

MEDIA FILE NOTE

Australian experts available for interview and comment on MITOCHONDRIAL DISEASE

[Professor John Christodoulou](#): clinician & researcher, mitochondrial disease in children

[Professor Carolyn Sue](#): clinician & researcher, mitochondrial disease in adults

[Assoc Professor David Thorburn](#): researcher, mitochondrial disease

[Assoc Professor Phillipa Lamont](#): clinician & researcher, mitochondrial disease in children

[Dr Doug Lingard](#): chairman, Australian Mitochondrial Disease Foundation; family affected by mitochondrial disease

For interviews with experts and patients and/or family members affected by mitochondrial disease, please contact:

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About mitochondrial disease

- Mitochondrial disease is a debilitating and potentially fatal genetic disorder that robs the body's cells of energy and can adversely affect many parts of the body, including the muscles, brain, heart, liver, digestive system, ears and eyes.
- Every year at least 50 children born in Australia will develop mitochondrial disease during their lifetime (estimated 1 in every 5,000 births). Up to 90,000 Australians (1 in 250 people) may carry the genetic changes that can cause mitochondrial disease. Many are symptomatic but undiagnosed or misdiagnosed, or are at risk of developing the disease or passing it on to their children.
- There are very few effective treatments and no cure for mitochondrial disease and much uncertainty regarding the progression of disease/symptoms and patients' prognosis.
- Mitochondrial disease is difficult to diagnose due to the widespread range, type and severity of symptoms and its varying onset and impact on patients' lives (from none to severe); there are more than 100 known subgroups of mitochondrial disease.
- The Australian Mitochondrial Disease Foundation was set up in 2009 by family members, friends and doctors of sufferers to fund essential research into the diagnosis, treatment and cure of mitochondrial disorders, and to support affected individuals and families.



John Christodoulou AM, MB BS PhD FRACP FRCPA FHGSA

John Christodoulou is a senior geneticist based at The Children's Hospital at Westmead, where he is Director of the Western Sydney Genetics Program, one of the few integrated clinical and laboratory diagnostic genetics services in Australia. He is also Professor, Disciplines of Paediatrics and Child Health and Genetic Medicine, in the Faculty of Medicine at the University of Sydney.

Prof Christodoulou's formal training includes paediatrics and clinical genetics, with his major clinical focus being the investigation and management of individuals with inborn errors of metabolism and neuro-developmental disorders. His research interests include the molecular pathogenesis of mitochondrial respiratory chain disorders, the biology of Rett syndrome and related disorders, and the development of new therapies for phenylketonuria (PKU). Prof Christodoulou is currently trialling whole body vibration therapy to assess its potential as a treatment to improve muscle mass and strength and bone structure in children and young adults with mitochondrial disorders.

Prof Christodoulou was appointed a member of the Order of Australia (AM) in 2010 for his services to human genetics as a researcher and clinician, particularly metabolic disorders of children. He is a board member of the Australian Mitochondrial Disease Foundation and former President of the Human Genetics Society of Australasia. He has also been a recent RCPA Examiner in Genetics and has published over 100 peer reviewed papers.



Carolyn Sue MBBS, PhD, FRACP

Carolyn Sue is a clinician-scientist, currently appointed as Professor at the University of Sydney, Director of Neurogenetics in the Department of Neurology at the Kolling Institute of Medical Research at Royal North Shore Hospital, Sydney, and Director of the National Centre for Adult Stem Cell Research (Sydney Node).

Prof Sue runs a tertiary referral clinic for adults with mitochondrial disorders at Royal North Shore Hospital. She has a major interest in understanding the pathophysiology involved in mitochondrial disease, with an emphasis on developing new treatment options for affected patients. She is also using stem cells as a cellular model to investigate mitochondrial disease. It was her vision, combined with the actions of Dr Doug Lingard, which led to the establishment in 2009 of the Australian Mitochondrial Disease Foundation, of which she is a board member.

