

**THE INTERNATIONAL PRIZE FOR SCIENTIFIC RESEARCH ARRIGO RECORDATI 2019**

**Norio Sakai, M.D. Ph.D**



**Child Healthcare and Genetic Science  
Laboratory, Division of Health Science,  
Osaka University, Japan**

Norio Sakai is a Professor at the Osaka University Graduate School of Medicine, in the Child Healthcare and Genetic Science Laboratory of the Health Science Division, in Osaka. Dr. Sakai graduated from the Department of Astronomy at Tokyo University in 1982 and the Osaka University Graduate School of Medicine in 1987. He completed his clinical training as a residency in 1990. He received a Ph.D from the Osaka University Graduate School of Medicine in 1994 with a paper on molecular cloning for Krabbe disease (*Krabbe disease: Isolation and characterization of a full-length cDNA for human galactocerebrosidase*).

He worked as a Research Fellow in the Department of Environmental Medicine, Res. Inst. Osaka Medical Center for Maternal and Child Health, Osaka for two years and moved to GSF-Institute for Mammalian Genetics in Germany.

He returned to Japan in 1998 to the Department of Paediatrics at the Osaka University School of Medicine and was appointed to the position of Assistant Professor in 2005, later working as Associate Professor from 2009. He led the clinical and research group for inborn errors of metabolic disease and medical genetics in the Pediatrics Department.

Dr. Sakai has published more than 60 articles in peer-reviewed international journals and has received several awards for his research on molecular analysis for inborn errors of metabolic diseases.

His clinical work focuses on the clinical treatment of lysosomal diseases with advanced methods including enzyme replacement therapy and hematopoietic cell transplantation. His more recent research has focused on disclosing the basic molecular pathology of lysosomal diseases and developing new treatments including chaperone therapy.

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**Francesco Emma M.D.**

**Director of the Division of Pediatric Nephrology  
Chief of the Department of Pediatric  
Subspecialties, IRCCS Children's Hospital  
Bambino Gesù, Rome, Italy**



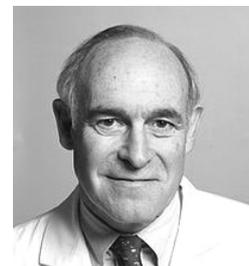
Francesco Emma received his medical degree from the Catholic University of Louvain, Brussels, Belgium, where he specialized in Pediatrics. He subsequently completed his training in Pediatric Nephrology at Boston Children's Hospital, Harvard Medical School, and moved to the Bambino Gesù Children's Hospital in Rome as a staff member in the Division of Nephrology and Dialysis. There, he served as the Head of the Rare Kidney Disease Clinic before being appointed Head of the Pediatric Nephrology Division in 2005, and has set-up the Nephrology Research Laboratory. He currently holds the position of Head of the Department of Pediatric Subspecialties.

Dr. Emma's primary research interests lie in rare renal diseases, in particular in cystinosis and nephrotic syndrome. He has authored more than 160 research articles in peer-reviewed journals and is the author of several textbook chapters. He has also served in the editorial board of several scientific journals, including Pediatric Nephrology, the Journal of Nephrology and Nephrology Dialysis and Transplantation. He is an editor for the textbook "Pediatric Nephrology".

Among his other appointments, he is the current chair of the Working Group on Inherited Renal Disorders of the European Society of Pediatric Nephrology (ESPN), has been past-secretary of the Working Group on Inherited Kidney diseases of the European Renal Association - European Dialysis and Transplant Association (ERA-EDTA), the past Chair of the Teaching Course Committee of the International Society of Pediatric Nephrology and the past-President of the Italian Society of Pediatric Nephrology.

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**Robert J. Desnick Ph.D., M.D.**



**Icahn School of Medicine at Mount Sinai  
Mount Sinai Health System, New York, NY**

Robert J. Desnick is Dean for Genetic and Genomic Medicine and Professor and Chairman Emeritus of the Department of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai.

In 1977, he joined the Mount Sinai faculty as the Arthur J. and Nellie Z. Cohen Professor of Pediatrics and Genetics, and Chief of Medical and Molecular Genetics. From 1993-2011, he was the first Chairman of the Department of Genetics and Genomic Sciences at Mount Sinai. In 2011 he became the Dean for Genetics and Genomic Medicine.

Dr. Desnick's research interests include lysosomal storage diseases (LSDs) and the inborn errors of heme biosynthesis, the porphyrias, and in particular, their treatment. His research efforts led to the Federal Drugs Administration (FDA) - and European Medicine Agency (EMA) - approval of enzyme replacement therapy (ERT) for Fabry disease (Fabrazyme) and on-going ERT clinical trials (FDA "Breakthrough" status) for Niemann-Pick B disease, both in partnership with Genzyme. In addition, he was a scientific founder of Amicus Therapeutics (NASDAQ; FOLD), which is developing oral pharmacologic chaperone therapy for Fabry disease (EMA-approved in 2016), Pompe disease, and other disorders. Currently, his laboratory is using gene editing technology to engineer gene therapy in the mouse model of Fabry disease with Sangamo Therapeutics.

For the porphyrias, he co-developed with Alnylam Pharmaceuticals an RNAi therapy for the acute hepatic porphyrias, performed the preclinical studies, and co-designed the on-going clinical trials. He also served as Principal Investigator for Clinuvel Pharmaceuticals' Phase 2 and 3 multisite clinical trials of afamelanotide, a "first-in-class" synthetic peptide for the treatment of the Erythropoietic Porphyrias, which was recently EMA-approved and pending FDA approval. He also served as the Chairman of the Scientific Advisory Committee (SAC) of Synageva Biopharma and currently serves as SAC Chair for Kiniksa Pharmaceuticals. In addition, his research includes genomics, pharmacogenomics, and personalized medicine. He has published over 740 research papers and chapters, including nine edited books. He is an elected Fellow of the American Association for the Advancement of Science and an elected member of the National Academy of Medicine.