Curriculum Vitae

Saadet Andrews (aka Mahmutoglu, Mercimek-Mahmutoglu), MD, PhD, FCCMG, FRCPC
Associate Professor, Division of Clinical and Metabolic Genetics, Department of Paediatrics, University of Toronto
Staff Physician, Clinical and Metabolic Genetics, The Hospital for Sick Children
Project Investigator, Genetics & Genome Biology, Research Institute, The Hospital for Sick Children
Associate Member, Institute of Medical Sciences, University of Toronto

A. Date Curriculum Vitae is Prepared: September 27, 2019

B. Biographical Information

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1. EDUCATION

Degrees

March 2018	Fellow of Royal College of Physicians and Surgeons of Canada (FRCPC)
Jan 2010-Jun 2013	Doctor of Philosophy (PhD), Creatine deficiency syndrome: contributions to selective and newborn screening and treatment, Department of Clinical Chemistry, VU Medical Center, Free University, Amsterdam, The Netherlands. Defense date June 5, 2013. Supervisors: Gajja S Salomons, Cornelis Jakobs.
May 2006-May 2008	Licentiate of the Medical Council of Canada, LMCC (#106310), Medical Council of Canada, Ottawa, Ontario, Canada. Certification Date June 17, 2008.
Sep 2005-May 2008	Fellow of Canadian College of Medical Geneticist (FCCMG), Biochemical Genetics, Department of Paediatrics, Department of Medical Genetics, Department of Laboratory Medicine, University of British Columbia, British Columbia Women's and Children's Hospital, Vancouver, British Columbia, Canada. Certification Date November 20, 2008. Supervisors: Hilary Vallance, Sylvie Langlois, Sylvia Stockler.
Jan-May 2002	Doctor of Medicine (Dr.), Medical Faculty, Leopold-Franzens Innsbruck University, Innsbruck, Austria. Certification Date June 25, 2002. Supervisor: Norbert Mutz (Dean).
Sep 1985-May 1992	Medical Doctor (MD), Licence # 6865, Medical Faculty, Ege University, Izmir, Turkey. Certification Date May 3, 1992. Supervisors: Sermet Akgun (Rector), Turan Ornek (Dean).

Postgraduate, Research and Specialty Training

Postgraduate Specialty Training

Jul 2008-Jun 2009	Clinical Fellow, Paediatric Neurology Fellowship, Division of Neurology, Department of Paediatrics, University of British Columbia, Vancouver, British Columbia, Canada. Supervisors: Mary Connolly, Kathy Selby, Elke Roland.
Sep 2005-May 2008	Biochemical Genetics Fellow, Canadian College of Medical Geneticist (CCMG) Accredited Biochemical Genetics Fellowship, Fellow of Canadian College of Medical Geneticist (FCCMG), Biochemical Genetics, Canadian College of Medical Geneticist, Department of Paediatrics, Department of Medical Genetics, Department of Laboratory Medicine, University of British Columbia, British Columbia Women's and Children's Hospital, Vancouver, British Columbia, Canada. Certification Date November 20, 2008. Supervisors: Hilary Vallance, Sylvie Langlois, Sylvia Stockler.
Jan 2003-Aug 2004	Paediatric Resident, Postgraduate Paediatric Residency Program, Department of Paediatrics, University of Vienna, Vienna, Austria. Certification Date September 25, 2004. Supervisors: Sylvia Stockler, Edith Schoeber, Radvan Urbanek.
Jul 2002-Dec 2002	Clinical Fellow, Division of Metabolic Diseases, Department of Paediatrics, University of Vienna, Vienna, Austria. Supervisors: Olaf Bodamer, Sylvia Stockler
Jun 2000-Jun 2001	Clinical Fellow, Division of Paediatric Nephrology, Department of Paediatrics, University of Vienna, Vienna, Austria. Supervisors: Christopher Aufricht, Thomas Mueller.
Oct 1993-Mar 1998	Paediatric Resident, Postgraduate Paediatric Residency Program, Department of Paediatrics, Cerrahpasa Medical Faculty, Istanbul University, Istanbul, Turkey. Certification Date September 3, 1998. Supervisor: Ozdemir Ilter. Thesis (October 1995- August 1998). Thesis Title: Prevalence of celiac disease in the first-degree relatives of Turkish patients with celiac disease. Supervisors: Gungor Tumay, Tufan Kutlu.

Research Training

May-Sep 2011	PhD student, Metabolic Laboratory, Department of Clinical Chemistry, VU Medical Center, Free University, Amsterdam, The Netherlands. Supervisors: Gajja S Salomons, Cornelis Jakobs.
Jul-Oct 2009	Research Fellow, Department of Paediatrics and Laboratory Medicine, University of Vancouver, Vancouver, British Columbia, CA. Supervisor: Marion Coulter-Mackie.
May-Jun 2005	Research Fellow, Metabolic Laboratory, Department of Clinical Chemistry, Free University, Amsterdam, The Netherlands. Supervisors: Gajja S Salomons, Cornelis Jakobs.
Sep 2004-Aug 2005	Research Fellow, Metabolic Laboratory, Department of Paediatrics, University of Vienna, Vienna, Austria. Supervisors: Birgit Rami, Edith Schober. Sylvia Stockler.

Oct 2004	Research Fellow. Metabolic Laboratory, Department of Clinical Chemistry, Free University, Amsterdam, The Netherlands. Supervisors: Nanda Verhoeven, Cornelis Jakobs.
Jul 2001-Dec 2002	Research Fellow and Clinical Fellow, Metabolic Laboratory and Metabolic Clinic, Department of Paediatrics, University of Vienna, Vienna, Austria. Supervisors: Olaf Bodamer, Sylvia Stockler

Specialty Training, Courses and Workshops

Nov 1, 2016	Workshop Attendee, Exam Chair Workshop, IMS, University of Toronto.
Nov 16 & Dec 2, 2016	Workshop Attendee, GLSE Faculty Development Workshop, IMS, University of Toronto. Supervisor Nana LEE.
Jun 2012	Workshop Attendee, LEAD 201 for Scientists, Management and Development Workshop, Leadership and Organizational Development, The Hospital for Sick Children. Supervisor: Lisa Schmidt.
Sep 2010	Course Attendee, Laboratory Biological Safety Course, Department of Health, Safety and Environment, University of British Columbia, Vancouver, British Columbia, CA.
Apr 2010	Course Attendee, Radionuclide Safety and Methodology (including receiving Class 7 Dangerous Goods) Course, Department of Health, Safety and Environment, University of British Columbia, Vancouver, British Columbia, CA
Sep-Oct 2009	Course Attendee, Understanding Statistics in Medicine: Basic Statistical Concepts Workshop, Center for Epidemiology and Evaluation, University of British Columbia, Vancouver, British Columbia, CA.
Oct 2008-Feb 2009	Course Attendee, Clinical Leadership Development Program, Coach Approach, Children's and Women's Hospital and Health Center of British Columbia, Vancouver, British Columbia, CA.
Jun 2008	Course Attendee, 7th International Postgraduate Course on Lysosomal Storage Diseases, Shire Human Genetic Therapies, Nierstein, Mainz-Birgen, Germany. Supervisor: Michael Beck.
April 2006	Course Attendee, New and Treatable Inborn Errors of Metabolism, CHU Ste-Justine, Department of Medical Genetics, University of Montreal, Montreal, Quebec, CA. Supervisor: Grant Mitchell.
April 2005	Course Attendee, 2nd focus course: Protein glycosylation in health and disease, Orphan Europe Academy, Athens, Greece.
Nov 2004	Course Attendee, 4th European Metabolic Course, Necker-Enfants Malades University Medical Centre, Paris, France. Supervisors: Jean-Marrie Saudubray, George Hoffmann.

Qualifications, Certifications and Licenses

Oct 2011-Present	Member, College of Physicians and Surgeons of Ontario, Toronto, Ontario, CA. License # 96871

Oct 2009-Sep 2011	Member, College of Physicians and Surgeons of British Columbia, Vancouver, British Columbia, CA. License # 25254
Nov 2008	FCCMG, The Canadian College of Medical Geneticist. License/ Membership Number not applicable.
Jun 2008	Member, Medical Council of Canada, Ottawa, Ontario, CA. License # 106310.
Jan 2003-Aug 2005	Member, Austrian Medical Chamber, Vienna, Austria. License # 22694/VI
Sep 1985-Jul 1999	Member, Turkish Medical Chamber, Istanbul, Turkey. License # 6865/ Membership # 26720

2. EMPLOYMENT

Current Appointments

CLINICAL HOSPITAL

Oct 2011-Present — Staff Metabolic Geneticist, Division of Clinical and Metabolic Genetics, Department of Paediatrics, The Hospital for Sick Children, Toronto, Ontario, CA.

I was appointed to develop a comprehensive Epilepsy Genetics Clinic and Neurometabolic Program at The Hospital for Sick Children. I consult on both inpatients and outpatients with complex epilepsy and complex neurodegenerative and neurometabolic disorders. I also became the point of reference for pyridoxine dependent epilepsy and its treatment.

CONSULTING HOSPITAL

May 2013-Present — Associate Staff, Neonatal Intensive Care Unit, Mount Sinai Hospital, Toronto, Ontario, CA.

I was appointed to provide consultations to the patients in the Neonatal Intensive Care Unit as consulting physician for metabolic genetic disorders.

May 2013-Present — Associate Staff, Department of Medicine, University Health Network, Toronto, Ontario, CA.

I was appointed to provide consultations to the patients with metabolic genetic disorders in the Emergency Department, Intensive Care Unit and Wards as consulting physician for metabolic genetic disorders.

CROSS APPOINTMENT HOSPITAL

Aug 2014-Present — Associate Staff, Movement Disorder Clinic, Division of Neurology, Toronto Western Hospital, University of Health Network, Toronto, Ontario, CA.

I was appointed to develop a comprehensive Movement Disorder Genetics Program for the diagnosis and treatment of patients with complex movement disorders in the Division of Clinical and Metabolic Genetics, The Hospital for Sick Children in collaboration with the Division of Paediatric Neurology.

Mar 2012-Present — Associate Staff, Division of Neurology, The Hospital for Sick Children, Toronto, Ontario, CA.

I was appointed to be a part of the epilepsy team to generate a complex epilepsy genetics clinic, in addition to strengthening neurometabolic team efforts at The Hospital for Sick Children.

RESEARCH

2011-Present — Project Investigator, Genetics and Genome Biology Research Program, Research Institute, The Hospital for Sick Children, Toronto, Ontario, CA.

I was appointed to the Research Institute in 2011 after my arrival, enabling me to be the Principle Investigator for research studies performed within The Hospital for Sick Children.

UNIVERSITY

Mar 2015-Present — Associate Member, Institute of Medical Sciences Graduate School, University of Toronto Toronto, Ontario, CA.

Oct 2011-Jun 2016 — Assistant Professor, Department of Paediatrics, Faculty of Medicine, University of Toronto, Toronto, Ontario, CA.

Jul 2016- Present — Associate Professor, Department of Paediatrics, Faculty of Medicine, University of Toronto, Toronto, Ontario, CA.

Previous Appointments

CLINICAL HOSPITAL

Oct 2009-Sep 2011 — Staff Biochemical Geneticist, Division of Biochemical Diseases, Department of Paediatrics, University of British Columbia, Vancouver, British Columbia, CA.

I was appointed to the Neurometabolic Clinic Program at The British Columbia's Children Hospital. I consulted on both inpatients and outpatients with complex neurodegenerative and neurometabolic disorders. I also become a point of reference for pyridoxine dependent epilepsy, creatine deficiency disorders and their treatment.

Jan-July 1999 — Pediatrician, Ömür Private Hospital, Istanbul, Turkey.

I was appointed as a paediatrician to provide clinical and emergency management.

Sep-Dec 1998 — Pediatrician, Neonatal Intensive Care Physician, International Private Hospital, Istanbul, Turkey.

I was appointed as a paediatrician to provide emergency management.

Jan-Sep 1992 — Family Physician, State Children's and Women's Hospital, Afyon, Turkey.

I was appointed as a family physician to provide emergency management for children.

UNIVERSITY

Oct 2009 – Sep 2011 — Assistant Professor, Division of Biochemical Diseases, Department of Paediatrics, University of British Columbia, Vancouver, British Columbia, CA.

WORK INTERRUPTIONS

Sep1999-May 2000	German School, University of Vienna, Vienna, Austria
	I attended to a full time German School after moving to Vienna to be able to do clinical fellowship at The Vienna Children's Hospital. I completed German School at proficiency level to be able to start with fellowship training.

3. HONOURS AND CAREER AWARDS

Distinctions and Research Awards

INTERNATIONAL

Received

March 2017	Movement Disorders "Nomenclature of Genetic Movement Disorders: Recommendations of the MDS Task Force", Authors Marras C, Lang A, van de Warrenburg BP, Sue CM, Tabrizi SJ, Bertram L, Mercimek-Mahmutoglu S, Ebrahimi-Fakhari D, Warner TT, Durr A, Assmann B, Lohmann K, Kostic V, Klein C. Best Review Article of 2016 (Award presented to Dr. Klein and Dr. Marras at the International Parkinson and Movement Disorder Society Annual Meeting in June 2017 in Vancouver, BC, Canada.
July 2016	Epilepsia Clinical Science Research Prize 2016 (Award Presented at American Epilepsy Society 70th Annual Meeting Houston, TX, December 2016) (http://www.ilae.org/Visitors/awards/Epilepsia-2015-Mahmutoglu.cfm).
May-June 2005	Society for the Study of Inborn Error of Metabolism (SSIEM) Exchange Grant to study 'Biochemical and molecular diagnosis of creatine deficiency syndromes'. Principal Author, Amsterdam, The Netherlands. *This was a grant to encourage exchange visits between institutions and provided junior trainees with the opportunity to train and learn in another institution". After I set-up urinary creatine, creatinine and guanidinoacetate measurements in the Metabolic Laboratory at The Vienna Children's Hospital, I was able to continue with the second part of the study in the Metabolic Laboratory, Department of Clinical Chemistry, Free University, Amsterdam, The Netherlands after receiving this exchange grant. The collaborative study results were published in Molecular Genetics and Metabolism in 2009 (*Mercimek-Mahmutoglu S et al: Screening for X-linked creatine transporter (SLC6A8) deficiency via simultaneous determination of urinary creatine to creatinine ratio by tandem mass-spectrometry. 2009;96:273-5.*). Total Amount: 2000 Euros*
Mar 2004	Paediatric Neurology Society Poster Award, Principal Author and Presenter, 30th Annual Meeting of the Paediatric Neurology Society, Bern, Switzerland. *I was the principal author for the poster (Mercimek-Mahmutoglu S et al) titled: Clinical, biochemical and molecular findings in 16 patients with GAMT deficiency, for which I won the second poster prize. This work was the foundation for a publication in Neurology in 2006 (see publications). Total Amount: 300 CHF*

Jan 2003-Aug 2005 — Scholarship, Paediatric Metabolic Training, awarded by Scientific Hospital Supplies (SHS).

I was awarded the Paediatric Metabolic Training Award at The Vienna Children's Hospital. Total Amount: 50,000 Euros

NATIONAL

Received

May 2009 — Linda Stevens Memorial Fund Travel Scholarship, Canadian College of Medical Geneticist, Principal Author and Presenter, Montreal, Quebec, CA.

This award was to support CCMG fellows-in-training by allowing them to participate in conferences to help enhance their educational experience. This travel scholarship award was provided to attend to the Garrod Association Annual Symposium 2009 to present my abstract entitled "A female patient with a medically refractory epilepsy and severe X-linked creatine transporter (SLC6A8) deficiency: successful treatment with l-arginine and l-glycine supplementation" as platform presentation. This work was published in Molecular Genetics and Metabolism in 2010 (see publications). Total amount $500 CND.

May 2006 — Garrod Association Trainee Award, Principal Author and Presenter, Halifax, Nova Scotia, Canada.

This award was to support CCMG fellows-in-training who attended the Garrod Symposium and presented an abstract, to help enhance the educational experience of trainees. I presented my abstract entitled "Long term outcome in patients with argininosuccinate lyase deficiency identified by newborn screening" as a platform presentation. Total amount $500 CND.

Oct 1998 — Dissertation Prize, Tümay Prize, Titled: Prevalence of Celiac disease in first degree relatives of Turkish patients with Celiac disease. Principal Author, Best dissertation prize, Cerrahpasa Medical Faculty, Istanbul University, Istanbul, Turkey.

This was an award received for my dissertation. I was required to complete my dissertation in order to finish my paediatric residency and be eligible for the board exam. Total Amount: 300,000 Turkish Liras.

LOCAL STUDENT AWARDS

Aug 2015 — SickKids Summer Student Research Program, Outstanding Poster Presentation, Toronto, Ontario, Canada

This award was awarded to Theodora Bruun for her poster presentation during the Sickkids Summer Student Research Program Symposium, under my mentorship as primary investigator. Theodora Bruun was hired as an undergraduate research student through funding I received from The Division of Clinical and Metabolic Genetics/Centre for Genetic Medicine Starbucks Clinical Genetics/Genomics Research Studentship Award. The title of the research project was "Neonatal encephalopathy in term newborns: identification of underlying genetic defects using whole exome sequencing".