Prof Sue's most recent research discovered that mitochondrial disease may affect 1 in 250 Australians (approx 90,000 people), many of them undiagnosed or at risk of developing the disease or passing it on to their children. Other research work has identified that hearing loss in patients with mitochondrial disease was due to cochlear impairment, not neural damage. This led to the first use of cochlear implants for the treatment of mitochondrial deafness, which has now been adopted worldwide. During her thesis work she established the use of hair follicles as a reliable source of mitochondrial DNA, thus avoiding muscle biopsy in relatives of affected individuals.

In 2003, she set-up Australia's first exercise laboratory dedicated to the treatment of mitochondrial muscle disease, and runs the only centre for the diagnosis, investigation and treatment of mitochondrial disorders. She is keen to establish a national patient registry to assess whether people at risk of mitochondrial disease could prevent or delay its onset through lifestyle changes. Prof Sue was named one of Sydney's Top 100 people in health and medicine in 2009 by *The Sydney Morning Herald's* the (Sydney) magazine.



David Thorburn BSc(Hons) PhD FHGSA

Associate Professor David Thorburn is an NHMRC Principal Research Fellow at the Murdoch Children's Research Institute, located in the Royal Children's Hospital in Melbourne. He holds honorary appointments in the Department of Paediatrics, University of Melbourne and with Genetic Health Services Victoria. He is the immediate-past

President of the Human Genetics Society of Australasia, a board member of the Australian Mitochondrial Disease Foundation and a former member of the Scientific Advisory Board for the United Mitochondrial Disease Foundation (USA).

Assoc Prof Thorburn's Mitochondrial Research Laboratory is primarily involved in research but also acts as the Australasian referral centre for diagnosis of mitochondrial disease in children. His lab has diagnosed more than 400 patients with mitochondrial disease and published over 100 scientific journal articles and reviews.

Assoc Prof Thorburn's research focuses on the genetic basis of mitochondrial energy generation diseases. He has a particular interest in understanding how mitochondrial DNA mutations are passed on from mothers to their children, and translating this knowledge into approaches for genetic counselling, prenatal diagnosis and prevention. He also studies the roles of nuclear genes, which are inherited from both parents. His lab has identified several novel "disease" genes using a range of gene mapping and molecular cell biology approaches. Recently, his lab has developed two mouse models of mitochondrial disease, which are being used to study the precise disease mechanisms and to trial treatment strategies.



Phillipa Lamont MBBS PhD FRACP

Associate Professor Phillipa Lamont is the Director of the Neurogenetic Unit at Royal Perth Hospital, and also is a paediatric neurologist at Princess Margaret Hospital for Children. Her formal training includes both paediatric and adult neurology. She completed her post-doctoral studies at the Centre for Neurological and Neurosurgical Diseases at Queen Square, London, in the laboratory of Professor Anita Harding.

This led to a continuing interest in mitochondrial disorders, among other neurogenetic disorders. The Neurogenetic Unit was set up in 2000 and continues, in collaboration with Professor Nigel Laing of WA Institute for Medical Research, to identify the genetic basis of neuromuscular disorders. Assoc Prof Lamont is a board member of the Australian Mitochondrial Disease Foundation.



Doug Lingard MBChB FRANZCR

Dr Doug Lingard is a founder and the chairman of the Australian Mitochondrial Disease Foundation (AMDF). Doug and his wife Margie are the parents of two children with mitochondrial disease; sadly, their son Alex died at the age of seven. The sudden illness and subsequent diagnosis of their daughter Rose 25 years later at the age of 20 spurred Doug's determination to set up the AMDF to raise funds for research into mitochondrial disease and provide support for patients and their families.

Doug is a radiologist and nuclear physician who has been active in both public and private medicine in Australia for over 30 years. He is a co-founder of the largest diagnostic imaging practice in Australia, Pittwater Radiology & Medical Imaging Australasia Ltd. Doug can be contacted through Moore Public Relations or directly on 0418 161 926.



Australian Mitochondrial Disease Foundation

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www.amdf.org.au | www.facebook.com

Stay in Bed Day (22 August 2010): www.stayinbedday.org.au

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