Participant Information Sheet

A Longitudinal Laboratory and Real-World Study of Gait and Balance in People with Friedreich's Ataxia

We would like to invite you to take part in our research study.

In this study we want to investigate different ways of measuring ataxia symptoms, especially balance control and walking (gait), in people with Friedreich's Ataxia. We also want to look at how these symptoms change over one year.

- Before you decide whether or not to take part, it is important that you understand why we are doing this research and what it will involve for you
- Please take time to read this information carefully. Feel free to discuss it with your family, friends or your GP.
- You are free to decide whether or not to take part in this study. If you decide not to take part, it will not affect the treatment you receive from your doctors.
- Please ask if you have any questions or if anything is unclear.

This information sheet contains several sections. In the first few sections, we explain what the study is about, why we are doing it and why you are invited to take part. We will then describe what will happen if you decide to participate.

In the final few sections of the information sheet, we explain what we will do with the data collected from this study and how the study is managed and overseen.

What is this study about?

Ataxia is a term used to describe several symptoms related to impairment of coordination, movement, walking, balance and speech.

Friedreich's ataxia is one of the most common recessively inherited disorders affecting movement and balance. It is thought to affect one in every 50,000 people.

People with Friedreich's ataxia experience balance and co-ordination difficulties. This causes clumsiness, unsteadiness and frequent falls. It can also cause speech and swallowing difficulties, weakness in the legs and sensory issues (e.g., numbness, tingling).

It is important to have effective ways of measuring symptoms of conditions like Friedreich's ataxia. The methods we currently use have limitations, for example they are not that good at detecting changes over a short period of time. In this study we want to look at new ways that are sensitive enough to detect ataxia symptoms (especially balance and gait) and their changes over one year. Being able to measure disease progression over a short period may be useful for designing future clinical trials of new treatments and will potentially reduce the number of participants needed for these trials.

We are planning to recruit 16 individuals (aged between 14 and 65 years) with a confirmed genetic diagnosis of Friedreich's ataxia into this study.

The assessment of ataxia will be performed using three different approaches:

- Assessment of signs and symptoms including physical examination and questionnaires.
- Analysis of posture and walking (in a special room called a gait laboratory).
- Assessment of movement and everyday activities via wearable activity monitor.

We will assess each person who takes part at three study visits over one year. These visits will be at the start of the study (Baseline), Month Six and Month 12 (End of Study).

Why am I being asked to take part?

You are being invited to take part in this study because you have a diagnosis of Friedreich's ataxia, and you are able to stand and walk without any aid for at least 20 metres.

Do I have to take part?

It is completely up to you to decide whether you want to join the study. If you agree to take part, we will ask you to complete a consent form.

If you take part, you are free to change your mind at any time.

What happens if I change my mind?

You can withdraw from this study at any time, without giving any reason and without your care being affected.

If you withdraw, we will keep all the information we have collected from you up until that point.

What happens if I agree to take part?

If, after reading this information sheet, you are interested in taking part in the study, we will arrange to talk to you about the study and what it will involve for you.

You will then be invited to the study site (Clinical Ageing Research Unit, CARU) in Newcastle for three study visits. Each visit is estimated to last approx. four hours.

Before we conduct assessments or collect information from you, we will ask you to sign a consent form. This confirms that you are happy to take part and fully understand what is involved.

We will also double check that you are suitable to take part. To be suitable you need to be able to stand and walk unaided for 20 metres and be happy to take part in all the study assessments. If you have any conditions other than Friedreich's ataxia which affect your balance or movement, it may not be possible for you to take part.

If you are not suitable to take part, we will explain this to you and your involvement in the study will then end.

If you are suitable to take part, you will undergo the following assessments:

- **Collection of medical history, demographic and lifestyle information:** We will ask you questions about your medical history, any medications you are taking and about your lifestyle.
- **Physical examination:** A doctor will perform general and neurological examinations. In addition, your height, weight, heart rate, respiration rate and blood pressure will be recorded.

