptoms

- Tremor
- Muscle stiffness
- Slow movements
- Problems with balance
- Memory loss in some cases

### How common is the condition?

Parkinson's disease affects people of all ethnicities. About 1-2% of people will develop Parkinson's disease during their lifetime.

### How it's treated

There is currently no known prevention or cure for Parkinson's disease. Certain medications may be used to delay or ease symptoms. Speech, physical, and occupational therapies may also help with symptom management.

**Read more at:** Mayo Clinic˅ National Institute of Neurological Disorders and Stroke˅ GeneReviews˅ The Michael J. Fox Foundation for Parkinson's Research˅

## It is important to discuss this result with a healthcare professional.

If you have a family history of this condition or think you have symptoms, consult with a healthcare professional.

Print report

If you have questions about your results or how they might affect you or your family, a genetic counselor may be able to help.

Learn more

See our Frequently Asked Questions for more information.

FAQs

# Parkinson's Disease

Parkinson's disease is characterized by tremor, muscle stiffness, and problems with movement. Many factors, including genetics, can influence a person's chances of developing Parkinson's disease. This test includes two genetic variants associated with increased risk of developing the condition.

Overview Scientific Details Frequently Asked Questions

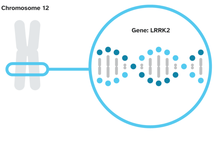
## Parkinson's disease is associated with variants in many genes.

This report includes one variant in the LRRK2 gene and one variant in the GBA gene. This test does not cover variants in other genes associated with Parkinson's disease.

LRRK2 GBA

The LRRK2 gene contains instructions for making an enzyme called leucine-rich repeat kinase 2, also called dardarin. This enzyme interacts with other proteins to turn them on and off. Certain variants in the LRRK2 gene cause this enzyme to be too active, which prevents it from properly controlling other proteins.

**Read more at Genetics Home Reference ˅**




## You have one of the two genetic variants we tested.

Variants Detected | View All Tested Markers

| Marker Tested | Genotype* | Additional Information |
|---|---|---|
| **G2019S**<br>**Gene:** LRRK2<br>**Marker:** rs34637584 | **A**<br>Variant copy from one of your parents<br>**G**<br>Typical copy from your other parent | ˅ **Biological explanation**<br>˅ **Typical vs. variant DNA sequence(s)**<br>˅ **Percent of 23andMe customers with variant**<br>˅ **References** [ 4, 9, 10, 12, 13, 14, 21, 22, 23, 24, 25, 36 ] \| ClinVar ˅ |

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

## Test Interpretation

This report provides risk estimates for people of European, Ashkenazi Jewish, and North African Berber descent. Estimates for other ethnicities are not currently available.

### Health Risk Estimates

Risk estimates are based on clinical studies that identify an association between a genotype and a health condition.

Risk estimates are not available for people who have two copies of the G2019S variant or a combination of the two tested variants. An interpretation of "increased risk" is provided to people with these results because it is likely that their risk is at least as high as for people with one copy of the G2019S variant alone.

Consider talking to a healthcare professional if you have any concerns about your results.

**References** [ 1, 4, 7, 8, 14, 15, 19, 29, 25, 30, 36 ]

**Lifetime risk** | Likelihood ratios

The lifetime risk estimates shown below represent the proportion of people expected to develop Parkinson's disease by age 89. These values are based on studies conducted in people of European, Ashkenazi Jewish, and North African Berber descent. In the general population, about 1-2% of people will develop Parkinson's disease during their lifetime, typically after the age of 55. However, some studies report that among the general population, the lifetime risk for Parkinson's disease may be approaching 4%.

| Genotype | Ashkenazi Jewish | European | North African Berber |
|---|---|---|---|
| One copy of G2019S variant | 26% | 24% | 91% |
| One copy of N3768S variant | 5.9% | Slightly increased risk (exact estimate not available) | Not available |
| Two copies of N3768S variant | 9.1% | Not available | Not available |

## Other Factors

Other factors besides the variants included in this test can influence your chances of developing Parkinson's disease.

This is not a complete list of other factors.

People with multiple risk factors may have a higher risk of developing Parkinson's disease.

Consult with a healthcare professional before making any major lifestyle changes.

