

## BIOGRAPHICAL SKETCH

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NAME Susan A. Slaughaupt	POSITION TITLE Associate Professor of Neurology (HMS) Assistant Geneticist (MGH)		
eRA COMMONS USER NAME sslaughaupt			
EDUCATION/TRAINING <i>(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)</i>			
INSTITUTION AND LOCATION	DEGREE <i>(if applicable)</i>	YEAR(s)	FIELD OF STUDY
Eckerd College, St. Petersburg, FL	B.S.	1985	Biology
University of Pittsburgh, Pittsburgh, PA	M.S.	1988	Human Genetics
University of Pittsburgh, Pittsburgh, PA	Ph.D.	1991	Human Genetics

### A. Positions and Honors

1991 - 1995 Research Fellow in Neurology, Massachusetts General Hospital and Harvard Medical School  
 1995 - 2000 Instructor in Neurology, Harvard Medical School  
 1997 - Director, Statistical Genetics Module, MGH Genomics Core Facility  
 2001 - 2006 Assistant Professor of Neurology, Massachusetts General Hospital and Harvard Medical School  
 2005 - Director, Genetics and Genomics Unit, Clinical Research Program, Mass General Hospital  
 2006 - Associate Professor of Neurology, Massachusetts General Hospital and Harvard Medical School

1981 - 1990 Wilbur C. Stauble Trust Scholarship, Western Pacific Industries  
 1982 - 1985 Honor Scholarship, Eckerd College  
 1986 Travel Award - Seventh International Congress of Human Genetics, Berlin  
 1988, 1990 Awardee - University of Pittsburgh Honors Convocation  
 1999 Golden Family Foundation Fellowship, 50<sup>th</sup> Anniversary Program for Scholars in Medicine, HMS  
 2005 William H. Kadel Medal for Outstanding Career Achievement, Eckerd College Alumni Association

### B. Selected peer-reviewed publications (selected from 88)

- Morizot DC, Slaughaupt SA, Kallman KD, Chakravarti A. 1991. Genetic linkage map of fishes of the genus *Xiphophorus* (Teleostei: Poeciliidae). Genetics 127:399.
- Lupski JR, Montes de Oca-Luna R, Slaughaupt SA, Pentao L, Guzzetta V, Trask BJ, Saucedo-Cardenas O, Barker D, Killian JM, Garcia CA, Chakravarti A, Patel PI. 1991. DNA Duplication Associated with Charcot-Marie-Tooth Disease Type 1A. Cell. 66:1.
- Blumenfeld A, Slaughaupt SA, Axelrod FB, Lucente DE, Maayan C, Liebert CB, Ozelius LJ, Trofatter JA, Haines JL, Breakefield XO, Gusella JF. 1993. Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. Nature Genetics. 4:160-164.
- Angrist M, Kauffman E, Slaughaupt SA, Matise TC, Puffenburger EG, Washington SS, Lipson A, Cass DT, Reyna T, Weeks DE, Sieber W, and Chakravarti A. 1993. A gene for Hirschsprung disease (megacolon) in the pericentromeric region of human chromosome 10. Nature Genetics. 4:451-356.
- Velinov M, Slaughaupt SA, Stoilov I, Scott CI, Gusella JF, and Tsipouras P. 1994. The achondroplasia gene maps to the telomeric region of chromosome 4p. Nature Genetics. 6(3):314-317.
- Slaughaupt SA, Blumenfeld A, Liebert CB, Mull J, Lucente DE, Monahan M, Breakefield XO, Axelrod FB, Maayan C, Parada L, and Gusella, JF. 1995. The human gene for neurotrophic tyrosine kinase receptor type 2 is located on chromosome 9 but is not the familial dysautonomia gene. Genomics. 25:730-732.
- Chadwick BP, Helbling LA, Angrist M, Chakravarti A, Gusella JF, Slaughaupt SA. 1998. Assignment of persephin (PSPN), a human neurotrophic factor, to chromosome 19p13.3 by radiation hybrid mapping and somatic cell hybrid PCR Cytogenet Cell Genet. 83(3-4):236-237.
- Chadwick BP, Mull J, Helbling LA, Gill S, Leyne M, Robbins CM, Pinkett HW, Makalowska I, Maayan C, Blumenfeld A, Axelrod FB, Brownstein M, Gusella JF, Slaughaupt SA. 1999. Cloning, mapping, and expression of two novel

