Date: 12/12/16

Neal John Sondheimer, M.D. Ph.D.

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The Hospital for Sick Children
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Toronto, ON, M5G 1X8

Education:

1994	A.B.	Harvard University (Biology)
2000	Ph.D.	University Of Chicago – Molecular Genetics and Cell Biology
2002	M.D.	University of Chicago
2009	Postdoc	The University of Pennsylvania – Genetics

Postgraduate Training and Fellowship Appointments:

2002-2007	Resident in Pediatrics, The Children's Hospital of Philadelphia
2002-2008	Resident in Genetics, The Children's Hospital of Philadelphia
2007-2009	Post-Doctoral Fellow, The University of Pennsylvania

Faculty Appointments:

2002-2005	Instructor-B, The University of Pennsylvania
2007-2009	Clinical Associate in Pediatrics, The University of Pennsylvania
2009-2015	Assistant Professor of Pediatrics, University of Pennsylvania School of Medicine
2015-present	Assistant Professor of Paediatrics, The University of Toronto School of Medicine

Hospital and/or Administrative Appointments:

2006-2015	Attending Physician, The Children's Hospital of Philadelphia
2012-2013	Co-Director, Palmieri Metabolic Lab - The Children's Hospital of Philadelphia
2013-2014	Assistant Program Director, Genetics Residency Programs
2014-2015	Program Director, Medical Genetics, The Children's Hospital of Philadelphia and The University of Pennsylvania
2014-2015	Training Director - Clinical Biochemical Genetics, The Children's Hospital of Philadelphia
2015-present	Staff Physician, SickKids
2015-2016	Member, Research Ethics Board, SickKids
2016-present	Residency Training Committee (Genetics), SickKids
2017	Interim Program Director (Genetics), SickKids

Other Appointments:

2008-2012	Co-Director, Mitochondrial Research Affinity Group, The Children’s Hospital of Philadelphia
2012-2014	Trustee Committee on Facilities and Campus Planning

Specialty Certification:

2006	American Academy of Pediatrics
2007	American Board of Medical Genetics - Clinical Genetics
2009	American Board of Medical Genetics - Clinical Biochemical Genetics

Licensure:

2004-2016	Commonwealth of Pennsylvania
2015-present	The College of Physicians and Surgeons of Ontario

Awards, Honors and Membership in Honorary Societies:

2012	Society for Pediatric Research
2013	Young Physician-Scientist Award - American Society for Clinical Investigation
2014	Outstanding Speaker Award - American Association of Clinical Chemistry
2015	Resident Teaching Prize (Genetics), SickKids

Memberships in Professional and Scientific Societies and Other Professional Activities:

International:

2013-Present	ASBMB
2016-Present	North American Metabolic Academy (Faculty)
2016-Present	SSIEM

National:

2006-present	Society for Inherited Metabolic Disorders

Major Academic and Clinical Teaching Responsibilities:

2005-2013	Lecturer - Pediatrics 200, UPenn SOM
2006-2013	Lecturer - MOD1006 (Medical Genetics), UPenn SOM
2010-2012	Research Mentor (Genetics residency) - Kristin D'Aco
2010	Center for Neurodegenerative Disease Research Lecture Series - "The influence of aging on mitochondrial heteroplasmy"
2010	Genes Genomes and Pediatric Disease Retreat Seminar - "Mitochondrial heteroplasmy and the influence of aging"
2011	Neonatology Grand Rounds - "Inheritance and Unpredictability:_Mitochondrial Heteroplasmy and Its Role in Disease"
2011	Genes Genomes and Pediatric Disease Seminar - "Use of the HSP2 promoter in mitochondrial transcription"
2012	Laura Dribin Mitochondrial Symposium - Course Faculty
2012	CHOP Research Scientific Symposium - Lecture
2012	Grand Rounds - The Children's Hospital of Philadelphia - "Exome Sequencing for Pediatric Disorders"

2013-2014	Lecturer - CAMB605, University of Pennsylvania SOM
2015-2016	Lecturer – MGY470, University of Toronto

Alternative Media:

1. The Health Show - #1310. The Health Show (NPR) May 2013.

2. "Chromosomal and Genetic Disorders" Board Review Pediatrics Series. Audio Digest Foundation, 13, 2014.
3. “Three Parent Embryos” – The National (CBC), September 27th, 2016

Patents:

Recombinant prion-like proteins and materials comprising same. USA Patent Number 7,569,660, 2009.

