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Education:

1994	A.B.	Harvard University (Biology)
2000	Ph.D.	University Of Chicago (Molecular Genetics and Cell Biology)
2002	M.D.	University of Chicago Pritzker School of Medicine
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-present	Assistant Professor of Pediatrics, University of Pennsylvania School of Medicine

Hospital and/or Administrative Appointments:

2006-present	Attending Physician, The Children's Hospital of Philadelphia
2012-2013	Clinical Signout, Palmieri Metabolic Lab - The Children's Hospital of Philadelphia
2013-2014	Assistant Program Director, Genetics Residency Programs
2014-present	Program Director, Medical Genetics, The Children's Hospital of Philadelphia and The University of Pennsylvania
2014-present	Training Director - Clinical Biochemical Genetics, The Children's Hospital of Philadelphia

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

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

Editorial Positions:

2010-Present	Reviewer - Developmental Disabilities Research Reviews
2010-Present	Reviewer - Human Mutation
2011-Present	Reviewer - BBA - Gene Regulatory Mechanisms
2011-Present	Reviewer - PLoS One
2012-Present	Reviewer - Clinical Chemistry
2012-Present	Reviewer - Mitochondrion
2012-Present	Reviewer - BMC Genomics
2012-Present	Reviewer - Pediatric Emergency Care
2012-Present	Reviewer - Nucleic Acids Research
2013-Present	Special Society Journal Reviewer - Alzheimer's & Dementia: the Journal of the Alzheimer's Association
2013-Present	Reviewer - The Journal of Pediatrics
2013-Present	Reviewer - Stem Cell Research
2014-Present	Reviewer - American Journal Of Obstetrics and Gynecology
2014-Present	Reviewer - Neurochemistry International
2015-Present	Reviewer - American Journal Of Human Biology

Major Academic and Clinical Teaching Responsibilities:

2005-2013	Lecturer - Pediatrics 200
2006-2013	Lecturer - MOD1006 (Medical Genetics)
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

Bibliography:

Research Publications, peer reviewed (print or other media):

1. Sokol RJ., Devereaux MW., Khandwala RA., Narkewicz MR., Sondheimer NJ.: Effect of vitamin E on transport processes in isolated rat hepatocytes. Journal of Pediatric Gastroenterology & Nutrition 11(2): 261-7, Aug 1990.

2. Sokol RJ., Taylor SF., Devereaux MW., Khandwala R., Sondheimer NJ., Shikes RH., Mierau G.: Hepatic oxidant injury and glutathione depletion during total parenteral nutrition in weanling rats. American Journal of Physiology 270(4 Pt 1): G691-700, Apr 1996.

3. Sondheimer N., Lindquist S.: Rnq1: an epigenetic modifier of protein function in yeast. Molecular Cell 5(1): 163-72, Jan 2000.

4. Satyal SH., Schmidt E., Kitagawa K., Sondheimer N., Lindquist S., Kramer JM., Morimoto RI.: Polyglutamine aggregates alter protein folding homeostasis in Caenorhabditis elegans. Proceedings of the National Academy of Sciences of the United States of America 97(11): 5750-5, May 23 2000. PMCID: PMC18505

5. Sondheimer N., Lopez N., Craig EA., Lindquist S.: The role of Sis1 in the maintenance of the [RNQ+] prion. EMBO Journal 20(10): 2435-42, May 15 2001. PMCID: PMC125465

6. Liu JJ., Sondheimer N., Lindquist SL.: Changes in the middle region of Sup35 profoundly alter the nature of epigenetic inheritance for the yeast prion [PSI+] Proceedings of the National Academy of Sciences of the United States of America 99 Suppl 4: 16446-53, Dec 10 2002. PMCID: PMC139907

7. Ernst LM., Sondheimer N., Deardorff MA., Bennett MJ., Pawel BR.: The value of the metabolic autopsy in the pediatric hospital setting. Journal of Pediatrics 148(6): 779-83, Jun 2006.

8. Biswas, Gopa. Tang, Weigang. Sondheimer, Neal. Guha, Manti. Bansal, Seema. Avadhani, Narayan G.: A distinctive physiological role for IkappaBbeta in the propagation of mitochondrial respiratory stress signaling. Journal of Biological Chemistry 283(18): 12586-94, May 2 2008. PMCID: PMC2335355

9. Deardorff, Matthew A. Gaddipati, Himabindu. Kaplan, Paige. Sanchez-Lara, Pedro A. Sondheimer, Neal. Spinner, Nancy B. Hakonarson, Hakon. Ficicioglu, Can. Ganesh, Jaya. Markello, Thomas. Loechelt, Brett. Zand, Dina J. Yudkoff, Marc. Lichter-Konecki, Uta.: Complex management of a patient with a contiguous Xp11.4 gene deletion involving ornithine transcarbamylase: a role for detailed molecular analysis in complex presentations of classical diseases. Molecular Genetics & Metabolism 94(4): 498-502, Aug 2008. PMCID: PMC2572572

10. Chapman KA, Bennett MJ, Sondheimer N.: Increased C3-carnitine in a healthy premature infant. Clinical Chemistry 54(11): 1914-7, Nov 2008. PMCID: PMC2891149

11. Guha Manti, Tang Weigang, Sondheimer Neal, Avadhani Narayan G: Role of calcineurin, hnRNPA2 and Akt in mitochondrial respiratory stress-mediated transcription activation of nuclear gene targets. Biochimica et biophysica acta 1797(6-7): 1055-1065, Feb 2010. PMCID: PMC2891149

