



National Institute of
Neurological Disorders
and Stroke

Dr. Kenneth (Kurt) Fischbeck Retirement Symposium

Lipsett Amphitheater | Bethesda, MD | November 9th, 2023

Gene identification to therapeutics development: Celebrating the scientific contributions of Kenneth Fischbeck, MD

Please join us to honor the career and scientific achievements of Dr. Kenneth 'Kurt' Fischbeck. The goal of this symposium will be to discuss research topics including disease gene identification and therapeutics development in neuromuscular disease. The agenda will include presentations from leading educators and researchers in the fields of polyglutamine and neuromuscular disease. Talks will review the impact of Dr. Fischbeck's research in neurogenetics and therapeutics development.

Funding for this event is sponsored by the NINDS Intramural Research Program and by the Foundation for Advanced Education in the Sciences (FAES).

Introduction:

8:00AM-8:25AM Registration and Reception

8:25AM-8:30AM Welcome and overview: *Christopher Grunseich, M.D., and Carsten Bönnemann, M.D. - NINDS, NIH*

8:30AM-8:35AM Opening remarks from NINDS Director: *Walter Koroshetz, M.D.*

8:35AM-8:40AM Opening remarks from NINDS Scientific Director: *Jeffrey Diamond, Ph.D*

Disease gene identification: Session Chair- Audrey Winkelsas, Ph.D, MD Candidate - University of Michigan

8:40AM-9:00AM Disease gene discovery and outreach in neurology: *Guida Landoure, M.D., Ph.D - University of Bamako, Mali*

9:00AM-9:20AM TRPV4 gene discovery exposes novel disease biology and a therapeutic opportunity: *Charlotte Sumner, M.D. - Johns Hopkins University*

9:20AM-9:40AM Hereditary Spastic Paraplegias: Advances from the NINDS Neurogenetics Branch: *Craig Blackstone, M.D., Ph.D – Massachusetts General Hospital*

Repeat expansion diseases and spinal and bulbar muscular atrophy: Session Chair- Laura Bott, Ph.D. - Northwestern University

9:40AM-10:00AM The Secret's Out: How the Hunt for a Rare Disease Gene led to the Discovery of an Entirely Novel Type of Genetic Mutation: *Albert La Spada, M.D., Ph.D.- UC Irvine*

10:00AM-10:20AM SBMA- more than just motor neurons: *Andy Lieberman, M.D. Ph.D. - University of Michigan*

10:20AM-10:40AM Spinal and bulbar muscular atrophy: *Diane Merry, Ph.D. - Thomas Jefferson University*

10:40AM-11:00AM Break

Polyglutamine diseases and beyond: Session Chair- Naemeh Pourshafie, Ph.D. - University of Pennsylvania

11:00AM-11:20AM Toward understanding and treating SCA3: *Henry Paulson, M.D., Ph.D. - University of Michigan*

11:20AM-11:40AM A Thirty Year Journey towards Understanding a Neurodegenerative Disease (Kurt- Thanks for being such an important part of the journey): *Harry Orr, Ph.D - University of Minnesota*

11:40AM-12:00PM The Presentation and Natural History of Spinocerebellar Ataxia, Type 7: *Brian Brooks, M.D., Ph.D. - NEI, NIH*

12:00PM-12:20PM Mitochondria and Repeat Expansion Disorders: *Srikanth Ranganathan, Ph.D. - NINDS, NIH*

12:20PM-1:40PM Picture and Lunch



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Patient advocacy: Session Chair- Deborah Kwon, Ph.D. - Biogen

1:40PM-1:50PM Foundation for Advanced Education in the Sciences (FAES)

1:50PM-2:10PM Leading the Search for a Cure: An Appreciation from the Kennedy's Disease Association: *Terry Thompson - Kennedy's Disease Association*

2:10PM-2:30PM Training a New Generation of Clinician Scientists to Treat Hereditary Neurological Diseases in Mali, W. Africa: *Katherine Meilleur, C.N.P., Ph.D. - Ionis*

2:30PM-2:40PM Coffee break

Disease mechanism and therapeutics development: Session Chair- Katherine Bricceno, Ph.D. - Biogen

2:40PM-3:00PM Targeting the proteasome in spinal muscular atrophy: *Barrington Burnett, Ph.D. - Uniformed Services University*

3:00PM-3:20PM A journey from mice to patients: Therapeutics development in SBMA: *Christopher Grunseich, M.D., - NINDS, NIH*

3:20PM-3:40PM Drug discovery and development in neuromuscular diseases: *Alexander McCampbell, Ph.D. - Roche*

3:40-3:50 Break

Finale:

3:50PM-4:20PM Keynote speaker Advancing Precision Medicines for Inherited Brain Disorders: *Beverly Davidson, Ph.D.- University of Pennsylvania*

4:20PM-4:40PM **Remarks from Dr. Kenneth Fischbeck**

4:40PM-5:00PM Closing remarks from NINDS Clinical Director: – *Avindra (Avi) Nath, M.D.*