



**PTNC**

Penn Medicine

**Translational Neuroscience**

Center

# **Neuro Gene Therapy Symposium**

**Thursday and Friday  
October 12 – 13, 2023**

**Gaulton Auditorium  
Biomedical Research Building**

**University of Pennsylvania  
Philadelphia, PA**



**Perelman**  
SCHOOL OF MEDICINE  
UNIVERSITY OF PENNSYLVANIA



# Neuro Gene Therapy Symposium

**Thursday and Friday  
October 12 – 13, 2023  
Gaulton Auditorium, BRB  
University of Pennsylvania**

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## **Day 1: Thursday, October 12, 2023**

**08:00 am – 09:00 am**      **Check-in & Breakfast**

**09:00 am – 09:15 am**      **Welcome & Introductory Remarks**  
**Frances Jensen, MD & Raquel Gur, MD, PhD; Co-Directors, PTNC**

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## **Session I: Introduction to Gene Therapy & Current Scope – Chair: Frances Jensen, MD**

**09:15 am – 09:45 am**      **Kenneth (Kurt) Fischbeck, MD**, NIH Distinguished Investigator, National Institute of Neurological Disorders and Stroke (NINDS)  
*Diagnosis and Treatment of Hereditary Neurological and Neuromuscular Diseases*

**09:45 am – 10:15 am**      **Daniel J. Rader, MD**, Seymour Gray Professor of Molecular Medicine; Chair of the Department of Genetics Chief of the Division of Translational Medicine and Human Genetics in the Department of Medicine, University of Pennsylvania  
*Translational Resources for Genetics at Penn*

**10:15 am – 11:00 am**      **Joint Talk:**

**Lauren Elman, MD**, Professor of Neurology, Director of the Muscular Dystrophy Association Clinic; Medical Center Director of the Penn Comprehensive Amyotrophic Lateral Sclerosis Center, University of Pennsylvania  
*Two Clinics Separated By 20 Years & a Cautionary Tale*

**Ingo Helbig, MD**, Assistant Professor, Division of Neurology, Children's Hospital of Philadelphia and the Clinical Director, Center for Epilepsy and Neurodevelopmental Disorders (ENDD) at University of Pennsylvania and Children's Hospital of Philadelphia  
*Clinical Trial Readiness and Natural History in Pediatric Epilepsies and Neurodevelopmental Disorders*

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**11:00 am - 11:15 am: Break**

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## **Session II: Successes & Current Landscape - Chair: Steven Scherer, MD, PhD**

- 11:15 am – 11:45 am**     **Jean Bennett, MD, PhD**, F.M. Kirby Professor of Ophthalmology, Department of Cell and Developmental Biology and Vice Chair for Basic Research, Department of Ophthalmology; Director of DNA, Tissue and Cell Line Bank for Retinal Degeneration & Molecular Diagnostic Studies Unit for Patients with Retinal Degeneration, F. M. Kirby Center for Molecular Ophthalmology, University of Pennsylvania  
*The Luxturna Experience: Insights Into Development of Gene-Based Treatments for Other Blinding Disorders* (pre-recorded talk)
- 11:45 am – 12:15 pm**     **Toby Ferguson, MD, PhD**, Head Neuromuscular and Movement Disorders Development Unit, Biogen  
*Use of Neurofilament in Development of Tofersen for SOD1 ALS*
- 12:15 pm – 12:45 pm**     **Steven Scherer, MD, PhD**, Ruth Wagner Van Meter and J. Ray Van Meter Professor of Neurology, Director of Penn Neurogenetics Therapy Center, University of Pennsylvania  
*Successful Treatments for Transthyretin Amyloidosis (ATTR)*
- 12:45 pm – 01:15 pm**     **Kevin Flanigan, MD**, Robert F. and Edgar T. Wolfe Foundation Endowed Chair in Neuromuscular Research; Director, Center for Gene Therapy at the Abigail Wexner Research Institute, Nationwide Children's Hospital  
*Next Directions in Dystrophinopathy Gene Therapy*

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***01:15 pm – 2:15 pm: Lunch/Break***

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## **Session III: Different Methodologies/Mechanisms – Chair: Frances Jensen, MD**

- 02:15 pm – 02:45 pm**     **Beverly L. Davidson, PhD**, Katherine A. High Chair in Cell and Gene Therapy, Director, Raymond G. Perelman Center for Cellular and Molecular Therapeutics, Chief Scientific Strategy Officer, Children's Hospital of Philadelphia and Professor of Pathology and Laboratory Medicine, University of Pennsylvania  
*Advancing Brain Gene Therapies*
- 02:45 pm – 03:15 pm**     **Drew Weissman, MD, PhD, Nobel Laureate**  
Roberts Family Professor in Vaccine Research; Director, Penn Institute for RNA Innovation; Director, Vaccine Research in the Infectious Disease Division; University of Pennsylvania  
**2023 Nobel Prize in Physiology or Medicine**  
*Nucleoside-Modified mRNA-LNP Therapeutics*
- 03:15 pm – 03:45 pm**     **Ricardo Dolmetsch, PhD**, President and Chief Scientific Officer, uniQure  
*Developing a Gene Therapy to Treat Huntington's Disease*