| Other Factors | References |
|---|---|
| **Age**<br>The risk of developing Parkinson's disease increases as a person ages. This condition is most often diagnosed in people over the age of 55. | [ 5 ] |
| **Sex**<br>Males have a higher chance of developing Parkinson's disease than females. The exact reason for this difference between the sexes is unknown. Researchers have proposed that it may be due to higher rates of occupation-related head injury and chemical exposure in males, or that females may be protected by higher estrogen levels. | [ 5, 27, 38 ] |
| **Family history**<br>First-degree relatives of an individual with Parkinson's disease have a higher chance of developing Parkinson's disease themselves. This may be primarily explained by genetic factors, but could also be explained by family members sharing a similar lifestyle and environment. | [ 5, 17, 29 ] |
| **Exposure to certain chemicals**<br>Exposure to certain chemicals increases the risk of developing Parkinson's disease. These include chemicals used in agriculture, such as pesticides, as well as chemicals used in industrial settings, such as metal degreasing and dry cleaning agents. | [ 5, 26, 29, 34 ] |
| **Physical activity**<br>People who engage in physical activity have a lower risk of developing Parkinson's disease compared to people who rarely or never do. This effect is strongest in people who report engaging in moderate or vigorous physical activity. Some studies suggest that the effect is stronger for males than it is for females. | [ 3, 35, 39 ] |
| **Head injury**<br>Some studies suggest an association between certain types of head injury and increased risk of developing Parkinson's disease later in life. Head injury that results in prolonged loss of consciousness appears to have the strongest association. Some researchers think that this might be due to inflammation in the brain after the injury. | [ 6, 16 ] |
| **Other genes**<br>There are other genes and variants that have been linked to Parkinson's disease. However, most of these variants are either rare or have only a small effect on risk. | [ 5 ] |
| **Other health conditions**<br>People with inflammatory bowel disease (IBD) have an increased risk of developing Parkinson's disease. This may be because intestinal inflammation caused by IBD may eventually lead to inflammation within the central nervous system and cause cells in the central nervous system to stop working and die. | [ 31, 37 ] |

## Test Details

### Indications for Use

The 23andMe PGS Genetic Health Risk Report for Parkinson's Disease is indicated for reporting of the G2019S variant in the LRRK2 gene and the N3768S variant in the GBA gene. This report describes if a person's genetic result is associated with an increased risk of developing Parkinson's disease, but it does not describe a person's overall risk of developing Parkinson's disease. This report is most relevant for people of European, Ashkenazi Jewish, and North African Berber descent.

**Special Considerations**

- Genetic testing for Parkinson's disease is not currently recommended by any healthcare professional organizations.

### Test Performance Summary

**Clinical Performance** [ 5, 11, 16, 30, 33 ]

The genes and variants included in this report only represent a subset of all those that have been associated with Parkinson's disease. The two variants tested are associated with an increased risk of developing Parkinson's disease. However, many people who have one or both of these variants do not develop the condition. In addition, most cases of Parkinson's disease do not have a genetic cause.

- 1-2% of people with Parkinson's disease have the G2019S variant in the LRRK2 gene. Among people with familial Parkinson's disease, 5-7% have this variant.
- 8-14% of people with Parkinson's disease have a variant in the GBA gene. The N3768S variant accounts for roughly half of those cases.

**Analytical Performance**

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

### Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

## References

1. Alcalay RN et al. (2014). "Comparison of Parkinson risk in Ashkenazi Jewish patients with Gaucher disease and GBA heterozygotes." JAMA Neurol. 71(6):752-7. ˅
2. Asselta R et al. (2014). "Glucocerebrosidase mutations in primary parkinsonism." Parkinsonism Relat Disord. 20(11):1215-26. ˅
3. Chen H et al. (2005). "Physical activity and the risk of Parkinson disease." Neurology. 64(4):664-9. ˅
4. Clark LN et al. (2006). "Frequency of LRRK2 mutations in early- and late-onset Parkinson disease." Neurology. 67(10):1786-91. ˅
5. Cook Shukla L et al. (2004). "Parkinson Disease Overview." [Accessed Aug 25, 2020]. ˅
6. Crane PK et al. (2016). "Association of Traumatic Brain Injury With Late-Life Neurodegenerative Conditions and Neuropathologic Findings." JAMA Neurol. 73(9):1062-9. ˅
7. Elbaz A et al. (2002). "Risk tables for parkinsonism and Parkinson's disease." J Clin Epidemiol. 55(1):25-31. ˅
8. Gan-Or Z et al. (2015). "Differential effects of severe vs mild GBA mutations on Parkinson disease." Neurology. 84(9):880-7. ˅
9. Garcia-Miralles M et al. (2015). "No dopamine cell loss or changes in cytoskeleton function in transgenic mice expressing physiological levels of wild type or G2019S mutant LRRK2 and in human fibroblasts." PLoS One. 10(4):e0118947. ˅
10. Goldwurm S et al. (2011). "Kin-cohort analysis of LRRK2-G2019S penetrance in Parkinson's disease." Mov Disord. 26(11):2144-5. ˅

See all references ˅

## Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

| Date | Change |
|---|---|
| April 17, 2017 | Parkinson's Disease report created. |

# Parkinson's Disease

Parkinson's disease is characterized by tremor, muscle stiffness, and problems with movement. Many factors, including genetics, can influence a person's chances of developing Parkinson's disease. This test includes two genetic variants associated with increased risk of developing the condition.



## Parkinson's Disease

What does this test do?

What does this test **not** do?

The report says the variants included in this test are most common and best studied in people of **European**, **Ashkenazi Jewish**, and **North African Berber** descent. What if I'm not of European, Ashkenazi Jewish, or North African Berber descent?

Where can I learn more about Parkinson's disease, support groups, and other resources?

My report says **one variant** called **G2019S** in the LRRK2 gene was detected. What does this mean?

What does **increased risk** mean?

My report says **one variant** called **G2019S** in the LRRK2 gene was detected. What are some things I could do?

How could my result affect my family?

