Cure SPG47 Advisory Meeting: 3/7/2017

Graciously hosted by BCH Translational Neuroscience Center

Meeting location: Center for Life Sciences
3 Blackfan Circle
12th floor conference room
Boston, MA 02115

Meeting Time: 2pm to 5pm

Contact phone: Lea Florentino, Administrative Assistant for Translational Neuroscience Center
(617) 919-6258

Parking: Corner of Longwood Avenue and Blackfan Circle (Boston Children's Hospital patient garage)

Adobe Connect: https://meeting.childrens.harvard.edu/r7e7gqt7eyg/

Once you are signed into Adobe Connect, telephone dial-in info will become available, and you will be able to view any slides being presented.

Introduction:

On behalf of the families affected by SPG47, we truly appreciate your willingness to participate in this first ever meeting of Cure SPG47 families, Medical Advisors, Board of Directors, and our extended team of Scientific Advisors and industry experts. We are extremely fortunate to have so many impressive people involved in the effort to cure and/or treat this devastating disease. Some of our goals for this meeting include:

- Building a deeper understanding of SPG47 and underlying AP-4 adaptor complex cell biology
- Building a deeper understanding of possible treatment hypotheses
- Building consensus around the best short and long-term approaches to treating/curing SPG47
- Coming away with a clearer strategy for moving forward, and establishing fundraising goals

Board of Directors:

Kira Dies, ScM, CGC: *Co-director of Clinical Research and Regulatory Affairs Service, Translational Neuroscience Center, Boston Children's Hospital*. Kira is a licensed genetic counselor at BCH. She has a deep understanding of the underlying genetics of neurodegenerative diseases like HSP. She manages multi-site clinical trials for neurogenetic conditions including tuberous sclerosis complex, Rett syndrome, and PTEN hamartoma syndrome.

Kevin Duffy: *Head Golf Professional, Riverton Country Club*. Kevin is Molly Duffy's father. He has been working in the golf industry for more than 15 years and is currently responsible for leading the golf operation at Riverton CC on both an operational and strategic level. His areas of expertise include marketing, relationship management, customer service, team building and coaching.

Chris Edwards: *Executive Director, Alternative Therapies Group*. Chris is Robbie Edwards' father. He has founded a series of startup companies during his career. He has extensive experience in building/managing teams with diverse skills sets, and with navigating complex governmental regulations and problem solving.

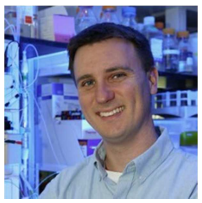
Erika M. Gill, MBA: *Senior Director, Asset Management, Biogen*. Erika has over 18 years' experience in healthcare and biotech, leading several cross-functional areas focusing on product program management, patient services programs, marketing and health care administration.

Medical Advisors:

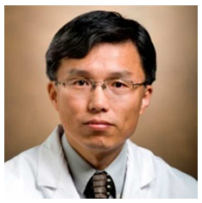
Dr. Craig Blackstone: *Senior Investigator, Cell Biology Section, National Institute of Neurological Disorders and Stroke, NIH*. Dr. Blackstone's laboratory investigates the cellular and molecular mechanisms underlying hereditary movement disorders. Craig is one of the most prominent HSP researchers in the world.

Dr. Basil Darras: *Associate Neurologist-in-Chief, Chief-Division of Clinical Neurology, Director- Neuromuscular Center, Boston Children's Hospital*. Dr. Darras' research is focused on the molecular genetics, diagnostics and therapeutics of pediatric neuromuscular diseases.

Dr. John Fink: *Professor, Department of Neurology, Director, Neurogenetic Disorders Program, University of Michigan*. In addition to being one of the world's foremost investigators of upper motor neuron disorders, Dr. Fink also maintains the largest clinic in the U.S. for persons with HSP or PLS.



Dr. Steven Gray: *Assistant Professor at UNC School of Medicine Gene Therapy Center*. Dr. Gray's core research focus is to develop adeno-associated virus (AAV) gene transfer vector systems, for clinically-relevant global gene transfer to the central and peripheral nervous system.



Dr. Jun Li: *Professor of Neurology, Vanderbilt University School of Medicine.* Dr. Li's research is focused on myelin biology and the pathogenesis and therapeutic development of inherited peripheral nerve diseases.

Extended CureSPG47 Scientific Team:

Dr. Irina Anselm: *Assistant Professor of Neurology, Harvard Medical School*. As the Director of the BCH Mitochondrial Program, Dr. Anselm applies the latest techniques to help patients and their families manage their disorders. One of her top concerns is improving the quality of life for patients through the study and development of new drugs and therapies.

