

## **GBA1 Gene Change in People of African Descent** *Frequently Asked Questions*

In August 2023, scientists from the Global Parkinson's Genetics Program (GP2) published a paper in *The Lancet Neurology*. The paper announces the discovery of a genetic variant that increases risk of Parkinson's disease in people of African and African admixed populations. The following frequently asked questions were developed for the Parkinson's community by The Michael J. Fox Foundation and GP2. You can learn more about this finding, *GBA1* and Parkinson's genetics on The Michael J. Fox Foundation website at [michaeljfox.org](https://michaeljfox.org).

### **Parkinson's & Genetics**

#### **Is Parkinson's genetic?**

Genes are passed down from parents. Differences in our genes make us who we are from hair color to height. Some changes in genes (a gene variant) increase the risk of having a disease.

Having a gene variant linked to Parkinson's does not mean you will absolutely develop disease. Scientists believe that genes, environment and aging together cause Parkinson's disease. Factors such as exposure to pesticides or toxins or a head injury can also raise risk.

Knowing more about genes can help develop better tests and new treatments for diseases.

Learn more at [michaeljfox.org/news/parkinsons-genetics](https://michaeljfox.org/news/parkinsons-genetics).

#### **Where can I learn more about genetics and Parkinson's?**

Genetic counselors can talk to you more about genetics and risk of disease. Because the *GBA1* finding is so new, they may not yet be aware of this finding. They can talk to you generally about genetic risk and what to consider.

Your doctor may be able to refer you to a genetic counselor. You also can find one on The National Society of Genetic Counselors website. Visit [findageneticcounselor.nsgc.org](https://findageneticcounselor.nsgc.org).

## **GBA1 Finding**

### **What is the new *GBA1* finding?**

The Global Parkinson's Genetics Program (GP2) study found a genetic variant in some people with Parkinson's disease. This is a variant of the *GBA1* gene. This change was found in people with African ancestry.

### **What is the GP2 study?**

The Global Parkinson's Genetics Program (GP2) studies genetics across global Parkinson's groups. This *GBA1* finding is an example of its aim to unite data across countries and communities toward new discoveries. The finding is from analysis of samples shared by people in the U.S. and in Nigeria, primarily.

GP2 is led by scientists at the U.S. National Institutes of Health. It partners with scientists and volunteers around the world. GP2 is a program of the Aligning Science Across Parkinson's (ASAP) initiative. The Michael J. Fox Foundation is a partner in GP2.

### **Why is this an important finding?**

This is an important but early step in understanding causes of Parkinson's in people of African descent. Right now, this finding does not impact care. In the future, you may be able to help test new treatments if you have this *GBA1* variant.

This finding also shows the importance of partnering with people of many backgrounds. Most genetics studies have been in people of European descent. This finding shows scientists have more to learn by working with diverse groups.

### **Will having this *GBA1* gene change impact my Parkinson's care?**

Right now, it will not change your Parkinson's care. In the future you may be able to enroll in certain studies looking at the *GBA1* pathway.

As scientists learn more about this variant, there may be other impacts. Some gene variants may be linked to faster or slower progression or to certain symptoms. That information could help doctors monitor disease more closely. It may help you make lifestyle choices as well. You may decide to exercise more or change your diet.

## **GBA1 and Parkinson's Disease**

### **What is the *GBA1* gene?**

The *GBA1* gene tells the body how to make the protein beta-glucocerebrosidase or GCase. This protein plays a role in the cell's "recycling center." This part of a cell removes damaged or faulty cell parts.

*GBA1* gene variants can reduce GCase activity. This means the cell's recycling center doesn't work correctly.

Without that function, damaged or faulty cell parts can build up, harming the cell. Scientists believe this build-up may lead to the cell death that causes Parkinson's symptoms in individuals with a *GBA1* change, but more research is needed.

### **Can doctors tell me if I have this *GBA1* change?**

Right now, your doctor cannot tell you if you have this variant. There are tests that share information on other changes in the *GBA1* gene. (Not all genetic tests look for all variants.)

Scientists can now learn more about this newly discovered *GBA1* variant. Soon it may be added to those existing *GBA1* tests.

### **If I was in one of the studies that contributed to the finding, can I know if I have this *GBA1* change?**

Right now, these studies cannot tell you if you have this gene variant. These studies remove your personal information from your study data and samples. This is to protect your identity. It also means they cannot tie your results back to you.

These and other studies may offer you your results in the future.

### **Do other people have this *GBA1* gene variant?**

In the study, this variant was rarely found in people with Parkinson's without African ancestry.

Scientists have found other changes in the *GBA1* gene. In fact, they have found more than 300 *GBA1* variants linked to Parkinson's disease. To date, *GBA1* gene variants have been found more commonly in people of Ashkenazi (Eastern European) Jewish descent.

### **Will my children inherit this *GBA1* genetic variant?**

It is possible they might inherit this variant. If they do, it does not mean they will absolutely get Parkinson's disease. There are many factors — aging, genetics, environmental exposure — that together influence Parkinson's risk. Also, the increased risk of Parkinson's with a *GBA1* change is relatively low compared to other known genetic links to Parkinson's disease.

Scientists will do more studies to learn more about how this *GBA1* variant in people of African descent raises Parkinson's risk.

A genetic counselor can speak more with you about passing down variants.

### **Are there treatments for *GBA1*?**

There are treatments currently in testing to correct *GBA1* and GCase function in Parkinson's disease. The scientists who found the variant in people of African descent are talking to the researchers testing these treatments. They may decide to open their trials to test for this new *GBA1* variant.