

ISN Forefronts Boston
Genetic Basis of
Renal Disease
September 11-14, 2014
Boston (MA), USA

Program

www.isnforefronts.org/boston

Advancing Nephrology Around the World



Program

Thursday

Welcome

17:00 - 17:15 Welcome and Opening Remarks

Session 1:

Moderator: Joseph Benventre (USA)

Personalized Genetic Medicine

17:15 - 18:00 **Keynote Lecture:**
Monogenic disease genes elucidate renal function
Richard Lifton, USA

18:00 - 19:00 *Welcome & Networking Reception*

Friday

Session 2:

Moderator: Jeffrey Kopp (USA)

Genome-Wide Association Studies and Epigenetics

- 08:30 - 09:15 **Keynote Lecture:**
The future of GWAS for the study of pathogenesis
Joel Hirschhorn, USA
- 09:15 - 09:45 GWAS in renal disease
Caroline Fox (USA)
- 09:45 - 10:15 Meta-analysis identifies multiple loci associated with kidney function-related traits in East Asian populations
Yukinori Okada (Japan)

10:15 - 10:45 *Coffee Break & Poster Viewing*

Session 3:

Moderators: Gregory Germino (USA) & Dorien Peters (The Netherlands)

Renal Cystic Ciliopathies: From Genes and Signaling Pathways to Treatment

- 10:45 - 11:15 Using broad genomic tools to identify mutations of *MUC1* as causing MCKD1
Mark Daly, USA
- 11:15 - 11:45 Polycystin-dependent inhibition and cilia-dependent activation of renal cyst growth
Stefan Somlo, USA
- 11:45 - 12:15 Defective glucose metabolism in polycystic kidney disease identifies a new therapeutic strategy
Alessandra Boletta (Italy)
- 12:15 - 12:45 Signaling pathways and therapeutic molecules in cystic kidney disease
Vicente Torres, USA

12:45 - 14:45 *Networking Lunch & Poster Viewing*

Session 4:

Moderator: Helen McNeill (Canada)

Animal Models of Genetic Renal Disease

- 14:45 - 15:15 Nephrotic Syndrome and soluble Flt1
Susan Quaggin (USA)
- 15:15 - 15:45 Lessons from mouse models of heart disease
Cecilia W. Lo (USA)

15:45 - 16:15 *Coffee Break & Poster Viewing*

Oral Presentations

Moderators: Corinne Antignac (France) and Friedhelm Hildebrandt (USA)

- 16:15 - 16:30 The ciliopathy protein ANKS6 is the critical activator of NEK8 kinase, regulating *situs* determination, cardiopulmonary development and renal morphogenesis
Peter Czarnecki (USA) Poster Board: P71
- 16:35 - 16:50 Mutations in *DCDC2* cause nephronophthisis with hepatic fibrosis
Markus Schueler (USA) Poster Board: P62
- 16:55 - 17:10 A high throughput mass spectrometry based test for medullary cystic kidney disease type 1
Daniel Gale (United Kingdom) Poster Board: P24
- 17:15 - 17:30 Robo2 forms a protein complex with Ilk, and loss of Robo2 alleviates the glomerular phenotype of Ilk podocyte-specific knockout mice
Hila Milo Rasouly (USA) Poster Board: P46
- 17:35 - 17:50 Podocyte MDM2 prevents P53 overactivation-related cell death (podoptosis), proteinuria, and glomerulosclerosis
Hans-Joachim Anders (Germany) Poster Board: P27
- 17:55 - 18:10 Genetic analysis in aging inbred strains and diversity outbred mice identifies novel genes and networks for age-related kidney dysfunction
Ron Korstanje (USA) Poster Board: P21

Saturday

Session 5:

Moderator: Weining Lu (USA)

Zebrafish as a model of (renal) disease

- 08:30 - 09:15 **Keynote Lecture:**
Small molecule screening in zebrafish models of disease
Leonard Zon (USA)
- 09:15 - 09:45 Genetic disease models in zebrafish for screening of therapeutic compounds
Randall Peterson (USA)
- 09:45 - 10:15 Zebrafish models of genetic nephrotic syndrome
Iain Drummond (USA)