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***03:45 pm – 4:00 pm: Break***

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#### **Session IV: Frontiers & Vision Forward – Chair: Raquel Gur, MD, PhD**

- 04:00 pm – 04:30 pm**     **Benjamin L. Prosser, PhD**, Associate Professor, Physiology; Director, Center for Epilepsy and Neurodevelopmental Disorders (ENDD); Associate Director, Pennsylvania Muscle Institute; Lead Coordinator, Leducq Transatlantic Network of Excellence (Leducq Cytoskeletal Network), University of Pennsylvania  
*Developing and Accelerating Therapeutics for Rare Neurodevelopmental Disorders* (pre-recorded talk)
- 04:30 pm – 05:00 pm**     **Steven E. Hyman, MD**, Core Institute Member, Director of the Stanley Center for Psychiatric Research, Broad Institute at MIT and Harvard  
*Schizophrenia: The Challenging Path from Genetics to Therapeutics*
- 

#### **Day 2: Friday, October 13, 2023**

- 08:00 am – 08:30 am**     **Check-in & Breakfast**
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#### **Session V: Academic Gene Therapy Centers - Chair: Colin Quinn, MD**

- 08:30 am – 09:00 am**     **Sara-Claude Michon, PhD**, Director of Operations, Penn Neurogenetics Therapy Center, University of Pennsylvania  
**Amy T. Waldman, MD, MSCE**, Associate Director, Clinical In Vivo Gene Therapy, Children's Hospital of Philadelphia  
*Joint talk: Setting Up an Academic Gene Therapy Center: Penn Neurogenetics Therapy Center and CHOP Clinical In Vivo Gene Therapy Center*
- 09:00 am – 09:30 am**     **James Wilson, MD, PhD**, Director, Gene Therapy Program; Rose H. Weiss Professor and Director, Orphan Disease Center; Professor of Medicine and Pediatrics, University of Pennsylvania  
*A Penn-Based Platform for Global Delivery of AAV Vectors to CNS*
- 09:30 am – 10:00 am**     **Pedro Gonzalez-Alegre, MD, PhD**, Head of Gene Therapy Research, Spark Therapeutics, Inc  
*From Academia to Industry, and Back*
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#### **10:00 am – 10:15 am: Break**

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#### **Session VI: Ethics & Regulatory Issues – Chair: Lauren Elman, MD**

- 10:15 am – 10:45 am**     **Holly Tabor, PhD**, Associate Professor of Medicine at Stanford University; Associate Director for Clinical Ethics and Education for the Stanford Center for Biomedical Ethics (SCBE), Co-Chair of the Ethics Committees at Stanford Hospital and Lucile Packard Children's Hospital  
*Ethical Issues of Gene Therapy*
- 10:45 am – 11:15 am**     **Peter Marks, MD, PhD**, Director of the Center for Biologics Evaluation and Research (CBER) at the Food and Drug Administration (FDA)  
*Facilitating the Development and Availability of Gene Therapy for Small Populations*

**11:15 am – 11:45 am**      **Allyson Berent, DVM, DACVIM**, Chief Science Officer, Foundation for Angelman Syndrome Therapeutics (FAST); Co-Director, Angelman Syndrome Biomarker and Outcome Measure Consortium; Chief Development Officer, Mahzi Therapeutics  
*A Journey Through Drug Development: How parents and patients are taking the lead: The Angelman Story of making the impossible possible*

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**11:45 am – 12:00 pm: Break**

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**Session VII: Future Directions & Emerging Targets – Chair: Stewart Anderson, MD**

**12:00 pm – 12:30 pm**      **David Irwin, MD**, Assistant Professor, Neurology; Co-Director, Penn Frontotemporal Degeneration Center, University of Pennsylvania  
*Hope Renewed: Gene Therapy Research for FTD*  
**Caregiver speaker** – recorded talk with live Q&A

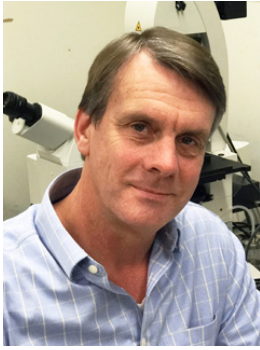
**12:30 pm – 01:00 pm**      **Erik Roberson, MD, PhD**, Professor, Rebecca Gale Endowed Professor, Vice Chair for Basic & Translational Research, Department of Neurology; Director, Alzheimer's Disease Center; Director, Center for Neurodegeneration and Experimental Therapeutics, University of Alabama at Birmingham  
*Optimizing Progranulin Gene Therapy for Frontotemporal Dementia Using Preclinical Models*

**01:00 pm – 01:30 pm**      **Michael Kaplitt, MD, PhD**, Professor of Neurological Surgery; Residency Director and Vice Chairman for Research, Weill Cornell Medical College  
*Opportunities and Challenges for Gene Therapies in Neurodegenerative Diseases*

