

## AGHA-AMDF Genetic Testing Partnership

We are pleased to announce that, in partnership with the Australian Genomics Health Alliance (AGHA; <https://www.australiangenomics.org.au>), a new research opportunity for people with suspected mitochondrial disease has become available.

Through this research project individuals who meet the appropriate entry criteria can gain access to state-of-the-art genetic testing for mitochondrial disease. The purpose of this study is to evaluate new molecular technologies (tests) which look for changes in your DNA (genetic code) responsible for causing mitochondrial disease. The results of this study will help determine the most effective approach to the diagnosis of mitochondrial disease and will hopefully aid earlier intervention and improved outcomes for patients and families with these disorders.

See <https://www.australiangenomics.org.au/our-research/rare-disease-flagship/> for more information on the AGHA.

This Australia-wide project will run for about 18 months. To be eligible, you will need to be referred to one of the following specialist clinics to see if you meet the entry criteria for the study:

### **South Australia and Northern Territory**

Dr Nick Smith – paediatric referrals

Please note: Only referrals by a neurologist or specialist paediatrician will be accepted at this clinic

Department of Neurology and Clinical Neurophysiology

Women's and Children's Hospital

Level 2, Clarence Rieger Building

Women's and Children's Hospital

72 King William Road North Adelaide, SA, 5006

Email: [nicholas.smith@health.sa.gov.au](mailto:nicholas.smith@health.sa.gov.au)

Ph: +61 (0)8 8161 7308

Fax: +61 (0)8 8161 6224

Dr Janice Fletcher – adult referrals

Please note: A referral must come from a physician, with all details of prior investigations included.

Clinical Director | Genetics & Molecular Pathology E [janice.fletcher@sa.gov.au](mailto:janice.fletcher@sa.gov.au)

SA Pathology

RAH: Margaret Graham Building,  
Frome Road, Adelaide

WCH: Level 4,  
Rieger Building,  
72 King William Road, North Adelaide

### **Queensland**

Prof David Coman – paediatric and adult referrals

Please note: Only referrals by a neurologist or a specialist paediatrician will be accepted at this clinic.

Lady Cilento Hospital and Wesley Hospital

501 Stanley St,

South Brisbane QLD 4101

Email [David.Coman@health.qld.gov.au](mailto:David.Coman@health.qld.gov.au)

Email [enquiries@drdavidcoman.com.au](mailto:enquiries@drdavidcoman.com.au)

### **Victoria and Tasmania**

Dr Joy Lee – paediatric referrals

Please note: Only referrals by a general paediatrician or paediatric sub-specialist will be accepted at this clinic.

Department of Metabolic Medicine

The Royal Children's Hospital

50 Flemington Rd

Parkville, Victoria 3052 AUS

Phone: 03 93456251

Fax Number: 03 93456740

Email: [metabolic@rch.org.au](mailto:metabolic@rch.org.au)

Professor Martin Delatycki – adult referrals

Please note for adult referrals: A referral must come from physician, with all details of prior investigations included.

Referrals can be sent to Melanie O'Loughlin

AGHA Associate Genetic Counsellor

Murdoch Children's Research Institute

The Royal Children's Hospital

Flemington Rd Parkville, Victoria 3052 AUS

Associate Professor Michael Fahey and Dr Maie Walsh - adult referrals

You will need a referral from your medical specialist with all details of prior investigations included.

Referrals should be addressed to:

Genetic Medicine & Familial Cancer Centre

The Royal Melbourne Hospital

Grattan Street, Parkville 3052

Contact: Ms Jessica Taylor

Ph: 03 9342 7151

Fax: 03 9342 4267

Email: [genetics@mh.org.au](mailto:genetics@mh.org.au)

### **Western Australia**

Prof Phillipa Lamont – adult referrals

Please note: A referral must come from physician, with all details of prior investigations included.

WA Neurogenetic Service

Royal Perth Hospital.

Phone 92243353

Dr Maina Kava – paediatric referrals

Please note: A referral must come from a paediatric neurologist with all details of prior investigations included.

Princess Margaret Hospital

Roberts Rd,

Subiaco WA 6008

Phone switchboard: 08 93408222

### **New South Wales and the ACT**

Dr Shanti Balasubramaniam – paediatric referrals

Please note: Only referrals by a specialist paediatrician or paediatric sub-specialty will be accepted at this clinic

Genetic Metabolic Disorders Service

Children's Hospital at Westmead

Cnr Hawkesbury Road & Hainsworth Street

Westmead, NSW

Phone: 02 9845 3654

Prof Carolyn Sue – adult referrals

Please note: A referral must come from neurologist, with all details of prior investigations included

Professor and Director of Neurogenetics

Head of Neuroscience Research, Kolling Institute

UNIVERSITY OF SYDNEY

Department of Neurology,

Royal North Shore Hospital,

Reserve Rd, St Leonards NSW 2065