4. PROFESSIONAL AFFILIATIONS AND ACTIVITIES

Professional Associations

2016- Present	**Member,** American Epilepsy Society, Membership Number is not applicable.
2015-Present	**Member,** International Movement Disorder Society, Membership Number is not applicable.
2014-Present	**Member**, Garrod Association. Membership Number is not applicable.
2012-Present	**Member,** Society for Inherited Metabolic Disorders (SIMD). Membership Number is not applicable.
2011-Present	**Member,** Ontario Medical Association (OMA). Membership Number 1180504
2011-Present	**Member,** The Canadian College of Physicians and Surgeons of Ontario. Membership Number 96871
2009-2011	**Member,** The Canadian College of Physicians and Surgeons of British Columbia. Membership Number 25254
2008-Present	**Member,** Fellow of Canadian College of Medical Geneticist (CCMG). Membership Number 741178
2003-Present	**Member,** Arbeitsgemeinschaft für pädiatricshe Stoffwechselerkrankungen (APS). Membership Number is not applicable.
2002-Present	**Member,** Society for the Study of Inborn Errors of Metabolism (SSIEM) Membership Number 54510716032015
2002-2005	**Member,** Austrian Paediatric Society. Membership Number is not applicable.

Administrative Activities

INTERNATIONAL

Sep 2019	**Invited Chair**, SSIEM Annual Symposium 2019, Primary Audience: Metabolic Genetics, Paediatricians, Paediatric Neurologist Rotterdam, The Netherlands. *I co-chaired the session for Parallel Session 4D: Novel Disease Genes (*https://ssiem2019.org/preliminary-program/
Apr-Sep 2019	**Organizing Committee Member**, Inborn cerebral creatine deficiency syndromes symposium (scientific and patient meeting), September 6-7, 2019, Rotterdam, The Netherlands *I worked as part of organizing committee for the program development, invitation of speakers*
May 2019	**Society for Inborn Errors of Metabolism (SSIEM) invited abstract review committee member** for SSIEM 2019 meeting in Rotterdam, The Netherlands. Reviewed and graded 125 abstracts.

April 2019	**International Inborn Errors of Metabolism and Nutrition Congress Scientific Committee Member**, April 10-14, Istanbul, Turkey (http://www.metabolizma2019.org/index_ing.php?sayfa=kurullar
March 2019	**Organizing Committee Member,** Cerebral Creatine Deficiency Syndrome (CCDS) Workshop, April 8, 2019 (https://creatineinfo.org/simd/)
2018-Present	**Board Member,** Association for Creatine Deficiencies (ACD), Scientific Medical Advisory Board (https://creatineinfo.org/smab/)
2018-Present	**Chair,** Cerebral Creatine Deficiency Syndromes Variant Curation Expert Panel, Clinical Genome Resources, (https://clinicalgenome.org/affiliation/50047/)
2018-2019	**Scientific Committee Member,** Society for Inborn Errors of Metabolism (SSIEM) Annual Symposium, International Scientific Committee Member, September 3-6, 2019, Rotterdam, The Netherlands
May 2018	**Committee Member,** Society for Inborn Errors of Metabolism (SSIEM) invited abstract review committee member for SSIEM 2019 meeting in Athens. Reviewed and graded 20 abstracts.
Dec 2017	**Expert panel member,** Biomarin Advisory Board Member, Pediatric-Onset Seizures: Best Practices for Molecular Genetic Testing, Washington DC, US. *I attended to a two day meeting with a group of international experts for epilepsy genetics.*
2014- Present	**Editorial Board Member**, Journal of Paediatric Genetics
Apr 2015	**Invited Co-Chair**, Energy Metabolism Defects, XIII. National Metabolic and Nutrition Congress, Primary Audience: Metabolic Genetics, Paediatricians, Paediatric Neurologist, Adana, Turkey. *I co-chaired the session for an international speaker Dr. Fukao, Ketolysis and Ketogenesis Defects.*
Jun 2014	**Invited Defense Committee Member**, PhD Thesis Defense Committee for Mr. Ndika, Department of Clinical Chemistry, VU Medical Centre, Free University. Title: Clinical, molecular and functional approaches to the understanding of creatine deficiency syndromes, Primary Audience: PhDs, MDs, Laboratory Technicians, Public, Amsterdam, The Netherlands. *The committee consisted of seven MD/PhDs. I was one of the defense committee members. I reviewed the PhD thesis and asked questions of the PhD candidate.*
Mar 2014	**Invited Member of PhD Thesis Review Committee**, PhD Candidate Mr. Ndika, Metabolic Laboratory, Department of Clinical Chemistry, VU Medical Centre, Free University. Title: Clinical, molecular and functional approaches to the understanding of creatine deficiency syndromes.

I was invited to review the PhD thesis of PhD candidate Mr. Ndika due to my expertise in creatine deficiency disorders. The review was performed in Toronto, Ontario, CA after receiving the PhD thesis by mail.

Jan-Jun 2014 **Invited Member of Organizing Committee** for the Symposium Updates on Neurometabolic Disorders, Amsterdam, The Netherlands.

I planned the scientific meeting together with Dr. Salomons and worked as secretary for scientific content of the meeting

Sep 2014 **Invited Co-Judge for Oral Presentation**, 3rd National Conference on Inborn Errors of Metabolism September 19-21, 2014, Primary Audience: Paediatricians, Metabolic Physicians, Clinical Geneticist, Trainees, Hyderabad, India.

I judged ten oral presentations related to various inherited metabolic disorders in an hour session according to significance, study design, innovation, and presentation quality.

Sep 2014 **Invited Co-Chair**, 3rd National Conference on Inborn Errors of Metabolism September 19-21, 2014, Primary Audience: Paediatricians, Metabolic Physicians, Clinical Geneticist, Trainees, Hyderabad, India.

I co-chaired a session for IEMs- Current Status, Dr. I. Verma, Epidemiology of IEMs and Dr. B.T. Poll-The, Peroxisomal Disorders.

Jan-Sep 2014 **Invited Guest Editor for Special Issue of Journal of Paediatric Epilepsy.**

I generated a list of topics and invited experts for the chosen topics to write review articles. This special issue was related to inherited metabolic disorders and epilepsy. I worked closely with the Editor of the Journal and editing team to complete edits of the manuscripts. The special issue is published online (2014, Volume 3, Number 4). http://content.iospress.com/journals/journal-of-pediatric-epilepsy/3/4.

NATIONAL

2018-2019 **President, Garrod Symposium 2019** organized the meeting with co-president.

2018-2019 **Scientific Committee Chairs, Garrod Symposium 2019** organized the meeting with co-president for scientific content and administrative responsibilities.

2017-2018 **Expert working group member,** Homocystinuria Task Force. *I was a member of this committee for decision making and review of the literature, if homocystinuria should be continued as part of Newborn Screening Ontario Program and how to improve to identify positives and decrease false positives to improve outcomes of patients by early treatment. We reviewed literature prepared the document as well as attended to 3 teleconferences to discuss.*

December 2017 **Invited Advisory Board Member,** Consultant, Canada Canadian Round Table on Homocystinuria and Cystadane, Recordati Rare Diseases Canada Inc. *I chaired and presented treatment of homocystinurias*

2015-Present **Laboratory Practice Committee,** Canadian College of Medical Geneticist (CCMG), Canada.

The Laboratory Practice Committee provides a forum for discussion of issues relevant to the laboratory investigations and statements. This committee consists of cytogeneticist, molecular geneticist and biochemical geneticists certified through CCMG for their speciality. As a member of the committee, I review documents generated by the Chair through e-mail communication and attend teleconferences or annual meetings.

2014-Present **Metabolics Committee Member**, Canadian College of Medical Geneticist (CCMG), Canada.

The Metabolics Committee provides a forum for discussion of issues relevant to the education and practice of Laboratory and Clinical Biochemical Genetics. This committee consists of laboratory and clinical biochemical geneticists certified through CCMG for this speciality. As a member of the committee, I review documents generated by the Chair through e-mail communication and attend teleconferences or annual meetings.

Jun 2014 Invited Advisory Board, Consultant, Hyperion Therapeutics Inc, Montreal, Canada

I sat on the Advisory Board of Hyperion to treat hyperammonemia for my medical and scientific advice.

2013-2018 **Credentials Committee Member**, Canadian College of Medical Geneticist (CCMG), Canada.

This committee consists of cytogeneticist, biochemical geneticist, molecular geneticist MD and PhDs and certified through CCMG for their respective subspecialities. The Credentials Committee is responsible for issues that pertain to applications by individuals for admission to the College. They review all applications from individuals, making recommendations as to the suitability of each candidate's background and training, before the examination process. They also liaise with the respective CCMG specialty committees (Clinical Practice, Molecular, Cytogenetics and Biochemical) in the review and subsequent implementation of training guidelines. I review 5-10 trainees documentation to make sure all training guidelines are met and inform chair of my assessment. I attend annual meetings and 1-2 teleconferences per year.

2011- 2017 **Program Director for Biochemical Genetics Fellowship,** Canadian College of Medical Geneticist (CCMG) Accredited Biochemical Genetics Fellowship, Toronto, Ontario, Canada

This is a CCMG accredited 3-year (shortened to 2-year in 2014) fellowship program and trains Canadian and International fellows specializing in biochemical genetics. I review applicants, arrange interviews, accept to the program, and apply for non-MOH funding or departmental funding. I register fellows to CCMG for Biochemical Genetics Fellowship. I arrange their rotations according to training requirements. I arrange supervisory meetings every 6 months throughout training and chair those meetings. I develop an agenda for supervisory meetings. I complete final training assessments for each fellow. I review their ITERs, FITERs, logbooks and write reference letters and attestation letters. I provide mentorship for electives, conference attendance, research and course attendance according to CCMG training requirements.

2010-2013 **Member,** Canadian College of Medical Geneticist (CCMG) Clinical Practice Committee, Toronto, Ontario, Canada

The Clinical Practice Committee (CPC) develops and/or reviews guidelines for the practice of Clinical Genetics and Prenatal Diagnosis. The CPC liaises with the Training committee in order to maintain and develop CCMG training standards and guidelines. The committee consists of clinical geneticist, biochemical geneticist, molecular geneticist, cytogeneticist. As a member of this committee, I reviewed documents distributed by the Chair.

May 2010 **Invited National Expert Consultation Board Member**, Gaucher National Consultative Committee, Toronto, Ontario, CA.

As an expert for the management of Gaucher disease, I was a member of the committee for the enzyme replacement therapy.

PROVINCIAL/REGIONAL

2018-2019 **Task Force Chair,** Isovaleric Aciduria Newborn Screening Review. *I was the chair of this committee to review newborn screening for isovaleric aciduria to revise newborn screening criteria to decrease false positive screening for this disease. I reviewed the literature, prepared the document as group chair and attended to 4 teleconferences to discuss with the group.*

2017-2018 **Task Force Committee Member,** X-linked adrenoleukodystrophy (X-ALD) Task Force. *I was a member of this committee for decision making and review of the literature, if X-ALD should be included into Newborn Screening Ontario Program. We reviewed literature prepared the document as a group and attended to 4 teleconferences to discuss points. I made connections with The Netherlands Newborn Screening Program to discuss their decisions.*

2018 **Primary Developer and Submitter**, Application of a protocol for "riboflavin supplementation therapy for riboflavin transporter deficiency*."* Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of riboflavin supplementation therapy in patients with riboflavin transporter deficiency. Approval of this application will allow all patients with riboflavin transporter deficiency to be treated in Ontario.

2018 **Primary Developer and Submitter**, Application of a protocol for "thiamine and biotin supplementation therapies for thiamin transporter deficiency*."* Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of thiamin and biotin supplementation therapies in patients with thiamin transporter deficiency. Approval of this application will allow all patients with thiamin transporter deficiency to be treated in Ontario.

2018 **Primary Developer and Submitter**, Application of a protocol for "serine and glycine supplementation therapies for serine biosynthesis disorders*."* Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of serine and glycine supplementation therapies in patients with serine biosynthesis disorders. Approval of this application will allow all patients with serine biosynthesis disorders to be treated in Ontario.

2015 **Primary Developer and Submitter**, Application of a protocol for "arginine and glycine supplementation therapy for creatine transporter deficiency*."* Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of arginine and glycine supplementation therapy in patients with creatine transporter deficiency. This application is currently under review. Approval of this application will allow all patients with creatine transporter deficiency to be treated in Ontario. I also published a case report to show that arginine and glycine supplementation therapy treats seizures in a patient with creatine transporter deficiency (see publication list).

2015 **Primary Developer and Submitter**, Application of a protocol for "tryptophan supplementation therapy for pyridoxine dependent epilepsy*."* Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of tryptophan therapy in patients with pyridoxine dependent epilepsy. This application is currently under review (document is submitted with dossier). Approval of this application will allow all patients with pyridoxine dependent epilepsy to be treated in Ontario. I also published a case report to show that if pyridoxine dependent epilepsy patients on lysine-restricted diet are not treated with tryptophan supplementation, they develop brain serotonin deficiency (tryptophan is precursor of serotonin) (please see publications).

2015 **Primary Developer and Submitter**, Application of a protocol for "Glycosade therapy for glucose transporter 1 deficiency*."* Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of Glycosade to the patients with glucose transporter 1 deficiency as an alternative therapy to the ketogenic diet. This application is currently under review (document is submitted with dossier). Approval of this application will allow all patients with glucose transporter 1 deficiency to be treated with Glycosade in Ontario. I also published a case report to show that Glycosade therapy is helpful to the patients who are not compliant with the ketogenic diet (please see publication list).

2014 **Primary Developer and Submitter**, Application of a protocol for "L-arginine supplementation therapy for pyridoxine dependent epilepsy*." to the* Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of L-arginine therapy in patients with pyridoxine dependent epilepsy. The cost of one-month therapy was about $300-400 CDN to the families.

Through my application, L-arginine has been approved to be covered by patient's drug plan for all patients with this disease in Ontario (document is submitted with dossier)

2014 **Primary Developer and Submitter**, Application of a protocol for "Pyridoxal-5-phosphate for pyridoxal phosphate dependent epilepsy." to the Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of pyridoxal-5-phosphate therapy (only effective treatment) in patients with pyridoxal phosphate dependent epilepsy. Through my application pyridoxal-5-phosphate has been approved to be covered by drug coverage plan for all patients with this disease in Ontario (document is submitted with dossier)

2013 **Primary Developer and Submitter**, Application of a protocol for "Lysine free formula coverage for pyridoxine dependent epilepsy*"* to the Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application documentation and provided information for the benefit of lysine-restricted diet and necessity of lysine-free formula treatment in patients with pyridoxine dependent epilepsy. Through my application lysine-free formula has been approved for coverage by drug plans for all patients with this disease in Ontario (document is submitted with dossier).

2012 **Primary Developer and Submitter**, Application of a protocol for "L-ornithine in GAMT deficiency." to the Inherited Metabolic Diseases Program, Exceptional Access Program Branch, Ontario Drug Benefit Program, Health and Long-term Care, Ministry of Health, Ontario, CA.

I prepared application document and provided information for the benefit of ornithine supplementation in patients with GAMT deficiency. Through my application ornithine has been approved coverage by drug plans for all patients with this disease in Ontario (document is submitted with dossier).

LOCAL

Research Institute, The Hospital for Sick Children

2013-2015 **Board Member**, Research Ethics Board, Research Institute, The Hospital for Sick Children, Toronto, Ontario, Canada.

I am one of the physician board members with the REB. I review 1-2 research protocols as primary or secondary reviewer monthly and present to the board. Depending on the project, review of one project lasts from 3-6 hours. I attend board meetings once a month for 2 hours and present protocols reviewed or contribute to the decisions taken by board members. Between 2013-2014, I reviewed the responses to the REB and approved final protocol for research application protocols for researchers as primary reviewer.

University of Toronto

August 2019	**Poster Judge,** Institute of Medical Sciences Research Day, Toronto, Canada
August 2019	**Poster Judge,** SickKids Summer Student Symposium Research Day, Toronto, Canada
June 20, 2019	**Exam Chair,** MSc Final oral exam, Institute of Medical Sciences, Title: Ultrasound-Targeted microRNA-26a Therapy for Abdominal Aortic Aneurysms, Toronto, ON, Canada
June 5, 2018	**Exam Chair,** MSc Final oral exam, Institute of Medical Sciences, Title: The impact of a childhood cancer diagnosis on their mother's mental healthcare use, Toronto, ON, Canada
February 6, 2018	**Poster Judge,** Medical Student Research Day, Toronto, Canada *I acted as poster judge with two other judges. We evaluated 6 posters after oral presentations according to set scoring and ranking criteria.*
June 16, 2017	**Exam Chair,** MSc Final oral exam, Institute of Medical Sciences, Title: Defects in sialylation cause glomerular injury, Toronto, ON, Canada
May 10, 2017	**Exam Chair,** MSc Final oral exam, Institute of Medical Sciences, Title: Characterizing the impact of penile-vaginal sex on the microbiome and immune parameters of HIV susceptibility in the female and male genital tracts, Toronto, ON, Canada
March 17, 2017	**Oral presentation and Poster Judge,** Division of Clinical and Metabolic Genetics Annual Research Day, March 17, 2017, Toronto, ON, Canada *I acted as oral presentation judge with two other judges. We evaluated 9 oral presentations according to set scoring and ranking criteria and chose the best oral research presentation.* *I acted as poster judge with two other judges. We evaluated 4 posters after oral presentations according to set scoring and ranking criteria.*
February 8, 2017	**Poster Judge,** Medical Student Research Day, Toronto, Canada *I acted as poster judge with two other judges. We evaluated 6 posters after oral presentations according to set scoring and ranking criteria.*
May 20, 2016	**Oral presentation judge**, The Institute of Medical Science Graduate School Scientific Day 2016, Laidlaw Manuscript Judge. *During the "Laidlaw Manuscript Competition", I act as oral presentation judge with three other judges. We evaluated 8 oral presentations according to set scoring and ranking criteria and chose the best oral research presentation.*
May 20, 2016	**Poster Judge**, The Institute of Medical Science Graduate School Scientific Day 2016, Alan Wu Poster Competition Judge. *During the Scientific Day, I act as poster judge with another judge. We evaluated 8 posters after oral presentations according to set scoring and ranking criteria.*
Aug 2015	**Poster Judge**, Undergraduate Research Day, Institute of Medical Sciences, Summer Undergraduate Research Day, University of Toronto, Ontario, Canada. *This is a yearly Research day. During the research day, I act as poster judge with other judges. We evaluated 8 posters after oral presentations according to set scoring and ranking criteria.*

2015-Present

Coordinator, Curriculum Development for Metabolic Genetics Teaching for Paediatric Neurology Residency Program according to Royal College Physicians and Surgeons Training requirements, Department of Paediatrics, University of Toronto, Ontario, Canada.

