Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) Research Project

Who are we?

- The Centre for Human Metabolomics (based at the North-West University, SA)
- We are an academic & service laboratory that **specialises** in disorders affecting energy production in babies, children, & adults

- In collaboration with the International Centre for Genomic Medicine in Neuromuscular Diseases (based at the University College London, UK)

What is the MADD Research Project about?

- MADD, aka Glutaric Aciduria Type II (GAII), is a disorder that affects the breakdown of fats, proteins & choline in the body
- In cells, a molecule called FAD helps to convert various nutrients into energy
- People with MADD/GAII do not have enough FAD due to one/more genetic mutation(s)

- In Southern African countries, genetic testing for MADD/GAII is very limited
- Genetically, the population also differs from other more researched populations
- **The MADD Research Project aims to provide free clinical assessments & genetic testing to better understand MADD/GAII in Southern Africa as part of an international research study**

You may partake in this study if:

- You have been, or are a family member of someone who has been, diagnosed with MADD/GAII
- You were born in a Southern African country
- You have an unknown or negative HIV status (Please note: The study will not test for this)

What do we ask of you?

- To store your urine & blood samples at the NWU/ICGNMD
- To collect your relevant medical & family history during a clinical consultation
- To be able to contact you for relevant updates

What are the risks?

- A small prick for blood samples

What do you get?

- Anonymity & data protection
- FREE:
  - Medical consultation with a neurologist
  - Follow-up MADD/GAII testing
  - Genetic testing
  - Genetic counselling
- The opportunity to help others affected by MADD/GAII

More info:

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NWU®

International Centre for Genomic Medicine in Neuromuscular Diseases

Centre for Human Metabolomics (CHM)

This research study has been approved by the Health Research Ethics Committee of the Faculty of Health Sciences of the North-West University (NWU-00966-19-A1-01) & will be conducted according to the ethical guidelines & principles of the international Declaration of Helsinki & the ethical guidelines of the National Health Research Ethics Council.

Date Prepared: April 2022
Further information is available upon request from the NWU