10:15 - 10:45 *Coffee Break & Poster Viewing*

Session 6:

Moderator: Paola Romagnani (Italy)

Genetics of congenital anomalies of the kidney

- 10:45 - 11:15 Autosomal dominant causes of congenital anomalies of the kidneys and urinary tract (CAKUT)
Ali Gharavi (USA)
- 11:15 - 11:45 miRNA139a
Dontscho Kerjaschki (Austria)
- 11:45 - 12:15 Uromodulin and kidney disease: A question of dosage
Olivier Devuyst (Switzerland)

12:15 - 14:15 *Networking Lunch & Poster Viewing*

Session 7:

Moderator: Ali Gharavi (USA)

Insights into podocyte biology and diseases

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| 14:15 - 14:45 | Insights into mechanisms from autosomal recessive forms of nephrotic syndrome: New concepts
<i>Corinne Antignac (France)</i> |
| 14:45 - 15:15 | Insights into mechanisms from autosomal dominant forms of FSGS
<i>Martin Pollack (USA)</i> |
| 15:15 - 15:45 | AKT2 is essential to maintain podocyte viability and function during chronic kidney disease
<i>Fabiola Terzi (France)</i> |
| 15:45 - 16:15 | Abatacept in B7-1-positive proteinuric kidney disease
<i>Peter Mundel (USA)</i> |

Sunday

Session 8:

Moderator: *Friedhelm Hildebrandt (USA)*

Stem cells, renal cancer and new therapeutic strategies in ciliopathies

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| 09:00 - 09:30 | Lin28 and stem cells
<i>George Daley (USA)</i> |
| 09:30 - 10:00 | Cdk inhibitors in ciliopathies
<i>Oxana Ibraghimov-Breskovnaya (USA)</i> |
| 10:00 - 10:30 | Primary cilia control cell fate in preadipocyte mesenchymal stem cells
<i>Peter Jackson, USA</i> |
| 10:30 - 11:00 | Genetic determinants of renal cancer
<i>Sakari Vanharanta (USA)</i> |

11:00 - 11:15 *Closing Remarks*

Biographies

Corinne Antignac



Corinne Antignac's research programs are mainly devoted to the identification of genes involved in rare hereditary renal diseases and to the characterization of the proteins encoded by these genes. Her lab's main results concern the identification of various genes underlying nephronophthisis, cystinosis and steroid-resistant nephrotic syndrome. In all cases, the genes appeared to encode new proteins of unknown functions, which have been (or still are) characterised in the laboratory. Animal models have been successfully generated, allowing better analysis of the disease phenotype and the future use of these models for testing emerging therapies. The group has also been involved in identifying genes by candidate gene approaches and in phenotype/genotype correlation in other hereditary renal disorders such as Bartter and Alport syndromes and renal tubular dysgenesis. More recently, the Antignac's group has used the next generation sequencing technologies not only to identify new genes involved in hereditary disorders, but also to implement new strategies for molecular diagnosis.

Alessandra Boletta



Alessandra Boletta graduated in Biology at the University of Pavia, Italy in 1992. She carried out her PhD-equivalent at the Mario Negri Institute for Pharmacological Research in Bergamo Italy, working on a project aimed at achieving gene delivery to the kidney. She carried out her postdoctoral training at the Johns Hopkins University School of Medicine, Baltimore, MD, USA working on Polycystic Kidney Disease. She returned to Italy to establish her own group in 2002 thanks to a Career Program of the Italian Telethon Foundation. Today she is head of Research Unit and Director of the Division of Genetics and Cell Biology at the San Raffaele Scientific Institute in Milan, Italy. Her main scientific interest is on Polycystic Kidney Diseases. More specifically she is focusing on studying the function of the PKD1 gene, mutated in the majority of cases of ADPKD, and of its product, Polycystin-1. A second scientific interest is in understanding the mechanisms leading to a progressive transformation of simple cysts into cystadenomas and carcinomas in syndromic forms of Renal Cell Carcinoma.