I generated a 2-month teaching program related to the metabolic genetics topics required by Royal College for Paediatric Neurology Residency. Teaching rounds are held every Wednesday morning between 8:00-9:00 AM. First one held April-June 2015. I arranged speakers according to the topics and liaised between Paediatric Neurology Training Admin Assistant, Speaker, Program Director of Teaching Rounds and Chief Resident. This program will be repeated every 2 years. This program will be repeated in 2016-2017 Academic Year.

2014-Present

Coordinator, Curriculum Development for Epidemiology and Critical Appraisal, Medical Genetics Residency Program Academic Half-Day Curriculum Committee, University of Toronto, Department of Paediatrics, Toronto, Ontario, Canada.

I prepare program and invite experts for the Academic Half Day teaching.

2013-Present

Member, Educational Committee, Medical Genetics Residency Program Academic Half-Day, Department of Paediatrics, University of Toronto, Toronto, Ontario, Canada.

This committee meets every other year to plan and update teaching curriculum for the Medical Genetics Residency Program. I am one of the members and coordinator for metabolic genetics and epidemiology and critical appraisal topics.

May 2013-Present

Poster Judge, Paediatrics Postgraduate Research Day, Department of Paediatrics Research Day, University of Toronto, Toronto, Ontario, Canada.

This is a yearly Research day. During the research day, I act as poster judge with my two co-judges. We evaluate 6 posters after oral presentations according to set scoring and ranking criteria.

2013-2016

Member, Clinical and Metabolic Genetics Observership Committee, University of Toronto, Department of Paediatrics, Toronto, Ontario, Canada.

I review observership applicants and meet every 6 months with committee members to discuss and make decision on who is accepted for observership.

2013-2016

Member, Clinical and Metabolic Genetics IMG Fellowship Selection Committee, University of Toronto, Department of Paediatrics, Toronto, Ontario, Canada.

I review IMG clinical and metabolic genetics fellowship applications. I meet with the committee to decide what funding route to take, the number of applicants to be accepted into the program, what program is suitable. The meetings are held 1-2/year according to the number of applicants.

2012-Present

Coordinator, Curriculum Development for Metabolic Genetics for Medical Genetics Residency Program Academic Half-Day, Department of Paediatrics, University of Toronto, Toronto, Ontario, Canada

I generated a 1-month teaching program for 3 years related to the metabolic genetics topics (12 topics in total) for Medical Genetics Training Program. Teaching rounds are held each year in January for 4 weeks 1:30-3:00 PM

during Academic Half Day. I arrange speakers according to the topics and liaise between Training Admin Assistant and Speaker.

2011-2017 **Program Director**, Clinical Metabolic Genetics Fellowship Program, Department of Paediatrics, University of Toronto, Toronto, Ontario, Canada.

This is a clinical fellowship consisting of 3-5 fellows/year. I developed program objectives for metabolic genetics fellowship. This is a 2-year program to train foreign specialists such as IMGs from various countries, i.e. Chile, Switzerland, United Emirate, Palestine, Saudi Arabia who specialize in paediatrics, paediatric neurology, endocrinology, internal medicine to become metabolic geneticists in their home country. As program director, I review fellowship applications and arrange skype or in-person interviews for candidates. I organized an interview committee consisting of three metabolic staff physicians including myself. I apply standardized interview processes, rank applicants accordingly and accept them into the program.
I liaise between CPSO, UFT, and SickKids during the hiring process. I apply to non-MOH and departmental funding for IMG fellowship applicants with no funding from their home country. I arrange academic year rotation plan for metabolic fellows. I mentor them for their elective rotations as well as courses. I arrange meetings to discuss their training and their needs during their training 1-2/year.

2012-2017 **Supervisor**, Teaching Curriculum Development for Metabolic/Biochemical Genetics Fellowship Program, Department of Paediatrics, University of Toronto, Toronto, Ontario, Canada.

This is a teaching round for metabolic/biochemical genetics fellows which occurs every Tuesday 4:00-5:00 pm. One of the fellows is responsible for arranging the program. I supervise the curriculum and help with the topics and topic presenters. I meet 1-2 times per year and review program through e-mails.

2011-2013 **Member**, Medical Genetics Residency Training Program CaRMs Selection and Interview Committee, Department of Paediatrics, University of Toronto, Toronto, Ontario, Canada.

I reviewed pre-selected candidates to choose 5 international and all Canadian candidates for an interview according to the criteria. Additionally, I was one of the interview committee members (consists of 4 members) interviewing all candidates according to criteria; occurring every Thursday for 4 weeks (mid Jan- mid Feb) for 2 years.

2011-2013 **Member, Metabolic Representative**, Medical Genetics Residency Training Committee, Department of Paediatrics University of Toronto, Toronto, Ontario, Canada.

This committee meets every other month to discuss Medical Genetics Residency training and related issues raised by trainees, assess trainees and graduate trainees. The committee discusses issues raised from resident retreat. I liaised between training committee and metabolic unit to find solutions to the problems related to the metabolic unit. As a member of this committee I was also involved in the accreditation of the program by Royal College as well as Royal College site visit for the accreditation of the program during my membership.

The Hospital for Sick Children

2018-Present **Committee Member,** Physician Representative for Project Horizon, The Hospital for Sick Children, Toronto, Canada.

2015-Present **Committee Member,** Credentials Committee, The Hospital for Sick Children, Toronto, Ontario, Canada.

I am one of the physician members as representative of the Department of Paediatrics. As a member of the Credentials Committee, I am responsible for reviewing the credentials and references of a subset of newly appointed medical surgical and scientific staff at The Hospital for Sick Children. The decisions of this committee are reported directly to the hospital's Medical Advisory Committee.

2015-Present **Committee Member,** Data Safety Monitoring Board for Dr. Feldman's Creatine Trial, The Hospital for Sick Children, Toronto, Ontario, Canada.

This is a study entitled: The effect of creatine supplementation on muscle function in childhood myositis. I review study documents, study assessment results and DSMB documents as a committee member and attend the committee meetings either in person or via teleconference every 4 months.

2013-2015 **Primary Developer**, Standardized needle muscle biopsy for metabolic investigations and neuromuscular disorders, The Hospital for Sick Children, Toronto, Ontario, Canada.

Diagnostic muscle biopsies were performed by general surgeons as an open muscle biopsy during surgical procedure until 2013. With my leadership, and in collaboration with Image Guide Therapy, Department of Radiology, needle muscle biopsy became standard procedure for diagnostic work-up. The price analysis showed that this is superior to open surgical muscle biopsy. It leaves a small scar compared to the open surgical muscle biopsy. The quality and quantity of sample size is appropriate to perform all necessary investigations. I also provided information as well as details of the process to Paediatric Neurology. Both divisions (Clinical and Metabolic Genetics and Neurology) have been using this procedure widely since the new technique of muscle biopsy was set up. We arranged various meetings including Departments of Pathology, Laboratory Medicine, Radiology to set-up this procedure. Policy-Procedure for The Hospital for Sick Children were prepared and under review in Policy Procedure Department, The Hospital for Sick Children. This protocol is a collaborative work between Vivian Cruz (Metabolic Nurse Practitioner) and myself. (Policy/Procedure is enclosed in the dossier).

2012-2013 **Primary Developer** (Policy/Procedure Development), Standardized CSF collection for Metabolic Investigations, The Hospital For Sick Children, Toronto, Ontario, Canada.

As neurometabolic physician, I use lumbar puncture as a diagnostic for neurodegenerative disorders. CSF collection methods as well as quality of samples are essential for correct diagnosis. For this reason, I set-up standardized lumbar puncture procedure in Image Guided Therapy, Radiology. We generated Policy/Procedure for the method. Paediatric Neurology and Clinical and Metabolic Genetics have been using this standardized LP procedure widely. This protocol is a collaborative work between Vivian Cruz (Metabolic Nurse Practitioner) and myself. (Policy/Procedure is enclosed in the dossier). (http://policies.sickkids.ca/published/Published/CLINS102/Main%20Document.pdf#search=CSF%20metabolic)

Division of Clinical and Metabolic Genetics

2019	**Developer,** Epic Order Set Development for CSF Neurometabolic Investigations
2019	**Committee Member,** Triage Working Group, Division of Clinical and Metabolic Genetics
2014-2016	**Committee Member,** Metabolic Genetics Staff Physician Search and Selection Committee, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada. *I review CVs of all applicants, attend the selection meeting for decision making of candidates to be invited for an interview and met candidate during interview process.*
2013	**Committee Member**, KidCare Metabolic Order Sets Committee, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada. *I prepared order sets for metabolic investigations for seizures, developmental delay, pyridoxine dependent epilepsy, creatine transporter deficiency, hypoglycaemia, dysmorphic features.*
2012	**Committee Member**, Metabolic Genetics Registered Nurse Search and Interview Committee, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada. *I reviewed CVs of all applicants, attended the selection meeting for decision making of candidates to be invited for an interview. I was one of the interview committee members during interview process.*
2012	**Committee Member**, Nurse Practitioner Search and Interview Committee, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada. *I reviewed CVs of all applicants, attended the selection meeting for decision making of candidates to be invited for an interview. I was one of the interview committee members during interview process.*
Oct 2011- Jan 2012	**Primary Developer,** Metabolic Nurse Practitioner Job Profile Development, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada. *I communicated with Metabolic Nurse Practitioners in US and Canada Metabolic Centers and reviewed their job profile. According to their job profile and the needs of our metabolic unit at The Hospital for Sick Children, I developed a job profile for a metabolic Nurse Practitioner. (This protocol is enclosed to the dossier).*
Sep 2012- Apr 2013	**Primary Developer**, Exercise Test Protocol for Metabolic Profiling of Rhabdomyolysis/Myopathy, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada. *I met with exercise physiologist at The Hospital for Sick Children and communicated with Dr. Tarnopolsky at McMaster University and developed an exercise test to apply to the patients with exercise induced muscle pain or rhabdomyolysis for metabolic profiling. This protocol is a collaborative work between Vivian Cruz (Metabolic Nurse Practitioner) and myself. This protocol is used by metabolic team members, metabolic fellows and staff*

physicians at The Hospital for Sick Children. (This protocol is enclosed to the dossier).

2011-Present **Triage Metabolic Staff Physician**, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada.

I review new referrals (5-10/week) and their investigations and make decisions on who will be seen and when patients are to be booked according to their chief complaint and urgency. This is performed on a weekly basis lasting 30-90 minutes/week.

2011-2013 **Committee Member**, Adult Metabolic Clinic Transition Committee, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada.

This committee made decisions for transition of metabolic patients to the Adult Metabolic Clinic and reviewed processes for a smooth transition according to patient needs and their continuity care.

2011-Present **Committee Member**, Newborn Screening Algorithm Committee, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada.

This committee meets 1-2/month for one hour to review and update algorithms for metabolic disorders screened in the Ontario Newborn Screening Program. We review literature and positive and negative controls and update cut offs in our center.

2011-2012 **Primary Developer**, composition of Metabolic Genetics Clinic information pamphlet and family history questionnaire, The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, Canada.

This is prepared in collaboration with genetic counsellors to give new patients information about the metabolic genetics clinic. A detailed history questionnaire is also prepared and completed prior to clinic in order for the most comprehensive information to be acquired during first clinic visit. This is enclosed.

Division of Biochemical Disease, British Columbia Children's Hospital

Nov 2009- May 2011 **Triage Metabolic Staff Physician**, British Columbia Children's Hospital, Division of Biochemical Diseases, Department of Paediatrics, Vancouver, British Columbia, Canada.

I reviewed new referrals (3-6/week) and their investigations and made decisions as to who will be seen and when patients are to be booked according to their chief complaint and urgency. This was performed on a weekly basis lasting about 30 minutes/week.

2009-2010 **Primary Developer**, Standardized order set development for hypoglycaemia diagnostic work-up for endocrinology and biochemical genetics, British Columbia Children's Hospital, Division of Biochemical Diseases, Department of Paediatrics, Vancouver, British Columbia, Canada.

I worked together with one of the endocrinology staff physicians and developed a single prolonged fasting challenge standing order to be used by endocrinology and biochemical genetics patients presenting with hypoglycaemia and undergoing prolonged fasting challenge test. This is approved through both divisions and hospital (enclosed).

2006-2010	**Primary Developer**, various inherited metabolic disorder specific follow-up protocol development, British Columbia Children's Hospital, Division of Biochemical Diseases, Department of Paediatrics, Vancouver, British Columbia, Canada.
	I developed follow-up protocols for various inherited metabolic disorders including CDG syndrome, X-linked adrenoleukodystrophy, creatine transporter deficiency, Fabry disease, Gaucher disease, glycogen storage disease type types Ia, Ib, III, IV, IX, mucopolysaccharidosis I, II, pyridoxine dependent epilepsy, tyrosinemia, very long chain fatty acid dehydrogenase deficiency (an example is enclosed).

Other

2014-2015	**Mentor**, WoodGreen Community Centre, International Medical Graduates (IMG), Toronto, Ontario, CA.
	I provide one-on-one mentorship for international medical graduates. This is a 10-week mentorship program for each mentor and we meet for an hour weekly.

Peer Review Activities

EXTERNAL REVIEWER

May 2017	Glycogen storage disease type VI and IX Diagnosis and Management Guidelines, American College of Medical Genetics and Genomics Practice Guideline

GRANT REVIEWS

Invited International Grant Reviews

2019	European Joint Program on Rare Diseases, Number of Reviews: 1
2019	National Research Agency, Number of Reviews: 1
2017	Action Medical Research For Children Grant, Number of Reviews: 1
2015	Mariani Foundation Grant, Number of Reviews: 1
2015- Present	Italian Telethon Foundation, Number of Reviews: 2
2015-Present	Swiss National Science Foundation, Number of Reviews: 2
2015	German Federal Ministry for Education and Research (BMBF), and the E-Rare 3 group of national research funding agencies, Number of Reviews: 1

Local Internal Grant Scientific Reviews prior to submission

Apr 2013	Carter M, Stravropoulos S, Number of Reviews: 1

ADJUDICATION

University of Toronto

April 2018	Institute of Medical Sciences scientific day Laidlaw Manuscript adjudication

March 2018 Review	CREMS 2018 Summer Research Scholarship Program Applicant Review
January 2018	Canada Graduate Scholarship Master's Program Applications
May 2017 Review	Ontario Graduate Scholarship Domestic Student Competition Applicant
April 2017	CREMS 2017 Summer Research Scholarship Program Applicant Review
Feb 2017 Review	Ontario Graduate Scholarship International Student Competition Applicant
May 2016 Review	Ontario Graduate Scholarship Domestic Student Competition Applicant
March 2016	CREMS 2016 Summer Research Scholarship Program Applicant Review

MANUSCRIPT REVIEWS

2019-Present	BMC Neurology, Number of Reviews: 2
2019-Present	ACS Chemical Neuroscience, Number of Reviews: 1
2019-Present	European Journal of Medical Genetics, Number of Reviews: 2
2019-Present	Expert Review Neurotherapeutics, Number of Reviews: 1
2019-Present	Nanomedicine, Number of Reviews: 2
2018-Present	Journal of Inherited Metabolic Disorders Reviews, Number of Reviews: 3
2018-Present	Scientific Reviews, Number of Reviews: 1
2018-Present	International Journal of Analytical Chemistry, Number of Reviews: 2
2018-Present	BMC Medical Genetics, Number of Reviews: 1
2018-Present	Journal de Pediatria, Number of Reviews: 1
2017-Present	GeneReviews Book, Number of Reviews: 1
2017-Present	BMC Medical Genomics, Number of Reviews: 1
2017-Present	Italian Journal of Pediatrics, Number of Reviews: 1
2017-Present	Seizure European Journal of Pediatric Neurology, Number of Reviews: 1
2017-Present	American Journal of Medical Genetics A, Number of Reviews: 2
2017-Present	Clinical Genetics, Number of Reviews: 1
2017-Present	JSM Neurosurgery and Spine, Number of Reviews: 1
2016-Present	Genetics in Medicine, Number of Reviews: 3
2016-Present	Expert Review of Endocrinology and Metabolism, Number of Reviews:1

2016-Present	Neurochemistry International, Number of Reviews:1
2016-Present	Epilepsia, Number of Reviews:6
2016-Present	Journal of Pediatric Research, Number of Reviews:4
2016-Present	Molecular & Cellular Probes, Number of Reviews: 2
2016-Present	Pediatrics, Number of Reviews: 3
2016-Present	European Journal of Neurology, Number of Reviews: 2
2015-Present	Neurology, Number of Reviews: 1
2015-Present	Biochimie, Number of Reviews: 2
2015-Present	Epilepsy Research, Number of Reviews: 2
2015-Present	Prenatal Diagnosis, Number of Reviews: 1
2015-Present	Human Genome Variation, Number of Reviews: 1
2015-Present	Journal of Developmental & Behavioral Pediatrics, Number of Reviews: 1
2015-Present	Neurobiology of Disease, Number of Reviews: 2
2015-Present	Journal of Medical Genetics, Number of Reviews: 1
2015-Present	Developmental Medicine & Child Neurology, Number of Reviews: 2
2015-Present	World Journal of Pediatrics, Number of Reviews: 1
2014-Present	Movement Disorder, Number of Reviews: 1
2014-Present	New England Journal of Medicine, Number of Reviews: 2
2014-Present	Metabolic Brain Disease, Number of Reviews: 5
2014-Present	Nutrition Research, Number of Reviews: 2
2014-Present	Canadian Journal of Neurological Sciences, Number of Reviews: 8
2014-Present	Orphanet Journal of Rare Diseases, Number of Reviews: 3
2014-Present	BioMed Research International, Number of Reviews: 1
2014-Present	International Journal of Emergency Mental Health, Number of Reviews: 1
2013-Present	Journal of Human Genetics, Number of Reviews: 1
2013-Present	Journal of Epilepsy, Number of Reviews: 1
2013-Present	PLOS ONE, Number of Reviews: 3
2013-Present	Molecular Genetics and Metabolism, Number of Reviews: 5
2012-Present	Gene, Number of Reviews: 2
2011-Present	Journal of Inherited Metabolic Disorders, Number of Reviews: 24

2011-Present	European Journal of Paediatrics, Number of Reviews: 1
2010-Present	Brain, Number of Reviews: 3
2008-Present	Human Mutation, Number of Reviews: 2

PRESENTATION REVIEWS

I review all power point presentations and poster presentations of trainees and research students for international, national and local meetings.

