



MicroRNA Autism Research Study

Do you or your family members have a diagnosis of autism?

Would you like to help scientists learn more about the
cause of autism?

We are looking for families to join our research
study.



What are we doing?

Autism Spectrum Disorders or Conditions (ASD/ASC) are often passed down in families, but there is no one gene that causes autism, and even in families with a high occurrence of autism, genetic testing doesn't always find a cause. It could be that ASD is not caused by a single gene, but in the way our bodies control how genes work. MicroRNAs in the blood may be important for this. In some families there are several autistic people but no known genetic cause found. For the first time, we would like to look at microRNAs in the blood of families, comparing people with and without autism in the same family.

What are microRNAs?

MicroRNAs are very small RNA molecules in the blood. There are thousands of different microRNAs that work in different ways. Some of these microRNAs can have an impact on how genes express themselves and can switch genes on and off in different brain regions. There are approximately two dozen microRNAs likely to be important to development of and connections between different areas of the brain.

Why are we doing this research?

It is estimated that 7.442 million people worldwide, and over 700,000 people in the UK (approximately 1% of the population) are autistic. Autism is a spectrum condition, and autistic people can show large differences in interaction and communication. There is no known biological marker for autism, and despite very strong genetic heritability there is no single gene contributing to more than 1% of cases. We are hoping to find out if microRNAs are biomarkers in ASD. This could help us understand more about Autism, and about what causes it. and that it might help tell us what causes autism.

Who can take part?

We are looking for six members of the same family (three with autism, three without) to take part in this research. The six family members need to be blood relatives and can include parents, grandparents, children, siblings, cousins, uncles or aunts.



What does it involve?

As a participant:

You will have a chance to talk to the research assistant about the whole process before you decide whether or not you will take part. If you choose to take part in this study the research assistant will

arrange visit you at home or at your local clinic (depending on which you prefer) at a time convenient to you. You will be asked to complete a consent form and fill in two questionnaires (a social communication questionnaire and the social responsiveness scale). The research assistant will then take two samples of blood from your arm. The amount of blood taken will be approximately one teaspoon per sample. The duration of this visit will vary for each person. This depends on how many of your family are participating, and how many of these family members are present at the visit. The visit could therefore take between 30 minutes (one participant) and 3 hours (6 participants) to complete.

One of the blood samples will go to a Genetics laboratory, for routine genetic screening to make sure that standard genetic testing does not identify a genetic cause for ASD. The other sample will go to the University of York for microRNA testing, and will analyse your blood sample using more detailed techniques.

As a parent/guardian of a participating child:

You and your child will have a chance to talk to the research assistant about the whole process before you decide whether or not you will take part. If you choose to take part in this study the research assistant will arrange visit you at home or at your local clinic (depending on which you prefer) at a time convenient to you. You will be asked to complete a consent form on behalf of your child and your child will also complete an assent form. You will then complete two questionnaires (a social communication questionnaire and the social responsiveness scale) for your child. An experienced research assistant will then take two samples of blood from your child's arm. The amount of blood taken will be approximately one teaspoon. The duration of this visit will vary for each person. This depends on how many of your family are participating, and how many of these family members are present at the visit. The visit could therefore take between 30 minutes and 3 hours to complete.

One of the blood samples will go to a Genetics laboratory, for routine genetic screening to make sure that standard genetic testing does not identify a genetic cause for ASD. The other sample will go to the University of York for microRNA testing, and will analyse the blood sample using more detailed techniques.

What happens if I change my mind?

This research is voluntary and you can change your mind about taking part at any time. If you decide to take part, and then change your mind, you will be withdrawn from the study, you don't need to tell us why you've changed your mind and it will not affect your future care.

If you are the parent or guardian of a child taking part, and they change their mind about participating in the study, they will be withdrawn. They don't need to tell us why they changed their mind and it will not affect their future care.



What are the benefits of taking part?

The research team hope that the research will lead to a better understanding around autism and the causes of autism.

What are the risks of taking part?

This study will involve a blood sample being taken. If you would like to be involved in this study, but have any anxieties about having a blood sample taken, the research assistant will work with you to prepare you for what will happen. We have videos and illustrations of what will happen, photographs of the research assistants and equipment that will be used, so you will have chance to prepare for the visit. If you change your mind about being involved, you can withdraw from the study. This will not affect your future care.



Will I find out the results?

This study is trying to find out whether we can find a biological marker for autism within families that have not found a genetic cause through traditional genetic testing. If we manage to find this in your family, we will inform you that we have. However, this is very new research, and we do not know exactly what this means. The research team are only testing a small piece of your DNA and would not know the significance of the results from these tests.

However, if we do find microRNA markers in your family, this will give us reason to do the same research on a large scale, and hopefully find out more about the cause of autism.