George Daley



George Q. Daley, M.D., Ph.D. is the Samuel E. Lux IV Professor of Hematology/Oncology, Director of the Stem Cell Transplantation Program at Children's Hospital Boston, Professor of Biological Chemistry and Molecular Pharmacology and Pediatrics at Harvard Medical School, and an investigator of the Howard Hughes Medical Institute. Dr. Daley received his bachelor's degree magna cum laude from Harvard University (1982), a Ph.D. in biology from MIT (1989), and the M.D. from Harvard Medical School summa cum laude (1991). He has served the International Society for Stem Cell Research (ISSCR) as past-President ('07-'08), led the special task forces that produced the ISSCR Guidelines for Stem Cell Research (2006) and Clinical Translation (2008), and is currently the ISSCR Clerk. Dr. Daley has been elected to the Institute of Medicine of the National Academies, American Society for Clinical Investigation, American Association of Physicians, American Pediatric Societies, American Academy of Arts and Sciences, and American Association for the Advancement of Science, and has received the NIH Director's Pioneer Award, the Judson Daland Prize from the American Philosophical Society, the E. Mead

Johnson Award from the American Pediatric Society, and the E. Donnal Thomas Prize from the American Society for Hematology. Dr. Daley's research exploits mouse and human disease models to identify mechanisms that underlie cancer and blood disease.

Mark Daly



Mark Daly is the founding chief of the Analytic and Translational Genetics Unit (ATGU) at Massachusetts General Hospital and an assistant professor in the Harvard Medical School. His research has historically focused on the development and application of statistical methods for the discovery and interpretation of genetic variation responsible for complex human disease and with the recent creation of the ATGU, he and other core faculty are now focused on the interpretation of genome sequence and the use of genome information in clinical settings. Mark is also a senior associate member and co-director of the Program in Medical and Population Genetics at the Broad Institute, where he leads many large scale genome sequencing studies in autism and inflammatory bowel disease. Daly's group has developed numerous methods and widely used software tools, including GENEHUNTER and HAPLOVIEW, genetic analysis tools used in thousands of laboratories worldwide; GRAIL and DAPPLE, web-based utilities for the interpretation of genome-wide association results; and have contributed to additional widely distributed tools developed in the Broad community such as PLINK and GATK. His earlier work at the Whitehead Institute and Whitehead/MIT Center for Genome Research (precursor to the Broad Institute) was instrumental in developing an understanding of patterns of variation in the human and mouse genomes and in the use of these patterns in disease gene mapping. While developing computational and statistical methods that can be broadly applied, his group has several primary medical genetics research foci. He has extensive research program in neuropsychiatric genetics - Particularly in autism, schizophrenia and ADHD - And has led large-scale GWAS And exome sequencing efforts in this area. His Lab serves as the analytic hub for the Psychiatric GWAS Consortium, and international consortium leading the largest collaborative GWAS Studies in 5 major psychiatric disorders. He also has a longstanding effort in the mapping of genes for Crohn's disease and ulcerative colitis where he helped found and lead an international effort that has identified more than 150 Genetic risk factors and, in collaboration with Dr. Ramnik Xavier's group, pursues the functional interpretation and clinical ramifications of these continued gene discovery efforts. More recently, the group also participates in numerous studies using exome sequencing to articulate the genetic origins of rare inherited diseases, early-onset and pediatric cancers, and severe adverse drug responses. Mark received his B.S. in physics from MIT and his PhD in human genetics from Leiden University, Netherlands.

