IRSF Spotlight: John Christodoulou, AM, MB, BS, PhD, FRACP, FFSc, FRCPA, CGHGSA, Children's Hospital at Westmead, Sydney, Australia

As we continue with our Investigator Spotlight series, it is our pleasure to highlight Dr. John Christodoulou this month. Dr. Christodoulou is the Director of the Western Sydney Genetics Program, Head of the Genetic Metabolic Disorders Service at the Children's Hospital at Westmead, Sydney, and Professor in the Disciplines of Paediatrics and Child Health and Genetic Medicine at the University of Sydney. He has also been appointed a member of the order of Australia (AM) for his services to human genetics as a researcher and clinician.

Dr. Christodoulou is an extremely active participant in IRSF's research program and has been funded by IRSF since 2002 for the IRSF MECP2 Mutation Database-RettBASE. This database has been constructed by merging mutation and polymorphism data on MECP2 from the published literature pertaining to Rett syndrome and related clinical disorders. This is a freely available resource to researchers and clinicians, with the overall goal of assisting health professionals in providing accurate genetic information to families of children with Rett syndrome.

Dr. Christodoulou also recently participated in the Data Blitz discussion on HDAC inhibitors at the 12th Annual Rett syndrome Symposium this past June. He runs an active laboratory-based and clinical research program, studying Rett syndrome, phenylketonuria (PKU), and the mitochondrial respiratory chain disorders. One of Dr. Christodoulou's current research projects includes the study of the HDAC6 inhibitor Tubastatin A as a potential therapy for Rett syndrome.

What prompted you to begin a career in research?
During my paediatric training, I encountered children with rare genetic disorders for which treatments were either suboptimal or nonexistent. I had always had an interest in basic biology, and in particular, the chemical processes that lead to disease. So I was quick to seize the opportunity to undertake PhD studies in biochemical genetics whilst training in genetics.

Provide a brief outline of your training and the work you have conducted:
I undertook my medical training at the University of Sydney. Following my intern year, I moved to the Royal Alexandra Hospital for Children in Sydney in 1982 where I began my paediatric training. In 1986, my wife and I moved to the Royal Children's Hospital in Melbourne were I undertook advanced training in clinical genetics and where I undertook PhD studies in the field of biochemical genetics. In 1990, we moved to Toronto where I rounded off my postdoctoral clinical and training in biochemical genetics. I returned to Sydney in 1992 to take up an academic position. A year or two later, a serendipitous meeting with Dr. Helen Leonard kindled my interest in the genetics and biology of Rett syndrome, and led me down a committed path of research into the disorder.

What is the single most rewarding aspect of conducting Rett syndrome research?
The notion that by improving our understanding of the biology of Rett syndrome, we may be able to develop specific targeted therapies to stop progression of the disorder with an ultimate aim of reversing the neurological manifestations of the disorder.

If you could pick any one symptom of Rett syndrome to prevent or to provide relief for, what would it be?
It would be spectacular to be able to stop progression of the neurological disorder, or even better, reverse its consequences.

What other disease(s) does your research focus on?
One of my other research interests is study of the biology of the mitochondrial respiratory chain disorders and the discovery of new disease causing genes. Also, we are studying the inborn error of metabolism phenylketonuria (PKU) where we are working on the development of a novel therapy that we hope will reduce the dependence of an onerous protein-restricted diet.

Besides your role as principal investigator on this project and as a Rett syndrome investigator, what other roles do you currently hold that are specific to the field of Rett syndrome research? (i.e. NIH Grant Reviewer, IRSF Grant Reviewer, member of specific Board or Panel, etc.)
I am a NHMRC (National Health and Medical Research Council of Australia) grant reviewer, and have sat on NHMRC genetics grant review panels. In addition, I am an IRSF grant reviewer and a member of the RettSearch executive committee.

Provide any other interesting information about yourself or your work that you would like the Rett syndrome community to know about you.
In my spare time (yeah right!) I try and keep fit by active participation in fencing at my Sydney University club. My favourite weapon is the foil, but will pick up and play with an epee from time to time. Nothing like stabbing at people to unwind!!

For more information on Dr. Christodoulou, please visit:br
/> sydney.edu.au/medicine/people/academics/profiles/johnch.php.

For a list of Dr. Christodoulou's publications, please visit:
www.ncbi.nlm.nih.gov/pubmed?term=christodoulou%20j.

For more information on the IRSF MeCP2 mutation database-RettBASE, please visit:
mecp2.chw.edu.au/.