

# Personalised Medicine

*Where is it now, where is it going and how will we pay for it?*

A SYMPOSIUM AND NETWORKING EVENT FOR ALL THOSE INTERESTED IN CURING GENETIC DISEASES

## Speaker Biographies



### ***Prof Richard Gibbs***

BSc, PhD

Professor Richard Gibbs is the Director of the Human Genome Sequencing Center and Chair of Molecular and Human Genetics at the Baylor College of Medicine (BCM) in Houston, US. He moved to the BCM from Melbourne in 1990 as a Postdoctoral Fellow to study the molecular basis of human X-linked diseases and develop technologies for rapid genetic analysis.

Prof Gibbs played a key role in planning and developing the Human Genome Project (HGP) and in 1996 established the Human Genome Sequencing Center (HGSC), one of the world's largest sequencing centres. The HGSC was subsequently one of five worldwide sites to complete the final phase of the Human Genome Project in 2005, contributing approximately ten per cent of the HGP.

In 2007, Prof Gibbs was involved in mapping the entire genome code of an individual: one of the fathers of DNA research, Nobel Prize winner Dr James Watson. Current research in the HGSC is focused on the genomics of cancer, heart disease and autism, and is sequencing single human genomes at an increasing rate. New molecular technologies are being developed for mapping and sequencing genes, exploring novel chemistries for DNA tagging, and manipulating DNA. The HGSC is also part of the Human Microbiome Project, developing tools to generate, manipulate and analyse genome data.



### ***Prof James Lupski***

MD, PhD, DSc (hon)

Professor James Lupski is Vice Chair of Molecular and Human Genetics at Baylor College of Medicine in Houston, US, and is renowned for his groundbreaking work in 'genomic disorders', a term he coined.

His research focuses on using human genetics, genomics, and molecular biological techniques to determine molecular mechanisms for diseases. His major focus is the molecular genetics of Charcot-Marie-Tooth (CMT) disease and related inherited demyelinating peripheral neuropathies, as well as determining the molecular mechanism for contiguous gene syndromes such as Smith-Magenis syndrome. Through these studies, Prof Lupski's laboratory has delineated the concept of genomic disorders and established the critical role of copy number variation (CNV) and gene dosage in conveying human disease phenotypes.

Prof Lupski's laboratory has also used chromosome engineering to develop mouse models for genomic disorders. The laboratory's CMT studies in collaborations with Prof Richard Gibbs and the Baylor Human Genome Sequencing Center resulted in the first personal genome sequence to identify a 'disease gene' by whole genome sequencing (WGS) and demonstrated the use of WGS for optimising patient management. His other human genetic disease gene mapping research involves collaborative efforts with Dr Richard A. Lewis to map rare, recessively inherited eye disorders.

For Prof Lupski's work on human genomics and the elucidation of genomic disorders, he was awarded an honorary Doctor of Science degree in 2011 from the Watson School of Biological Science at the Cold Spring Harbor Laboratory. He has co-authored more than 500 scientific publications and is a co-inventor on more than 20 patents.

8:00am – 11:00am  
Thursday 15<sup>th</sup> March 2012  
Offices of PwC  
Level 10, 201 Sussex St, Sydney

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### ***Prof John Christodoulou, AM***

MB BS PhD FRACP FFS<sub>c</sub> FRCPA FHGSA

Professor 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 Sydney Medical School at the University of Sydney.

Prof Christodoulou is a member of the National Health and Medical Research Council's Human Genetics Advisory Committee, which advises the NHMRC on high-level technical and strategic issues in human genetics, and on the social, ethical and legal implications of human genetics and related technologies.

Prof Christodoulou's major clinical focus is 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). He 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 its Scientific & Medical Advisory Panel, and former President of the Human Genetics Society of Australasia. He has also been a recent RCPA Examiner in Genetics and has published over 140 peer reviewed papers.



### ***Prof David Thorburn***

BSc(Hons) PhD FHGSA FFS(RCPA)

Professor David Thorburn is an NHMRC Principal Research Fellow; Head of Mitochondrial Research, Genetic Disorders Theme; and Leader, Genomics & Personalised Medicine Research Affinity Group at the Murdoch Childrens Research Institute at 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. In addition, he chairs one of several writing committees of the Royal College of Pathologists of Australasia developing professional guidelines for the use of Next-Generation DNA Sequencing in clinical practice.

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 and has diagnosed more than 500 patients with these debilitating disorders. His research focuses on the genetic basis of mitochondrial energy generation diseases, with recent research focused on the use of Next Generation Sequencing for investigating these complex disorders. In a recent publication in *Science Translational Medicine*, he described the promise and challenges of transferring these technologies from research to diagnostic use in a study sequencing 1000 genes in 42 patients with severe infantile mitochondrial disease.

Prof Thorburn is a past President of the Human Genetics Society of Australasia, a board member of the Australian Mitochondrial Disease Foundation and its Scientific & Medical Advisory Panel, and a former member of the Scientific Advisory Board for the United Mitochondrial Disease Foundation (USA). He has published over 100 scientific journal articles and reviews.

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### *Dr Clive Morris*

Clive Morris is Head of the Research Group at the National Health and Medical Research Council (NHMRC), Australia's major governmental funding body for health and medical research.

The Research Group develops and manages strategies for NHMRC's health and medical research funding schemes, manages the peer review process and manages the post award activities of grants and their associated payments. In addition, the Research Group supports and advises administering institutions and supports the Harmonisation of Multicentre Ethical Review (HoMER) Initiative.

During his time with NHMRC, Clive has undertaken a number of roles, including looking after NHMRC's health ethics and health advisory activities and corporate support functions. His previous Commonwealth appointments include heading the Molecular Biology Section at the Therapeutic Goods Administration, and as a senior toxicologist with Food Standards Australia and New Zealand (FSANZ).



### *Dr Doug Lingard*

MBChB FRANZCR

Dr Doug Lingard is a founder and the chairman of the Australian Mitochondrial Disease Foundation (AMDF) and member of its Scientific & Medical Advisory Panel.

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.



### *Prof Carolyn Sue*

MBBS, PhD, FRACP

Prof Sue is currently appointed as Professor at the University of Sydney, the Director of the Department of Neurogenetics at Royal North Shore Hospital and the Director of the National Centre for Adult Stem Cell Research (Sydney Node). She completed her post-doctoral studies at Columbia University in New York, and returned to Australia in 2000 to establish her own research team, currently located at the Kolling Institute of Medical Research, at Royal North Shore Hospital.

She has a major interest in understanding the disease processes involved in mitochondrial disorders, 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.

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