Issue 35

November 2015

UPBEAT

The Newsletter for People with Early Onset Parkinson's
Understanding Parkinson's by Belief in Education, Attitude and Treatment

Genetic testing and Parkinson's

You may have heard of people getting a genetic test for a specific condition. You might have even begun to consider whether you should take part in genetic testing. Genetic testing has the power to let you know what used to be unknowable, but genetic risk assessment is very complex and ambiguous. You may go through the process of genetic testing and feel confused and less confident with the results. The question now is do you want to know?

What is genetics?

Genetics is the branch of biology that deals with heredity. Genes are units of heredity carried in your DNA. They are like recipes that make proteins. Genes determine the traits that pass down from parent to child. You inherit about 20,000 genes from your parents coded by about 3 billion pairs of coding units on your DNA. They determine the colour of your eyes and how tall you may be in conjunction with the powerful influence of the environment from the moment you are conceived. In some instances genes can determine the risk you have in developing certain diseases or conditions in your lifetime, sometimes very strongly.

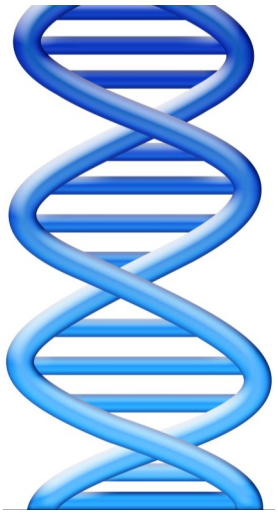
Scientists and researchers are interested in the role played by genetics in health and illness. They believe that both environmental factors and genetics contribute to the development of illness. And the degree of influence that your genes have varies depending on the disease or condition.

Genetics and Parkinson's

In Parkinson's the vast majority of cases are called sporadic (or idiopathic), meaning the cause is unknown. These non-familial cases usually mean no other family members have Parkinson's. Researchers believe onset is due to a combination of genetic and environmental factors. However, approximately 10% of Parkinson's cases have been linked to genes or genetic mutations with strong effects. In these familial cases the person with Parkinson's will usually have a parent, sibling or child also living with Parkinson's.

Genetic abnormalities that lead to Parkinson's are rare. In these cases the mutated genes associated with Parkinson's can be inherited either dominantly or recessively. Scientists have currently identified at least six gene mutations that are associated with Parkinson's (SNCA, PARKIN, PINK1, DJ1, LRRK2 and GBA). More genetic risk factors are being discovered, but some people have mutations in a gene and never develop Parkinson's symptoms.

A dominant Parkinson's gene is common in families with many relatives affected by Parkinson's through different generations. The genetic mutation SNCA is an example of this type of inheritance. SNCA codes for alpha-synuclein, a protein that is found in the brain and clumps together in people with Parkinson's.



DNA

Genetic testing

LRRK2 is another genetic mutation that is inherited dominantly. Mutations in the LRRK2 gene account for a greater number of Parkinson's cases in people from Ashkenazi (Eastern European) Jewish and North African backgrounds than in the general population. It is not yet known why this is.

In contrast, recessive Parkinson's genes are found in cases within one generation, such as in siblings. The genetic mutations PARKIN, PINK1 and DJ1 genes are examples of this type of inheritance.

In addition, there are Parkinson's risk genes where a mutation means an increased likelihood of Parkinson's but is not a direct causal link. Researchers are looking for other factors (environmental factors or genetic mutations, for example) that either push or protect someone with a gene mutation to/from having Parkinson's.

Genetics testing—providing answers or generating more questions?

New sophisticated tests are being developed to help you find out whether your genes play a role in certain conditions. Genetic testing might be ordered through a health care professional. There is also personal direct-to-consumer (DTC) genetic testing, in which you place an order with a private company, send in a bit of saliva or blood and receive results about your personal genome directly from the company.

Your doctor may suggest genetic testing if:

- you have been diagnosed with Parkinson's before the age of 40
- many relatives in your family also have been diagnosed with Parkinson's, or
- you are at risk for familial Parkinson's based on your ethnicity.

For some people, the knowledge gained from genetic testing may be important for your family planning. Genetic testing can give you information about the likelihood of someone inheriting Parkinson's. This does not mean that Parkinson's will develop if a gene is passed down, but the risk of someone inheriting Parkinson's is higher in dominant genetic mutations versus recessive ones.

Who benefits from genetic tests?

At the present time learning about your Parkinson's genetic status from genetic testing will not change your Parkinson's prevention or treatment plan. In the future when there are treatments to slow down or prevent the onset of Parkinson's, genetic testing will be important to help identify people who are at risk.

You may not benefit directly from genetic testing, but individuals with certain genetic mutations—both with and without Parkinson's—can help speed discovery in Parkinson's research. For example, the study of the SNCA gene has led to knowledge about alpha-synuclein, which most likely applies to everyone with Parkinson's.