- actin genes, Actin-Like-7A (ACTL7A) and Actin-Like-7B (ACTL7B) from the Familial dysautonomia candidate region on 9q31. Genomics. 58:302-309.
- Chadwick BP, Leyne M, Gill S, Liebert CB, Mull J, Mezey E, Makalowska I, Robbins C, Frischauf AM, Maayan C, Blumenfeld A, Axelrod FB, Brownstein M, Gusella JF, Slaugenhaupt SA. 1999. Cloning, mapping, and expression of a novel brain specific transcript in the Familial Dysautonomia candidate region on 9q31. Mamm. Genome. 11:81-83.
- Sun M, Goldin E, Stahl S, Falardeau JL, Kennedy JC, Acierno JS, Bove C, Kaneski CR, Nagle J, Bromley MC, Colman M, Schiffmann R, Slaugenhaupt SA. Mucopolidosis type IV is caused by mutations in a gene encoding a novel transient receptor potential channel. Hum Molec Genet 2000; 9(17):2471-2478.
- Slaugenhaupt SA, Blumenfeld A, Gill SP, Leyne M, Mull J, Cuajungco MP, Liebert CB, Chadwick B, Idelson M, Reznik L, Robbins CM, Makalowska I, Brownstein MJ, Krappmann D, Scheidereit C, Maayan C, Axelrod FB, Gusella JF. Tissue-specific expression of a splicing mutation in the *IKBKAP* gene causes familial dysautonomia. Am J Hum Genet 2001; 68:598-605.
- Smoller JW, Acierno JS, Rosenbaum JF, Biederman J, Pollack MH, Meminger S, Pava J, Helbling L, White C, Bulzacchelli M, and Slaugenhaupt SA. Targeted genome screen of panic disorder and anxiety disorder proneness using homology to murine QTL regions. 2001; Neuropsychiatric Genetics 105:195-206.
- Smoller JW, Rosenbaum JF, Biederman J, Susswein LS, Kennedy J, Kagan J, Snidman NC, Laird N, Tsuang MT, Faraone SV, Schwarz A, and Slaugenhaupt, SA. Genetic association analysis of behavioral inhibition using candidate loci from mouse models. Neuropsychiatric Genetics 2001; 105:226-235.
- Acierno JS jr, Kennedy JC, Falardeau JL, Leyne M, Bromley MC, Colman MW, Sun M, Bove C, Ashworth LK, Chadwick LH, Schiripo T, Ma S, Goldin E, Schiffmann R, Slaugenhaupt SA. A physical and transcript map of the *MCOLN1* gene region on human chromosome 19p13.3-p13.2. Genomics. 2001 Apr 15;73(2):203-10.
- Cuajungco MP, Leyne M, Mull J, Gill SP, Gusella JF, Slaugenhaupt SA. 2001. Cloning, characterization, and genomic structure of the mouse *Ikbkap* gene. DNA and Cell Biology 20(9):579-86.
- Slaugenhaupt SA. 2002. Genetics of familial dysautonomia: Tissue-specific expression of a splicing mutation in the *IKBKAP* gene. Clin Auton Res 12(Suppl 1):15-19.
- Falardeau JL, Kennedy JC, Acierno JS, Sun M, Stahl S, Goldin E, Slaugenhaupt SA. 2002. Cloning and characterization of the mouse *Mcoln1* gene reveals an alternatively spliced transcript not seen in humans. BMC Genomics 3:3
- Slaugenhaupt SA. 2002. The Molecular basis of Mucopolidosis Type IV. Curr Molec Med. 2(445-450).
- LaPlante JM, Falardeau J, Sun M, Kanazirska M, Brown EM, Slaugenhaupt SA, Vassilev PM. 2002. Identification and characterization of the single channel function of human mucolipin-1 implicated in mucopolidosis type IV, a disorder affecting the lysosomal pathway. FEBS Lett. 532(1-2):183-7.
- Cuajungco M, Leyne M, Mull J, Gill S, Lu W, Zagzag D, Axelrod FB, Maayan C, Gusella J, Slaugenhaupt SA. 2003. Tissue-specific reduction in splicing efficiency of *IKBKAP* due to the major mutation associated with Familial Dysautonomia. Am J Hum Genet. 72(3):749-58.
- Freed LA, Acierno JS Jr, Dai D, Leyne M, Marshall JE, Nesta F, Levine RA, Slaugenhaupt SA. 2003. A locus for autosomal dominant mitral valve prolapse on chromosome 11p15.4. Am J Hum Genet. 72:1551-9.