Scientific Reviewer for Research Ethics Board Applications:

November 2018	Inbar-Feigenberg, Number of Reviews: 1
August 2018	Schulze A, Number of Reviews: 1
July 2018	Sondheimer N, Number of Reviews: 1
Feb 2018	Mendoza R, Number of Reviews: 1
Jan 2018	Schulze A, Number of Reviews: 1
Aug 2017	Kannu P, Number of Reviews: 1
Mar 2015	Gassas A, Number of Reviews: 1
May 2012	Schulze A, Number of Reviews: 1
Feb 2012	Carter M, Vincent J, Number of Reviews: 1
Feb 2012	Minassian B, Number of Reviews: 1

Other Research and Professional Activities

2017-2018	**Principal Investigator**, Retrospective review of all patients seen in the Metabolic Genetics Clinic with the diagnosis of inherited neurotransmitter disorders, Toronto, Ontario, Canada. *I am PI for this retrospective study to review diagnostic yield of genetic investigations in patients with neuronal ceroid lipofuscinosis.*
2017-2018	**Principal Investigator**, Neuronal Ceroid Lipofuscinosis: diagnostic yield of molecular genetic testing and phenotype, genotype, natural history of the patients diagnosed with neuronal ceroid lipofuscinosis, Toronto, Ontario, Canada. *I am PI for this retrospective study to review diagnostic yield of genetic investigations in patients with neuronal ceroid lipofuscinosis.*
2016-2017	**Principal Investigator**, Retrospective review of all patients seen in the Metabolic Genetics Clinic for diagnostic work-up of movement disorders, Toronto, Ontario, Canada. *I am PI for this retrospective study to review diagnostic yield of genetic investigations in patients with movement disorders.*

2016-2017 **Principal Investigator,** Disorders of intracellular cobalamin metabolism: phenotype, genotype and long-term treatment outcome, Toronto, Ontario, Canada. Collaborators: Lizbeth Mellin, Garrett Bullivant, Vivian Cruz.

I am PI for this retrospective collaborative study to review outcome of patients with cobalamin disorders.

2016-Present **Principal Investigator** SickKids. International Network on Neurotransmitter Disorders (iNTD) Registry. International Collaborative Study, Investigators: Dr. Opladen, Dr. Kurian, Dr. Garcia-Cazorla

I am PI for SickKids participants.

2016-2017 **Principal Investigator** SickKids. Exploring the natural history of Alexander disease. International collaborative study, Investigator Dr. Waldman USA

I am PI to register SickKids participants.

2016-2017 **Principal Investigator,** SickKids. USP9X and Neurodevelopmental Disorders. International Collaborative Study, Investigator Dr. Jolly Australia.

I am PI to register SickKids participants.

2016-2017 **Principal Investigator,** SickKids. Vanishing white matter: A study of the phenotypic variation and the relationship between genotype and phenotype. International Collaborative Study, Investigator Dr. M. van der Knaap, The Netherlands.

I am PI to register SickKids participants.

2016-2017 **Principal Investigator,** SickKids. Evaluation of Sequence Variants: Epilepsy and developmental delay caused by likely pathogenic variant in PPP3CA. International Collaborative Study, Investigator Dr. Weimin BI, USA.

I am PI to register SickKids participants.

2016-2017 **Principal Investigator,** SickKids. Genetic study of musculoskeletal and neurological rare diseases. National Collaborative Study, Investigator Dr. P. Campeau, Sainte Justine, Montreal, Canada

I am PI to register SickKids participants.

2015-2016 **Principal Investigator.** Retrospective review of patients on the ketogenic diet seen between July 2012 and December 2014, Toronto, Ontario, Canada. Collaborators: Donner, E, Hewson S, Zak M.

I am PI for this retrospective collaborative study to review genetic diagnosis in patients on the ketogenic diet. This study is a part of the prospective study investigating prevalence of GLUT1 deficiency in patients on the ketogenic diet.

2015-2016 **Principal Investigator**. Retrospective review of patients with congenital disorders of glycosylation diagnosed at The Hospital for Sick Children, Toronto, Ontario, Canada. Collaborators: Al Teneiji, Cordeiro D.

This study will review all patients diagnosed with congenital disorders of glycosylation (CDG) type I at The Hospital for Sick Children for disease manifestations and long-term outcome. CDG type I is a rare disorder and the

knowledge for some subgroups is very sparse. This review study will guide physicians for the diagnostic work-up.

2015-2016 **Principal Investigator**. Retrospective review of patients with pyridoxine dependent epilepsy diagnosed at The Hospital for Sick Children. Toronto, Ontario, Canada. Collaborators: Al Teneiji, Amal / Cordeiro, Dawn.

Pyridoxine dependent epilepsy is a rare disorder and our center has the majority of patients diagnosed in Canada (>10 patients followed in my clinic). This study will shed light on the disease severity from biochemical and clinical phenotype, which has not been reported previously. I hypothesize that high neurotoxic metabolite accumulation is related to severe phenotype, which I will be able to prove with this study. I will also publish long-term treatment outcome of patients with pyridoxine dependent epilepsy.

2015-2016 **Principal Investigator**. Retrospective review of all patients diagnosed with pyruvate dehydrogenase complex deficiency at The Hospital for Sick Children. Toronto, Ontario, Canada. Collaborators: Inbar-Feigenberg, M, Hewson, S, Herd, S.

I diagnosed a patient with pyruvate dehydrogenase (PDH) complex deficiency based on the low enzyme activity level on skin fibroblasts who had normal lactate levels. This disorder is known to cause severe lactic acidosis and patients are investigated only if they have lactic academia/acidosis. Reporting on patients diagnosed at The Hospital for Sick Children, including my patient, will guide physicians to investigate this disorder in patients with normal lactate levels as well. I also review all PDH complex enzyme activity measurements performed at SickKids for this review article.

2014-2015 **Principal Investigator**. Retrospective registry of patients with creatine transporter deficiency using Research Electronic Data Capture (REDCap) software for evaluation of long-term treatment outcome of patients. The Hospital for Sick Children, Toronto, Ontario, Canada. Collaborators: Salomons G and van de Kamp J.

I developed a RedCAP questionnaire for this study and invited more than 30 physicians to contribute. This study will be the major study to report treatment outcome of this rare disease as well as give treatment recommendations.

2012-2015 **Principal Investigator**. Retrospective study of patients with X-linked adrenoleukodystrophy at The Hospital for Sick Children. Toronto, Ontario, Canada. Collaborators: Tran, C, Blaser, S, Raiman, J.

This study reviewed 45 patients diagnosed and followed at The Hospital for Sick Children. We reviewed outcome of these patients.

2012-2015 **Principal Investigator**. Intractable epilepsy: retrospective review of all patients with intractable epilepsy at The Hospital for Sick Children to generate a database. Toronto, Ontario, Canada. Collaborators: Minassian B, Donner E, Zak M, Stavropoulos J.

In this study, we developed a database including 1250 patients with epilepsy seen in the epilepsy clinics at SickKids. The diagnoses for all patients were recorded. The study results will shed light on the diagnostic yield of genetic studies.

2012-2015 **Principal Investigator**. Neonatal epileptic encephalopathy: identification of underlying metabolic epilepsies using next generation sequencing. Toronto, Ontario, Canada. Collaborators: Wilson D.

As a hypothesis, I believe that newborns with neonatal brain injury have an underlying genetic disease which contributes to their brain injury. In this study I enrolled 11 newborns with encephalopathy and found a potential genetic diagnosis in >60% of the babies. We are in the process of publishing our results to be able to request further funding for this interesting study.

2012-2015 **Principal Investigator.** Expanding the phenotypic spectrum of GLUT1 deficiency to the patients with intractable epilepsy on ketogenic diet. Toronto, Ontario, Canada. Collaborators: Donner E, Hewson S, Zak M, Salomons G.

This study will look at prevalence of GLUT1 deficiency in patients on the ketogenic diet. GLUT1 deficiency is one of the rare disorders responding to the ketogenic diet therapy. The study results shed light on whether patients with good response to the ketogenic diet should be screened for GLUT1 deficiency by genetic testing. The study enrolled 70 patients and all genetic tests are completed. We are analyzing the data and preparing the manuscript.

C. Academic Profile

1. RESEARCH STATEMENTS

Neurometabolic research program

1) Clinical neurometabolic research program:

a. Epilepsy Genetics Research Program

i. I generated an epilepsy database enrolling 1298 patients seen between 2012-2013 in epilepsy clinics. The genetic diagnosis yield was 14% in this retrospective study. This study was funded through various small grants in total of $11,800. The study results were reported as poster and oral platform presentations by research students and trainees in various international and national meetings (please see CV under abstracts and presentations). This study results will be submitted in early 2016.

ii. I am the principal investigator for the epilepsy genetics study to discover genes underlying epilepsy and intellectual disability at The Hospital for Sick Children. So far about 100 patients are enrolled for whole exome or genome sequencing. I received McLaughlin Centre, Accelerator Grants in Genomic Medicine ($75,000) in 2013. I collaborate with Dr. Minassian, one of the Co-Leader of Canadian Epilepsy Network “Personalized medicine in the treatment of epilepsy” Granted $5.0M by Genome Canada, to Dr. Cosette, Dr. Michaud and Dr. Minassian. I am the SickKids lead and enrolled 65 patients from SickKids for this national study. We are expecting to receive results from Genome Quebec for data analysis. I also collaborate with Genome Clinic for usage of epilepsy genetic panels vs whole genome sequencing (WGS). We have been analyzing data together with Genome Clinic researchers. We have confirmed a genetic diagnosis in at least 7 patients.

iii. I initiated a prospective cohort study to look at the prevalence of GLUT1 deficiency, one of the neurometabolic disorders responding to the ketogenic diet therapy, in the ketogenic diet clinic in collaboration with Dr. Donner. The sequencing of the SLC2A1 gene is performed by Dr. Salomons (former PhD supervisor) in collaboration. The goal of the study was to investigate, if this treatable neurometabolic disorder was under diagnosed. The prevalence of GLUT1 deficiency is 2% in this small cohort. The study is finalized and will be submitted at the end of 2015. This study was the foundation of future study to investigate patients on the ketogenic diet using whole exome sequencing to identify underlying causes as well as using metabolomics, which I am applying for funding.

iv. I am the principal investigator for a pilot study “Neonatal epileptic encephalopathy: identification of underlying genetic causes using next generation sequencing”. My hypothesis is that term newborns with NE have genetic defects that contribute to neonatal brain injury. To prove my hypothesis, I enrolled 10 patients and finished data analysis and validation of study results. The

genetic diagnostic yield was 60% in this pilot study. We diagnosed 4 patients with known genetic disorders and found 2 novel genes (AACS and NRCA). Using molecular and biochemical studies, we will prove that both genetic defects are the causative. This study was funded through Starbucks Summer Student Research Program. ($4,800). We reported our preliminary results at the SickKids Summer Research Program in August 2015, Theodora Bruun, my research student funded through Starbuck awarded for the Outstanding Poster Presentation. We will publish study results and submit grants to expand this study in 2016. This study was also invited for full proposal in 2014 March of Dimes foundations Basil O'Connor Starter Scholar Research Award.

b. Treatable neurometabolic disorders research program: International Collaborations:

[1] I completed an international treatment registry study as principal investigator enrolling 22 patients with GAMT deficiency to report their treatment outcome and update treatment recommendation. This manuscript will be submitted in 2 months. This study is funded through Rare Disease Foundation Microgrant Program, Vancouver ($3,500).

[2] I started an international treatment registry study as principal investigator to report treatment outcome of patients with CRTR deficiency.

[3] I am principal and co-senior author for an international genotype phenotype study as well as functional characterization of missense variants in the GAMT gene published in Human Mutation in 2014.

[4] I am collaborator for the international study for the functional analysis of variants in the SLC6A8 gene in CRTR deficiency published in Molecular Genetics and Metabolism in 2012.

2) Laboratory neurometabolic disorders research program:

a. Carrier frequency of creatine deficiency disorders (GAMT, AGAT and CRTR deficiencies) in the general population: I developed expression systems for GAMT and CRTR deficiencies. Estimated carrier frequency of GAMT deficiency was 1 in 812 individuals (0.123%) in the general population. This study is published in Molecular Genetics and Genomics in 2015. Estimated carrier frequency of CRTR deficiency in females was 1 in 4060 females. This study is published in Gene in 2015. We developed expression system for AGAT (arginine glycine amidinotransferase (AGAT) deficiency for the first time and published our study results in Human Mutation in 2016. These studies are the foundation of newborn screening for treatable neurometabolic creatine deficiency disorders.

b. Drug screening for PDE: After following patients with PDE and applying innovative therapies, I realized that these treatments are very cumbersome for families and there is no 100% cure despite very strict and cumbersome treatments. I decided to develop PDE (caused by mutations in the ALDH7A1 gene) zebrafish knock-out and knock-down model for drug screening to discover new treatment modalities for PDE. We have ALDH7A1 zebrafish model. We have been developing an enzyme assay for PDE for robust drug screening. If the accumulating neurotoxic metabolite normalizes enzyme activity, we will try those compounds on ALDH7A1 zebrafish disease model that we developed in Research Institute at SickKids.

2. TEACHING PHILOSOPHY

As an Academic Clinician who works in a rapidly changing subspecialty, I have had the opportunity to hone my teaching style and teach many different levels of learners. I recognize the importance of a metabolic genetic being taught in any specialty (and subspecialty) and have been a driving force for the integration of and development of curriculum that includes genetics in the Neurology Department. I recognize that not everyone is interested in learning about metabolic genetics in the same manner so I have adopted a case-based teaching style for differential diagnosis for inherited metabolic disorders during ward consults and outpatient clinics. I try to engage the learner and guide them during my teaching, rather than just lecturing to them.

My teaching emphasizes the partnership between the clinician and the patient and their family in the decision making process of genetic testing. I encourage students to reflect on aspects of their clinical interactions with patients and families in order for them to recognize family dynamics, social

issues and religious beliefs that often influence decisions. I help trainees to look for non-verbal as well as verbal cues as they develop their clinical acumen. I believe that in order to gain trust and respect from patients and families, which are pillars of any clinician-patient relationship, they must respect the patients and their beliefs.

I have the pleasure of teaching not only undergraduate students, but a vast array of postgraduate trainees and fellows who have an interest in metabolic genetics. I have developed a number of teaching aids and I spend many hours one-on-one teaching. As Research is also a big part of our Residency Program, I take great pride in supervising the trainees and fellows with their research projects and case reports. I provide them countless hours of guidance on ways to enhance their project and am able to watch proudly as I see them present their research at various conferences.

I am currently looking at expanding my knowledge in the diagnosis and management of paediatric movement disorders and have secured a one-month observership with Dr. J. MINK a pediatric neurologist and movement disorder specialist at University of Rochester Medical Center. I believe in always learning new things and keeping the mind active. Upon my return from my observership, I will be able to impart my knowledge on our trainees and fellows at SickKids and help our knowledge base expand.

3. CREATIVE PROFESSIONAL ACTIVITIES STATEMENT

This dossier has been developed to support my promotion to Associate Professor at the Department of Paediatrics, University of Toronto on the basis of my creative professional activity with emphasis on professional innovation. As an Academic Clinician, I developed a Neurometabolic Disorders Program in the Division of Clinical and Metabolic Genetics, Department of Paediatrics, University of Toronto, The Hospital for Sick Children.

I was appointed as an Academic Clinician, Staff Metabolic Geneticist, Neurometabolic Physician and as an Assistant Professor at the University of Toronto in 2011. My job profile was that of an Academic Clinician with 60% of my time devoted to clinical care, specifically neurometabolic disorders due to my subspecialty fellowship trainings in metabolic (biochemical) genetics and paediatric neurology as well as experience in neurometabolic disorders. As I was the only Canadian College of Medical Geneticist (CCMG) certified biochemical geneticist, I was given responsibility and job of being program director for biochemical genetics fellowship program. For this reason 10% of my time was devoted to education. I had 5% of my time for administrative activities and 25% of my time for my research activities.

Neurometabolic disorders are an important group of disorders presenting in the neonatal, infantile or early childhood periods. Neurological manifestations include seizures, epilepsy, movement disorder, severe hypotonia and regression in neurodevelopmental milestones. Seizures are often refractory to antiepileptic drugs. Some neurometabolic disorders result from genetically determined abnormalities of enzymes or transport proteins affecting the development or functioning of the central nervous system. Some of these disorders can be treated with protein-restricted diet or with vitamins and cofactors to prevent disease progression and treat seizures and movement disorder. Therefore, evaluation and diagnosis of patients with neurometabolic disorders to identify underlying genetic causes is essential. It is also crucial to give patients a chance of treatment. However, the confirmation of the diagnosis requires extensive and esoteric, sometimes invasive investigations as well as multiple genetic tests to arrive at a diagnosis. With the developments in next generation targeted sequencing and whole exome sequencing, the confirmation of a genetic diagnosis has been tremendously improved in the recent years.