PUBLICATIONS (2012-2016):

1. Ahrens-Nicklas, R, Umanah, G., **Sondheimer, N.**, Deardorff, M., Wilkens, A., Conlin, L., Santani, A., Nesbitt, A., Juulsola., J et al. (2016). Precision therapy for a new disorder of AMPA receptor recycling due to mutations in ATAD1. Neurol. Genet. *in press*.
2. Assoum, M., Philippe, C., Isidor, B., Perrin, L., Makrythanasis, P., **Sondheimer, N**., Paris, C., Douglas, J., Lesca, G., Antonarakis, S., et al. (2016). Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. Am. J. Hum. Genet. *in press* (10.1016/j.ajhg.2016.10.009)
3. Bhoj, E., Li, M., Ahrens-Nicklas, R., Pyle, L., Wang, J., Zhang, V., Clarke, C., Wong, L., **Sondheimer, N**., Ficicioglu, C., et al. (2015). Pathologic Variants of the Mitochondrial Phosphate Carrier SLC25A3: Two New Patients and Expansion of the Cardiomyopathy/Skeletal Myopathy Phenotype With and Without Lactic Acidosis. JIMD Rep. *19*, 59–66.
4. Clarke, C., Xiao, R., Place, E., Zhang, Z., **Sondheimer, N**., Bennett, M., Yudkoff, M., and Falk, M.J. (2013). Mitochondrial respiratory chain disease discrimination by retrospective cohort analysis of blood metabolites. Mol. Genet. Metab. *110*, 145–152.
5. D'Aco, K.E., Manno, M., Clarke, C., Ganesh, J., Meyers, K.E.C., and **Sondheimer, N**. (2013). Mitochondrial tRNA(Phe) mutation as a cause of end-stage renal disease in childhood. Pediatr. Nephrol. *28*, 515–519.
6. Gai, X., Ghezzi, D., Johnson, M.A., Biagosch, C.A., Shamseldin, H.E., Haack, T.B., Reyes, A., Tsukikawa, M., Sheldon, C.A., Srinivasan, S., **Sondheimer, N**. et al. (2013). Mutations in FBXL4, encoding a mitochondrial protein, cause early-onset mitochondrial encephalomyopathy. Am. J. Hum. Genet. *93*, 482–495.
7. Ganetzky, R., Finn, E., Bagchi, A., Zollo, O., Conlin, L., Deardorff, M., Harr, M., Simpson, M., McGrath, J., Zackai, E., Lemmon, MA., **Sondheimer, N**. (2015). EGFR mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. Mol. Genet. Genomic Med. *3*, 452–458.
8. Grant, S.F.A., Glessner, J.T., Bradfield, J.P., Zhao, J., Tirone, J.E., Berkowitz, R.I., Hakonarson, H., and **Sondheimer, N**. (2012). Lack of relationship between mitochondrial heteroplasmy or variation and childhood obesity. Int. J. Obes. (Lond). *36*, 80–83.
9. Schadt, E.E., Banerjee, O., Fang, G., Feng, Z., Wong, W.H., Zhang, X., Kislyuk, A., Clark, T.A., Luong, K., Keren-Paz, A., **Sondheimer, N.** et al. (2013). Modeling kinetic rate variation in third generation DNA sequencing data to detect putative modifications to DNA bases. Genome Res. *23*, 129–141.
10. Seifert, E.L., Ligeti, E., Mayr, J.A., **Sondheimer, N**., and Hajnóczky, G. (2015). The mitochondrial phosphate carrier: Role in oxidative metabolism, calcium handling and mitochondrial disease. Biochem. Biophys. Res. Commun. *464*, 369–375.
11. Seifert, E.L., Gál, A., Acoba, M.G., Li, Q., Anderson-Pullinger, L., Golenár, T., Moffat, C., **Sondheimer, N**., Claypool, S.M., and Hajnóczky, G. (2016). Natural and induced mitochondrial phosphate carrier loss: differential dependence of mitochondrial metabolism and dynamics, and cell survival, on the extent of depletion. J. Biol. Chem. jbc.M116.744714.
12. **Sondheimer, N**. (2013). Newborn Screening by Sequence and the Road Ahead. Clin. Chem. *59*, 1011–1013.
13. **Sondheimer, N**., Soundararajan, S., Koutzaki, S.H., and Doyle, A.M. (2014). Kidney transplantation from a deceased donor with metachromatic leukodystrophy. Transplantation *97*, e42-4.
14. **Sondheimer, N**., Zollo, O., Van Deerlin, V., and Trojanowski, J.Q. (2014). Analysis of