**Concluding Remarks**      **Raquel Gur, MD, PhD & Frances Jensen, MD; Co-Directors, PTNC**

**take away lunch available**

**The PTNC would like to thank the efforts of our symposium organizing committee listed here:**



Stewart Anderson, MD  
Professor of Psychiatry  
Associate Chair for Research, Child Psychiatry  
Associate Director, Lifespan Brain Institute  
Children's Hospital of Philadelphia and UPenn School of Medicine  
<https://www.research.chop.edu/anderson-laboratory>  
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Professor Departments of Psychiatry, Neurology & Radiology  
Perelman School of Medicine  
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and  
Sr. Vice Chair for Research and Penn-CHOP Research Integration  
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Director, Lifespan Brain Institute,  
Children's Hospital of Philadelphia and Penn Medicine  
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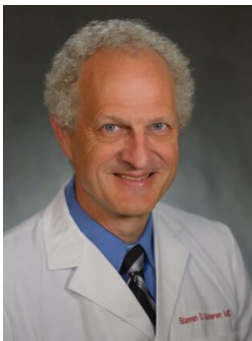
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Chair of the Department of Neurology  
Co-Director Penn Medicine Translational Neuroscience Center  
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## Speaker Bios in Order on Agenda

Thursday, October 12, 2023

### Session I



**Kenneth (Kurt) Fischbeck, MD**

**NIH Distinguished Investigator, National Institute of Neurological Disorders and Stroke (NINDS)**

Dr. Fischbeck trained at Harvard, Hopkins, and UCSF. He joined the Penn Neurology Department in 1982 and in 1998 came to NINDS as Neurogenetics Branch Chief. He has received awards from AAN and ANA and was elected to the National Academy of Medicine in 2000. His research group studies the causes and mechanisms of hereditary neuromuscular diseases in cell culture models and does natural history and clinical proof of concept studies to develop safe and effective treatment. He has worked on

Duchenne muscular dystrophy, spinal muscular atrophy, Charcot-Marie-Tooth disease, Friedreich's ataxia, and particularly spinal and bulbar muscular atrophy (SBMA, Kennedy's disease), the first repeat expansion neuromuscular disease to be identified.



**Daniel J. Rader, MD**

**Seymour Gray Professor of Molecular Medicine; Chair of the Department of Genetics; Chief of the Division of Translational Medicine and Human Genetics in the Department of Medicine; Associate Director of Penn's Institute for Translational Medicine and Therapeutics; Co-Director of the Penn Medicine Biobank, Perelman School of Medicine at the University of Pennsylvania**

Dr. Rader trained in internal medicine at Yale-New Haven Hospital and in human genetics and physiology of lipoprotein metabolism at the National Institutes of Health. He has been on the Penn faculty since 1994. He is a recipient of several national awards, including the Clinical Research Award from the American Heart Association. He is currently the deputy editor of the journal Arteriosclerosis, Thrombosis and Vascular Biology, the Chief Scientific Advisor to the Familial Hypercholesterolemia Foundation, and serves on the Board of Directors of the International Society for Atherosclerosis, the Board of External Experts of the National Heart Lung and Blood Institute, and the Advisory Board for Clinical Research for the NIH. Dr. Rader has been elected to the American Society of Clinical Investigation, the Association of American Physicians, and the Institute of Medicine of the National Academy of Sciences (now the National Academy of Medicine).



**Lauren Elman, MD**

**Professor of Neurology; Director of the Muscular Dystrophy Association (MDA) Center; Director of the Penn Comprehensive Amyotrophic Lateral Sclerosis (ALS); University of Pennsylvania**

Lauren Elman, MD got her undergraduate degree from Cornell University and her medical degree from Cornell University Medical College. She has been at Penn since then where she did her internship, residency and neuromuscular training. She currently serves as the Director of the MDA Center and the Penn Comprehensive ALS Center. Patient care is focused on those with ALS and adults with inherited disorders of the motor nerve and muscle. Her research interests include phenotype-genotype correlations and early phase clinical trials.



**Ingo Helbig, MD**

**Assistant Professor, Division of Neurology, Children's Hospital of Philadelphia and the Clinical Director, Center for Epilepsy and Neurodevelopmental Disorders (ENDD) at University of Pennsylvania and Children's Hospital of Philadelphia**

Dr. Helbig is the Director of Genomic Science at the Epilepsy NeuroGenetics Initiative (ENGIN) at Children's Hospital of Philadelphia, one of the largest epilepsy genetics centers in the US. He is also the Clinical Director of the recently established Center for Epilepsy and Neurodevelopmental Disorders (ENDD) at Children's Hospital of Philadelphia and the University of Pennsylvania where he is currently

focusing on novel tools to map the natural history of STXBP1-related disorders and SYNGAP1-related disorders for clinical trial readiness. Dr. Helbig went to medical school in Heidelberg and Mannheim, Germany, and Lexington, Kentucky, USA. He trained at the Epilepsy Research Centre, Melbourne, Australia, and was Assistant Professor at the Department of Neuropediatrics, Kiel, Germany. Between 2011 and 2015, he co-headed the EuroEPINOMICS-RES Consortium, the European counterpart of the NIH-funded Epi4K consortium involved in collaborative genomic studies to identify genes for human epilepsies. He was part of the Genetics Commission of the International League Against Epilepsy (ILAE) from 2014-2017 and currently leads the Epilepsioime Task Force of the ILAE Genetics Commission, which aims at increasing genetic literacy in the epilepsy community. After heading the epilepsy genetics group at the University of Kiel, Germany, he transferred to the Children's Hospital of Philadelphia (CHOP) in 2014 and became faculty in the Division of Neurology in July 2017. The main focus of his prior work was to understand how genetic changes lead to severe epilepsies in both children and adults, contributing to several new gene findings in the field in the last seven years including GRIN2A, CHD2, KCNA2, HCN1, and DNMT1. Dr. Helbig uses clinical and research expertise to curate epilepsy-related genes in variants within his leadership role of the Epilepsy Clinical Domain Working Group.

