

# New biobank provides insights into Parkinson's disease

May 26 2021, by Liselotte Fritz



Kajsa Brolin, doctoral student in Translational Neurogenetics at Lund University, is using the biobank in her doctoral thesis. Credit: Kennet Ruona

Around 20,000 Swedes are currently suffering from Parkinson's disease, yet there are still only treatments to alleviate symptoms. Through a new biobank, researchers gain access to the genetic profiles of 1000 Parkinson's patients together with comprehensive information about their lifestyles and medical histories.



"I am convinced that many researchers will use the <u>biobank</u> over the next few years, which could lead to the development of new treatments," says Andreas Puschmann, associate professor of neurology at Lund University Hospital.

An estimated 20,000 Swedes suffer from Parkinson's disease and 2,000 new cases are added every year. In the early stages of the disease, the patient's mobility is most affected. In later stages, patients can have problems such as sleep disorders, worry, low mood, depression and dementia. The treatments currently available only address symptom alleviation, mainly by providing the brain with new dopamine. There is a great need to find new drugs that could combat this serious disease.

In the Multipark strategic research area at Lund University, a large research team is working on mapping the development of Parkinson's disease to find new treatments. Over four years, the researchers have built up a biobank of blood samples. Half of all Parkinson's patients in Skåne, 1,000 people, and an equally large control group matched in terms of gender, age and location, have submitted blood samples and answered questions in a broad questionnaire which captures everything from lifestyle factors, symptoms, course of the disease and use of medication.

### Many types of analysis

The new biobank contains various kinds of samples from the patients—serum, plasma, DNA and RNA—which are preserved deepfrozen underground in Lund's Biobank Syd.

"We chose to take blood samples that enable many types of analysis depending on whether researchers in the future will want to look at biomarkers or genetic variations. The patient surveys we conducted enable the study of biomarkers and genetics in the blood and their



connection with environmental factors. Access to the Swedish Parkinson's register also enables patients to be monitored over time, and the study of their medical history and <u>use of medication</u>," explains Maria Swanberg, associate professor of translational neurogenetics at the Department of Experimental Medical Sciences. She is the biobank coordinator and the person to contact if you want to use it in your research and have questions and applications for access to samples.

Maria Swanberg herself has studied the lifestyle factors that can have a direct or indirect impact on whether or not a person will suffer from Parkinson's disease. These factors include coffee and tobacco consumption as well as exposure to herbicides.

"One strength of the case control study is that we see how it reproduces the known research results about Parkinson's disease. As in several previous studies, we can observe that smokers and coffee-drinkers are affected to a lesser extent whereas exposure to pesticides increases the risk of the disease."

### Most common genetic mutations

Andreas Puschmann, associate professor of neurology at Lund University Hospital, together with researchers at the Karolinska Institute and the Universities of Gothenburg and Umeå, has used the biobank in a research project. 658 of a total of 2206 DNA samples were retrieved from the biobank in Lund. The aim of the study was to find out how often the most commonly identified genetic mutations behind the disease are found in Sweden compared with other population groups in the world. It emerged that only 13 patients, or 0.6 % of the Swedish patients, had any of the mutations that have aroused a lot of attention and for which treatments are currently under development.

"We also discovered another aspect. It turned out that 21 percent of all



patients had a relative with Parkinson's, despite there being so few mutation carriers. We suspect that there is a significant genetic link to the disease, but that there may also be other underlying genetic factors and mechanisms. Instead of a single gene, there may be an interaction between two or three genes," says Andreas Puschmann.

The researchers are now rapidly finding new methods to understand how genetics affects which diseases we get. Biobanks, such as the one built up by Multipark, with <u>blood samples</u> and patient information, are a prerequisite for making progress with regard to Parkinson's disease. Besides DNA samples, the researchers can also study RNA—as an intermediary step between our DNA and the proteins that are to be encoded. If the DNA is the plan itself, the RNA is the transcriptionand the protein is the final product.

"The RNA samples in the biobank enable researchers to observe the effect of the mutations on the genetic expression, for example. We will be able to do a lot in the next few years. Progress is being made step by step, which could lead to the development of new treatments," says Andreas Puschmann.

## Combining enormous amounts of data

Kajsa Brolin, doctoral student in Translational Neurogenetics at Lund University, is using the biobank in her doctoral thesis.

"It is fantastic that so many people have volunteered, half of all Parkinson's patients in Skåne. This is unique and makes it one of the largest case control studies in Sweden. For me, it feels important to give something back."

In an attempt to answer the question of why people get Parkinson's disease, Kajsa Brolin has combined information about patients' lifestyles



with analysis of the genetic material. Why do smokers not get the disease as often as non-smokers? Can the explanation be found in the smallest components of DNA?

The basic building block in DNA is a large molecule called a nucleotide, which in turn is made up of nitrogenous bases—adenine, cytosine, guanine and thymine. Each individual has millions of variations of these nitrogen bases.

By compiling enormous amounts of data, combining them and conducting statistical analyses, Kajsa Brolin is trying to determine the significance of the genetic material at this microlevel for whether or not an individual will be affected by Parkinson's disease.

"I combine data to see whether there is any connection between the nitrogenous bases, lifestyle habits and whether or not a person gets the disease. I conduct specific analyses on a group level but one can also use the biobank to examine the cell or protein level."

# What do you believe the biobank will contribute in future?

"The major goal is to understand the disease. The main focus is on the genetic causes of the disease and on understanding the underlying biological processes. The big objective is to find new drugs; I hope that the biobank can be one piece of that particular puzzle," says Kajsa Brolin.

#### **Biobank facts**

Multipark's biobank is a unique case control study in Sweden which includes half of all Parkinson's disease patients in Skåne. Finding new



treatments for Parkinson's disease requires international collaborations. Multipark is now working to get the cohort included in a larger context and in studies initiated by the International Parkinson's Disease Genomics Consortium (IPDGC) and the Global Parkinson's Genetic Programme (GP2), among others.

#### Provided by Lund University

Citation: New biobank provides insights into Parkinson's disease (2021, May 26) retrieved 3 May 2024 from <a href="https://medicalxpress.com/news/2021-05-biobank-insights-parkinson-disease.html">https://medicalxpress.com/news/2021-05-biobank-insights-parkinson-disease.html</a>

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