- **Ataxia and disease severity assessment scales:** These are specific clinical examination tools assessing the severity of ataxia and other Friedreich's ataxia-related symptoms.

- **Assessment of upper limb co-ordination:** We will ask you to perform two tasks to measure your hand co-ordination. These involve using a keyboard and putting pegs in holes in a board whilst being timed.

- **Assessments of cognitive function:** We will ask you to complete some tests of your memory and the way you understand things.

- **Questionnaires:** You will complete several questionnaires related to balance, physical and mental well-being, and quality of life. These questionnaires can either be completed online (via computer or smartphone) from your home or can be completed on paper.

- **Walking and balance assessments:** These assessments will take place in a special area called a gait laboratory. Wearable sensors will be placed on different parts of your body (e.g. lower back and/or feet) (see the below Figure for examples of the types of sensor we may use) and we will ask you to walk at different speeds. We will also assess your balance whilst standing in a variety of stances (different standing positions). We will record you on video during the assessments in the gait lab so that we can check the data we collect. You will be recognisable in these videos however they will be stored securely by the gait lab team at Newcastle University. We may wish to use these videos in the future for teaching and training purposes however you can choose on the informed consent form whether you would like your videos to be used for these purposes.

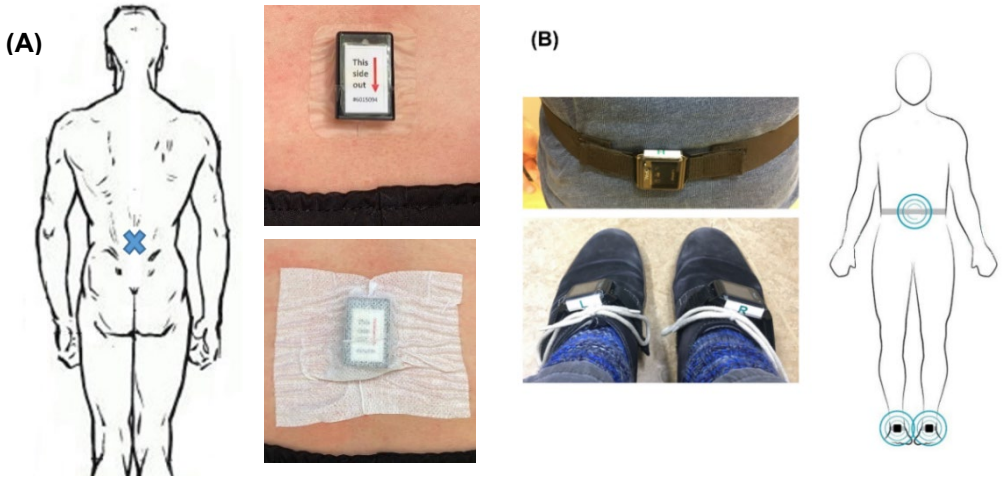

Figure. (A) Axivity, AX6 (single sensor). (B) Opal sensors, one placed in the lower back and two others on feet.

- **Collection of Information from Medical Records:** We will ask your permission to collect any relevant clinical information from your medical records from before and during the time you are in the study (this includes results from any clinical tests or investigations).

- **Wearable technology for seven-day monitoring:** During your visit, the Axivity AX6 wearable sensor will be placed on your body (as described above). We will ask you to keep it on for the next seven days to measure your physical activity in your usual environment. You will need to keep the sensor on at all times unless you are doing activities in water such as swimming. At the end of this time, you will post the sensor back (we will provide you with a special pre-paid envelope to return the sensor). We will give you full instructions on wearing and using the sensor and a separate instruction sheet will also be provided.

The above assessments will be repeated at each of the following study visits (Month Six and Month 12) at CARU.

Once you have completed the Month 12 Visit (and returned your wearable sensor) your participation in the study will be complete. At the end of the study, we will ask you for feedback on your experience of taking part.

Completion of Questionnaires Online

As mentioned above, you will have the option to complete the study questionnaires online (via computer or smartphone). We will ask you to provide an email address which will be used to send you a link to a webpage where you can complete the questionnaires.