## Session II



**Jean Bennett, MD, PhD**

**F.M. Kirby Professor of Ophthalmology, Department of Cell and Developmental Biology and Vice Chair for Basic Research, Department of Ophthalmology; Director of DNA, Tissue and Cell Line Bank for Retinal Degeneration & Molecular Diagnostic Studies Unit for Patients with Retinal Degeneration, F. M. Kirby Center for Molecular Ophthalmology, University of Pennsylvania**

Jean Bennett joined University of Pennsylvania's School of Medicine in 1992 and has spent the past 3 decades developing gene-based strategies for treating inherited retinal degenerations. She has run a true bench-to-bedside translational research program and, in the process, has trained hundreds of physician-scientists, many of whom are now leaders in translational research around the globe. She was the scientific leader of a team that translated reversal of blindness in animal models to demonstration of efficacy and safety of gene therapy in children and adults. She was the scientific director of clinical studies at the Children's Hospital of Philadelphia that led to first FDA-approved gene therapy for a genetic disease (blindness due to RPE65 deficiency, studies that were sponsored by a company that she co-founded (but in which she waived financial benefit), Spark Therapeutics). She helped develop the primary outcome measure for that trial that led in 2017 to the first US FDA-approved gene therapy product. This was subsequently approved by the European Medicines Agency. In 2023, Jean became Professor Emeritus but continues to develop gene-based therapies for blinding diseases at UPenn and with other biotechnology and academic groups. Jean graduated from Yale College, received her PhD (Zoology, Cell and Developmental Biology) from University of California, Berkeley and her MD from Harvard Medical School. She then received post-graduate training at Yale University and Johns Hopkins in Human Genetics and Developmental Genetics. An internationally recognized expert in gene therapy, Dr. Bennett has authored more than 120 peer-reviewed papers. She has received many awards including the Smithsonian Ingenuity Award and was a Co-recipient of the Champalimaud Award, the Sanford Lorraine Cross Award, the Harrington Prize, and the Helen Keller Prize in Vision Research. She is a member of the National Academy of Medicine, National Academy of Science, the Association of American Physicians and the American Academy of Arts and Sciences.



**Toby Ferguson, MD, PhD**

**Head Neuromuscular and Movement Disorders Development Unit, Biogen**

Toby is a neuromuscular neurologist and neuroscientist who joined Biogen in 2013. His professional experience has focused on developing treatments for neuromuscular disease, with a focus on ALS and SMA as well as a more recent focus on movement disorders. He is currently the Head of the Neuromuscular and Movement Disorders Development Unit at Biogen. He plays a key role in developing clinical trials across his therapeutic areas and in driving clinical and preclinical strategy within neurodegenerative disease more broadly. His group also works closely with the preclinical scientific and biomarker teams as well as with external collaborators to identify novel disease targets and to develop the needed tools for efficient clinical development.

At Biogen, he has advanced multiple programs into the clinic for ALS, SMA, Myotonic Dystrophy, and Parkinson's disease. He has also helped to successfully develop Qalsody (tofersen) an ASO indicated for the treatment SOD1 ALS. He is dedicated to the successful development of meaningful therapeutics for



neurologic disease and strongly believes that collaboration across industry, academia, and advocacy organizations is crucial to developing meaningful therapies.

Prior to Biogen, Toby had a clinical neuromuscular neurology practice and a lab focused on peripheral axon injury and regeneration at Shriners Research Center and Temple University in Philadelphia. Toby trained in neurology and neuromuscular neurology at the University of Pennsylvania. He obtained an MD and PhD (Neuroscience) at the University of Florida.



**Steven Scherer, MD, PhD**

**Ruth Wagner Van Meter and J. Ray Van Meter Professor of Neurology, Director of Penn Neurogenetics Therapy Center, University of Pennsylvania**

Dr. Scherer is the Ruth Wagner Van Meter and J. Ray Van Meter Professor of Neurology at the University of Pennsylvania, and the Director of the Penn Neurogenetics Therapy Center. Finding the causes of peripheral neuropathy has been the focus of his clinical and laboratory research, and is a collaborative effort involving physicians and scientists from around the world, as well as patients

and patient-led organizations. His current work includes preclinical investigations of animal models of inherited neuropathies, as well as natural history and clinical trials of people with inherited neuropathies.