Due to my training and specialization in neurometabolic disorders, after my arrival I met with pediatric neurology colleagues and epileptologist at The Hospital for Sick Children to discuss the needs for diagnostic work-up in patients with neurometabolic disorders and complex epilepsy and complex movement disorders with developmental delays. There was a clear, unmet need for a metabolic geneticist to support the paediatric neurologist and developmental paediatricians for metabolic and genetic investigations for neurometabolic disorders presenting with epilepsy and developmental disabilities as well as complex movement disorders in Greater Toronto Area.

For these reasons, under my leadership, the Neurometabolic Disorders Program has been established in the Division of Clinical and Metabolic Genetics at The Hospital for Sick Children. This program includes Complex Epilepsy Genetics, Combined Movement Disorder and Treatable Neurometabolic Disorders Clinics. My Neurometabolic Disorders Program has been grown quickly receiving referrals from clinical genetics, pediatric neurology, developmental paediatric clinics and from community pediatric neurologists in Greater Toronto Area. My stepwise diagnostic approach using metabolic and genetic investigations increased diagnostic yield in patients with epilepsy and developmental delays from less than 15% to more than 30%. I developed a diagnostic algorithm, which is published in Epilepsia in 2015. Combined Movement Disorder Clinic was established in 2012 and staffed by metabolic geneticist and paediatric movement disorder specialist, a genetic counsellor and a metabolic nurse. This clinic provided a multidisciplinary approach to the patients with movement disorders from diagnostic and treatment point of view. I developed Treatable Neurometabolic Disorders Clinic for the patients with a genetically confirmed neurometabolic disorder including creatine transporter deficiency (CRTR), pyridoxine-dependent epilepsy (PDE), guanidinoacetate methyltranferase (GAMT) deficiency and glucose transporter 1 (GLUT1) deficiency. I have been applying REB approved innovative therapies to the patients. I have been reporting out the treatment outcomes of patients on these therapies including arginine and glycine supplementation in CRTR deficiency (Mercimek-Mahmutoglu 2009), lysine-restricted diet in PDE (Mercimek-Mahmutoglu 2014), arginine supplementation therapy in PDE (Mercimek-Mahmutoglu 2014), low glycemic index diet and Glycosade therapy in GLUT1 deficiency (Al Muqbil 2015) and lysine supplementation therapy in GAMT deficiency (patient in Alberta). Due to my expertise in these disorders, I receive requests from international colleagues or parents for CRTR deficiency, GAMT deficiency and pyridoxine dependent epilepsy for my treatment recommendations.

As an extremely exciting addition to my clinical academic career, I have been leading the epilepsy genetics research program for diagnostic yield and gene identification in the Research Institute, The Hospital for Sick Children as Project Investigator since early 2012. I collaborate with Canadian Epilepsy Network “Personalized medicine in the treatment of epilepsy” Granted $5.0M by Genome Canada, to Dr. Cosette, Dr. Michaud and Dr. Minassian. I receive referrals from paediatric neurology and clinical genetics clinics to enrol patients into my epilepsy research study. I also collaborate with Genome Clinic to evaluate use of epilepsy genetic panels versus whole genome sequencing as diagnostic tool.

I have been invited as speaker to give talks for genetic diagnosis of epilepsy, creatine deficiency disorders and PDE to local, national and international meetings. The clinical diversity of the Neurometabolic Program also attracts elective trainees and fellows for metabolic genetics training at The Hospital for Sick Children since my arrival.

D. Research Funding

1. Grants, Contracts and Clinical Trials

FUNDED PEER-REVIEWED GRANTS

2019	**Principal Investigator.** Urine creatine panel in the diagnosis of creatine deficiency disorders and its diagnostic yield. Starbucks Summer Studentship Research Program. The Hospital for Sick Children. Amount: $5,800 CAD. Scholarship Grant.
2018	**Principal Investigator.** Neuronal ceroid lipofuscinosis: diagnostic yield of molecular genetic testing and phenotype, genotype and natural history of the patients diagnosed with neuronal ceroid lipofuscinosis. Rare Disease Foundation, Microgrant Program. Amount: $5,000 CAD. Funding type: Grant.

2018 **Principal Investigator.** Neuronal ceroid lipofuscinosis: diagnostic yield of molecular genetic testing and phenotype, genotype and natural history of the patients diagnosed with neuronal ceroid lipofuscinosis. Starbucks Summer Studentship Research Program. The Hospital for Sick Children. Amount: $5,800 CAD. Scholarship Grant.

2018-2020 **Principal Investigator.** Neonatal encephalopathy: identification of underlying genetic causes. Physicians' Services Incorporated Foundation, Health Research Grant. Principal Investigator: Andrews S. Co-Investigators: Marshall C, Vann C, Wilson D. Amount: $191,500 CAD. Funding type: Operating Grant.

2016 **Principal Investigator**. Disorders of intracellular cobalamin metabolism: phenotype, genotype and long-term treatment outcome. Starbucks Summer Studentship Research Program. The Hospital for Sick Children. Principal Investigator: Mahmutoglu, Saadet. Collaborators: Mellin L, Cruz V, Bullivant G. Funding Amount: $4,800 CAD. Funding type: Grant for summer student salary support.

This research study reviewed 33 patients with disorders of intracellular cobalamin metabolism. The study results were presented at the SickKids Summer Student Research Symposium in August 2016.

2015 **Principal Investigator**. Neonatal encephalopathy in term newborns: identification of underlying genetic defects using whole exome sequencing. Starbucks Summer Studentship Research Program. The Hospital for Sick Children. Principal Investigator: Mahmutoglu S. Collaborators: DesRoches C, Marshall C. Funding Amount: $4,800 CAD. Funding type: Grant for summer student salary support.

This research screened 10 patients presenting with neonatal encephalopathy. The results showed a diagnosis in 6 families identifying 4 known genetic disorders and two novel genes as likely cause. The study results were presented at the SickKids Summer Student Research Symposium.

2014-2015 **Co-Investigator**. Identification of genes involved in epilepsy and epilepsy pharmacoresistance. McLaughlin Centre, Accelerator Grants in Genomic Medicine. Principal Investigator: Minassian B. Collaborators: Mahmutoglu S, Andrade D. Amount: $75,000 CAD. Funding type: Grant.

I am co-investigator for this grant and enrol patients in this study for whole exome sequencing.

2013-2015 **Principal Investigator**. Identifying the genes and underlying causes for intractable epilepsies by next generation sequencing. McLaughlin Centre, Accelerator Grants in Genomic Medicine. Grant number MC-2013-08. Principal Investigator: Mahmutoglu S. Co-Investigators: Minassian B. Amount: $75,000 CAD. Funding type: Grant.

I designed the study to investigate the underlying causes of complex epilepsy and cognitive dysfunction. So far more than 100 patients have been enrolled into the study. Data analysis and validation of results are completed for 20 families.

2012-2014 **Principal Investigator**. First steps to the improvement of long-term outcome of GAMT deficiency by using an international database. Rare Disease

Foundation, Microgrant Program. Principal Investigator: Mahmutoglu S. Collaborator: Salomons G. $3,500 CAD. Funding type: Grant.

I designed this international registry study as well as a RedCAP questionnaire for the study. I enrolled 22 patients from European and North American countries. Data analysis is completed and manuscript is underway for submission in September 2015.

2012-2014 **Principal Investigator**. Is low creatine kinase a nonspecific-screening marker for creatine deficiency syndromes? Rare Disease Foundation, Microgrant Program. Principal Investigator: Mahmutoglu S. Co-Investigator: Kyriakopoulou L. Funding amount: $7,000 CAD. Funding type: Grant.

I designed this study as a collaborative study between the Department of Laboratory Medicine and the Department of Paediatrics to evaluate specificity and sensitivity of CK as a screening test of creatine deficiency disorders. These disorders are only diagnosed if physicians request specific urine tests (urine guanidinoacetate and creatine measurements). These tests are only performed in tertiary care hospitals like The Hospital for Sick Children. If I am able to find another marker, this will help physicians to think of these as treatable disorders.

2012-2014 **Co-Investigator**. Pilot study for newborn screening for guanidinoacetaat methyltransferase deficiency. MetaKids. The Netherlands. Principal Investigator: Salomons, G. Co-Investigators: Mahmutoglu S, Loeber RIVM, Struys E, Jakobs C. Funding amount: $55,500 EUR. Funding type: Grant.

I am co-investigator for this study and planned this study as a follow-up study of my recent project (Mercimek-Mahmutoglu et al 2012, Molecular Genetics and Metabolism, see publication list). This study sequenced DNA samples (anonymized) of 500 newborns for GAMT gene to investigate carrier frequency of this disease in the newborn population. The study results helped to set-up newborn screening for GAMT deficiency in The Netherlands as this is a treatable disease if identified in the neonatal period and treatment results in normal neurodevelopmental outcome. The study results are submitted to Gene in April, 2015 and tentatively accepted for publication as of August 20, 2015.

2012-2013 **Principal Investigator**. Myoclonic epilepsy: generating a database to identify treatable causes. Rare Disease Foundation, Microgrant Program. Principal Investigator: Mahmutoglu, S. Collaborator: Minassian B. Funding Amount: $3,500 CAD. Funding type: Grant.

I designed this study to develop a database and look at diagnostic yield. This study increased awareness of diagnostic investigations at The Hospital for Sick Children and became the foundation for a prospective study for whole exome and genome sequencing in patients with epilepsy.

2012-2013 **Principal Investigator**. Expanding the phenotypic spectrum of GLUT1 deficiency: a treatable metabolic epilepsy. Rare Disease Foundation, Microgrant Program. Principal Investigator: Mahmutoglu S. Collaborator: Minassian B. Funding Amount: $3,500 CAD. Funding type: Grant.

I designed this study to develop a database and increase awareness of GLUT1 deficiency at The Hospital for Sick Children. This study became the foundation for the prospective study to investigate patients on the ketogenic diet for GLUT1 deficiency using direct sequencing.

2012 **Principal Investigator**. Retrospective review of patients with Myoclonic epilepsy to generate a database: first steps to identify underlying aetiology. Starbucks Summer Student Research Program. The Hospital for Sick Children. Principal Investigator: Mahmutoglu S. Collaborators: Minassian B. Funding Amount: $4,800 CAD. Funding type: Grant for summer student salary support.

This research generated a database for prospective whole exome sequencing study. The study results were presented on the SickKids Summer Student Research Day.

2011-2012 **Principal Investigator**. A pilot study to assess prevalence of GAMT deficiency in British Columbia. Rare Disease Foundation, Microgrant Program. Principal Investigator: Mahmutoglu S. Co-Investigator: Sinclair G. Funding amount: $7,000 CAD. Funding type: Grant.

I designed this study to investigate carrier frequency of GAMT deficiency in the newborn population in British Columbia. This study was the foundation for another grant (Metakids) and resulted in establishment of newborn screening in the Netherlands as well as pilot newborn screening of GAMT deficiency in British Columbia (by Dr. Sinclair). The study results are published in 2012 in Molecular Genetics and Metabolism.

2010-2012 **Principal Investigator**. Prevalence of carpal tunnel syndrome in British Columbia; finding the treatable disorders as a cause of CTS. Rare Disease Foundation, Microgrant Program. Principal Investigator: Mahmutoglu S. Co-Investigators: Wong P, Verchere C. Funding Amount: $7,000 CAD. Funding type: Grant.

This research study was designed to look at prevalence of carpal tunnel syndrome in individuals underwent nerve conduction study. This study was the foundation of a prospective follow-up study to screen patients for mucoploysaccharidosis. The study is completed and became the foundation for other prospective studies leading to funding through Industry at The Hospital for Sick Children (see non-peer reviewed grants, funded)

2010-2011 **Principal Investigator**. Prevalence of myoclonic epilepsy in British Columbia; development of a diagnostic flowchart to help physicians. Rare Disease Foundation, Microgrant Program. Principal Investigator: Mahmutoglu S. Co-Investigator: Connolly M. Funding Amount: $3,500 CAD. Funding type: Grant.

This research evaluated the genetic diagnosis of myoclonic epilepsy as retrospective review. I designed the study to develop a database to arrange prospective study for whole exome sequencing to identify underlying causes.

FUNDED NON-PEER-REVIEWED GRANTS

2015-2016 **Principal Investigator**. Prevalence of MPS in individuals undergoing nerve conduction study for carpal tunnel syndrome. Principal Investigator: Mahmutoglu S. Co-Investigators: Raiman J, Vijsar,J. Funding Amount: $25,000 CAD. Funding type: Industry sponsored (Genzyme), PI initiated study.

This research will evaluate prevalence of mucopolysaccharidosis in individuals undergoing nerve conduction study. If the prevalence is high, urine MPS screening could be part of clinical care to be able to identify patients with mucopolysaccharidosis for enzyme replacement therapy.

2015-2016 **Principal Investigator.** Prevalence of mucopolysaccharidoses in individuals with musculoskeletal system and connective tissue manifestations in the Rheumatology Clinics. Principal Investigator: Mahmutoglu S. Co-Investigators: Raiman J, Laxer R, Levy D. Funding Amount: $20,000 CAD. Funding type: Industry sponsored (Genzyme) PI initiated study.

This research will evaluate prevalence of mucopolysaccharidosis in individuals with musculoskeletal problems. If the prevalence is high, urine MPS screening could be part of clinical care to be able to identify patients with mucopolysaccharidosis for enzyme replacement therapy.

2014-2015 **Principal Investigator**. Prevalence of MPS in individuals undergoing nerve conduction study for carpal tunnel syndrome. Principal Investigator: Mahmutoglu S. Co-Investigators: Raiman J, Vijsar J. Funding Amount: $25,000 CAD. Funding type: Industry sponsored (Genzyme), PI initiated study.

This research will evaluate prevalence of mucopolysaccharidosis in individuals undergoing nerve conduction study. If the prevalence is high, urine MPS screening could be part of clinical care to be able to identify patients with mucopolysaccharidosis for enzyme replacement therapy.

CLINICAL TRIALS

2016-2022 **Principal Investigator**. 024PKAN15004, A randomized, double-blind, placebo-controlled study with an open-label extension. Pediatric site investigator Canada-wide. Funding type: Industry sponsored Clinical Trial.

2016-2019 **Principal Investigator**. LUM-001-C-01, Observational study of males with creatine transporter deficiency. Funding type: Industry sponsored study.

2016-2018 **Principal Investigator**. ALXN1101-MCD-202: A Phase 2/3, Multicenter, Multinational, Open Label Study to Evaluate the Efficacy and Safety of ALXN1101 in Neonates with Molybdenum Cofactor Deficiency (MoCD) Type A. Principal Investigator: Mahmutoglu S. Accepted as site investigator for Canada on July 31, 2015. Information for the amount of funding and contracts are expected to arrive soon. Funding type: Industry sponsored Clinical Trial.

I was selected as site investigator (after full day site visit selection completed in July 2015) and principal investigator for this 5 year industry sponsored study for a new drug for molybdenum cofactor deficiency type A. This disease was not a treatable disease presenting with neonatal onset seizures and neurodegenerative disease leading to death in infancy. The drug, if started in the neonatal period treats seizures. If the drug is started before symptoms within the first few hours of life this may result in normal neurodevelopmental outcome. This disease is biochemically diagnosed at The Hospital for Sick Children. This trial will give us the opportunity to be the center for North America for this disorder.

2011-2013 **Co-Investigator**. A multicenter, multinational, extension study to evaluate the long-term efficacy and safety of BMN 110 in patients with mucopolysaccharidosis IVA (Morquio A Syndrome) 005. Biomarin Pharmaceutical. Principal Investigator: Raiman J. Co-Investigator: Mahmutoglu S, Fagfourty H. Funding amount: $2,136,164.80 CAD. Funding type: Clinical Trial, Industry funded.

I was co-investigator for the study and saw patients during their weekly infusions and had outcome assessments.

2011-2012 **Co-Investigator**. A phase 3, randomized, double-blind, placebo-controlled, multinational clinical study to evaluate the efficacy and safety of 2.0 mg/kg/week and 2.0 mg/kg/every other week BMN 110 in patients with mucopolysaccharidosis IVA (Morquio A Syndrome) 004. Biomarin Pharmaceutical. Principal Investigator: Raiman J. Co-Investigator: Mahmutoglu S, Fagfourty H. Funding amount: $191,129.40 CAD. Funding type: Clinical Trial, Industry funded.

I was co-investigator for the study and saw patients during their weekly infusions and had outcome assessments.

2. SALARY SUPPORT AND OTHER FUNDING

Other Funding

2015-2017 **Principal Investigator**. Development of zebrafish model for drug screening for pyridoxine dependent epilepsy caused by ALDH7A1 genetic defect. Division of Clinical and Metabolic Genetics, Department of Paediatrics Start-up funding, The Hospital for Sick Children. Funding amount: $120,000. Funding type: Operating Grant. Toronto, Ontario, Canada

2013-2015 **Principal Investigator**. Expression and characterization of GAMT, GATM and SLC6A8 missense variants in biological systems to identify carrier frequency of creatine deficiency disorders in the general population: first steps towards newborn screening for treatable neurometabolic disorders. Department of Paediatrics Start-up funding, The Hospital for Sick Children. Funding amount: $150,000. Funding type: Operating Grant. Toronto, Ontario, CA.

