

What happened to my UK MND Collections sample?

Information Sheet J

This information sheet aims to answer questions about what happened to your clinical information and sample after you participated in the MND Association's UK MND Collections database. Originally called the UK MND DNA Bank, in 2017 it was renamed UK MND Collections and includes the DNA and Cell Banks and an Epidemiology Dataset.

The content is split into the following sections:

- 1: What is UK MND Collections?**
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Disclaimer: Please note that information provided in this information sheet is based on a review of the currently available literature. This information sheet was written by the MND Association staff who are not clinicians and so any information provided in this sheet should not be considered as clinical advice. You should always discuss potential treatments with your clinician.



This symbol is used to highlight **our other publications**. To find out how to access these, see *Further information* at the end of this sheet.

1: What is UK MND Collections?

MND is thought to be caused by a combination of subtle genetic, lifestyle and environmental factors. The UK MND Collections database was designed to help identify the subtle genetic factors involved in the disease by studying the DNA (or genetic material) of people with MND.

Over 3,000 people contributed blood and data samples to the UK MND Collections over the ten years of the collection/recording phase of this project.

Two hundred people with MND also completed an epidemiology questionnaire. Together with questionnaire data from age and gender matched controls, these data form the Epidemiology Dataset part of the UK MND Collections. This provides genetic, clinical and lifestyle information in order to identify potential environmental components contributing to disease in people with MND.

2: Where is my clinical information stored?

Information about every single participant and the sample they have given is stored on a secure database. This is stored following the guidelines of the Data Protection Act.

When the details were recorded on the database, they were anonymised using a unique ID number. Participants are identifiable only by their date of birth. No other information can be accessed through the database.

Confidential information, including the participant's personal and contact details, will never leave the central hospital where the clinical information was entered into the database.

The Epidemiology dataset was established to provide genetic, clinical and lifestyle information in a group of individuals in order to identify the association of environmental risks with aspects of MND such as age of onset, survival and interactions between risk factors.

Everyone who participated in the epidemiology study was given an ID and invited to take part in the DNA and Cell Bank Study. Where appropriate, this data is linked to a sample ID from the DNA and Cell Banks.

All of the data collected can now be used to investigate what environmental factors might be associated with the onset and progression of MND.

3: What happened to my blood sample?

Each blood sample taken was divided into two sets. Together with the accompanying, anonymised information on the participants, these have been used to create an important resource of DNA samples and cell lines for MND researchers to use.

DNA

One set of samples were sent to the Centre for Integrated Genomic Medical Research Biobank (CIGMR Biobank), based at the University of Manchester. The DNA was extracted from the blood sample using state-of-the-art technology and is then tested for quantity and quality (as only 'top notch' samples can be used for research).

These DNA samples are being used in research to tell us more about the genetic causes of the disease.

Cell lines

The other set of samples were sent to the European Collection of Authenticated Cell Cultures (ECACC) at Porton Down in Wiltshire. They were used to create an 'everlasting' source of DNA.

White blood cells, which contain DNA, were taken from the blood and 'treated' with a harmless virus. This treatment allows the white blood cells to divide continually, creating an 'everlasting' supply of DNA. After the treatment they were plunged into liquid nitrogen, freezing the cells for storage. We are storing both treated and untreated white blood cells.

Originally these cell lines were created in order to replenish the DNA in Manchester, should this ever run low. This will ensure that the UK MND Collections is an everlasting resource for researchers.

The cells are also being used to create models to understand the causes of MND. White blood cells can be used to study why motor neurones die in MND – modelling what happens in people with MND.

4: How are the samples being used?

The samples are being used for genetic research, to understand the way that variations in our DNA may contribute to why people develop MND. The samples are also being used to create models of MND – providing ways to understand why motor neurones die and to develop new treatments for MND.

Genetic research

Samples from the UK MND Collections are helping scientists identify new genes that cause inherited MND or those that influence susceptibility to sporadic MND. This will

offer crucial insights into the causes of MND giving us a better understanding of the disease that may lead to the development of new treatments.

All forms of MND may have a small genetic component. In about 5-10% of cases however, there is a strong family history of MND, indicating that the disease may be inherited. This is sometimes known as 'familial MND'.

The more common form of MND (accounting for the remaining 90-95% of cases) is known as 'sporadic MND', so-called as the disease appears for no apparent reason.