Most studies that involve genetic testing do not reveal the results to those who participate. Providing a sample for Parkinson's genetics research is an easy way to help find the proteins and cellular pathways involved in Parkinson's, and move closer towards tackling this condition.

Caution around new DNA testing kit

Genetic testing is a very personal decision but a note of caution. It is important to consider what you may experience before buying a test. The

Will genetics help us crack the Parkinson's code?

Look for the World Parkinson Coalition programme on genetics. Renowned experts will have a discussion about how the inherited forms of Parkinson's are helping us better understand the underlying structure of this condition. Available for streaming 5 January 2016. Take advantage of this amazing learning opportunity on www.worldpdcoalition.org

Genetic testing

results of personal DTC tests can be confusing or misleading. The DTC tests that are currently available only analyse a few Parkinson's related genes. If the tests turn out negative a person may still develop Parkinson's. However, a positive test does not necessarily mean that someone will develop Parkinson's. You should also know how your personal information will be kept private.

Low costs are making genetic tests more ubiquitous. A personal genetic testing kit from US company 23andMe is a spit test that claims to access more than 100 pieces of genetic information. It also tests for genetic variants that may reveal risk factors for Parkinson's and Alzheimer's.

You can use 23andMe's genetic testing kit without consulting a doctor:

- 23andMe's test tries to determine whether you are at risk of developing Parkinson's and other health conditions, or
- if you have already been diagnosed with Parkinson's, 23andMe's test tries to determine which genes may have been risk factors.

We recommend that you seek advice

Deciding to undergo genetic testing is a very personal decision. You may want to better understand your condition or to make informed life choices. However, genetic testing will not change your Parkinson's prevention or treatment plan. Any time genetic testing is considered, particularly in a condition such as Parkinson's where there is no available prevention or treatment based on the genetic findings, we recommend that you speak to your doctor or Community Educator and get a referral to a genetics counsellor.

It is important to consider what you may experience before buying a genetic test and seek advice to discuss the impact the information will have on you and your family. When you use a DTC genetic test kit, you may get information you do not want, are not expecting or are not prepared to know. You also need to consider whether or not your family is ready or willing to know about genetic testing information that might possibly have implications for them. Some people may worry unnecessarily if they know that there is an increased risk of developing Parkinson's or other conditions.

"It may be appealing to better understand your genetic makeup, but the genetics of Parkinson's is complex. You may carry a gene linked to Parkinson's, but it does not mean you will go on to develop the condition. We encourage anyone considering genetic testing to speak to your doctor or Parkinson's Community Educator and make sure you have a referral to a genetics counsellor. Nobody should face genetic testing alone," says Deirdre O'Sullivan, Chief Executive, Parkinson's New Zealand.

Selected references

www.pdf.org
www.parkinsons.about
www.thelancet.com
www.michaeljfox.org
www.parkinsons.org.uk

Like us on Facebook

We would love to hear from you on Facebook. Our Facebook page is a place where you can connect to ask questions, talk to each other and share ideas on living with Parkinson's. Like our page on Facebook.com/Parkinsonsnz to see who is fundraising for Parkinson's and find out about upcoming events around the country. Some recent posts on our Awareness Week advert, World Parkinson Congress updates and Brain Research New Zealand (BRNZ) PhD Scholarship applications.

2016 UPBEAT weekend

2016 UPBEAT weekend

We are very excited that our Waikato branch will be hosting our March 2016 UPBEAT weekend in Hamilton. The weekend is open to people diagnosed before the age of 60 who are currently under 65 years of age. This is an opportunity to meet new people who are also living with Parkinson's, learn from each other and find out more about managing this condition. In the feedback we have received from the 2014 UPBEAT weekend many people have said meeting new people was the highlight of weekend.

2014 UPBEAT weekend in Napier

Tell us what you want

UPBEAT is *your* weekend, so we want to know what information interests you.
Please give us a call or email UPBEAT@parkinsons.org.nz to tell us what you would like to see on the programme. We are looking for people to let us know about

- a topic you would like covered at the weekend
- a topic you would be prepared to present
- your suggestions for workshop programmes any other information that interests you.

Feedback survey

If you have been to an UPBEAT weekend, we would be grateful if you could let us know your opinions and any improvements we could make for the future by participating in the survey on our website—if you haven't already:

www.parkinsons.org.nz/upbeat.

Parkinson's on the Big Screen: Awareness Week 1-7 November

Parkinson's Awareness Week is 1-7 November. This is a time when we focus on showcasing the great work we do, make people aware of the services we provide and raise much needed funds.

The wonderful teams at Val Morgan Cinemas and FCB New Zealand have donated their time and media for us to have a commercial running on over 360 screens during Awareness Week. If you haven't seen our commercial yet, have a look on our YouTube channel on youtube.com/ParkinsonsNewZealand.

UPBEAT newsletter is online

To reduce printing and postage costs, we would love it if more people received the UPBEAT newsletter by email. For those of you with email we will no longer send a printed copy unless you request one. If we don't have your email please let us know by emailing UPBEAT@parkinsons.org.nz.