12. Sondheimer Neal, Fang Ji-Kang, Polyak Erzsebet, Falk Marni J, Avadhani Narayan G: Leucine-rich pentatricopeptide-repeat containing protein regulates mitochondrial transcription. Biochemistry 49(35): 7467-73, Sep 2010. PMCID: PMC2932791

13. Sondheimer Neal, Glatz Catherine E, Tirone Jack E, Deardorff Matthew A, Krieger Abba M, Hakonarson Hakon: Neutral mitochondrial heteroplasmy and the influence of aging. Human molecular genetics 20(8): 1653-9, Apr 2011. PMCID: PMC3063991

14. Glatz Catherine, D'Aco Kristin, Smith Sabrina, Sondheimer Neal: Mutation in the mitochondrial tRNA(Val) causes mitochondrial encephalopathy, lactic acidosis and stroke-like episodes. Mitochondrion 11(4): 615-9, Jul 2011. PMCID: 21540128

15. Grant S F A, Glessner J T, Bradfield J P, Zhao J, Tirone J E, Berkowitz R I, Hakonarson H, Sondheimer N: Lack of relationship between mitochondrial heteroplasmy or variation and childhood obesity. International journal of obesity 36(1): 80-3, Jan 2012.

16. Zollo Ornella, Tiranti Valeria, Sondheimer Neal: Transcriptional requirements of the distal heavy-strand promoter of mtDNA. Proceedings of the National Academy of Sciences of the United States of America 109(17): 6508-12, Apr 2012. PMCID: PMC3340101

17. Schadt Eric E, Banerjee Onureena, Fang Gang, Feng Zhixing, Wong Wing H, Zhang Xuegong, Kislyuk Andrey, Clark Tyson A, Luong Khai, Keren-Paz Alona, Chess Andrew, Kumar Vipin, Chen-Plotkin Alice, Sondheimer Neal, Korlach Jonas, Kasarskis Andrew: Modeling kinetic rate variation in third generation DNA sequencing data to detect putative modifications to DNA bases. Genome research 23(1): 129-41, Jan 2013. PMCID: PMC3530673

18. D'Aco Kristin E, Manno Megan, Clarke Colleen, Ganesh Jaya, Meyers Kevin E C, Sondheimer Neal: Mitochondrial tRNA(Phe) mutation as a cause of end-stage renal disease in childhood. Pediatric nephrology (Berlin, Germany) 28(3): 515-9, Mar 2013. PMCID: PMC3557766

19. Vergano Samantha A, Crossette Jonathan M, Cusick Frederick C, Desai Bimal R, Deardorff Matthew A, Sondheimer Neal: Improving surveillance for hyperammonemia in the newborn. Molecular genetics and metabolism 110(1-2): 102-5, May 2013. PMCID: PMC3755016

20. Spinale Joann M, Laskin Benjamin L, Sondheimer Neal, Swartz Sarah J, Goldstein Stuart L: High-dose continuous renal replacement therapy for neonatal hyperammonemia. Pediatric nephrology (Berlin, Germany) 28(6): 983-6, Jun 2013. PMCID: PMC3633740

21. Clarke Colleen, Xiao Rui, Place Emily, Zhang Zhe, Sondheimer Neal, Bennett Michael, Yudkoff Marc, Falk Marni J: Mitochondrial respiratory chain disease discrimination by retrospective cohort analysis of blood metabolites. Molecular genetics and metabolism 110(1-2): 145-52, Sep-Oct 2013. PMCID: PMC3812452

22. Gai Xiaowu, Ghezzi Daniele, Johnson Mark A, Biagosch Caroline A, Shamseldin Hanan E, Haack Tobias B, Reyes Aurelio, Tsukikawa Mai, Sheldon Claire A, Srinivasan Satish, Gorza Matteo, Kremer Laura S, Wieland Thomas, Strom Tim M, Polyak Erzsebet, Place Emily, Consugar Mark, Ostrovsky Julian, Vidoni Sara, Robinson Alan J, Wong Lee-Jun, Sondheimer Neal, Salih Mustafa A, Al-Jishi Emtethal, Raab Christopher P, Bean Charles, Furlan Francesca, Parini Rossella, Lamperti Costanza, Mayr Johannes A, Konstantopoulou Vassiliki, Huemer Martina, Pierce Eric A, Meitinger Thomas, Freisinger Peter, Sperl Wolfgang, Prokisch Holger, Alkuraya Fowzan S, Falk Marni J, Zeviani Massimo: Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. Am J Human Genet 93(3): 482-495, Sep 2013. PMCID: PMC3769923

23. Neal Sondheimer, Ornella Zollo, Vivianna Van Deerlin and John Q. Trojanowski: Analysis of CSF mitochondrial DNA levels in Alzheimer disease. Annals of Neurology 75(3): 458-460, Mar 2014. PMCID: in process

24. Sondheimer, Neal; Soundararajan, Suganthi; Koutzaki, Sirma H.; Doyle, Alden M.: Kidney Transplantation From a Deceased Donor With Metachromatic Leukodystrophy. Transplantation 15(97): e42-4, Apr 2014 Notes: in press. PMCID: in process

25. E.J. Bhoj, M. Li, R. Ahrens-Nicklas, L.C. Pyle, J. Wang, V.W. Zhang, C. Clarke, L.J. Wong, N. Sondheimer, C. Ficicioglu, M. Yudkoff: 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 reports 2015 Notes: Epub ahead of print.

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