Congenital anomalies of the kidney and urinary tract (CAKUT) are the major cause of end-stage renal disease in children and young adults. Genetic factors play a critical role in CAKUT determination, but high phenotypic and genetic heterogeneity, variable expression, and complex modes of inheritance hamper gene discovery. We previously demonstrated that copy number variations (CNVs) are a major source of genetic variation underlying CAKUT. In a large cohort of patients and controls, we show that the different categories of CAKUT have distinct genetic architecture, we identify CNVs that show high genotype-phenotype correlation, while others show marked pleiotropic effect on urinary and extraurinary development. Finally, by conducting multidisciplinary approaches, we provide evidence for the identification of the main genetic drivers underlying the chromosomes 22q11.2 and 16p11.2 microdeletion syndromes.

SPEAKER: A/Prof Simone Sanna-Cherchi
Assistant Professor of Medicine, Division of Nephrology
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HOST: Professor Stuart Cook
Cardiovascular & Metabolic Disorders Programme
Duke-NUS Medical School

DATE / TIME: Monday, 24 April 2017 — 12:00 to 1:00 PM
(Lunch will be served by 11:30 am)

VENUE: Amphitheatre, Level 2
Duke-NUS Medical School

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Dr. Sanna-Cherchi completed medical school, as well as his training in internal medicine and nephrology at the University of Parma, Italy. He then moved to Columbia University for his post-doctoral training in genetics and nephrology under his mentor, Ali Gharavi. He then repeated his residency training in internal medicine at St. Luke’s - Roosevelt Hospital in New York, and his fellowship in nephrology at Columbia University. He has been a tenure-track faculty member in the Division of Nephrology since then. Dr. Sanna-Cherchi has had independent funding since 2009 (American Society of Nephrology, American Heart Association, Department of Defense, as well as NIH R21 and R01 awards, and many others). His work on congenital kidney defects and pediatric renal diseases has been published on the most prestigious journals, including the New England Journal of Medicine (2013 and 2017), the Journal of Clinical Investigation (2015), the American Journal of Human Genetics (2007, 2012, 2015), the Journal of the American society of Nephrology (2005, 2009, 2014), Kidney International (2011, 2015) and many others.