

Citation for the Award of the Doctor of Medical Science *honoris causa* to Professor Kathryn North AM

Professor Kathryn North is one of Australia's leading and most influential physician scientists. Trained as a paediatrician, neurologist and clinical geneticist, Kathryn is recognised internationally for her leadership in genomic medicine and her research into inherited myopathies and genes that influence human muscle performance.

Professor North's achievements in bringing genomics medicine into clinical practice have transformed the lives of thousands of children across Australia through major innovations in the areas of gene discovery, improved diagnosis and prevention, understanding of disease mechanism and the development and evaluation of novel therapies through clinical trials. She is also leading national and international initiatives focused on genomic data sharing and the implementation of genomic medicine into clinical practice.

Kathryn established the first paediatric Neurogenetics Clinic in Australia in 1996, and over 15 years cared for over 2500 children and adolescents affected by rare neuromuscular disorders. Partnering her research program with the clinic provided patients with access to a rapidly increasing number of disease genes they could be tested for, using new gene sequencing technology that could quickly and accurately provide a diagnosis. Kathryn's research team has discovered over 20 disease genes for inherited myopathies, spinal muscular atrophy and congenital muscular dystrophies. She has led the world in improving the diagnostic rate for these disorders through introduction of new, accurate and fast, genetic sequencing technology: in 1999, less than 10% of patients received a diagnosis, by 2016 this had risen to over 60%.

Since taking up the leadership of the Murdoch Children's Research Institute in 2013, Kathryn has focused on making genomic medicine part of standard care for all Australians. She is leading a consortium of over 70 institutions around Australia to integrate genomic medicine into healthcare, supported by an NHMRC grant of \$25M – the second largest grant ever awarded by the federal government. This consortium, Australian Genomics, brings together the diagnostic pathology and clinical genetics services of all Australian States and Territories, along with the major research and academic institutions, with the aim to shorten diagnosis times, enable early intervention and provide access to treatment for people with genetic disorders.

In recognition of her international leadership in genomic medicine, Kathryn was appointed as Vice Chair of the Global Alliance for Genomics and Health (GA4GH), an international consortium of more than 500 institutions across 46 countries. As part of her role in GA4GH, Kathryn is leading a network of national genomics initiatives – including Genomics England and the US Precision Medicine Initiative - to link and share information, and develop rigorous standards and protocols related to ethics, privacy and data sharing.

Kathryn has published over 330 research articles in highly ranked journals including *Nature Genetics*, *American Journal of Human Genetics*, *Journal of Clinical Investigation*, *Annals of Neurology*, *Brain*, and *Science*, and her body of work has been cited over 10,000 times. She has been invited to speak at over 170 national and international conferences, including highly distinguished invitations such as Visiting Professor to the Harvard University Genetics Program, the Third Morgan-Hughes Thomas Distinguished Lecturer to the Medical Research Council Neuromuscular Translational Research Conference in the UK, and as the 2017 Prichard Lecturer Hospital for Sick Kids in Toronto. She has been awarded over \$50 million in total career funding (as chief or lead investigator) from competitive national and international funding bodies including NHMRC, ARC, and US Department of Defense.

Her work has been recognised through numerous prizes and awards, including the GSK Australia Award for Research Excellence, the Ramaciotti Medal for Excellence in Biomedical Research, and Member of the Order of Australia for service to medicine in the field of neuromuscular and neurogenetics research, paediatrics and child health as a clinician and academic, and to national and international professional associations.

Kathryn is a Founding Fellow and Council member of the Australian Academy of Health and Medical Sciences. She is currently a member of the NHMRC Research Council as Principal Committee Chair, and is serving a second three-year term as Chair of the NHMRC Research Committee. Her national and international standing is further reflected through her roles as board member of the Victorian Comprehensive Cancer Centre and as Chair of the International Advisory Board undertaking a review of the UCL Great Ormond Street Institute of Child Health (UK).

For over 25 years Kathryn North has made outstanding contributions to improving the health of the Australian people through her research excellence and leadership in neuromuscular biology and her advocacy for those affected by neuromuscular and rare inherited genetic disorders. In recognition of the excellence of her contributions to medical science Professor Kathryn North is awarded the Doctor of Medical Science *honoris causa*.