

Dr. Neal Sondheimer
Assistant Professor of Paediatrics

A. Date Curriculum Vitae is Prepared: 2018-08-15

B. Biographical Information

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1. EDUCATION

Degrees

June 1995 - June 2002	Doctor of Medicine (M.D.), University of Chicago, Pritzker School of Medicine, Chicago, Illinois, United States.
June 1995 - Dec 2000	Doctor of Philosophy (Ph.D.), Molecular Genetics and Cell Biology, University of Chicago, Chicago, Illinois, United States. Dissertation mentor: Lindquist, Susan Thesis Title: Identification of novel prion elements in <i>Saccharomyces cerevisiae</i>
Sep 1990 - June 1994	Bachelor of Arts (A.B.), Biology, Harvard University, Cambridge, Massachusetts, United States.

Postgraduate, Research and Specialty Training

July 2007 - June 2009	Post-Doctoral Fellow, Genetics, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, United States. Supervisor(s): Narayan Avadhani.
July 2007 - June 2008	Fellow in Clinical Biochemical Genetics, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, United States.
June 2002 - June 2007	Resident in Paediatrics, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, United States.
June 2002 - June 2008	Resident in Genetics, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, United States.

Qualifications, Certifications and Licenses

2007 - Present	Specialty Certification, American Academy of Pediatrics, Elk Grove Village, Illinois, United States.
2007 - Present	Specialty Certification - Clinical Genetics, American Board of Medical Genetics and

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2009 - Present Specialty Certification - Clinical Biochemical Genetics, American Board of Medical Genetics and Genomics, Bethesda, Maryland, United States.
2017 – Present Canadian College of Medical Genetics, Affiliate Member

2. EMPLOYMENT

Current Appointments

HOSPITAL

Sept 2015 - Present Staff Physician, Clinical and Metabolic Genetics, The Hospital for Sick Children, Toronto, Ontario.

Jan 2017 – Present Morbidity and Mortality Divisional Leader. Division of Clinical and Metabolic Genetics, The Hospital for Sick Children, Toronto, Ontario

RESEARCH

Sept 2015 - Present Associate Scientist, Genetics & Genome Biology, Research Institute, The Hospital for Sick Children, Toronto, Ontario.

UNIVERSITY

July 2018 - Present Member, Institute of Medical Sciences, University of Toronto, Toronto, Ontario.
July 2018 - Present Associate Professor, Departments of Paediatrics and Molecular Genetics, University of Toronto, Toronto, Ontario.

Previous Appointments

HOSPITAL

June 2014 - Aug 2015 Training Director, Clinical Biochemical Genetics, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania.
June 2014 - Aug 2015 Program Director, The Children's Hospital of Philadelphia and Medical Genetics, University of Pennsylvania, Philadelphia, Pennsylvania.
July 2013 - June 2014 Assistant Program Director, Genetics Residency Programs, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania.
Nov 2006 - Aug 2015 Attending Physician, Biochemical Genetics, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania.
Jan 2017 – Aug 2017 Interim Program Director, Postgraduate Medical Education, Department of Paediatrics, University of Toronto, Toronto, Ontario.

RESEARCH

Oct 2008 - June 2012 Co-Director, Mitochondrial Research Affinity Group, Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, United States.

UNIVERSITY

Sept 2015 – Jun 2018 Assistant Professor, Department of Paediatrics, University of Toronto, Toronto, Ontario.

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Sept 2016 – Jun 2018	Associate Member, Institute of Medical Sciences, The University of Toronto
Jan 2008 – June 2018	Assistant Professor, Molecular Genetics, The University of Toronto
June 2009 - Aug 2015	Assistant Professor of Pediatrics (Tenure track), Department of Paediatrics, Perelman School of Medicine, The University of Pennsylvania, Philadelphia, Pennsylvania
July 2007 - June 2009	Clinical Associate in Pediatrics, Department of Paediatrics, The University of Pennsylvania, Philadelphia, Pennsylvania..
June 2002 - June 2005	Instructor-B, Department of Paediatrics, The University of Pennsylvania, Philadelphia, Pennsylvania.

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3. HONOURS AND CAREER AWARDS

Distinctions and Research Awards

INTERNATIONAL

Received

- October 2016 **North American Metabolic Academy**, Faculty.
- April 2013 **Young Physician-Scientist Award**, American Society for Clinical Investigation, Ann Arbor, Michigan, United States.
- May 2012 **Society for Pediatric Research**, The Woodlands, Texas, United States. Honorary Society.

Teaching Awards

LOCAL

Received

- May 2016 Margaret W. Thompson Award – Medical Genetics Education

Professional Associations

- Apr 2017 – Present **Member**, Garrod Society
- Sept 2016 - Present **Member**, Society for the Study of Inborn Errors of Metabolism
- Sept 2016 - Present **Member**, Society for Inherited Metabolic Disorders
- Aug 2015 - Present **Member**, The Canadian College of Physicians and Surgeons of Ontario

LOCAL

- Sept 2016 - Present **Committee Member**, STRAD Advisory Committee, Research Training Centre, The Hospital for Sick Children, Toronto, Ontario.
- Jan 2015 - Dec 2016 **Board Member**, Research Ethics Board, Research Institute, The Hospital for Sick Children, Toronto, Ontario.

Peer Review Activities

EDITORIAL BOARDS

- July 2017 - Present **Editorial Board**, Molecular Case Studies, Cold Spring Harbor

GRANT REVIEWS

- Reviewer**. OGS 2017-2018 Scholarships

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MANUSCRIPT REVIEWS

Reviewer. Cold Spring Harbor Molecular Case Studies, HighWire

Reviewer. American Journal of Obstetrics and Gynecology, Elsevier

Reviewer. Biochimica et Biophysica Acta (BBA) - Gene Regulatory Mechanisms, MGM, Elsevier

Reviewer. Congenital Heart Disease, Wiley

Reviewer. PLOS ONE, Public Library of Science

Reviewer. Journal of Inherited Metabolic Disease

Reviewer. Congenital Heart Disease

Reviewer. npj: Genomic Medicine

Other Research and Professional Activities

Developer – MitoTIP – an algorithmic determination of mitochondrial tRNA mutation pathogenicity at www.mitomap.org

Innovations and Development in Teaching and Education

MGY470 Curriculum – Ethics in Clinical Genetics

C. Academic History

1. RESEARCH STATEMENTS

My Research focuses on the regulation of mitochondrial gene expression and the impact of mitochondrial mutations in common and rare disease.

- Strongly pathogenic mitochondrial mutations exist in a state of heteroplasmy, a mixture of normal and mutated genomes. This state provides opportunities for therapy, as the increase of wild type mitochondrial DNA or the suppression of mutated mitochondrial DNA could improve health. My team and I are investigating mechanisms that could allow shifts in heteroplasmic ratios.
- Mitochondrial inheritance has the potential to impact high-prevalence phenotypes by its interactions with the mitochondrial genome. We are studying this question using common diseases and phenotypes such as Alzheimer disease, aging and preterm birth. In addition to mitonuclear coordination low prevalence heteroplasmic mutations in the mitochondrial DNA may have profound effects over time.
- The mitochondrial genome is the small, densely coding, matrilineally inherited DNA that encodes core subunits of the electron transport chain. Many features of gene regulation are more similar to bacterial and phage systems than they are to gene regulation in the nucleus. Defects in the maintenance of mitochondrial DNA and in the translation of gene products are known causes of disease. Myself and my team are investigating the dysregulation of mitochondrial transcription as another possible avenue to bioenergetic failure.

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