

Researcher awarded the AM for services to human genetics

Professor John Christodoulou has been appointed a member of the Order of Australia (AM) for his services as a researcher and clinician to human genetics, particularly the metabolic disorders of children.

“Developing a better understanding of diseases is the only way to have effective treatments,” said Professor Christodoulou.

Prof Christodoulou’s research identified a second gene which causes a clinical picture that overlaps with Rett Syndrome, a devastating progressive disease in girls. He also led the research which identified the gene that is responsible for Arts syndrome, a very rare, but potentially lethal, inherited disease that affects boys.

‘This translated immediately into oral therapy that has made a difference to our patients,’ said Professor Christodoulou. Brothers, Thomas and Bradley, spent many weeks a year in intensive care after catching a cold that progressed to pneumonia and total paralysis of their breathing muscles. In the past two years, they have had very few days in hospital.

There is no curative treatment yet for Rett Syndrome,” he explained. “By understanding better the biology, we will be in a position to design specific treatments to slow or halt the progression of the disorder, or perhaps even a cure”.

Prof. Christodoulou developed an interest in inborn errors during his training in paediatrics. He was a resident at the Hospital in 1982, when it was located at Camperdown. He then worked in the field of biochemical genetics at The Murdoch Research Institute from 1986-90 and then worked at The Hospital for Sick Kids in Toronto for 2 years, before returning to The Children’s Hospital at Westmead as senior lecturer.

Now he is the Director of The Western Sydney Genetics Program, a comprehensive and integrated clinical and laboratory genetics service at The Children’s Hospital at Westmead. His clinical practice is in the management of children with inborn errors of metabolism. He is also Professor in the disciplines of Paediatrics and Child Health and Genetic Medicine, Faculty of Medicine, University of Sydney.

“I have a fabulous team to work with”, said Prof Christodoulou. “There are a number of kids we see and don’t have an effective therapy for them. We aim to have a better understanding and design new treatments to benefit kids.”

“I also have an interest in mitochondrial respiratory chain disorders. The mitochondria is the powerhouse of the cell, it is responsible for generating most of the energy of the cell. We have trial going at the moment, but it is too early to know if there will be a benefit. “

“ PKU is another of my research interests. Newborn screening has allowed us to identify it and begin treatment in the first month of the baby’s life. This prevents mental retardation. PKU is controlled with a strict diet. We are working on a new therapy that could allow kids to have a more relaxed diet.”

Professor Christodoulou has been widely recognised for his contribution to the field of human genetics. Now he has been honoured with an AM. Says Christodoulou: “I was deeply honoured to receive this award, which is really a reflection of the wonderful people, those in the clinical and diagnostic arms of the WSGP, my enthusiastic research team, and my colleagues at the University, with who I have had the great privilege to work.