**Kevin Flanigan, MD**

**Robert F. and Edgar T. Wolfe Foundation Endowed Chair in Neuromuscular Research; Director, Center for Gene Therapy at the Abigail Wexner Research Institute, Nationwide Children's Hospital**

Dr. Flanigan is the Director of the Center for Gene Therapy at the Abigail Wexner Research Institute of Nationwide Children's Hospital (NCH), where he holds the Robert F. & Edgar T. Wolfe Foundation Endowed Chair in Neuromuscular Research. He is also Director of the Neuromuscular Division at NCH, and Professor of Pediatrics and Neurology at the Ohio State University. He trained in Neurology and Neuromuscular Disease at the Johns Hopkins Hospital, followed by a

post-doctoral fellowship in Human Molecular Biology and Genetics at the University of Utah. He has been a member of the Executive Board of the World Muscle Society since 2001, and is a past Chair of the Executive Committee of TREAT-NMD, the international alliance directed toward establishing the infrastructure to ensure that promising new therapies reach patients as quickly as possible. His laboratory work is directed toward the molecular characterization and therapy of neuromuscular diseases using both gene replacement on RNA-modifying therapies, and toward the identification of genetic modifiers of disease. He has conducted multiple clinical trials of gene modifying gene transfer therapies in Duchenne muscular dystrophy (DMD) as well as the childhood neurodegenerative disorders mucopolysaccharidosis types 3A and 3B.



### **Session III**



**Beverly L. Davidson, PhD**

**Katherine A. High Chair in Cell and Gene Therapy, Director, R.G. Perelman Center for Cellular and Molecular Therapeutics; Chief Scientific Strategy Officer, Children's Hospital of Philadelphia and Professor of Pathology and Laboratory Medicine, Perelman School of Medicine at the University of Pennsylvania**

The Davidson lab is focused on genetic diseases that affect the brain, including how mutant gene products contribute to disease, and why certain brain regions are more susceptible. The team employs advanced molecular methods, sequencing and imaging modalities in animal models, and uses a variety of molecular tools to test various hypotheses. The lab is also engaged in the development of next generation therapeutics for inherited disorders, including the engineering of novel gene therapy vector capsids and cargo to approach tissue and cell type specific treatments.

Recent honors include election into the American Academy of Arts and Sciences and the National Academy of Medicine, the Hereditary Disease Foundation's Leslie Gehry Brenner Prize for Innovation in Science, and recipient of the Dr. John W. Schut Research Achievement Award from the National Ataxia Foundation. She is the past president of the American Society of Gene and Cell Therapy, the largest international association of gene and cell therapy research.



**Drew Weissman, MD, PhD, Nobel Laureate**

**Roberts Family Professor in Vaccine Research; Director, Penn Institute for RNA Innovation; Director, Vaccine Research in the Infectious Disease Division; University of Pennsylvania**

**Winner of the 2023 Nobel Prize in Physiology or Medicine**

Drew Weissman, M.D., Ph.D. is a professor of Medicine at the Perelman School of Medicine, University of Pennsylvania. He received his graduate degrees from Boston University School of Medicine. Dr. Weissman, in collaboration with Dr. Katalin Karikó, discovered the ability of modified nucleosides in RNA to suppress activation of innate immune sensors and increase the translation of mRNA containing certain modified nucleosides. The nucleoside-modified mRNA-lipid nanoparticle vaccine platform Dr. Weissman's lab created is used in the first 2 approved COVID-19 vaccines by Pfizer/BioNTech and Moderna. They continue to develop other vaccines that induce potent antibody and T cell responses with mRNA-based vaccines. Dr. Weissman's lab also develops methods to replace genetically deficient proteins, edit the genome, and specifically target cells and organs with mRNA-LNPs, including lung, heart, brain, CD4+ cells, all T cells, and bone marrow stem cells.

Dr. Weissman and Dr. Karikó were jointly awarded the 2023 Nobel Prize in Physiology or Medicine "for their discoveries concerning nucleoside base modifications that enabled the development of effective mRNA vaccines against COVID-19." <https://www.nobelprize.org/>



**Ricardo Dolmetsch, PhD**

**President and Chief Scientific Officer, uniQure**

Dr. Ricardo Dolmetsch joined uniQure in September 2020. In his role as President and Chief Scientific Officer, he leads their gene therapy research activities, as well as nonclinical development, process development, analytical development and vector development. His previous work also focused on leading the development of treatments for neurodegenerative and neuropsychiatric diseases.

He holds a bachelor's degree from Brown University and Ph.D. in neuroscience from Stanford University. He conducted his postdoctoral training at Harvard University Medical School and Children's Hospital Boston.