Oliver Devuyst



Olivier Devuyst, MD, PhD, is Full Professor in the Institute of Physiology of the UZH and invited Professor at the Université catholique de Louvain (UCL) Medical School in Brussels, Belgium. He has a joint appointment in the Division of Nephrology of the Universitäts Spital (Zurich) and the Saint-Luc Academic Hospital (Brussels). Dr. Devuyst and his group investigate the molecular mechanisms of membrane transport and the pathophysiology of inherited renal tubular diseases, using a translational approach from human genetics to model systems. These studies, which have generated more than 250 peer-reviewed articles, are funded by national and international agencies, including the European Union (FP6, FP7), the Baxter Extramural Foundation and the National Institutes of Health. Dr. Devuyst has given more than 200 invited lectures and has been the laureate of several international prizes (including the Galien Prize in 2003, the International Spa Foundation Prize in 2007, and the Prix de la Fondation du Rein in 2009). He serves in the Editorial Board of *Kidney International*, *Pflügers Archiv* and *Frontiers in Renal and Epithelial Physiology*, and he is Associate/Speciality Editor of *Peritoneal Dialysis International* and *Nephrology Dialysis Transplantation*.

Dr. Devuyst has been elected at the Royal Academy of Medicine of Belgium in 2005. Since 2010, he coordinates the Working Group on Inherited Kidney Disorders (WGIKD) of the European Renal Association (ERA-EDTA). In 2014, he co-chaired (with Prof. V. Torres) the KDIGO conference on autosomal dominant polycystic kidney disease.

Iain Drummond



Iain Drummond received his PhD. from the University of California Berkeley for work on ionic signaling and the *Drosophila* heat shock response. His postdoctoral work at Northwestern University Medical School and the University of Chicago focused on gene regulation in *Dictyostelium* and the Wilms tumor suppressor in human cells. Dr. Drummond is currently an Associate Professor in Medicine and Genetics at Harvard Medical School / MGH. His lab established the zebrafish as a genetic system for discovering mechanisms of kidney development and disease. His work has revealed new genes required for cilia function in organogenesis and in human Primary Ciliary Dyskinesia, established collective cell migration as an essential element of nephron morphogenesis, discovered new pathways underlying nephrotic syndrome, revealed new cellular mechanisms linked to Polycystin gene functions, and developed the zebrafish pronephros as an *in vivo* system to evaluate pathogenicity of candidate human disease gene sequence polymorphisms.

Caroline Fox



Dr. Fox is a population scientist who brings together the techniques of traditional epidemiology, biomarkers, genetics and genomics, and radiographic imaging towards the pursuit of uncovering metabolic risk factors for cardiovascular disease. She has created a world-renowned computed-tomography body composition database, using standard imaging techniques in novel ways to create protocols for the assessment of compartments of body fat and pioneering the exploration of the concept that fat can have locally toxic effects. Dr. Fox also studies chronic kidney disease, and leads a large international consortium dedicated to uncovering genes for kidney function.

Ali Gharavi



Ali Gharavi MD is Professor of Medicine and Chief of the Division of Nephrology at the Columbia University College of Physicians and Surgeons. He obtained his M.D. degree at George Washington University, and completed training in internal medicine, hypertension and nephrology at the Mount Sinai Medical Center before pursuing post-doctoral studies in genetics at Yale University. His research is focused on understanding the genetic basis for kidney structure and function, and specifically, investigation of IgA nephropathy, the most common form of glomerulonephritis, and congenital malformations of the urinary tract, the main cause of pediatric end-stage kidney failure.

Joel Hirschhorn



Joel Hirschhorn is the Concordia Professor of Pediatrics and Professor of Genetics at Boston Children's Hospital and Harvard Medical School. He is a senior associate member of the Broad Institute, and co-directs the Broad Institute's Metabolism Program.

Dr. Hirschhorn's research focuses on using human genetics and genomics to identify genes that influence common diseases and quantitative traits, including obesity and height. Hirschhorn was an early advocate of collaboration, meta-analysis, and stringent statistical thresholds, which were essential elements of many recent discoveries in human genetics. He is a co-PI of the GENIE consortium studying the genetics of diabetic kidney disease. He continues to lead a large international consortium (GIANT) that has discovered almost all of the common genetic variants that are known to influence human height and weight. His laboratory also develops and implements novel computational methods to gain biological insights from human genetic data.

