



**2011 International
Familial Dysautonomia
Research Conference**



**Sponsored by
Dysautonomia Foundation, Inc.**

**Location
Renaissance Hotel 57
130 E 57th St., New York, NY**

Program – Thursday October 27th 2011

Morning Session 9 AM – 12 PM

- 9:00 AM **Welcome and Opening Remarks**
David Brenner, Executive Director of the
Dysautonomia Foundation
- 9:10 AM **Introduction and Goals of the Conference**
Mike Brownstein, Chairman of the Scientific Advisory
Board of the Dysautonomia Foundation

FD PHENOTYPE AND NOVEL TREATMENTS

Co-Chairs: Horacio Kaufmann, MD and Felicia Axelrod, MD

- 9:20 AM **Familial Dysautonomia: An afferent disorder**
Horacio Kaufmann, MD (New York, USA)
- 9:40 AM **Optic Atrophy in FD**
Carlos Mendoza, MD (Boston, USA)
- 10:00 AM **Von Economo Neurons in FD**
Patrick Hof, MD (New York, USA)
- 10:20 AM **Coffee Break**
- 10:40 AM **Loss of Muscle Spindle Afferents Explain the
Ataxic Gait in Riley- Day Syndrome**
Vaughan Macefield, PhD (Sydney, Australia)
- 11:00 AM **Is Dopa Decarboxylase Inhibition Useful in FD?**
Lucy Norcliffe-Kaufmann, PhD (New York, USA)
- 11:20 AM **Open discussion**
- 12:00 PM **Lunch with posters 12:00 – 12:45 PM**
- 12:45 PM *After lunch, there will be an opportunity to hear from several people
with FD:*
Living with FD – A Patient Perspective
Felicia Axelrod, MD (New York, USA)

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Afternoon Session 1 PM – 5 PM

ADVANCEMENTS IN BASIC SCIENCE: Molecules & Mice Chair: Susan Slaugenhaupt, PhD

- 1:00 PM **Insights into the Role of Elongator in Cerebral Cortical Neurogenesis**
Laurent Nguyen, PhD (Liege, Belgium)
- 1:20 PM **Involvement of IKAP/Elp1 in Microtubule Organization**
David Cheishvili, PhD (Tel Aviv, Israel)
- 1:40 PM **Transcriptional Analysis of FD with Patient Olfactory Ecto-Mesenchymal Stem Cells**
El-Cherif Ibrahim, PhD (Marseille, France)
- 2:00 PM **Peripheral Nervous System Development: Which Steps Go Awry to Cause FD?**
Frances Lefcort, PhD (Bozeman, MT USA)
- 2:20 PM **Coffee Break**
- 2:40 PM **Modifying IKAP expression using transgenic mice: insights into Elongator function during development**
Susan Slaugenhaupt, PhD (Boston, USA)
- 3:00 PM **Mouse Models for FD: Insights into Mechanisms and Therapeutic Strategies**
Ioannis Dragatis, PhD (Memphis, TN USA)
- 3:20 PM **Transgenic Mice**
Gil Ast, PhD (Tel Aviv, Israel)
- 3:40 PM **The Role of IKAP/hELP1 in PNS Development of the Chick Embryo and in Human ESC Derived PNS Neurons as Models to Study FD Phenotype**
Miguel Weil, PhD (Tel Aviv, Israel)
- 4:00 PM **Open discussion**

Cocktails, Dinner and Entertainment 5:30 PM

Program – Friday October 28th 2011

Morning Session 9 AM – 12:30 PM

STRATEGIES FOR SPLICING THERAPIES & CLINICAL TRIALS **Co-Chairs: Wayne Bardin, MD and Michael Brownstein, MD,PhD**

9:00 AM **Modification of mRNA splicing as a treatment for FD**
Susan Slaughter, PhD (Boston, USA)

9:20 AM **Phosphatidylserine Increases IKBKAP Levels**
Gil Ast, PhD (Tel Aviv, Israel)

9:40 AM **Identification of Compounds that Increase *IKBKAP* Expression in Familial Dysautonomia-iPS cells**
Gabsang Lee, PhD (Baltimore, USA)

10:00 AM **Strategies for Drug Screening and Therapy of Familial Dysautonomia**
Miguel Weil, PhD (Tel Aviv, Israel)

10:20 AM ***Coffee Break***

10:40 AM **Clinical Trials in FD**
Lucy Norcliffe-Kaufmann, PhD (New York, USA)

11:00 AM **Cutaneous Innervation, a Biomarker**
Gwenn Wendelschafer-Crabb, MS (Minneapolis, USA)

11:20 AM **Retinal Fibers (OCT), a Non-Invasive Biomarker**
Carlos Mendoza, MD (Boston, USA)

11:40 AM **Point-Counter-Point Discussion**

12:15 PM ***Lunch***

Conference Ends – 2:00 PM

List of Attendees

Gil Ast, PhD - Tel Aviv University, Israel
Felicia Axelrod, MD - NYU School of Medicine, USA
C. Wayne Bardin, MD - Bardin LLC, USA
Italo Biaggioni, MD - Vanderbilt University School of Medicine, USA
Michael Brownstein, MD, PhD - Scientist Emeritus, NIH, USA
Mark Chapleau, PhD - University of Iowa, USA
David Cheishvili, PhD - Hebrew University / Hadassah, Israel
Gwen Wendelschafer-Crabb, MS - University of Minnesota, USA
Gayatri Devi, MD - NYU School of Medicine, USA
Paula Dietrich, PhD - University of Tennessee, USA
Ioannis Dragatsis, PhD - University of Tennessee, USA
Roy Freeman, MD - Harvard Medical School, USA
David Goldstein, MD, PhD – NIH/NINDS, USA
Robert Gross, PhD - Dartmouth College, USA
James Gusella, PhD - MGH / Harvard Medical School, USA
Patrick Hof, MD - Mount Sinai School of Medicine, USA
El Chérif Ibrahim, PhD - Université de la Méditerranée, France
Giris Jacob, MD, DSc - Tel Aviv Sourasky Medical Center, Israel
Horacio Kaufmann, MD - NYU School of Medicine, USA
Gabsang Lee, PhD - John Hopkins University School of Medicine, USA
Frances Lefcort, PhD - Montana State University, USA
Phillip Low, MD - Mayo Clinic in Rochester, USA
Yvonne Lui, MD - NYU School of Medicine, USA
Vaughan Macefield, PhD - University of Western Sydney, Australia
Jose Martinez, MS - NYU School of Medicine, USA
Carlos Mendoza-Santiesteban, MD - Tufts School of Medicine, USA
Laurent Nguyen, PhD - University of Liège, Belgium
Mats Nilbratt, PhD - MGH / Harvard Medical School, USA
Lucy Norcliffe-Kaufmann, PhD - NYU School of Medicine, USA
Sathiya Ramakrishnan, PhD - Massachusetts General Hospital, USA
Dusan Roncevic, MD - NYU School of Medicine, USA
Susan Slaugenhaupt, PhD - MGH / Harvard Medical School, USA
Gail Sonenshein, PhD - Boston University School of Medicine, USA
Sara Sunshine - MGH / Harvard Medical School, USA
Miguel E. Weil, PhD - Tel Aviv University, Israel
Michael Weiser, MD, PhD – Actin Biomed, USA