This is my major wet laboratory research project to set up expression systems at The Hospital for Sick Children for creatine deficiency disorders (3 different genetic disorders in the same biochemical pathway of creatine synthesis and transport). Through this funding support, I hired a research technician and developed expression systems for three disorders. Two of the studies are completed and published in 2015 (DesRoches et al, in Gene and in Molecular Genetics and Genomics, see publication list). Using this funding support, I developed a zebrafish model for pyridoxine dependent epilepsy to use the model for drug screening (please see research statement)

2015-2016 **Faculty Development Program Award.** Division of Clinical and Metabolic Genetics, Department of Paediatrics, University of Toronto, The Hospital for Sick Children. Amount: $10,000 CAD. Toronto, Ontario, Canada.

This funding was provided in order to support a one month observership with Dr. J. MINK pediatric neurologist, movement disorder specialist at University of Rochester Medical Center, Rochester, NY, US to gain experience in the diagnosis and management of paediatric movement disorders. This visit will happen in early 2016. To increase my knowledge prior to this visit, I started working in the Movement Disorder Clinic, Toronto Western Hospital under Dr. A. Lang's supervision, who is the well-known movement disorder specialist worldwide for adult Parkinson disease and adult movement disorders.

E. Publications

1. MOST SIGNIFICANT PUBLICATIONS

1. Bruun TUJ*, DesRoches CL, Wilson D, Chau V, Nakagawa T, Yamasaki M, Hasegawa S, Fukao T, Marshall C, **Mercimek-Andrews S.** Prospective cohort study for identification of underlying genetic causes in neonatal encephalopathy using whole-exome sequencing. Genet Med 2018;33(3):875-884. Impact factor 7.329. *Trainee publication: I supervised Bruun undergraduate student for the study. Senior Responsible Author.*

I am the principal and senior responsible author for this study. To identify underlying genetic defects, we applied whole-exome sequencing (WES) in term newborns with neonatal encephalopathy as a prospective cohort study. The diagnostic yield was 36%.

2. Khaikin Y*, Sidky S*, Abdenur J, Anastasi A, Ballhausen D, Buoni S, Chan A, Cheillan D, Dorison N, Goldenberg A, Goldstein J, Hofstede FC, Jacquemont ML, Koeberl DD, Lion-Francois L, Lund AM, Mention K, Mundy H, O'Rourke D, Pitelet G, Raspall-Chaure M, Tassini M, Billette de Villemeur T, Williams M, Salomons GS, **Mercimek-Andrews S**. Treatment outcome of twenty-two patients with guanidinoacetate methyltransferase deficiency: An international retrospective cohort study. Eur J Paediatr Neurol. 2018;22(3):369-379. Impact Factor: 2.362. **Senior Responsible Author**.

I am the principal and senior responsible author for this clinical retrospective study. In this study, I applied clinical severity score to 22 patients from different countries using RedCAP external database and development of extensive questionnaires to assess treatment outcome. The study was funded through Rare Disease Microgrant Program. I supervised two research students for the questionnaire development and data analysis.

3. Bruun TUJ*, Sidky S*, Bandeira AO, Debray FG, Ficicioglu C, Goldstein J, Joost K, Koeberl DD, Luísa D, Nassogne MC, O'Sullivan S, Õunap K, Schulze A, van Maldergem L, Salomons GS, **Mercimek-Andrews S**. Treatment outcome of creatine transporter deficiency: international retrospective cohort study. Metab Brain Dis. 2018;33(3):875-884. Impact Factor: 2.441. **Senior Responsible Author**.

I am the principal and senior responsible author for this clinical retrospective study. In this study, I developed and applied clinical severity score to 17 patients from different countries using RedCAP external database and development of extensive questionnaires to assess treatment outcome. I supervised two research students for the questionnaire development and data analysis.

4. Hamdan FF, Myers CT, Cossette P, Lemay P, Spiegelman D, Laporte AD, Nassif C, Diallo O, Monlong J, Cadieux-Dion M, Dobrzeniecka S, Meloche C, Retterer K, Cho MT, Rosenfeld JA, Bi W, Massicotte C, Miguet M, Brunga L, Regan BM, Mo K, Tam C, Schneider A, Hollingsworth G; Deciphering Developmental Disorders Study, FitzPatrick DR, Donaldson A, Canham N, Blair E, Kerr B, Fry AE, Thomas RH, Shelagh J, Hurst JA, Brittain H, Blyth M, Lebel RR, Gerkes EH, Davis-Keppen L, Stein Q, Chung WK, Dorison SJ, Benke PJ, Fassi E, Corsten-Janssen N, Kamsteeg EJ, Mau-Them FT, Bruel AL, Verloes A, Õunap K, Wojcik MH, Albert DVF, Venkateswaran S, Ware T, Jones D, Liu YC, Mohammad SS, Bizargity P, Bacino CA, Leuzzi V, Martinelli S, Dallapiccola B, Tartaglia M, Blumkin L, Wierenga KJ, Purcarin G, O'Byrne JJ, Stockler S, Lehman A, Keren B, Nougues MC, Mignot C, Auvin S, Nava C, Hiatt SM, Bebin M, Shao Y, Scaglia F, Lalani SR, Frye RE, Jarjour IT, Jacques S, Boucher RM, Riou E, Srour M, Carmant L, Lortie A, Major P, Diadori P, Dubeau F, D'Anjou G, Bourque G, Berkovic SF, Sadleir LG, Campeau PM, Kibar Z, Lafrenière RG, Girard SL, **Mercimek-Mahmutoglu S**, Boelman C, Rouleau GA, Scheffer IE, Mefford HC, Andrade DM, Rossignol E, Minassian BA, Michaud JL. High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. Am J Hum Genet. 2017;101(5):664-685. Impact factor 11.202. *Contributed with cases.* **Co-author**.

I am the principal investigator to enroll patients from SickKids for this international study.

5. Zabinyakov N*, Bullivant G*, Cao F, Fernandez Ojeda M, Jia ZP, Wen XY, Dowling JJ, Salomons GS, **Mercimek-Andrews S**. Characterization of the first knock-out aldh7a1 zebrafish model for pyridoxine-dependent epilepsy using CRISPR-Cas9 technology. PLoS One. 2017 Oct 20;12(10):e0186645. doi: 10.1371/journal.pone.0186645. eCollection 2017. Impact factor 2.806. **Principal Author, Senior Responsible Author.**

I am the principal and senior responsible author for this research study. I developed knock-out aldh7a1 zebrafish with a homozygous 5 base pair (bp) mutation in ALDH7A1. Knock-out aldh7a1 embryos have spontaneous rapid increase in locomotion and a rapid circling swim behavior earliest 8-day post fertilization (dpf). Electroencephalogram revealed large amplitude spike discharges compared to wild type. Knock-out aldh7a1 embryos have elevated alpha-AASA, piperideine 6-carboxylate and pipecolic acid compared to wild type embryos at 3 dpf. Knock-out aldh7a1 embryos showed no aldh7a1 protein by western blot compared to wild type. Our knock-out aldh7a1 zebrafish is a well characterized model for large-scale drug screening using behavioral and biochemical features and accurately recapitulates the human PDE-ALDH7A1 disease.

2. PEER-REVIEWED PUBLICATIONS

The names of all trainees and research students working under my supervision are underlined and marked with star.

Journal Articles

1. Jilani A*, Matviychuk D, Blaser S, Dyack S, Mathieu J, Asuri N. Prasad, Prasad C, Kyriakopoulou L, **Mercimek-Andrews S.** High diagnostic yield of direct Sanger sequencing in the diagnosis of neuronal ceroid lipofuscinoses. JIMD Reports 2019; 1-11 (online since September 2019). Impact Factor: none. *Trainee publication: I supervised A. Jilani (medical student, research student) for literature review, data collection, entry, data analysis and manuscript preparations.* **Senior Responsible Author**.
2. Costain G, Cordeiro D, Matviychuk D, **Mercimek-Andrews S**. Clinical Application of Targeted Next-Generation Sequencing Panels and Whole Exome Sequencing in Childhood Epilepsy. Neuroscience. 2019 Sep 2. pii: S0306-4522(19)30572-X. doi: 10.1016/j.neuroscience.2019.08.016. [Epub ahead of print]. Impact Factor: 3.504. *Trainee publication: I supervised G. Costain (medical genetics resident) for literature review and manuscript preparations.* **Senior Responsible Author**.
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Abstracts

The names of all trainees and research students working under my supervision are underlined and marked with star.

1. Zabinyakov N*, Kungurova Y*, Jansen E, Cameron J, MacNeil L, Salomons G S, **Mercimek-Andrews S**. Gamma aminobutyric acid and tricarboxylic acid pathways in knock-out aldh7a1 zebrafish for pyridoxine dependent epilepsy. JIMD September 2019, Vol 42-Supp 1. SSIEM Symposium 2019, Rotterdam, The Netherlands. (Platform Presentation). **Senior Responsible Author.**

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12. Bullivant G*, Cao F, Fernandez M, Jia ZP, Wen XY, Dowling JJ, Salomons G, **Mercimek-Mahmutoglu S**. Characterization of aldh7a1 knock-out zebrafish using CRISPR-Cas9 technology. Genetics & Genome Biology Program Annual Retreat, May 3, 2017, Toronto, Canada (Poster Presentation). **Senior Responsible Author.**
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Book Chapters

1. Colin Wilbur, **Saadet Mercimek-Andrews**, Anthony Lang. The Importance Of Inborn Errors Of Metabolism For Movement Disorders in Inherited Metabolic Movement Disorders: Recognition, Understanding, Improving Outcomes. Editors: Darius Ebrahimi-Fakhari & Phillip L. Pearl. 2019 (in press)
2. **Saadet Mercimek-Andrews**. Disorders of Creatine Metabolism: Creatine Deficiency Syndromes and Movement Disorders. in Inherited Metabolic Movement Disorders: Recognition, Understanding, Improving Outcomes. Editors: Darius Ebrahimi-Fakhari & Phillip L. Pearl. 2019 (in press)
3. Stoeckler-Ipsiroglu S, **Mercimek-Mahmutoglu S**, Salomons G. Creatine Deficiency Syndromes. In: Inborn Metabolic Diseases. Diagnosis and Treatment. Fernandes J, Saudubray JM, van den Berghe G (eds). Springer 6th ed. 2016:243-248. **Co-author**.
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10. **Mercimek-Mahmutoglu S**, Stöckler-Ipsiroglu S. Cerebral creatine deficiency and movement disorders. In Paediatric movement disorders Progress in understanding edited by E. Fernandez-Alvarez, A. Arzimanoglou, E. Tolosa. 2005;223-230. **Principal Author**.
11. **Mercimek-Mahmutoglu S**, Stöckler-Ipsiroglu S. Clinical and Biochemical Characteristics of Creatine Deficiency Syndromes. In J. Klepper, Brain metabolism revisited- concepts and treatments- metabolic epileptic encephalopathies, 1st edition, Int. Symposium 'Focus on Neuropaediatrics'. 2004;58-68, Fulda, Germany. **Principal Author**.

3. SUBMITTED PUBLICATIONS

Book Chapters in press

1. Bruun T, **Mercimek-Mahmutoglu S**. Inherited neurotransmitter disorders. Clinical Child Neurology Editor: Dr. Salih (in press).
2. Bruun T, **Mercimek-Mahmutoglu S**. Treatable Metabolic Epilepsies. Clinical Child Neurology Editor: Dr. Salih (in press).

4. MANUSCRIPTS UNDER PREPARATION

1. Pyridoxine-dependent epilepsy zebrafish model

F. Patents and Copyrights

No patents or copyrights.

G. Presentations and Special Lectures

1. International

Abstracts and Other Papers- Oral Platform Presentations

September 2019	**Senior Responsible Author**. Parallel Session 1D: Disorders of haem biosynthesis, vitamins, purines and pyrimidines. Title: Gamma aminobutyric acid and tricarboxylic acid pathways in knock-out aldh7a1 zebrafish for pyridoxine-dependent epilepsy. SSIEM Annual Symposium 3-6 September 2019. Presenter: Mercimek-Andrews S. Authors: Zabinyakov N*, Kungurova Y*, Jansen E, Cameron J, MacNeil L, Salomons G S, **Mercimek-Andrews S.**
Nov 2015	**Senior Responsible Author**. Concurrent Session. Title: Diagnostic yield of genetic testing in epileptic encephalopathy in childhood. World Neurology Conference, Santiago, Chile, October 31-November 5, 2015. Presenter: Mahmutoglu S. Authors: Patel J*, Cordeiro D, Hewson S, Cohn R, Kannu P, Kobayashi J, **Mercimek-Mahmutoglu S**.
Sep 2013	**Senior Responsible Author.** Concurrent Session. Title: Natural history of X-linked adrenoleukodystrophy at the hospital for sick children and Toronto General Hospital: twenty-three year experience. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 3-6, 2013. Presenter: Tran C*. Authors: Tran C*, Patel J*, Mamak E, Hewson S, Faghfoury H, Raiman J, Blaser S, Clarke JTR, **Mercimek-Mahmutoglu SM**. Trainee Presentation.
Sep 2010	**Principal Author**. Concurrent Session. Title: Atypical presentation of antiquitin deficiency in a female with neonatal hypoglycemia, hyperalacticacidemia and intractable myoclonic epilepsy. Society for the Study of Inborn Errors of Metabolism Annual Symposium, Istanbul, Turkey, October 31-September 3, 2010. Presenter: **Mahmutoglu S**. Authors: **Mercimek-Mahmutoglu S**, Horvath GA, Coulter-Mackie M, Connolly M, Waters PJ, Jakobs C, Stockler-Ipsiroglu S.
Sep 2010	**Principal Author. Concurrent Session**. Title: Treatment of intractable epilepsy in a female with X-linked cerebral creatine transporter (SLC6A8) Deficiency. Society for the Study of Inborn Errors of Metabolism Annual Symposium, Istanbul, Turkey, October 31-September 3, 2010. Presenter: Mahmutoglu S. Authors: **Mercimek-Mahmutoglu S**, Connolly M, Poskitt K, Lowry N, Salomons GS, Casey B, Sinclair G, Jakobs C, Stockler-Ipsiroglu S.
Jun 2008	**Principal Author**. **Plenary session**. Title: Progression of organ manifestations upon enzyme replacement therapy in a patient with mucopolysaccharidosis type I/Hurler prior to hematopoietic stem cell transplantation. 7th International Postgraduate Course on Lysosomal Storage Diseases. Nierstein, Germany.

Presenter: Mahmutoglu S. Authors: **Mercimek-Mahmutoglu S,** Lillquist Y, Davis J, Human D, Paschke E, Clarke LA, Stockler –Ipsiroglu S.

Sep 2004 **Principal Author**. **Concurrent Session**. Title: Persistierende hyperinsulinämische Hypoglykämie (PHHI), Erfahrungsbericht von 1978 bis 2000 an den Kinderkliniken Wien und Salzburg.100th Annual Meeting of the Society for German Paediatrics and 42nd Annual Meeting of the Society for Austrian Paediatrics, Berlin, Germany. Presenter: Rami B. Authors: **Mercimek-Mahmutoglu S**, Rami B, Herle M, Rittinger O, Stöckler- Ipsiroglu S, Konstantopoulou V, Schober E.

Invited Lectures and Presentations

Sep 2019 **Invited Plenary Speaker**. Title: Treatment Outcomes of Cerebral Creatine Deficiency Syndromes. Inborn Cerebral Creatine Deficiency Syndromes Symposium, September 6-7th, 2019, Rotterdam, The Netherlands.

Sep 2019 **Invited Plenary Speaker**. Title: Epilepsy in Cerebral Creatine Deficiency Syndromes. Inborn Cerebral Creatine Deficiency Syndromes Symposium, September 6-7th, 2019, Rotterdam, The Netherlands.

April 2019 **Invited Plenary Speaker**. Title: Creatine metabolism disorders; diagnostic methods and current treatments options. International Inborn Errors of Metabolism and Nutrition Congress, April 10-14, 2019, Istanbul, Turkey.

March 2019 **Invited Speaker.** Title: Neurometabolic disorders and their diagnostic odyssey. Department of Pediatric Metabolism Diseases, Aegean University, Izmir, Turkey.

July 2018 **Invited Plenary Speaker**. Title: CTD and AGAT novel therapies. CCDS Scientific and Patient Symposium, July 27-29, 2019, Austin, USA.

Jun 2017 **Invited Plenary Speaker.** Title: Epilepsy in Christianson syndrome. 2nd International Basic and Clinical Conference on Christianson Syndrome. June 28-July 1, 2017, Montreal, Canada

Jun 2016 **Invited Plenary Speaker.** Title: Creatine deficiency disorders. Portuguese Society of Inherited Metabolic Diseases, Porto, Portugal.

Nov 2015 **Invited Plenary Speaker**. Title: Inherited metabolic disorders presenting with epilepsy. The Chilean Society of Childhood and Adolescence Psychiatry and Neurology XXXIII Annual Meeting. La Serena, Chile

Nov 2015 **Invited Plenary Speaker**. Title: Inherited metabolic disorders as mimickers of hypoxic-ischemic encephalopathy. The Chilean Society of Childhood and Adolescence Psychiatry and Neurology XXXIII Annual Meeting. La Serena, Chile

Nov 2015 **Invited Plenary Speaker**. Title: Inherited metabolic diseases presenting with movement disorders. The Chilean Society of Childhood and Adolescence Psychiatry and Neurology XXXIII Annual Meeting. La Serena, Chile

Apr 2015 **Invited Plenary Speaker**. Title: Creatine deficiency disorders. XIII. National Metabolic and Nutrition Congress. Adana, Turkey.

Sep 2014 **Invited Plenary Speaker**. Title: Inherited metabolic disorders in childhood epilepsy.3rd International Conference on Inborn Errors of Metabolism. Hyderabad, India.

