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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.