Dr. Hirschhorn received his A.B. summa cum laude in biochemistry from Harvard College and later earned his M.D. and Ph.D. in genetics from Harvard Medical School. As a graduate student, he employed yeast genetics to study chromatin structure and transcription with Fred Winston. He completed his postdoctoral training with Eric Lander at the Whitehead Institute/MIT Center for Genome Research, where he developed tools to interpret genetic association studies including genotyping technologies and analytic methods. He started his own laboratory at Boston Children's Hospital in 2001. In 2011, Hirschhorn received, from the Society of Pediatric Research, the E. Mead Johnson Award, a prestigious honor in pediatric research.

Oxana Ibraghimov-Breskovnaya



Dr. Oxana Ibraghimov-Beskovnaya is Head of Renal Research and Distinguished Scientific Fellow at Genzyme, a Sanofi company. Her laboratory investigates molecular pathogenesis of PKD and other renal disorders, and applies this knowledge to the development of novel treatments targeting key disease-promoting pathways. More recently she focused on the effects of dysfunctional cilia-cell cycle regulation and differentiation in PKD and other ciliopathies. This work resulted in discovery of cyclin dependent kinase

(CDK) inhibition as an effective approach for the treatment of PKD in preclinical trials. At present, the laboratory explores the effects of glycosphingolipid modulation on cystogenesis and fibrosis. Dr. Ibraghimov-Beskovnaya has also established Renal Biomarkers group focusing on discovery of urinary biomarkers for translational studies. She received her M.S. degree in genetics from the Moscow State University and her Ph.D. in molecular biology from the Institute for Genetics and Selection, Moscow, Russia. She completed her post-doctoral training at the University of Iowa, where she studied Duchenne Muscular Dystrophy and the role of dystrophin and dystrophin-associated glycoproteins in DMD and other neuromuscular diseases.

Peter Jackson



Dr. Jackson's lab focuses on enzyme mechanisms controlling cell growth and division, and cancer. At Stanford, he studied cell and centrosome cycles, SCF and APC/C E3 ubiquitin ligases and their inhibitors, cyclin-dependent and Polo kinases, and the Cdc14 and PP2A phosphatases. At Genentech, he has developed drugs targeting cell cycle checkpoints (including the Chk1 inhibitor currently in clinical trials), cellular stress pathways, and tumor metabolism. His lab studies protein modifying and regulatory enzymes

including acetyltransferases and E3 ubiquitin ligases controlling cell regeneration, cancer, and signaling in primary cilia. His lab has developed methods for proteomic network building and metabolomics to discover new human disease genes in ciliopathies, human degenerative diseases, and cancer.

Dontscho Kerjaschki



Dr. Dontscho Kerjaschki is the Chair of the Department of Pathology at the Medical University of Vienna. Dr. Kerjaschki received his MD and his license for Pathology and Cytology from the University of Vienna in the 1970s. He was Visiting Professor (1980-1989) at the Departments of Cell Biology at Yale and the University of California, San Diego. He has received several distinguished awards, and was elected a fellow of the Royal College and of the German National Academy of Sciences. He has served on the editorial boards of the Journal of the American Society of Nephrology, Journal of Clinical Investigation, and as Associate Editor of American Journal of Pathology. His own publications currently number 240. His research is focused on the biology and pathology of kidney glomerular diseases and on lymphatic vessels.