Sep 2014	**Speaker**, Title: Vitamin B6 metabolism disorders. 3rd International Conference on Inborn Errors of Metabolism. Hyderabad, India.
Jun 2014	**Invited Plenary Speaker**. Title: GAMT deficiency: new insights for the treatment recommendations and incidence. Symposium Updates on Neurometabolic Disorders. Amsterdam, The Netherlands.
Jul 2013	**Invited Plenary Speaker**. Title: Identifying Treatable Metabolic Causes of Neonatal Seizures. 3rd Biennial Conference on Brain Injury in Children, Toronto, Ontario, CA.
Jun 2013	**Key Note Speaker**. **Principal Author**, **PhD Defenses Presentation**. Title: Creatine deficiency syndromes: contributions to selective and newborn screening and treatment. PhD Defense. VU Medical Centre, Amsterdam, The Netherlands.
Mar 2013	**Invited Plenary Speaker**. Title: Inherited metabolic disorders presenting with epilepsy. Neurosantiago 2013, Neurology & Neurosurgery Conference. Santiago, Cuba.
Apr 2004	**Invited Plenary Speaker**. Title: Per-operative Management of patients with inborn errors of metabolic diseases. 9th Paediatric Inborn Errors of Metabolic Disease Symposium. Graz, Austria.
Nov 2003	**Invited Plenary Speaker**. Gaucher disease in Austria. 4th Gaucher Meeting. Austria, Vienna.

Abstracts and Poster Presentations

September 2019	**Senior Responsible Author.** Poster Presentation. Title: Genetic landscape of inherited metabolic disorders identified by next generation sequencing in childhood epilepsy. Authors: Costain G*, Cordeiro D, Matviychuk D, Mercimek-Andrews S. SSIEM Annual Symposium 3-6 September 2019. Trainee presentation.
September 2019	**Senior Responsible Author.** Poster Presentation. Title: Phenotypic and genotypic spectrum of mitochondrial aminoacyl-tRNA synthetase deficiencies from a single clinic. Authors: Al Balushi A*, Matviychuk D, Jobling R, Blaser S, **Mercimek-Andrews S**. SSIEM Annual Symposium 3-6 September 2019. Trainee presentation.
September 2019	**Senior Responsible Author.** Poster Presentation. Title: High diagnostic yield of direct Sanger sequencing in the diagnosis of neuronal ceroid lipofuscinoses. Authors: Jilani A*, Matviychuk D, Blaser S, Dyack S, Mathieu J, Prasad N, Prasad C, Kyriakopoulou L, **Mercimek-Andrews S**. SSIEM Annual Symposium 3-6 September 2019. Trainee presentation.
September 2019	**Senior Responsible Author.** Poster Presentation. Title: Genetics landscape of pediatric neurometabolic leukodystrophies. Authors: Damseh N*, Inbar-Feigenberg M, Blaser S, **Mercimek-Andrews S.** SSIEM Annual Symposium 3-6 September 2019. Trainee presentation.
September 2019	**Senior Responsible Author.** Poster Presentation. Title: Phenotypic spectrum of glucose transporter type 1 (GLUT1) deficiency syndrome in the pediatric

population. Authors: Nimmo G*, **Mercimek-Andrews S**. SSIEM Annual Symposium 3-6 September 2019. Trainee presentation.

April 2019 **Senior Responsible Author.** Poster Presentation. Title: Diagnostic Yield of Neuronal Ceroid Lipofuscinoses Molecular Genetic Tests: Phenotype and Genotype of Neuronal Ceroid Lipofuscinoses. Authors: Matviychuk D*, Jilani A*, Blaser S, Dyack S, Mathieu J, Prasad N, Prasad C, Kyriakopoulou L, **Mercimek-Andrews S**. ACMG Annual Clinical Genetics Meeting, April 2-6, 2019. Trainee presentation.

Nov 2015 **Senior Responsible Author**. Title: Phenotypic and biochemical features of pyruvate dehydrogenase complex deficiency: a retrospective cohort study at The Hospital for Sick Children. November 1-6, 2015, World Neurology Conference, Santiago, Chile. Authors: Inbar-Feigenberg M*, Cameron JM, Clark JT, Feigenbaum A, Hewson S, Siriwardena K, Robinson BH, **Mercimek-Mahmutoglu S**. Trainee Presentation.

Mar 2014 **Principal Author**. Title: Thirteen new patients with guanidinoacetate methyltransferase deficiency and functional characterization of nineteen missense variants in the GAMT gene. Society for Inherited Metabolic Disorders (SIMD) 37th Annual Meeting, March 9-12, 2014, Asilomar Conference Center, Pacific Grove, California, US. Authors: **Mercimek-Mahmutoglu S**, Ndika J, Kanhai W, Billette de Villemeur T, Cheillan D, Christensen E, Dorison N, Hannig V, Hendriks Y, Hofstede FC, Lion F, Lund A, Mundy H, Pitelet G, Raspall MC, Scott-Schwoerer J, Valayannopoulos V, Williams M, Salomons GS.

Mar 2014 **Principal Author**. Title: Retrospective review of cerebral spinal fluid catecholamine and serotonin metabolites for indications and diagnosis of neurotransmitter disorders. SIMD 40th Annual Meeting, March 9-12, 2014, Asilomar Conference Center, Pacific Grove, California, USA. Authors: **Mercimek-Mahmutoglu S**, Sidky S, Hyland K, Patel J, Donner EJ, Logan W, Mendoza-Londono R, Moharir M, Raiman J, Schulze A, Siriwardena K, Yoon G, Kyriakopoulou L.

Oct 2013 **Senior Responsible Author**. Title: Underlying genetic etiologies in epilepsy patients: a retrospective large single center study. 63rd American Society of Human Genetics Annual Meeting, Boston, MA, US, October 22-26, 2013. Authors: Guerin A*, Imhof E, Zak M, **Mercimek-Mahmutoglu S**. Trainee presentation.

Sep 2013 **Senior Responsible Author**. Title: Prevalence of inherited metabolic disorders in epilepsy patients: a large, single center study. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 2-7, 2013. Authors: Imhof E*, Zak M, Hewson S, Feigenbaum A, Kobayashi J, Minassian B, Raiman J, Siriwardena K, Tein I, Clarke J, **Mercimek-Mahmutoglu S**. Research Student Presentation.

Sep 2013 **Senior Responsible Author**. Title: Low cerebrospinal fluid catecolamine, serotonin and 5-methyltetrahydrofolate levels in a patient with infantile onset CBLG deficiency. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 2-7, 2013. Authors: Jobling R*, **Mercimek-Mahmutoglu S**. Trainee Presentation.

Sep 2013 **Senior Responsible Author**. Title: Two new patients with pyridoxine dependent epilepsy caused by ALDH7A1 genetic defect: long-term follow-up and normal neurodevelopmental outcome. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 2-7, 2013. Authors: Cordeiro D*,

	Mamak E, Donner E, Feigenbaum A, Siriwardena K, **Mercimek-Mahmutoglu S**. Metabolic Nurse Presentation.
Sep 2013	**Senior Responsible Author**. Title: Menkes disease caused by a novel frameshift mutation masquerading as an early onset mitochondrial encephalopathy. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 2-7, 2013. Authors: Mecija M*, Gonzalez S, **Mercimek-Mahmutoglu S**. Metabolic Nurse Presentation.
Sep 2013	**Senior Responsible Author**. Title: Atypical clinical response and normal CSF neurotransmitters in a patient with pyridoxamine-5-phosphate oxidase deficiency caused by a novel deletion in the PNPO gene. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 2-7, 2013. Authors: Guerin A*, Tran C*, Lewis J, Go CY, Salomons GS, **Mercimek-Mahmutoglu S**. Trainee Presentation.
Sep 2013	**Principal Author**. Title: Retrospective review of cerebrospinal fluid catecholamine and serotonin metabolites for indications and diagnosis of neurotransmitter disorders. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 2-7, 2013. Authors: **Mercimek-Mahmutoglu S**, Hyland K, Kyriakopoulou L, Sidky S, Allam N, Schulze A, Siriwardena K, Yoon G, Logan W, Soman T.
Sep 2013	**Senior Responsible Author**. Title: Retrospective review of all GSD type VI and IX patients at the hospital for sick children. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 2-7, 2013. Authors: Roscher A*, Hewson S, Feigenbaum A, Kronick J, Raiman J, Schulze A, Siriwardena K, **Mercimek-Mahmutoglu S**. Trainee Presentation.
Sep 2013	**Senior Responsible Author**. Title: Short-chain acyl-CoA dehydrogenase deficiency (SCADD) in twin brothers presenting with non-alcoholic steatohepatitis. 12th International Congress of Inborn Errors of Metabolism, Barcelona, Spain, September 2-7, 2013. Authors: Hewson S*, Nagy L, Ling S, **Mercimek-Mahmutoglu S**. Genetic Counsellor Presentation.
Oct 2012	**Senior Responsible Author**. Title: Severe infantile onset phenotype of CblG deficiency in a patient with abnormal neurotransmitter metabolites. 62th American Society of Human Genetics Annual Meeting, San Francisco, CA, US, November 6-10, 2012. Authors: Jobling R*, Hewson S, **Mercimek-Mahmutoglu S**. Trainee presentation.
Mar-Apr 2012	**Principal Author**. Etiology of myoclonic epilepsy in children and adolescents in British Columbia. Society for Inherited Metabolic Disorders (SIMD) 34th Annual Meeting, March 31- April 3, 2012, Charlotte, North Carolina, US. Authors: **Mercimek-Mahmutoglu S**, Lafek M, Demos M, Connolly M.
Mar-Apr 2012	**Principal Author**. Title: Carpal tunnel syndrome in children and adolescents in British Columbia. Society for Inherited Metabolic Disorders (SIMD) 34th Annual Meeting, March 31- April 3, 2012, Charlotte, North Carolina, US. Authors: **Mercimek-Mahmutoglu S**, Verchere C, Wong P.
Mar-Apr 2012	**Principal Author**. Title: Carrier frequency of GAMT deficiency: first steps to newborn screening for a treatable neurometabolic disease. Society for Inherited Metabolic Disorders (SIMD) 34th Annual Meeting, March 31- April 3, 2012, Charlotte, North Carolina, US. Authors: **Mercimek-Mahmutoglu S**, Sinclair G, SJM van Dooren, W. Kanhai, J. Nelson, O. Betsalel, O. Abdul-Hamid, Jakobs C, Salomons GS.

Aug-Sep 2011 **Principal Author**. Title: Carpal tunnel syndrome in children and adolescents in British Columbia. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Geneva, Switzerland, August 30-September 2, 2011. Authors: **Mercimek-Mahmutoglu S**, Maric B, Verchere C, Wong P.

Aug-Sep 2011 **Principal Author**. Title: Two novel compound heterozygous mutations in the Twinkle Helicase (C100rf2) gene causing early onset mitochondrial encephalomyopathy. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Geneva, Switzerland, August 30-September 2, 2011. Authors: **Mercimek-Mahmutoglu S**, Niederhoffer K, Selby KS.

Aug-Sep 2011 **Principal Author**. Title: Mucolipidosis type II (I-cell disease) masquerading as congenital infections in a newborn. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Geneva, Switzerland, August 30-September 2, 2011. Authors: **Mercimek-Mahmutoglu S**, Ting JT, Hochwald OH.

Mar 2011 **Principal Author**. Title: Antiquitin deficiency: Atypical neonatal presentation with severe hypoglycemia, hyperlacticacidemia, myoclonic epilepsy and hypoxic ischemic encephalopathy. Society for Inherited Metabolic Disorders (SIMD) 34th Annual Meeting, February 27-March 2, 2011, Asilomar Conference Center, Pacific Grove, California, USA. Authors: **Mercimek-Mahmutoglu S**, Horvath GA, Coulter-Mackie M, Connolly M, Waters PJ, Nelson T, Jakobs C, Stockler-Ipsiroglu S.

Mar 2011 **Principal Author**. Title: Six new patients with creatine deficiency syndromes identified by selective screening in British Columbia. Society for Inherited Metabolic Disorders (SIMD) 34th Annual Meeting, February 27-March 2, 2011, Asilomar Conference Center, Pacific Grove, California, USA.

Mar 2011 **Principal Author**. Title: Two different clinical phenotype in two siblings with 3-methylglutaconic aciduria type I. Society for Inherited Metabolic Disorders (SIMD) 34th Annual Meeting, February 27-March 2, 2011, Asilomar Conference Center, Pacific Grove, California, USA. Authors: **Mercimek-Mahmutoglu S**, Bhanji N, Waters P, Stockler-Ipsiroglu S.

Sep 2010 **Senior Responsible Author**. Title: Juvenile neuronal lipofuscinosis in a patient with compound heterozygous CLN3 mutations: a 9 year follow-up. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Istanbul, Turkey, August 31-September 3, 2010. Authors: Al-Thihli K*, Matsuba C, Roland E, Stockler-Ipsiroglu S, **Mercimek-Mahmutoglu S**. Trainee presentation.

Sep 2010 **Principal Author**. Title: Two different clinical phenotype in two siblings with 3-methylglutaconic aciduria type 1. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Istanbul, Turkey, August 31-September 3, 2010. Authors: **Mercimek-Mahmutoglu S**, Bhanji N, Waters P, Stockler-Ipsiroglu S.

Sep 2010 **Senior Responsible Author**. Title: Intermittent Choreoathetosis in a 9 year old boy with late onset NKH caused by a novel homozygous missense mutation in the GLDC Gene. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Istanbul, Turkey, August 31-September 3, 2010. Authors: Brunel-Guitton C*, Casey B, Hewes D, Vallance H, Stockler-Ipsiroglu S, **Mercimek-Mahmutoglu S**. Trainee presentation.

Sep 2010 **Principal Author**. Title: Six new patients with creatine deficiency syndromes identified by selective screening in British Columbia. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Istanbul, Turkey, August 31-September 3, 2010. Authors: **Mercimek-Mahmutoglu S**, Roland E, Huh L,

	Steinraths M, Connolly M, Salomons GS, Sinclair G, Jakobs C, Stockler-Ipsiroglu S.
Sep 2009	**Principal Author**. Title: Progression of cardiac manifestation on enzyme replacement therapy in a 4-6/12-year-old patient with mucopolysaccharidosis type II, Hunter syndrome. 11th International Congress of Inborn Errors of Metabolism, San Diego, California, USA, August 29-September 2, 2009. Authors: **Mercimek-Mahmutoglu S**, Human D, Stockler-Ipsiroglu S.
Sep 2009	**Principal Author**. Title: Progression of organ manifestations upon enzyme replacement therapy in a patient with mucopolysaccharidosis type I / Hurler prior to hematopoietic stem cell transplantation. 11th International Congress of Inborn Errors of Metabolism, San Diego, California, USA, August 29-September 2, 2009. Authors: **Mercimek-Mahmutoglu S**, Lillquist Y, Davis J, Human D, Paschke E, Clarke LA, Stockler-Ipsiroglu S.
May 2007	**Principal Author**. Title: Outcome of enzyme replacement therapy in a patient with mucopolysaccharidosis type I/Hurler prior to bone marrow transplantation. 4th Annual MPS I Symposium, May 18-19, 2007, Salt Lake City, Utah. Authors: **Mercimek-Mahmutoglu S**, Lillquist Y, Davis J, Human D, Paschke E, Clarke LA, Stockler-Ipsiroglu S.
Aug-Sep 2004	**Principal Author**. Title: Vanishing white matter disease - A new leukoencephalopathy: case report of 3 patients. Society for the Study of Inborn Errors of Metabolism 41st Annual Symposium, Amsterdam, The Netherlands, August 31-September 3, 2004. Authors: **Mercimek-Mahmutoglu S**, van der Knaap M, Ipsiroglu O, Scheper G, Konstantopoulou V, Stöckler-Ipsiroglu S.
Aug-Sep 2004	**Principal Author**. Title: Hypomyelination and atrophy of the basal ganglia and cerebellum, a new syndrome: case report. Society for the Study of Inborn Errors of Metabolism 41st Annual Symposium, Amsterdam, The Netherlands, August 31-September 3, 2004. Authors: **Mercimek-Mahmutoglu S**, van der Knaap M, Baric I, Konstantopoulou V, Prayer D, Stöckler-Ipsiroglu S.
Aug-Sep 2004	**Principal Author**. Title: Evidence of neurological manifestations in patients with type I Gaucher disease Society for the Study of Inborn Errors of Metabolism 41st Annual Symposium, Amsterdam, The Netherlands, August 31-September 3, 2004. Authors: **Mercimek-Mahmutoglu S**, Gruber S, Wöber C, Moser E, Stöckler-Ipsiroglu S.
Aug-Sep 2004	**Collaborator**. Title: Treatment and outcome of patients with fatty acid oxidation defects identified by expanded newborn screening in Austria. Society for the Study of Inborn Errors of Metabolism 41st Annual Symposium, Amsterdam, The Netherlands, August 31-September 3, 2004. Authors: Konstantopoulou V, Möslinger D, **Mercimek-Mahmutoglu S**, Bodamer OA, Erwa W, Stöckler-Ipsiroglu S.
Aug-Sep 2004	**Collaborator**. Title: Expanded newborn screening for inborn errors of metabolism by tandem mass spectrometry in Austria. Society for the Study of Inborn Errors of Metabolism 41st Annual Symposium, Amsterdam, The Netherlands, August 31-September 3, 2004. Authors: Möslinger D, Mühl A**, Mercimek-Mahmutoglu S**, Konstantopoulou V, Stöckler-Ipsiroglu S, Bodamer OA.
Mar 2004	**Principal Author**. Title: Clinical, biochemical and molecular findings in 16 patients with GAMT deficiency. 30th Annual Meeting of the Society for

Neuropaediatrics, Bern, Switzerland, March 25-28, 2004. Authors: **Mercimek-Mahmutoglu S**, Edlinger-Horvat C, Item C et al.