- Slaugenhaupt SA, Mull J, Leyne M, Cuajungco MP, Gill SP, Hims M, Quintero F, Axelrod FB, Gusella JF. 2004. Rescue of a human mRNA splicing defect by the plant cytokinin kinetin. Hum Mol Genet; 13(4):429-36
- Axelrod FB, Goldberg JD, Rolnitzky L, Mull J, Mann SP, Gold von Simson G, Berlin D, Slaugenhaupt SA. 2005. Fludrocortisone in patients with familial dysautonomia--assessing effect on clinical parameters and gene expression. Clin Auton Res.;15(4):284-91.
- Nesta F, Leyne M, Yosefy C, Simpson C, Dai D, Marshall JE, Hung J, Slaugenhaupt SA\*, Levine RA\*. 2005. New locus for autosomal dominant mitral valve prolapse on chromosome 13: clinical insights from genetic studies. Circulation. 112(13):2022-30.\*co-senior authors
- Close P, Hawkes N, Cornez I, Creppe C, Lambert CA, Rogister B, Siebenlist U, Merville MP, Slaugenhaupt SA, Bours V, Svejstrup JQ, Chariot A. 2006. Transcription impairment and cell migration defects in elongator-depleted cells: implication for familial dysautonomia. Mol Cell. May 19;22(4):521-31.
- Laplante JM, Sun M, Falardeau J, Dai D, Brown EM, Slaugenhaupt SA\*, Vassilev PM\*. Lysosomal exocytosis is impaired in mucopolidosis type IV. Mol Genet Metab. 2006 Dec;89(4):339-348. \*co-senior authors
- Ibrahim EC, Hims MM, Shomron N, Burge CB, Slaugenhaupt SA\*, Reed R\*. 2007. Weak definition of *IKBKAP* exon 20 leads to aberrant splicing in familial dysautonomia. Hum Mutat. 28(1):41-53. \*co-senior authors
- Hims MM, Ibrahim EC, Leyne M, Mull J, Liu L, Lazaro C, Shetty RS, Gill S, Gusella JF, Reed R, and Slaugenhaupt SA. 2007. Therapeutic potential and mechanism of kinetin as a treatment for the human splicing disease familial dysautonomia. J Mol Med. 85(2):149-61.
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- Venugopal B, Browning MF, Curcio-Morelli C, Varro A, Michaud N, Nanthakumar N, Walkley SU, Pickel J, Slaugenhaupt SA. 2007. Neurologic, Gastric, and Ophthalmologic Pathologies in a Murine Model of Mucopolidosis Type IV. *Am J Hum Genet*, 81:1070-1083.
- Gold-von Simson G, Leyne M, Mull J, Rolnitzky LM, Goldberg JD, Berlin D, Axelrod FB, Slaugenhaupt SA. *IKBKAP* mRNA in peripheral blood leukocytes: a molecular marker of gene expression and splicing in familial dysautonomia. *Pediatr Res*. 2008 Feb;63(2):186-90.
- Simson GG, Goldberg JD, Rolnitzky LM, Mull J, Leyne M, Voustianiouk A, Slaugenhaupt SA, Axelrod FB. Kinetin in familial dysautonomia carriers: implications for a new therapeutic strategy targeting mRNA splicing. *Pediatr Res*. 2008 63(2):186-90.
- Chen YT, Hims MM, Shetty RS, Mull J, Liu L, Leyne M, Slaugenhaupt SA. Loss of mouse *Ikbkap*, a subunit of Elongator, leads to transcriptional deficits and embryonic lethality that can be rescued by human *IKBKAP*. *Mol Cell Biol*. 2009. 29(3):736-44.
- Simson GG, Goldberg JD, Rolnitzky LM, Mull J, Leyne M, Voustianiouk A, Slaugenhaupt SA, Axelrod FB. Kinetin in familial dysautonomia carriers: implications for a new therapeutic strategy targeting mRNA splicing. *Pediatr Res*. 2008. Nov. 19 (Epub ahead of print)
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- Micsenyi MC, Dobrenis K, Stephney G, Pickel J, Vanier MT, Slaugenhaupt SA, Walkley SU. Neuropathology of the *Mcoln1*<sup>-/-</sup> knockout mouse model of Mucopolidosis type IV. *J Neuropathol Exp Neurol*. 2009. 68(2): 125-135.