Any relevant results obtained will be fed back to you through a follow up clinic to discuss relevant information. Given the nature of results that can potentially be uncovered through DNA testing, any relevant information may also be passed on to the local Genetics service for further discussion. Your GP will be copied into clinically relevant information so they are aware of progress.

Will my expenses be covered?

In most instances research visits will take place at a location convenient for you, such as at your home or a local clinic. If necessary, visits may be arranged elsewhere and your expenses for these research visits will be reimbursed.

Who has approved this research study?

This work was part-funded by the Wellcome Trust [ref: 204829] through the Centre for Future Health (CFH) at the University of York and approved by Yorkshire & The Humber - Bradford Leeds Research Ethics Committee on the 28th of September 2018.

Data storage arrangements

The main database will not contain any person identifiable details, all data will be stored with a unique participant identifier. Personal data and the link to the unique identifier will be kept in a locked filing cabinet in a separate building. Only the Chief Investigator and Trial Coordinator will have access to personal data. For all other databases and documents we will use the unique

identifier only. This means that if these files or databases were to fall into anyone else's hands there will not be a way of linking any information/data with the person. Blood samples will be stored with a unique identifier in a locked research facility in the Biology department at University of York. We may store the blood samples for up to 5 years. We are hoping to develop this into a larger study, and the blood samples collected in this study may be needed to inform this. After 5 years, blood samples will be disposed of in accordance with the Human Tissue Authority's Code of Practice, and the link to the unique participant identifiers will be destroyed.

How will my data be used in the future

This is a small study to see whether the initial data supports doing a larger study. If we find that microRNAs are a biomarker for ASD, we are hoping to join together experts in the field of clinical academic research around autism spectrum disorders, and genetic expertise around microRNA technology, and carry out a large scale research trial. If that happens we may include the data we have collected from you and your family in this study. The same data storage arrangements would apply, keeping all your personal data separate from the blood sample/genetic data, with unique identifiers that keep it anonymous.

The data collected as a result of this study (questionnaire data and blood samples) will only be used in this study and in future research about autism spectrum disorder carried out by the same research team.

General Data Protection Regulation (GDPR) Information

This study is a collaboration between Leeds and York Partnership Foundation Trust (LYPFT) and University of York. The University of York is the sponsor for this study based in England and will act as the data controller for the data collected during the study. This means that the University of York is responsible for looking after your information and using it properly. Under the direction of the University of York, LYPFT will keep identifiable information about you (your name and contact details, your/your child's date of birth) from this study for 5 years after the study has finished (until January 2024).

Your rights to access, change, or move your information are limited, as we need to manage your information in specific ways in order for the research to be reliable and accurate. If you withdraw from the study, we will keep the information about you that we have already obtained. To safeguard your rights, we will use the minimum personally-identifiable information possible.

You can find out more about how we use your information on <https://www.york.ac.uk/records-management/dp/> and on the specific GDPR information sheet provided.

For compliments or concerns about this research:

If you have any general comments or compliments about the research, or conversely, any complaints about the study or the way you've been approached and treated during it, you can contact the researcher (Catarina Teige) or the Chief Investigator (Professor Barry Wright) in the first instance, who will do their best to answer your questions.

Trial Coordinator:

Dr. Catarina Teige:
Tel: 01904 294279
Email: c.teige@nhs.net

Chief Investigator:

Professor Barry Wright
Tel: 01904 294244
Email: barry.wright1@nhs.net

If you remain dissatisfied, you can contact the Research and Enterprise Office of the University of York:

Dr Michael Barber
Innovation Centre, York Science Park, York, YO10 5DG, UK
Tel: +44 (0)1904 435154.

The normal National Health Service complaints mechanisms will also be available to you.

In the event that something does go wrong and you are harmed during the research and this is due to someone's negligence then you may have grounds for legal action for compensation against the University of York or the NHS Trust.

If you would like to make a complaint, please contact our Patient Advice and Liaison Service (PALS) using the following details:

Freephone: 0800 052 5790

E-mail: pals.lypft@nhs.net

Write to: The PALS Office,
Becklin Centre, Alma Street,
Leeds, LS9 7BE

Visit: www.leedsandyorkpft.nhs.uk/service_users/PALS

How can I get involved?

If you are interested in finding out more information please contact:

Megan Garside, Research Assistant on:

Telephone: 01904 294244

Email: research.comic@nhs.net

Write to: COMIC Research
IT Centre,
Innovation Way,
Heslington, York,
YO10 5NP

Visit: <https://www.comic.org.uk/>



For further support:

National Autistic Society <http://www.autism.org.uk/>

NHS Choices <https://www.nhs.uk/conditions/autism/>

Child Autism UK <https://www.childautism.org.uk/>

Scope <https://www.scope.org.uk/support/families/diagnosis/autism>