Dr. Mimoun Azzouz: *Chair of Translational Neuroscience, ERC Advanced Investigator, Director of Research and Innovation, University of Sheffield*. Dr. Azzouz has a long-standing interest in developing gene therapy approaches for neurodegenerative diseases. His team utilizes viral-based gene transfer systems both for research and gene therapy applications.

James T. Bennett, MD, PhD: *Pediatric Geneticist, Seattle Children's Hospital Genetics Care Team, Associate Editor American Journal of Medical Genetics*. Dr. Bennett is board certified in Clinical Molecular Genetics. He has initiated a project of collecting and reporting pertinent medical information for known SPG47 patients.

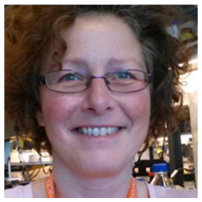
Dr. Gerard Berry: *Director, Metabolism Program, Boston Children's Hospital, Specialist in Genetics and Genomics, Professor of Pediatrics*. Dr. Berry specializes in metabolic and genetic disorders, and is certified in pediatrics, biochemical genetics and pediatric endocrinology.

Rittik Chaudhuri, MD, PhD: *Medical Doctor and a PhD in Cell Biology, currently working at a Hospital in Boston*. Dr. Chaudhuri did a joint PhD between the Robinson and Bonifacino labs, in which the AP-4 adaptor complex was jointly discovered, and his PhD involved investigating adaptor complex assembly.

Wendy K. Chung, MD, PhD: *Associate Professor of Pediatrics (in Medicine) Columbia University Institute for Genomic Medicine*. Dr. Chung is a board certified clinical geneticist with a PhD in molecular genetics. She is the director of the clinical genetics program at Columbia University, a co-director of the molecular genetics diagnostics lab, and heads a research laboratory in the division of molecular genetics investigating the genetic bases for a variety of Mendelian and complex traits.

Alexandra Davies: *PhD candidate, jointly supervised by Dr. Borner (Max Planck Institute of Biochemistry, Germany) and Professor Robinson, Cambridge Institute for Medical Research*. The focus of Alex's PhD research involves studying the AP-4 adaptor complex using CRISPR CAS9 and other tools.

Darius Ebrahimi-Fakhari, MD, PhD: *Resident physician in the combined Pediatrics & Child Neurology Program at Boston Children's Hospital / Harvard Medical School*. Dr. Ebrahimi-Fakhari has a long-standing interest in childhood-onset neurometabolic-, neurodegenerative-, and movement disorders.



Dr. Jennifer Hirst: *Principal Research Associate, Robinson lab, Cambridge Institute for Medical Research*. Dr. Hirst is a cell biologist who discovered the AP-4 and AP-5 adaptor complexes and has been studying their function and link with Hereditary Spastic Paraplegia.

Robin Kleiman, PhD: *Research Associate, Translational Neuroscience Center, Boston Children's Hospital*. Dr. Kleiman is a research scientist specializing in neuroscience and systems biology. She has 14 years of experience in drug discovery which includes new target identification and validation, biological pathway mining, biomarker discovery and drug development.

Professor Margaret (Scottie) Robinson: *Principal Investigator, Cambridge Institute for Medical Research*. Prof Robinson has worked on identifying and characterizing adaptor protein complexes for 30 years.

Mustafa Sahin, MD, PhD: *Director, Translational Neuroscience Center, Professor in Neurology, Harvard Medical School*. Dr. Sahin's lab investigates the normal cellular functions of signaling pathways implicated in neurological disease. His research is focused on proteins affected in TSC and SMA.

Meeting Agenda:

1. Brief introductions
2. Presentations:
    a. Dr. Darras: SPG47 clinical overview
    b. Dr. Blackstone: Outcome measurement
    c. Dr. Hirst: AP-4 cell biology
    d. Dr. Kleiman: Fibroblast/drug screening overview
    e. Dr. Azzouz: Presentation of gene therapy proposal
3. Discussion topics:
    a. Is SPG47 solvable?
    b. What are the best short and long term strategies?
4. Priorities for research funding
5. Fundraising and growing the Cure SPG47 organization
6. Open discussion
7. Conclusion and review of next steps

Dinner:

Refreshments will be available during the meeting.

You are also invited to join us for dinner at the Longwood Grille & Bar at 6:00 – our treat! Please RSVP to info@curespg47.org so that we can provide the restaurant with an accurate head count.

http://www.longwoodgrilleandbar.com/
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