In sporadic MND, normal, inherited, genetic differences may contribute to the risk of developing MND, but they are not the single cause of the disease. It is only in combination with environmental and lifestyle factors (e.g. smoking, intense exercise) that these variations may affect the chances of someone developing MND.



For further information about inherited MND, see: Information sheet B1 – *Introduction to inherited MND*.

Models of MND

The white blood cells are being used to create cellular models of MND.

For some studies, researchers can learn what goes wrong in motor neurones by studying what happens in white blood cells, so the cell lines in the UK MND Collections will be used directly.

However, for other studies, researchers can learn most by actually studying motor neurones themselves. In a step that was unimaginable when the UK MND Collections was first created, it is possible to create motor neurones from blood samples.

White blood cells from the UK MND Collections are being converted into 'induced pluripotent stem cells' (iPSCs), which in turn can be turned into motor neurones and their support cells known as 'glia'.

It is important to note that cells derived from the UK MND Collections will not be used as a basis for future treatments and will only be used to study MND and related neurodegenerative disorders, including frontotemporal dementia.



For further information about stem cells, see: Information sheet F – *Stem cells and MND*.

5: Who can use the samples and information from the UK MND Collections?

If a researcher would like to use the DNA or the cell lines for their study, they can apply to the MND Association for access to the UK MND Collections samples. Depending on their research needs, some may decide to use DNA taken directly from the blood sample and subsequently stored in Manchester; but others may request to use the cell lines from the set stored at ECACC.

Strict rules and regulations have been adopted by the MND Association to ensure that UK MND Collections samples and accompanying information are not misused.

Our Biomedical Research Advisory Panel (BRAP) guide us in the decision making of applications that wish to use these samples. The University of Manchester also use their Technical Access Committee to determine whether the right amount of sample has been requested for the intended use. The samples requested are then sent out to the researcher, if it is decided that the proposed study is: relevant to MND, novel (new to science), scientifically justified, fully funded, and ethically approved.

Alongside the samples, researchers will be provided with clinical information, such as age when the sample was taken, gender, type of participant (e.g., person with MND, unaffected 'control' or family member).

For participants with MND this includes information about their symptoms at the time of giving a sample. This information can be used in combination with the sample to learn more about the underlying causes and any factors that may affect the development of MND. Researchers are asked to explain why they need to use the clinical information as well as the samples.

All these details are given as an anonymous code, no personal information about the participants, such as their name or address, are shared.

UK MND Collections samples are currently being used in a large number of studies investigating the causes of inherited and sporadic MND.



To find out more information on all the projects we fund, including those that use our UK MND Collections samples, see Information Sheet E - *Research we fund*.

6: Can I find out how my blood sample and information has been used?

Due to your clinical information being stored anonymously it is not possible for us to find out what has happened to your specific blood sample or the information you shared, which studies it may have been used in or what the results of the studies specifically mean for you or your family.

7: Withdrawing a sample from the UK MND Collections

If you would like to withdraw a sample from the UK MND Collections please contact the Research Development team on 01604 611 880 or research@mndassociation.org.

To find out more about the UK MND Collections, please visit our website: www.mndassociation.org/dna-bank-sample-how-has-it-been-used

8: How do I find out more?

Useful links

MND and Stem cells

www.mndassociation.org/stemcells

European Medicines Agency (EMA)

www.mndassociation.org/inheritedmnd

Further information

You may find these information sheets from the MND Association helpful:

B1 – *Introduction to inherited MND*

E – *Research we fund*

F – *Stem cells and MND*

We also provide the following guides:

Living with motor neurone disease – our main guide to help you manage the impact of the disease

Caring and MND: support for you – comprehensive information for unpaid or family carers, who support someone living with MND

Caring and MND: quick guide – the summary version of our information for carers

You can download most of our publications from our website at www.mndassociation.org/publications or order in print from the MND Connect helpline, who can provide further information and support.

MND Connect can also help locate external services and providers, and introduce you to our available services, including your local branch, group, Association visitor or regional care development adviser.



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MND Association website and online forum

Website: www.mndassociation.org

Online forum: forum.mndassociation.org or through the website

We welcome your views

Your feedback is really important to us, as it helps improve our information for the benefit of people living with MND and those who care for them. If you would like to provide feedback on any of our information sheets, you can access an online form at: www.surveymonkey.co.uk/r/infosheets_research

You can request a paper version of the form or provide direct feedback by email: research@mndassociation.org.