

NTSAD Research Initiative

Year Awarded	Investigator(s)	Institution	Project	Type of Project	NIH follow-up success
2011	Florian Eichler, MD	Massachusetts General Hospital	Clinical Outcome Measures for a Gene Therapy Trial in Infantile and Juvenile GM2	natural history study (TSGT Consortium)	
	Guangping Gao, PhD	University of Massachusetts Medical School	Optimization of Efficacious Gene Therapy for Canavan Disease	gene therapy	
	Yu-Tah Li, PhD	Tulane University	Studies of Taurine-Conjugated GM2 in Tay-Sachs Disease	novel marker	
2010	Fran Platt, PhD	University of Oxford	Optimizing the Therapeutic Potential of Anti-inflammatory Therapy in Tay-Sachs and Related Diseases: Targeting IL-1 β Generated by Aberrant NLRP3 Inflammasome	small molecules	
	Maria Traka, PhD	University of Chicago	Development of an in vitro approach to identify molecular pathways of Canavan disease	basic research	
	Jean-Pyo Lee, PhD / Evan Y. Snyder, MD, PhD	Tulane University	The Therapeutic Potential of Human Induced Pluripotent Stem Cells (iPSCs) in the Sandhoff Disease Mouse Model of Lysosomal Storage Disorders.	stem cell therapy	
	Gustavo Maegawa, PhD	Johns Hopkins University	Developing a High Throughput Screening Assay to Identify Potential Drugs for Metachromatic Leukodystrophy	small molecules	
	Doug Martin, PhD	Auburn University	Sheep as a Model of Tay-Sachs Disease	gene therapy (TSGT Consortium)	
2009	Alexey Pshzhetsky, PhD	Universite of Montreal	Novel therapy for Tay-Sachs disease, sialidosis and galactosialidosis using a metabolic bypass catalyzed by the lysosomal sialidase Neu4	novel marker	
	Mark Sands, PhD	Washington University	Combination Therapy for Krabbe Disease	combination therapy	
	Joe Clarke, MD, PhD	Hospital for Sick Children	Proposed Investigator-Initiated Clinical Trial of Pyrimethamine as a Treatment for Late-Onset GM2 gangliosidosis (Tay-Sachs and Sandhoff Disease)	phase I clinical trial	
	Florian Eichler, MD	Massachusetts General Hospital	A Biomarker for Disease Progression in GM2 and other Neurolipidoses	biomarker	
	Edwin Kolodny, MD	NYU	Proposed Investigator-Initiated Clinical Trial of Pyrimethamine as a Treatment for Late-Onset GM2 gangliosidosis (Tay-Sachs and Sandhoff Disease)	phase I clinical trial	
2008	Stephanos Kyrkanides, PhD	Stony Brook University	Retrograde transfer of therapeutic vectors enabled by the trigeminal sensory system	gene therapy	
	Angela Gritti, PhD / Alessandra Biffi, PhD	San Raffaele	Evaluation of Combined Approaches Using Hematopoietic and Neural Stem Cells for the Treatment of Globoid Cell Leukodystrophy	combination therapy (stem cells)	
2007	Florian Eichler, MD	Massachusetts General Hospital	The Natural History of Tay-Sachs Disease	gene therapy (TSGT Consortium)	
	Miguel Sena-Esteves, PhD	Massachusetts General Hospital	AAV-mediated Gene Therapy for Tay-Sachs Disease: Vector Selection for Preclinical Development	gene therapy (TSGT Consortium)	yes
	Timothy Cox, MD M. Begoña Cachón-	University of Cambridge	Pre-Clinical/Clinical Research Program: Tay-Sachs and Related Diseases	gene therapy (TSGT Consortium)	yes
	Douglas Martin, PhD	Auburn University	Pre-Clinical Studies of AAV Gene Therapy in Feline GM2 Gangliosidosis	gene therapy (TSGT Consortium)	yes
	Thomas Seyfried, PhD	Boston College	Neurochemical and Immunological Evaluation of AAV Gene Therapy Strategies	gene therapy (TSGT Consortium)	yes
2007	Susan L. Cotman, PhD	Massachusetts General Hospital	Small molecule screening to identify modifiers of lysosomal trafficking, a putative therapy for Batten disease	small molecules	yes
	Doug Martin, PhD	Auburn University	Pre-clinical gene therapy for GM2 in a feline model	gene therapy (TSGT Consortium)	yes
	Miguel Sena-Esteves, PhD	Massachusetts General Hospital	AAV-mediated gene therapy for Tay-Sachs: Vector selection for pre-clinical development	gene therapy (TSGT Consortium)	yes
	Aryan Namoodiri, PhD	Uniformed Services University of the Health Sciences	Preclinical Research toward Acetate Supplementation Therapy for Canavan Disease	small molecules	yes
2004	James A. Shayman, MD	University of Michigan	High throughput screening for inhibitors of ganglioside GM2 synthase	small molecules	
2003	Jean-Pyo Lee, PhD/Evan Y. Snyder, MD, PhD	Beth Israel Deaconess Medical Center/Burnham Institute	Therapeutic Potential of Neural Stem Cells in the Gangliosidoses (Tay-Sachs & Sandhoff Diseases)	stem cell therapy	
	Cynthia Tift, MD, PhD	Children's Research Institute of Children's National Medical Center	Comprehensive Biochemical Analysis of Cerebrospinal Fluid in Patients with GM2 Storage Disorders:Molecular Pathogenesis of Disease Progression	biomarkers	
2002	Bruce A. Bunnell, PhD	Tulane University	In utero Gene Therapy of Sandhoff Disease in a Murine Model	gene therapy	
	Stephanos Kyrkanides., PhD	University of Rochester School of Medicine & Dentistry	Perinatal Gene Therapy for β -hexosaminidase disorders (Tay-Sachs and Sandhoff diseases)	gene therapy	yes
	Paola Leone, PhD	University of Medicine and Dentistry of New Jersey	Neuroprotective Effect of Minocycline in Sandhoff Disease	small molecules	
	Thomas N. Seyfried, PhD	Boston College	Therapeutic evaluation of NB-DGJ for ganglioside storage diseases	substrate reduction	yes
NOTE: There are 13 additional Lysosomal Storage Disease Reseach Consortium projects funded 2006-2008. See LSDRC page in Research We Fund section for more information.					