cerebrospinal fluid mitochondrial DNA levels in Alzheimer disease. Ann. Neurol. *75*, 458–460.
15. Spinale, J.M., Laskin, B.L., **Sondheimer, N**., Swartz, S.J., and Goldstein, S.L. (2013). High-dose continuous renal replacement therapy for neonatal hyperammonemia. Pediatr. Nephrol. *28*, 1–4.
16. Vergano, S.A., Crossette, J.M., Cusick, F.C., Desai, B.R., Deardorff, M.A., and **Sondheimer, N**. (2013). Improving surveillance for hyperammonemia in the newborn. Mol. Genet. Metab. *110*, 102–105.
17. Zollo, O., Tiranti, V., and **Sondheimer, N**. (2012). Transcriptional requirements of the distal heavy-strand promoter of mtDNA. Proc. Natl. Acad. Sci. U. S. A. *109*, 6508–6512.

Lectures by Invitation (Last 5 years):

Oct, 2012	"The Somatic Stability of MtDNA in Aging and Disease," Department of Epidemiology, Harvard School of Public Health
Oct, 2012	"Regulation of Mitochondrial Transcription - Many Promoters and More Players," Department of Pharmacological Sciences, Stony Brook University
Jun, 2013	"Regulating the Power Supply through Mitochondrial Transcription", Department of Biochemistry and Molecular Biology, The Pennsylvania State University
May, 2014	"Defects of mitochondrial phosphate transport," Thomas Jefferson University
Jul, 2014	"Newborn Screening in the Era of Genomic Medicine" - American Association of Clinical Chemistry Meetings - Chicago
Nov, 2014	"Mechanisms of Mitochondrial Disease", Division of Human Genetics, SickKids, Toronto

GRANTS HELD (2002-2006)

Ongoing Research Support

Transdisciplinary Research Center for Preterm Birth 11/01/14-10/31/19
PI: Deborah Driscoll
The Transdisciplinary Research Center for Preterm Birth investigates metabolic and environmental cues leading to preterm birth. I lead the team responsible for examining the effect of somatic and inherited mitochondrial mutations upon the risk for preterm birth.

Department of Paediatrics Funding 9/01/15-8/31/18
PI: Neal Sondheimer
Unrestricted startup fund.

SickKids Center For Genomic Medicine Award 9/01/15-8/31/18
PI: Neal Sondheimer
Funding for the development of techniques for the analysis of mitochondrial sequence.

Completed Research Support

R01ES021733 Andrea Baccarelli 09/01/12-06/30/16
Molecular and Epigenetic Mitochondriomics of Air Particles, Lead and Cognition
This subproject will investigate mitochondrial heteroplasmy and DNA damage in the MOBILIZE cohort as part

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