### **C. Research Support**

#### **Ongoing Research Support**

R21 NS058318 Slaugenhaupt (PI)

9-15-07 to 9-14-09

NIH/NINDS

“Development of kinetin as a treatment for familial dysautonomia”

The goal of this grant is to test kinetin in an animal model of FD in order to move towards clinical trials in patients.

R01 NS39995 Slaugenhaupt (PI)

2-15-03 to 2-14-08 (no cost extension)

NIH/NINDS

“Molecular Analysis of Mucopolidosis IV”

The major goal of this grant is determine the function of mucopolin-1, the gene that causes Mucopolidosis Type IV.

R01 NS36326-05 Slaugenhaupt (PI)

1-15-04 to 12-31-09 (no cost extension)

NIH/NINDS

“Mechanism of Familial Dysautonomia”

The major goal of this grant is to characterize the splicing defect in *IKBKAP* that causes familial dysautonomia.

Leducq Foundation, Transatlantic Network of Excellence for Cardiovascular Research 10-1-07 to 9-30-12

“Mitral Valve Disease: From Genetic Mechanisms to Improved Repair” Levine (PI)

This grant funds an international network of investigators working on mitral valve disease.

Role: Core Member, US Scientific Director

ML4 Foundation

7-1-08 to 6-30-09

“Development of a neuronal cell line and testing of potential therapies in mucopolidosis type IV murine cellular models”

Role: Mentor

This grant provides partial salary support for Cyntia Morelli to work on a project aimed at testing compounds in an MLIV cellular model.

Dysautonomia Foundation Slaugenhaupt (PI) 10-1-08 to 9-30-09

"High-throughput luminescence-based assay for identifying therapeutic agents that alter *IKBKAP* expression and splicing"

This goal of this grant is to generate an assay for high-throughput screening of splicing modulators.

UL1 RR 025758-01

05/19/08- 04/30/2013

NIH

"Harvard Clinical and Translational Science Center (UL1)"

P.D/ P.I.: Lee M Nadler, MD

Co-P.D.: Steven D Freedman, MD, PhD

Role: Genetics Support Team

Goals: Provide enriched resources to educate and develop the next generation of researchers trained in the complexities of translating research discoveries into clinical trials and ultimately into practice. Design new and improved clinical research informatics tools for analyzing research data and managing clinical trials. Support outreach to underserved populations, local community and advocacy organizations, and health care providers. Assemble interdisciplinary teams and forge new partnerships with private and public health care organizations.