PARTICIPANT INFORMATION SHEET

Invitation to Participate in Research Project: *A Qualitative Study Exploring Patient and Family Experiences of Diagnosis in Mitochondrial Disease*

You are being invited to take part in a research project. Before you decide whether or not to take part, it is important for you to understand why the research is being undertaken and what it will involve. Please take time to read the following information carefully and discuss it with others, if you wish. Thank you for reading this.

What is the purpose of this research project?

The purpose of this student research project is to gain greater understanding of the patient and family experience of diagnosis in mitochondrial disease (MD). The aims are to explore patient and family experiences and impact of the process or journey to diagnosis, including genetic testing and receiving genetic results (where applicable). In doing so this project will add to research evidence to guide MD support groups and healthcare professionals in understanding what diagnosis is like for patients and families, increasing awareness of MD, and identifying what patients and families feel is currently done well, and what requires improvement, in MD diagnosis.

Why have I been invited to take part?

You have been invited because you are part of a patient support group for mitochondrial disease (The Lily Foundation or My Mito Mission UK), you are an adult aged 18+ who can communicate in spoken and written English, and you identify that you, or a member of your family (or someone you care for), has a diagnosis of mitochondrial disease.

Do I have to take part?

No, your participation in this research project is entirely voluntary and it is up to you to decide whether or not to take part. If you decide to take part, the researcher will discuss the research project with you and ask you to sign a consent form. The patient support group through which you heard about the project will not be told whether or not you choose to take part (unless you ask us to refer you back to them for support).

If you decide not to take part, you do not have to explain your reasons and it will not affect your legal rights or the support you receive from the patient support group you are part of. You are free to withdraw your consent to participate in the research project at any time, without giving a reason, even after signing the consent form.

What will taking part involve?

Participation will involve one online interview of 45-60 minutes on the secure video-conferencing platform Zoom. The interview will be arranged at a time to suit you between November 2022 to March 2023, and will be audio recorded to ensure that a transcript of the interview can be faithfully and accurately reproduced. Due to the virtual nature of the interview you will require an internet connection and access to a device (e.g. smartphone or computer) capable of connecting online to Zoom for a video call. The researcher will arrange the Zoom meeting and provide access to you for the meeting. There is no financial compensation for participation in this study.

During the interview you will be asked about your or your family's experiences of diagnosis of mitochondrial disease. If there are any questions in the interview that you do not wish to answer, you can let the researcher know and this will be respected.

What are the possible benefits and risks of taking part?

There will be no direct advantages or benefits to you from taking part, but your participation will contribute to improving understanding and awareness of mitochondrial disease, and what diagnosis is like for patients and families, and may help guide services to better support people in the mitochondrial disease community in the future.

It is possible that reflecting on or discussing your experiences of mitochondrial disease or diagnosis may be upsetting. If this happens, the researcher can refer you to The Lily Foundation or My Mito Mission UK for further support if you wish, or you can contact them or another rare disease or mental health support organisation directly which may include:
Genetic Alliance UK https://geneticalliance.org.uk/
Mind https://www.mind.org.uk/
Centre for Mental Health UK – a list of helplines and crisis contacts
https://www.centreformentalhealth.org.uk/helplines-and-crisis-contacts

If you become distressed at any point you will be reminded of your right to discontinue your participation in the project or interview, with no negative consequences to you.

Will my taking part in this research project be kept confidential?

Yes. All information collected from (or about) you during the research project will be kept confidential and any personal information you provide will be managed in accordance with data protection legislation. Please see '**What will happen to my Personal Data?**' (below) for further information.

In exceptional circumstances, the research team may over-ride confidentiality if they feel they have a legal and/or professional obligation to do so, for example if they have reason to believe that there is a risk to your safety, or the safety of others. If this does happen, where appropriate, the research team will aim to notify you of the need to over-ride confidentiality.

What will happen to my Personal Data?

All correspondence will take place through the password protected Cardiff University email account. The researcher will collect your name, contact details, gender and age as part of the recruitment process (including your name and contact details on the consent form), and allocate you a participant ID number under which your data will later be anonymised.

Completed consent forms will be stored on the Cardiff University secure electronic storage, and deleted from other devices following transfer. The audio recordings of interviews will be uploaded onto Cardiff University's secure media storage system following each interview, and deleted from other devices following transfer. Audio recordings will be transcribed verbatim and anonymised with pseudonyms (including for place names and the names of other people) by the researcher as soon as possible. Anonymised personal data including transcripts will be stored on a password-protected computer belonging to the researcher during the project. When the project is completed, the anonymised data will be transferred to Cardiff University secure electronic storage and deleted from the researcher's own device. If participants request withdrawal from the project at any point before anonymisation, they can also request that their stored data be deleted and this request will be actioned.

Cardiff University is the Data Controller and is committed to respecting and protecting your personal data in accordance with your expectations and Data Protection legislation. Further information about Data Protection, including your rights, the legal basis under which Cardiff University processes your personal data for research, Cardiff University's Data Protection Policy, how to contact the Cardiff University Data Protection Officer and how to contact the Information Commissioner's Office may be found at https://www.cardiff.ac.uk/public-information/policies-and-procedures/data-protection.

What happens to the data at the end of the research project?

Following completion of the project your data will be retained according to Cardiff University's Records Retention Schedule and relevant GDPR legislation. Consent forms and interview recordings will be retained in Cardiff University secure electronic storage for a minimum of five years following completion of the project or after publication of any findings based on the data collected, whichever is later. Anonymised personal data including interview transcripts will be retained for a minimum of five years, but may be published in support of the research project and/or retained indefinitely, where it is likely to have continuing value for research purposes.

What will happen to the results of the research project?

The results of the project will be written up and submitted in a student MSc Dissertation to Cardiff University in summer 2023. The findings will also be put into a report which will be available to participants and to the patient support groups The Lily Foundation and My Mito Mission UK later in 2023. The researcher also hopes to present these findings as part of World Mitochondrial Disease Awareness Week 2023 or similar conference events, and publish them in an academic journal. You will not be identified in any publication, report or presentation of this research, and any quotations from your interview will be anonymised.

Who is organising and funding this research project?

The research is organised by Dr Stella Johnson, MSc student in Genetic and Genomic Counselling at the Cardiff University School of Medicine, and supervised by Dr Rebecca Dimond, staff member at Cardiff University School of Social Sciences. The study does not receive any funding.

Who has reviewed this research project?

This research project has been reviewed and given a favourable opinion by the Cardiff University School of Medicine Research Ethics Committee (SMREC).

What if there is a problem?

If you wish to complain, or have grounds for concerns about any aspect of the manner in which you have been approached or treated during the course of this research, please contact Dr Stella Johnson (johnsonsl1@cardiff.ac.uk) or Dr Rebecca Dimond (dimondr1@cardiff.ac.uk). If your complaint is not managed to your satisfaction, please contact the Ethics Officer for the Cardiff University School of Medicine Research Ethics Committee Dr Ned Powell at powellng@cardiff.ac.uk.

If you are harmed by taking part in this research project, there are no special compensation arrangements. If you are harmed due to someone's negligence, you may have grounds for legal action, but you may have to pay for it.

To ask questions or to register your interest in taking part in the project:
Please contact the Principal Investigator Dr Stella Johnson on JohnsonSL1@cardiff.ac.uk .

Thank you for considering taking part in this research project. If you decide to participate, you will be given a copy of your signed consent form to keep for your records.