Cecilia Lo



Dr. Cecilia Lo is a developmental biologist with a passion for cardiovascular development and congenital heart disease. She received her BS degree from the Massachusetts Institute of Technology and her PhD from Rockefeller University. After postdoctoral training at Harvard Medical School, she relocated to University of Pennsylvania, where she rose through the ranks from Assistant Professor to full Professor in the Department of Biology. In 2001, she moved to the intramural research program at the National Heart Lung and Blood Institute as Chief of the Laboratory of Developmental Biology and in 2004, further appointed as the Director of the Genetics and Developmental Biology Center. Dr. Lo relocated in 2009 as the founding Chair and Professor of the new Department of Developmental Biology in the University of Pittsburgh School of Medicine, and holds the F. Sargent Cheever Chair. Her research interests has been focused on investigations into the developmental etiology of congenital heart disease, with her recent work focused on the use of forward genetic screen in mice to identify genes and pathways that cause structural heart defects. In addition, her laboratory has embarked upon clinical translational work in collaboration with clinician colleagues to leverage findings from her mouse modeling to investigate the developmental mechanisms and genetic etiology of human CHD. These studies have brought her unexpectedly to an appreciation of the important role of cilia biology in congenital heart disease, currently a central focus of research program in both her mouse modeling and human clinical translational studies.

Peter Mundel



Peter Mundel holds an MD from the University of Heidelberg, Germany, where he also began his career as a faculty member in the Department of Anatomy. Dr. Mundel is considered an international authority in podocyte biology, and has been working on the role of podocytes in kidney disease for more than 25 years. A major focus of Dr. Mundel's research is the development of novel treatments for glomerular kidney diseases through a detailed mechanistic understanding of the cell biology and pathology of podocytes.

For the past two decades, aided by sustained NIH funding, Dr. Mundel has worked on the validation of the kidney podocyte as a therapeutic target for proteinuria. Accordingly, his work published in *Nature Medicine* in 2008 showed for the first time that the anti-proteinuric effect of Cyclosporine A results not from its classically known immunomodulatory effect, but rather, from stabilizing the podocyte actin cytoskeleton, leading to a commentary in the *New England Journal of Medicine* titled "Proteinuria and immunity--an overstated relationship?"

The Mundel laboratory was also the first to report a role for B7-1/CD80 in podocytes in the pathogenesis of proteinuria, as B7-1 expression had been believed to be restricted to antigen presenting cells. Recently, in a paper published in the *New England Journal of Medicine*, Dr. Mundel and his team showed that the B7-1 inhibitor CTLA4-Ig induced remission of proteinuria in patients with severe and refractory nephrotic syndrome, a first-in-human discovery described by the associated editorial as a "A New Era of Podocyte-Targeted Therapy for Proteinuric Kidney Disease."

Dr. Mundel has also devoted substantial scientific effort to the study of synaptopodin, a molecule he discovered 25 years ago. Highly expressed in kidney podocytes and neurons, synaptopodin regulates the actin cytoskeleton. This work has led Dr. Mundel and his team into diverse areas of investigation, including studies of neurons, cardiac myocytes and cancer cells, as well as model organisms such as knockout mice and *Drosophila*. Similarly, the need for an appropriate podocyte cell culture model to study synaptopodin led Dr. Mundel to the development of the mouse immortalized podocyte cell line, an essential tool now freely shared with several hundred laboratories worldwide.

Dr. Mundel has held leadership roles in various settings throughout his career, as Vice Chair for Research in the Department of Medicine and Chief of Molecular Medicine at the University of Miami, and as Associate Chief for Translational Research in the Nephrology Division at MGH.

Dr. Mundel has given more than 100 invited lectures nationally and internationally and he is the recipient of numerous awards including the Heisenberg Award from the German Research Foundation, the Wolfgang-Bargman Prize from the German Anatomical Society, and the Young Investigator Award from the American Society of Nephrology and the American Heart Association.

Yukinori Okada



Dr. Yukinori Okada is a Tenure-track Junior Associate Professor at Tokyo Medical and Dental University, Japan. He received M.D. and Ph.D. from University of Tokyo. He has multiple professional backgrounds as a rheumatologist, a statistician, and a bioinformatician. Through international collaboration partnerships, he has identified more than a hundred of novel disease risk loci of multiple human complex traits by large scale genome-wide association studies (GWAS). His recent research interests includes

genetic and bioinformatics analysis integrating big data obtained by technologies in the post-GWAS era, such as next generation sequencing or typing of rare functional variants, and its application to novel drug discovery (Okada et al. Genetics of rheumatoid arthritis contributes to biology and drug discovery. *Nature* 2014).