The system used for these questionnaires is secure and only authorised members of the research team will be able to see your responses.

The questionnaires you complete will not identify you by name. You will be sent a new link before each of the study visits which will take you to the relevant questionnaires.

We can send you a text message alert as a reminder to fill in any incomplete questionnaires if you opt in to provide your mobile phone number. Your phone number will be stored on the same secure system as your email address and will not be shared with anyone else.

If you do not have access to the internet, or would prefer to complete your questionnaires on paper, we can arrange this for you.

Who is the sponsor and data controller for this research study?

Newcastle University is the Sponsor for this study.

Newcastle University and The Newcastle upon Tyne Hospitals NHS Foundation Trust (Newcastle Hospitals) will use information from you to undertake this study and will act as the data controllers. This means that Newcastle University and Newcastle Hospitals are responsible for looking after your information and using it properly.

The lawful basis for carrying out this study under the General Data Protection Regulation (GDPR) is Task in the Public Interest, (Article 6,1e). This is because research is seen as part of the University's and Trust's duties.

This study is part of a larger project involving researchers from the Hertie Institute for Clinical Brain Research and Centre for Integrative Neuroscience in Tubingen (Germany). Data collected as part of this study will be shared with the research team in Tubingen however this data will be de-identified. This means that the researchers in Tubingen will not know who you are (instead of your name you will be identified by a study-specific code only).

Representatives from the Sponsor, Newcastle Hospitals, or the regulatory authorities may request access to the study records to check that the study is being conducted correctly. This may include accessing identifiable information about you. Any staff accessing this information will work to strict codes of confidentiality.

Identifiable data from this study will be kept by Newcastle Hospitals for at least five years after the study has ended.

De-identified data (e.g., where you are not directly identifiable) will be held by Newcastle University and the project partners (Germany) for longer (20 years or more).

How will you use information about me?

We will need to use information from you for this study.
This information will include:

- Your full name
- Your date of birth
- Your NHS number
- Your postal address
- Your email address (optional)
- Your telephone number (including mobile phone number for text message alerts; optional)
- Details of your medical history, genetic diagnosis and demographic information (e.g., sex, ethnicity, employment status and educational level)
- Videos of your assessments in the gait laboratory

We will use this information to do the research or to check your records to make sure that the research is being done properly.

As described above, people who do not need to know who you are will not be able to see your name or contact details. Your data will have a code number instead.
We will keep all information about you safe and secure.

Once we have finished the research, we will keep some of the data so we can check the results. We will write our reports in a way that no-one can work out that you took part in the study.

The study data will be de-identified and may in future be shared with other researchers in the UK and overseas.

It may also be made available as “open data” through a research data repository. This means the de-identified study data will be publicly available and may be used for purposes not related to this study. We will make every attempt to ensure that it is not possible to identify you from this open data.

With your permission, we will notify your general practitioner (GP) so they are aware you are taking part in this study.

What are my choices about how my information is used?

You can stop being part of the study at any time, without giving a reason, but we will keep information about you that we already have.

If for any reason you lose the ability to continue to consent to take part, we will withdraw you from further participation in the study but will keep all the data we have collected from you up until that point.

We need to manage your records in specific ways for the research to be reliable. This means that we won’t be able to let you see or change the data we hold about you.

Where can I find out more about how my information is used?

You can find out more about how we use your information

- at https://www.ncl.ac.uk/data.protection/ and/or by contacting the Newcastle University Data Protection Officer rec-man@ncl.ac.uk or the Newcastle Hospitals Data Protection Officer nuth.dpo@nhs.net.
- at www.hra.nhs.uk/information-about-patients/
- via a leaflet available from www.hra.nhs.uk/patientdataandresearch
- by asking one of the research team
- by sending an email to **nuth.mitoresearch@nhs.net** or
- by ringing us on **0191 2083105**

If you have any concerns about the way your data is handled as part of this study, you have the right to lodge a complaint with the Information Commissioners Office (ICO), https://ico.org.uk/.