**Session IV**



**Benjamin (Ben) L. Prosser, PhD**

**Associate Professor, Physiology; Director, Center for Epilepsy and Neurodevelopmental Disorders (ENDD); Associate Director, Pennsylvania Muscle Institute; Lead Coordinator, Leducq Transatlantic Network of Excellence (Leducq Cytoskeletal Network), University of Pennsylvania**

Dr. Benjamin Prosser is an Associate Professor of Physiology at the University of Pennsylvania Perelman School of Medicine. He earned his Ph.D. in Molecular Medicine from the University of Maryland School of Medicine in 2009 and started his own lab at Penn Medicine in 2014, with an initial focus on developing novel therapeutics for heart failure. In recognition of the lab's cardiac work, Dr. Prosser was named the Outstanding Early Career Investigator by the American Heart Association in 2017 and received the Outstanding Investigator Award by the International Society of Heart Research in 2022. In 2018, Dr. Prosser's daughter Lucy was born and diagnosed with STXBP1 encephalopathy, a rare, genetic neurodevelopmental disorder. Dr. Prosser started a separate research arm in his lab focused on developing new therapies for STXBP1 and related disorders. This work ultimately spurred the formation of the Center for Epilepsy and Neurodevelopmental Disorders (ENDD), which Dr. Prosser directs with his colleagues Drs. Beverly Davidson and Ingo Helbig of the Children's Hospital of Philadelphia (CHOP). ENDD represents a cross-institute, interdisciplinary collaboration of fundamental, translational and clinical researchers focused on developing genetic therapies for STXBP1, SYNGAP1, and related neurodevelopmental disorders.

Here Dr. Prosser will provide an overview of the challenges and strategies taken to achieve these translational and clinical research goals and highlight one of ENDD's lead therapeutic strategies – the use of antisense-oligonucleotides (ASOs) to target and manipulate mRNA processing in order to restore expression of STXBP1 and SYNGAP1. [bpros@pennmedicine.upenn.edu](mailto:bpros@pennmedicine.upenn.edu)



**Steven E. Hyman, MD**

**Core Institute Member of the Broad Institute of MIT and Harvard and Director of the Stanley Center for Psychiatric Research; Harvard University Distinguished Service Professor and Harald McPike Professor of Stem Cell and Regenerative Biology**

From 2001 to 2011 Hyman served as Provost (chief academic officer) of Harvard University, where he had a special focus on developing and supporting collaborative, interdisciplinary, and cross institutional efforts in the sciences, arts, and humanities. From 1996 to 2001 he served as Director of the US National Institute of Mental Health (NIMH), where he invested in neuroscience and emerging genomic technologies and initiated a series of large clinical effectiveness trials to inform practice. He has served as Editor of the Annual Review of Neuroscience (2002-2016), founding President of the International Neuroethics Society (2008-2013), President of the Society for Neuroscience (2015), and President of the American College of Neuropsychopharmacology (2018). He is a fellow of the American Academy of Arts and Sciences, a fellow of the American Association for the Advancement of Science, a distinguished life fellow of the American Psychiatric Association, and a member of the National Academy of Medicine (NAM). At the National Academies of Sciences, Engineering, and Medicine he has served on the NAM Council (2012-2018), on the Governing Board of the National Research Council (2016-2019) and chaired the National Academies' Forum on Neuroscience and Nervous System Disorders (2012-2018), which brings together industry, government, foundations, patient groups, and academia. He is currently a member of the Committee on Science, Technology, and the Law (CSTL). He chairs the Boards of Directors of the Charles A. Dana Foundation (NY) and the Wyss Center for Bio and Neuroengineering (Geneva, Switzerland), and is a Director of Annual Reviews Inc (Palo Alto), a nonprofit scientific publisher. In the private sector he is a Director of Voyager Therapeutics, Cyclarion Therapeutics, and Vesalius Therapeutics. He serves on the scientific advisory boards of Janssen Research and Development and F-Prime Capital. He received his BA, summa cum laude, from Yale, an MA from the University of Cambridge, which he attended as a Mellon fellow studying History and Philosophy of Science, and an MD, cum laude, from Harvard Medical School.

**Session V**



**Sara-Claude Michon, PhD**

**Director of Operations, Penn Neurogenetics Therapy Center,  
University of Pennsylvania**

Sara-Claude earned her PhD in Neuroscience in 2013 and has over 10 years of experience with clinical research for neurodegenerative disorders. Since joining Penn in 2018, she specialized in the operationalization and management of early phase clinical trials for neurological disorders. In her role, she collaborates with Penn investigators and biomedical companies, and leverages Penn resources to deliver early phase clinical trials of novel genetic-based therapies to patients with neurological disorders.



**Amy T. Waldman, MD, MSCE**

**Associate Director, Clinical In Vivo Gene Therapy, Children's  
Hospital of Philadelphia and Associate Professor of Neurology at  
the University of Pennsylvania**

Dr. Waldman is an Associate Professor of Neurology at the Perelman School of Medicine at the University of Pennsylvania. She is also the Associate Director of Clinical In Vivo Gene Therapy at the Children's Hospital of Philadelphia (also known as CHOP) and the Medical Director of the Leukodystrophy Center of Excellence at CHOP. She serves on the Neurologic and Ophthalmic Gene and Cell Therapy Committee for ASGCT.

Dr. Waldman received her Medical Doctorate from Jefferson Medical College (Thomas Jefferson University). She completed her pediatrics residency at The Children's Hospital of Philadelphia (CHOP) and child neurology residency at both CHOP and the Hospital of the University of Pennsylvania. In 2005, she co-founded the Pediatric MS Center at CHOP, and in 2015, she co-founded the Leukodystrophy Center of Excellence at The Children's Hospital of Philadelphia where she now serves as the Medical Director. Dr. Waldman's primary research focuses on the development and interpretation of outcome measures for clinical trials in neuroinflammatory and neurodegenerative diseases. In 2021, she was appointed the Associate Director of Gene Therapy at CHOP.