Randall Peterson



Randall Peterson received his PhD from Harvard University where he studied as a Howard Hughes Medical Institute predoctoral fellow in the laboratory of Stuart Schreiber. As a graduate student, he pioneered the first *in vivo* high-throughput chemical screens using zebrafish. Following graduation, he completed a postdoctoral fellowship with Mark Fishman at Massachusetts General Hospital. Dr. Peterson is currently Associate Professor of Medicine at Harvard Medical School, Scientific Director of the MGH Cardiovascular Research Center, and Senior Associate Member of the Broad Institute. He is the recipient of a EUREKA award from the National Institutes of Health and is the Charles and Ann Sanders MGH Research Scholar.

Martin Pollack



Dr. Pollack received a bachelors degree from of Princeton University and an M.D. from New York University School of Medicine. He completed a residency in Internal Medicine at Columbia-Presbyterian Medical Center in New York, and clinical training in nephrology at the Brigham and Women's Hospital, Boston. After a research fellowship in the Department of Genetics of Harvard Medical School, Dr. Pollak joined the nephrology faculty at Brigham and Women's Hospital and Harvard Medical School. In 2010, he assumed the role of Chief of Nephrology at Beth Israel Deaconess Medical Center where he is also Professor of Medicine at Harvard Medical School.

Susan Quaggin



Susan Quaggin, MD graduated from the Faculty of Medicine at the University of Toronto in 1988 and received her specialty degree in Internal Medicine in 1992. She completed her sub-specialty training in Nephrology in 1993 at U of T and did a post-doctoral fellowship at Yale University where she studied the genetic basis of kidney development.

In 1997, she returned to Toronto to do a second post-doctoral fellowship in mouse genetics in the laboratory of Janet Rossant. From 1997 until 2012, she was at the University of Toronto where she was a Senior Scientist at the Samuel Lunenfeld Research Institute, a practicing Nephrologist at St. Michael's Hospital and the Gabor-Zellerman Professor in Renal Medicine.

Quaggin has served as an elected councillor of the American Society for Clinical Investigation (ASCI), is a member of the ISN Council and was elected to the American Association of Physicians (AAP) in 2013. She received the Kidney Foundation of Canada 2009 Award for Research and a Finnish Distinguished Professorship in 2012. In addition, Quaggin sits on the editorial boards of several journals, is the Deputy Editor of the Journal of the American Society of Nephrology and has organized a number of international renal and vascular meetings.

In January 2013, Quaggin joined Northwestern University Feinberg School of Medicine as the Charles Horace Mayo Professor of Medicine, where she serves as the director of the Feinberg Cardiovascular Research Institute (FCVRI) and chief of the Division of Nephrology.

Quaggin's research program focuses on genetic pathways required to establish and maintain the integrity of microvascular beds including the glomerular filtration barrier – a highly selective filter that separates the blood from the urinary space. To understand the pathways and interactions between perivascular cells and the endothelium, her research team has developed a number of genetic models that permit cell and time-specific manipulation of gene expression.

Stefan Somlo



Dr. Somlo is the C. N. H. Long Professor of Medicine (Nephrology) and Professor of Genetics and serves as the Chief of the Section of Nephrology at the Yale University School of Medicine. Dr. Somlo's research interests are the genetic bases of human polycystic kidney and liver diseases with the goal of understanding the role cilia and polycystin signaling pathways in both normal adult tissue homeostasis and in progression of cystic diseases. His laboratory has discovered genes for human autosomal dominant polycystic kidney disease (PKD2), isolated polycystic liver disease (PRKCSH, SEC63) and recessive polycystic kidney disease (PKHD1). They have used these discoveries to define both genetic and cellular mechanisms underlying the respective human diseases. Dr. Somlo is a graduate of Harvard College and the College of Physicians and Surgeons of Columbia University. He did his clinical training at the Albert Einstein College of Medicine and Yale and was on faculty at Albert Einstein before returning to Yale. He is a member of the American Society for Clinical Investigation and Association of American Physicians for which he is also a Councilor. He is a recipient of the Lillian Jean Kaplan International Prize for Polycystic Kidney Disease Research and the Homer W. Smith Award from the American Society of Nephrology.