Are there any benefits to participating in this study?

There will be no direct benefits from taking part in this study. However, we hope that the information we collect will help us to identify tools that can robustly measure the severity of ataxia and track the progression in people with Friedreich's ataxia. The data collected from this study will inform the design of future studies looking at treatments for Friedreich's ataxia.

Are there any disadvantages to taking part?

The research study visits will last much longer than your usual clinic appointments. We will provide you with assistance with your travel and parking costs for the research visits. We will also provide you with refreshments/reimbursement for the cost of light refreshments during the visits.

The questionnaires we ask you to complete at home will take approximately one hour and, if you choose to complete them online, will require the use of a smartphone or computer and internet access.

The study assessments that will be performed are safe and well-tolerated by most people and these will be performed by researchers who are experienced in delivering them. We will ensure before you enter the study, and again before each assessment, that you are suitable to undergo the assessment.

You may also feel unsteady when you do some of the balance assessments, or when you are asked to walk at a faster pace in the gait laboratory, however trained staff will be with you at all times during these assessments to ensure that you are safe and comfortable.

If you find any of the study assessments physically or emotionally challenging, a member of the research team will be on hand to provide support. If you would prefer to discuss any difficulties with a member of your usual clinical care team, we will arrange this for you.

For the wearable sensors, you may experience slight irritation from the adhesive (glue) used to attach the sensors to your body. Any irritation should not last long and you will be provided contact details for the research team so that you can seek advice if you have any concerns.

We do not expect to find any area of clinical concern (i.e., indicating a potential problem) or any health issues that you were not previously aware of, during the gait and balance assessments. If anything is found which may need further investigation or treatment, the study doctors and Principal Investigator will discuss this with you and inform your GP or refer you to the appropriate specialists for further follow-up.

What happens once this study has finished?

Once you have attended your three study visits, you will have completed the study.

We think that it will take us about 18 months to complete the study for all participants.

Once the study has been completed and the results analysed, we hope to publish the findings in medical journals and present them at conferences. We will also publicise the findings through patient groups and charities.

It will not be possible to provide you with your individual results from the study assessments. However, once all the study data have been analysed, a summary of the overall study findings will be available. We will send a copy of this summary to you along with a thank you letter for taking part.

Who has reviewed this study?

All research in the NHS is looked at by an independent group of people, called a Research Ethics Committee, to protect your interests. This study has been reviewed and granted a favorable opinion by East of England -Cambridge South Research Ethics Committee.

Who will be managing this study?

Newcastle University are sponsoring this study which means they are responsible for its conduct and management.

The Chief Investigator for this study is Dr Yi Shiau Ng. Dr Ng has responsibility for the overall conduct of the study.

Newcastle University work closely with Newcastle Hospitals, where the study visits will be taking place.

How is this study funded?

The French Friedreich's Ataxia Association (AFAF) have provided funding for this study.

Who should I contact if I have any concerns?

Chief/Principal Investigator: Dr Yi Shiau Ng
Email: **nuth.mitoresearch@nhs.net**
Study Contact Name: Isabel Barrow
Tel: 0191 2083105
Email: **nuth.mitoresearch@nhs.net**

If you wish to discuss the study or have any concerns, you can contact a member of the study team on the details above.

If you wish to discuss the study or raise a complaint with someone independent of the study team, please contact Newcastle University at:
Email: sponsorship@newcastle.ac.uk

If you wish to raise a complaint on how your personal data is handled, you can contact the Newcastle University Data Protection Officer who will investigate the matter by emailing rec-man@ncl.ac.uk

If you are not satisfied with their response you can complain to the Information Commissioner's Office (ICO): https://ico.org.uk/

If you have received this information sheet via your clinical team and would prefer to raise your concerns with someone not involved in your care, you can contact the local Patient Advice and Liaison Service (PALS).

This service is confidential and can be contacted on:

Freephone: **0800 032 0202**

Email: NorthofTynePALS@northumbria-healthcare.nhs.uk

Address: Freepost PALS

Thank you for reading this information sheet