**James M. Wilson, MD, PhD**

**Rose H. Weiss Professor and Director, Orphan Disease Center; Professor of Medicine and Pediatrics; Director, Gene Therapy Program, Perelman School of Medicine, University of Pennsylvania**

Co-Founder and Scientific Advisor, Scout Bio  
Co-Founder and Chief Scientific Advisor, Passage Bio  
Co-Founder, G2 Bio  
Co-Founder, Chief Scientific Officer, and Board Member of Institute for Life-Changing Medicine (ILCM)  
Co-Founder and Chief Scientific Advisor, iECURE  
Co-Founder and Chief Scientific Advisor, Center for Breakthrough Medicines

James M. Wilson, MD, PhD, is a Professor in the Perelman School of Medicine at the University of Pennsylvania where he has led an effort to develop the field of gene therapy. His research career spanning over 40 years has focused on rare diseases and ways to treat them by gene therapy. Dr. Wilson has published over 600 papers and is named on more than 1200 patents worldwide. The Wilson lab identified a new type of vector based on novel isolates of adeno-associated viruses which have become best in class for gene therapy. More recently Dr. Wilson's laboratory has focused on improved vectors for gene therapy and clinical applications of genome editing and mRNA therapy.



**Pedro Gonzalez-Alegre, MD, PhD**

**Head of Gene Therapy Research, Spark Therapeutics, Inc.**

Dr. Gonzalez-Alegre is a physician-scientist with subspecialty training on movement disorders and clinical neurogenetics. He received his MD, PhD (Neuroscience) from the University of Malaga in Spain. After completing a neurology residency and fellowship on Movement disorders/Neurogenetics at the University of Iowa Hospitals and Clinics (UI), he joined the faculty at that institution where he remained for over a decade. Dr. Gonzalez-Alegre joined the Department of Neurology at the

University of Pennsylvania in 2014, where he was the founding Director of the of the Huntington's Disease Center of Excellence and the Translational Center of Excellence on Neurogenetics Therapy at the Department of Neurology, and Director of Clinical Programs at the Raymond G. Perelman Center for Cellular & Molecular Therapeutics at the Children's Hospital of Philadelphia. During his academic career, Dr. Gonzalez-Alegre provided clinical care to patients suffering from Parkinson's disease, HD, ataxia and other neurogenetic disorders while being an active researcher and educator. His research efforts were geared towards the development of novel nucleic acid-based therapeutic strategies for neurological diseases, including development of innovative technologies to improve delivery of therapeutic agents to the human brain. In 2021, he joined Spark® Therapeutics as the Head of Gene Therapy Research, where he leads all therapeutic areas and immunology within the Research & Technology organization.

## Session VI



**Holly Tabor, PhD**

**Associate Professor of Medicine at Stanford University  
Associate Director for Clinical Ethics and Education for the  
Stanford Center for Biomedical Ethics (SCBE)  
Co-Chair of the Ethics Committees at Stanford Hospital and Lucile  
Packard Children's Hospital**

Holly Tabor, PhD, is Associate Professor of Medicine at Stanford University. She is the Associate Director for Clinical Ethics and Education for the Stanford Center for Biomedical Ethics (SCBE) and is Co-Chair of the Ethics Committees at Stanford Hospital and Lucile Packard Children's Hospital. Her research focuses on ethical issues in genetics and genomics and inclusive health care for people with disabilities, especially people with intellectual and/or developmental disabilities. She has received the Stanford School of Medicine Henry J. Kaiser Award for Excellence in Preclinical Teaching, the Stanford Faculty Women's Forum Allyship Award and the Outstanding Community Engaged Faculty Award from the Stanford Medicine Office of Community Engagement.



**Peter Marks, MD, PhD**

**Director, Center for Biologics Evaluation and Research (CBER), U.S. Food  
and Drug Administration (FDA)**

Peter Marks received his graduate degree in cell and molecular biology and his medical degree at New York University and completed Internal Medicine residency and Hematology/Medical Oncology training at Brigham and Women's Hospital in Boston. He has worked in academic settings teaching and caring for patients and in industry on drug development and is an author or co-author of over 100 publications. He joined the FDA in 2012 as Deputy Center Director for CBER and became Center Director in 2016. Over the past several years he has been integrally involved in the response to various public health emergencies, and in 2022 he was elected a member of the National Academy of

Medicine.