Fabiola Terzi



Fabiola Terzi, M.D, PhD, is Director of Research at INSERM (Institut National de la Sante et de la Recherche Medicale) and head of the team « Mechanisms and Therapeutic Strategies of Chronic Kidney Disease » at Paris Descartes University, Necker Hospital, France. She obtained a MD from the University of Milan and certified in Pediatrics and Nephrology at the Universities of Milan and Paris Descartes. She received her PhD in Physiopathology at Paris 7 University. Since 2014, she co-head the Department of « Cell & Growth » at the Institut Necker Enfants Malades, in Paris. Her major research interests focus on the mechanisms that control the progression of chronic kidney disease (CKD). Towards this goal, she has developed several experimental models of CKD in mice from different genetic background and genetically modified animals. She has also fostered several translational studies aiming at identifying novel biomarkers and targets of CKD progression in humans. Dr Terzi has been awarded the « Grand Prix de l'Academie Nationale de Medecine » and the « Prix de la Fondation du Rein ».

Vicente Torres



Vicente Torres received MD and Doctoral degrees from the University of Barcelona and moved to the Mayo Clinic in 1972 for research training and residencies in Internal Medicine and Nephrology. He joined the faculty at this institution in 1979 and became Professor of Medicine in 1991. He has served as chair of the Division of Nephrology at Mayo, Director of the NIH funded Mayo Kidney Disease Research Training Grant, on the Scientific Advisory Board of the PKD Foundation, and on several NIH study sections and committees. His research in the last twenty years has focused on PKD. He directs the NIH funded Mayo PKD Translational Center, is a principal investigator for the NIH funded CRISP imaging and HALT-PKD clinical trial consortia, and for Industry funded clinical trials of vasopressin V2 receptor antagonists. He was awarded the 2007 Lillian Jean Kaplan International Prize for Advancement in the Understanding of Polycystic Kidney Disease.

Sakari Vanharanta



Sakari Vanharanta completed his undergraduate studies at the University of Helsinki, Finland, where he attended the MD/PhD programme. After a short period of clinical training, he moved to New York to work as a postdoctoral fellow at the Memorial Sloan-Kettering Cancer Center. In 2014, he joined the Medical Research Council Cancer Unit at the University of Cambridge as a junior group leader.

Leonard Zon



Dr. Zon is the Grousbeck Professor of Pediatric Medicine at Harvard Medical School, Investigator at Howard Hughes Medical Institute, and Director of the Stem Cell Program, Children's Hospital Boston. Dr. Zon received a B.S. degree in chemistry and natural sciences from Muhlenberg College (1979) and an M.D. degree from Jefferson Medical College (1983). He subsequently did an internal medicine residency at New England Deaconess Hospital (1986) and a fellowship in medical oncology at Dana-Farber Cancer Institute (1989). His postdoctoral research was in the laboratory of Stuart Orkin (1990). Dr. Zon is internationally recognized for his pioneering work in the fields of stem cell biology and cancer genetics. Dr. Leonard Zon has been the pre-eminent figure in establishing the zebrafish as an invaluable genetic model for the study of the blood and hematopoietic development. He is founder and former president of the International Society for Stem Cell Research and chair of the Executive Committee of the Harvard Stem Cell Institute (HSCI). In 2005, he completed a term as President of the American Society for Clinical Investigation. In that same year, Dr. Zon was elected to the Institute of Medicine of the National Academies. In 2008, Dr. Zon was elected to the American Academy of Arts & Sciences. In 2010, Dr. Zon was awarded the E. Donnall Thomas Lecture and Prize from American Society of Hematology. In 2013, Dr. Zon received the ISEH Donald Metcalf Lecture Award.