**Allyson Berent, DVM, DACVIM**

**Chief Science Officer, Foundation For Angelman Syndrome  
Therapeutics (FAST)  
Co-Director, Angelman Syndrome Biomarker and Outcome Measure  
Consortium  
Chief Development Officer, Mahzi Therapeutics**

Dr. Berent is a veterinary internal medicine specialist/ interventionalist who graduated from Cornell University and completed her residency at the University of Pennsylvania, where she served as an Adjunct Assistant Professor before joining the Animal Medical Center in NYC. She is the Director of the Interventional Endoscopy Service, focusing on clinical trials researching medical devices particularly for ureteral and biliary obstructions in animals with naturally occurring diseases. In 2014 Dr. Berent's daughter, Quincy, was diagnosed with Angelman syndrome. In 2015 she joined the Board of Directors for the Foundation for Angelman Syndrome Therapeutics (FAST), becoming the Chief Science Officer. Dr. Berent serves as the co-director of the Angelman Syndrome Biomarker and

Outcome Measure Consortium, to co-director for the International Angelman Syndrome Research Council (INSYNC-AS), and is an advisor to numerous pharmaceutical companies working on therapeutic candidates for rare neurodevelopmental disorders. Dr. Berent co-founded GeneTx Biotherapeutics, a company focused on advancing an antisense oligonucleotide therapy for AS, where she was the Chief Operating Officer. GeneTx was acquired in 2022 by Ultragenyx Pharmaceuticals, after launching the Phase 1/2 clinical trial, and she now serves as a consultant for Ultragenyx. Dr. Berent is currently the Chief Development Officer at Mahzi Therapeutics advancing gene therapies and disease modifying therapies for rare neurodevelopmental disorders.

## **Session VII**



**David Irwin, MD**

**Assistant Professor, Neurology; Co-Director, Penn Frontotemporal Degeneration Center, University of Pennsylvania**

Dr. David Irwin co-directs the Penn Frontotemporal Degeneration Center at the University of Pennsylvania Perelman School of Medicine and is the PI of the Penn Digital Neuropathology Lab. He has dual training in cognitive neurology and neuropathology and his lab focuses on integrating human brain histopathology and molecular techniques with imaging methods to discover

therapeutic targets and develop tissue-sensitive biomarkers to facilitate clinical trials for emerging therapies for FTD, LBD, AD and related disorders.



**Erik Roberson, MD, PhD**

**Rebecca Gale Endowed Professor  
Vice Chair for Basic & Translational Research, Department of Neurology  
Director, Alzheimer's Disease Center  
Director, Center for Neurodegeneration and Experimental Therapeutics  
Associate Director, Integrative Center for Aging Research  
University of Alabama at Birmingham**

Dr. Roberson is a physician-scientist with a focus on age-related neurodegenerative diseases. He earned his A.B. with highest honors from Princeton University, then his M.D. and Ph.D in neuroscience at Baylor College of Medicine where he studied molecular mechanisms of learning and memory. He completed a residency in neurology at the University of California San Francisco, where he also served as Chief Resident in Neurology. After residency, he completed a clinical fellowship in behavioral neurology at UCSF and resumed basic research at the Gladstone Institute of Neurological Disease. He joined the faculty at UAB in 2008.

The Roberson lab studies the neurobiology of Alzheimer's disease (AD) and frontotemporal dementia (FTD), with a focus on understanding the cellular and molecular mechanisms of these disorders and identifying new therapeutic strategies. Focus areas include tau and its binding partners in neuronal dysfunction in AD, the mechanisms by which genetic risk factors drive AD, and how progranulin deficiency causes FTD.

Dr. Roberson directs the UAB Alzheimer's Disease Research Center and the Center for Neurodegeneration and Experimental Therapeutics, serves as Vice Chair for Basic and Translational Research in the Department



of Neurology, and cares for patients at the UAB Brain Aging and Memory Clinic.  
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**Michael Kaplitt, MD, PhD**  
**Professor of Neurological Surgery; Residency Director and Vice**  
**Chairman for Research; Weill Cornell Medical College**

Michael Kaplitt, M.D., Ph.D., is a tenured Professor of Neurological Surgery, Vice-Chairman for Research, and Residency Program Director in the Department of Neurological Surgery at Weill Cornell Medical College. Vice-Chairman for Research, and Residency Program Director in the Department of Neurological Surgery at Weill Cornell Medical College. Dr. Kaplitt graduated from Princeton University in 1987 with a degree in Molecular Biology and Russian Studies. Through the Tri-Institutional M.D.-Ph.D. Program, he received his Ph.D. in Molecular Neurobiology in 1993 from The Rockefeller University and his M.D. in 1995 from Cornell Medical College. After his neurosurgery residency at The New York-Presbyterian Hospital, he completed a fellowship in Stereotactic and Functional Neurosurgery at the University of Toronto prior to joining Weill Cornell in July 2001. He has pioneered the use of gene therapy in the brain, having published on the first use of adeno-associated virus (AAV) as a gene delivery vehicle for the brain in 1994. In 2003, he performed the first human clinical trial of gene therapy for Parkinson's disease, which led to the first successful double-blind, controlled study of gene therapy for a neurological disease. His lab continues to focus upon use of gene transfer to generate new models and novel potential therapies for Parkinson's disease, psychiatric disorders and pain. He has also pioneered the use of non-invasive focused ultrasound in patients with tremor and Parkinson's disease, and is currently performing studies in rodents, primates and human subjects exploring novel applications of focused ultrasound in Parkinson's and Alzheimer's diseases. Dr. Kaplitt is the recipient of numerous honors and awards, including the Young Investigator Award from the American Society for Gene Therapy, and is a member of the American Society for Clinical Investigation.