Mar 2004 **Principal Author**. Title: Vanishing white matter disease, a new leukoencephalopathy. Case report of 3 patients. 30th Annual Meeting of the Society for Neuropediatrics, Bern, Switzerland, March 25-28, 2004. Authors: **Mercimek-Mahmutoglu S**, Ipsiroglu OS, Prayer D, van der Knaap MS et al.

Feb 2003 **Principal Author**. Title: Different clinical outcome in monozygotic twins with Adrenoleukodystrophy. 29th Annual Meeting of the Society for Neuropaediatrics, Vienna, Austria, February 13-15 2003. Authors: **Mercimek-Mahmutoglu S**, Bodamer OA, Stöckler-Ipsiroglu S.

Feb 2003 **Principal Author**. Title: Tyrosine Hydroxylase Deficiency, a rare defect in neurotransmitter metabolism. Case report and review of the literature. 29th Annual Meeting of the Society for Neuropaediatrics, Vienna, Austria, February 13-15, 2003. Authors: **Mahmutoglu S**, Möslinger D, Bodamer OA, Stöckler-Ipsiroglu S.

Sep 2002 **Principal Author**. Title: Tyrosine Hydroxylase Deficiency, a rare defect in neurotransmitter metabolism in a 8 month old patient. 98th Annual Meeting of the Society for German Paediatrics, Leipzig, Germany, September 18-21, 2002. Authors: **Mahmutoglu S**, Bodamer OA, Möslinger D, Stöckler- Ipsiroglu S.

Sep 2002 **Principal Author**. Title: Different clinical outcome in monozygotic twins with adrenoleukodystrophy. Society for the Study of Inborn Errors of Metabolism 40th Annual Symposium, Dublin, Ireland, September 3-6, 2002. Authors: **Mercimek-Mahmutoglu S**, Bodamer OA, Stöckler-Ipsiroglu S.

Other Presentations

Sep 2011 **Speaker**. Title: Creatine deficiency syndromes and their treatment. Academic Rounds. Department of Clinical Chemistry, VU Medical Center. Amsterdam, Netherlands, September 8, 2011.

Mar 2002 **Speaker.** Title: A new patient with tyrosine hydroxylase enzyme deficiency, case report. Seminar for Inborn Errors of Metabolic Diseases, Fulda, Germany, March 8-9, 2002.

2. National

Abstracts and Other Papers- Oral Platform Presentations

May 2019 **Senior Responsible Author.** Platform Presentation. Title: Genetic landscape of inherited metabolic disorders identified by next generation sequencing in childhood epilepsy. Authors: Costain G*, Cordeiro D, Matviychuk D, Mercimek-Andrews S. Garrod Symposium 2019, May 9-11, 2019. Trainee presentation.

May 2019 **Senior Responsible Author.** Poster Presentation. Title: Phenotypic and genotypic spectrum of mitochondrial aminoacyl-tRNA synthetase deficiencies from a single clinic. Authors: Al Balushi A*, Matviychuk D, Jobling R, Blaser S, Mercimek-Andrews S. Garrod Symposium 2019, May 9-11, 2019. Trainee presentation.

May 2019 **Senior Responsible Author.** Poster Presentation. Title: High diagnostic yield of direct Sanger sequencing in the diagnosis of neuronal ceroid lipofuscinoses. Authors: Jilani A*, Matviychuk D, Blaser S, Dyack S, Mathieu J, Prasad N, Prasad C, Kyriakopoulou L, Mercimek-Andrews S. Garrod Symposium 2019, May 9-11, 2019. Trainee presentation.

May 2019 **Senior Responsible Author.** Poster Presentation. Title: Genetics landscape of pediatric neurometabolic leukodystrophies. Authors: Damseh N*, Inbar-Feigenberg M, Blaser S, Mercimek-Andrews S. Garrod Symposium 2019, May 9-11, 2019. Trainee presentation.

May 2019 **Senior Responsible Author.** Title: Phenotypic spectrum of glucose transporter type 1 (GLUT1) deficiency syndrome in the pediatric population. Authors: Nimmo G*, Mercimek-Andrews S. Garrod Symposium 2019, May 9-11, 2019. Trainee presentation.

April 2019 **Senior Responsible Author.** Title: Diagnostic Yield of Neuronal Ceroid Lipofuscinoses Molecular Genetic Tests: Phenotype and Genotype of Neuronal Ceroid Lipofuscinoses. Authors: Matviychuk D*, Jilani A*, Blaser S, Dyack S, Mathieu J, Prasad N, Prasad C, Kyriakopoulou L, Mercimek-Andrews S. ACMG Annual Clinical Genetics Meeting, April 2-6, 2019. Trainee presentation.

May 2017 **Senior Responsible Author.** Plenary session. Disorders of intracellular cobalamin metabolism: phenotype, genotype and long-term treatment outcome. Garrod Symposium May 4-6, 2017, Montreal, Canada. Presenter Lizbeth Mellin-Sanchez*, Authors: Garrett Bullivant*, Vivian Cruz, Julian Raiman, Andreas Schulze, Komudi Siriwardena, **Saadet Mercimek-Mahmutoglu**. Trainee presentation

Nov 2013 **Senior Responsible Author**. Plenary session. Title: Underlying genetic etiologies in epilepsy patients: a retrospective large single center study. 37th CCMG Annual Scientific Meeting, Toronto, Ontario, Canada, November 7-9, 2013. Presenter Guerin A. Authors: Guerin A*, Imhof E*, Zak M, **Mercimek-Mahmutoglu S**. Trainee presentation.

Jun 2011 **Senior Responsible Author**. Concurrent session. Title: Aetiology of carpal tunnel syndrome in children in British Columbia. Canadian Neurological Sciences Federation 46th Annual Congress, Vancouver, British Columbia, Canada, June 15-17, 2011. Presenter Maric B. Authors: **Mercimek-Mahmutoglu S**, Maric B*, Verchere C, Wong P. Research student presentation.

Jun 2009 **Principal Author**. Concurrent session. Title: A female patient with a medically refractory epilepsy and severe X-linked creatine transporter (SLC6A8) deficiency: successful treatment with l-arginine and l-glycine supplementation. Canadian Neurological Sciences Federation 44th Annual Congress, Halifax, Nova Scotia, Canada June 9-12, 2009. Presenter: **Mahmutoglu S**. Authors: **Mercimek-Mahmutoglu S**, Connolly M, Lowry N, Stockler S.

May 2009 **Collaborator**. Plenary session. Title: Selective Screening for Pyridoxine Dependent Epilepsy (Antiquitin Deficiency). Annual Meeting of the Garrod Association, Montreal, Quebec, Canada. May 1-2, 2009. **Presenter: Mahmutoglu S.** Authors: Stockler S, Connolly M, **Mahmutoglu S,** Coulter-Mackie M.

May 2009 **Principal Author**. Plenary session. Title: A female patient with a medically refractory epilepsy and severe X-linked creatine transporter (SLC6A8) deficiency:

successful treatment with l-arginine and l-glycine supplementation. Annual Meeting of the Garrod Association, Montreal, Quebec, Canada, May 1-2, 2009. Presenter: **Mahmutoglu S**. Authors: **Mercimek-Mahmutoglu S**, Connolly M, Lowry N, Stockler S.

May 2006 — **Principal Author**. Plenary session. Title: Long term outcome in patients with argininosuccinate lyase deficiency identified by newborn screening. Annual Meeting of the Garrod Association: Treatment of Metabolic Disease, Halifax, Nova Scotia, Canada, May 12-13, 2006. Presenter: **Mahmutoglu S**. Authors: **Mercimek-Mahmutoglu S**, Haeberle J, Strobl W, Moeslinger D, Stockler-Ipsiroglu S.

Apr 2004 — **Principal Author.** Plenary session. Title: Hypomyelination and atrophy of the basal ganglia and cerebellum syndrome, case report. 9th National Paediatric Inborn Errors of Metabolic Disease Symposium, Graz, Austria, April 22-24, 2004. Presenter: **Mahmutoglu S**.

Apr 2004 — **Principal Author**. Plenary session. Title: Vanishing white matter disease, a new leukoencephalopathy, case reports of 3 patients. 9th National Paediatric Inborn Errors of Metabolic Disease Symposium, Graz, Austria, April 22-24, 2004. Presenter: **Mahmutoglu S**.

May 1999 — **Principal Author**. Concurrent session. Title: Prevalence of Celiac disease in first-degree relatives of turkish patients with celiac disease. 35th Turkish Pediatric Association Congress, Ankara, Turkey, May19-23, 1999. Presenter: **Mahmutoglu S**.

Invited Lectures and Presentations

June 2019 — **Invited Speaker.** Title: Creatine deficiency disorders: phenotypes, genotypes, diagnosis and treatment outcomes**.** Genetic and Neurology Grand Rounds, Department of Medical Genetics, University of Alberta, Stollery Children's Hospital, Edmonton, Alberta, Canada.

May 2019 — **Invited Speaker.** Title: Pyridoxine-dependent epilepsies: phenotypes and treatment outcomes. Garrod Symposium 2019, May 9-11, 2019.

Jan 2011 — **Invited Speaker.** Title: Treatable epilepsies caused by inborn errors of metabolism. Genetic Grand Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada.

Apr 2004 — **Invited speaker.** Plenary session. Title: Per-operative Management of patients with inborn errors of metabolic diseases. 9th paediatric inborn errors of metabolic disease symposium, 22-24th April, 2004, Graz, Austria.

Other Presentations, Abstracts- Poster Presentations

May 2017 — **Senior Responsible Author.** Riboflavin transporter deficiency mimicking mitochondrial encephalomyopathy caused by complex II deficiency. Garrod Symposium May 4-6, 2017, Montreal, Canada. Presenter Nimmo GAM*, Authors Chitayat D, **Mercimek-Mahmutoglu S**. Trainee Presentation.

Nov 2013 — **Principal Author**. Title: Thirteen new patients with guanidinoacetate methyltransferase deficiency and functional characterization of nineteen missense variants in the GAMT gene. Canadian College of Medical Geneticist (CCMG) 37th Annual Scientific Meeting, Toronto, Ontario, Canada, November 7-

9, 2013. Authors: **Mercimek-Mahmutoglu S**, Ndika J, Kanhai W, Billette de Villemeur T, Cheillan D, Christensen E, Dorison N, Hannig V, Hendriks Y, Hofstede FC, Lion F, Lund A, Mundy H, Pitelet G, Raspall MC, Scott-Schwoerer J, Valayannopoulos V, Williams M, Salomons GS.

Nov 2013 **Senior Responsible Author**. Title: Atypical clinical response and normal CSF neurotransmitters in a patient with pyridoxamine-5-phosphate oxidase deficiency caused by a novel deletion in the PNPO gene. Canadian College of Medical Geneticist (CCMG) 37th Annual Scientific Meeting, Toronto, Ontario, Canada, November 7-9, 2013. Authors: Guerin A*, Tran C*, Lewis J, Go CY, Salomons GS, **Mercimek-Mahmutoglu S**. Trainee Presentation.

Nov 2013 **Senior Responsible Author**. Title: Short-chain acyl-CoA dehydrogenase deficiency (SCADD) in twin brothers presenting with non-alcoholic steatohepatitis. Canadian College of Medical Geneticist (CCMG) 37th Annual Scientific Meeting, Toronto, Ontario, Canada, November 7-9, 2013. Authors: Hewson S*, Nagy L, Ling S, **Mercimek-Mahmutoglu S**. Genetic Counsellor Presentation.

Nov 2013 **Principal Author**. Title: Retrospective review of cerebrospinal fluid catecholamine and serotonin metabolites for indications and diagnosis of neurotransmitter disorders. Canadian College of Medical Geneticist (CCMG) 37th Annual Scientific Meeting, Toronto, Ontario, Canada, November 7-9, 2013. Authors: **Mercimek-Mahmutoglu S**, Kyriakopoulou L, Sidky S, Hyland K.

Nov 2013 **Senior Responsible Author**. Title: Retrospective review of all GSD type VI and IX patients at the hospital for sick children. Canadian College of Medical Geneticist (CCMG) 37th Annual Scientific Meeting, Toronto, Ontario, Canada, November 7-9, 2013. Authors: Roscher A*, Hewson S, Feigenbaum A, Kronick J, Raiman J, Schulze A, Siriwardena K, **Mercimek-Mahmutoglu S**. Trainee Presentation.

Jun 2008 **Principal Author**. Title: Follow-up of a patient with MPS type II on ERT. Advances in the Treatment of Lysosomal Storage Disorders, Halifax, Nova Scotia, CA, June 6-8, 2008. Presenter: **Mahmutoglu S**.

May 2006 **Principal Author**. Title: Determination of urinary creatine to creatinine ratio as a screening method for creatine transporter (SLC6A8) deficiency. Annual Meeting of the Garrod Association: Treatment of Metabolic Disease, Halifax, Nova Scotia, CA, May 12-13, 2006. Authors: **Mercimek-Mahmutoglu S**, Muehl A, Neophytou B, Jakobs C, Salomons GS, Verhoeven N, Stoeckler-Ipsiroglu S.

Oct 2003 **Principal Author**. Title: Clinical, biochemical and molecular findings in 15 patients with GAMT deficiency. 41st Annual Meeting of the Society for Austrian Paediatrics, Salzburg, Austria, October 1-4, 2003. Authors: **Mercimek-Mahmutoglu S**, Stromberger C, Item C.

Oct 2003 **Principal Author**. Title: Vanishing white matter disease, a new leukoencephalopathy. Case report of 3 patients. 41st Annual Meeting of the Society for Austrian Paediatrics, Salzburg, Austria, October 1-4, 2003. Authors: **Mercimek-Mahmutoglu S**, Ipsiroglu OS, Paky F, et al.

Oct 2003 **Principal Author**. Title: Clinical, biochemical and molecular findings in 15 patients with GAMT deficiency. 41st Annual Meeting of the Society for Austrian Paediatrics, Salzburg, Austria, October 1-4, 2003. Authors: **Mercimek-Mahmutoglu S**, Stromberger C, Item C.

Oct 1996 **Principal Author**. Title: Idiopathic juvenile osteoporosis. XL. National Pediatric Association Congress, Gaziantep, Turkey, October 14-17, 1996. Authors: **Mercimek S,** Tuysuz B.

Oct 1996 **Principal Author**. Title: Osteoectasia with hyperphosphatasia (Juvenile Paget's Disease). XL. National Pediatric Association Congress, Gaziantep, Turkey, October 14-17,1996. Authors: **Mercimek S**, Tuysuz B.

Other presentations oral presentations

May 2006 **Principal Author.** Concurrent session. Title: Gaucher Disease type I with L444P homozygous mutation: case presentation. Guidelines for treatment of Gaucher Disease with miglustat, Halifax, Nova Scotia, Canada May 11, 2006. Authors: **Mahmutoglu S**, Stockler-Ipsiroglu S.

Jan 2004 **Principal Author**. Title: A patient with therapy resistant epilepsy, ataxia and developmental delay, case report: In Unsolved Cases in Neuropediatrics. Austrian National Society of Neuropediatrics, Austria, January 9-11, 2004. Presenter: **Mahmutoglu S**.

Jan 2004 **Principal Author**. Title: A new patient with H-ABC syndrome, case report in Unsolved Cases in Neuropediatrics. Austrian National Society of Neuropediatrics, Austria, January 9-11, 2004. Presenter: **Mahmutoglu S**.

Jun 2003 **Principal Author**. Title: Different outcome in 2 siblings with vanishing white matter disease: In Unsolved Cases in Neuropediatrics. Austrian National Society of Neuropediatrics, Vienna, Austria, June 27-28, 2003. Presenter: **Mahmutoglu S**.

3. Provincial/Regional

Invited Lectures and Presentations

March 2019 **Invited Speaker:** Title: Genetic landscape of pediatric movement disorders & childhood epilepsy. Genetic Grand Rounds, Medical Genetics Program of Southwestern Ontario London Health Sciences Centre – Victoria Hospital, London, Ontario, Canada

Apr 2013 **Invited Speaker**. Title: Investigating Metabolic Causes of Developmental Delay, Genyzme Rare Disease Night, Toronto, Ontario, Canada, April 10, 2013.

Jan 2013 **Invited Speaker**. Title: Developmental delay and inherited metabolic disorders. Clinical Genetics Grand Rounds, North York General Hospital, Toronto, Ontario, Canada

Dec 2012 **Invited Speaker**. Title: Metabolic disorders presenting with seizures in childhood. East York General Hospital Grand Rounds, East York General Hospital, Toronto, Ontario, Canada December 13, 2012. Presenter: **Mahmutoglu S**.

4. Local

Abstracts and Other Papers- Oral Platform Presentations

March 2017 **Senior Responsible Author.** Plenary session. Disorders of intracellular cobalamin metabolism: phenotype, genotype and long-term treatment outcome. Division of Clinical and Metabolic Genetics Research Day, March 17, 2017, Toronto, Canada. Presenter Lizbeth Mellin-Sanchez*, Authors: Bullivant G*, Cruz V, Raiman J, Schulze A, Siriwardena K, **Mercimek-Mahmutoglu S**. Trainee presentation.

March 2017 **Senior Responsible Author.** Plenary session. Characterization of aldh7a1 knock-out zebrafish using CRISPR-Cas9 technology. Division of Clinical and Metabolic Genetics Research Day March 17, 2017, Toronto, Canada. Presenter Garrett Bullivant*. Authors: Cao G, Fernandez M, Jia ZP, Wen XY, Dowling JJ, Salomons G, **Mercimek-Mahmutoglu S**. Trainee presentation.

March 2017 **Senior Responsible Author.** Diagnostic yield of genetic testing in pediatric movement disorders. Presenter **Mercimek-Mahmutoglu S.** Division of Clinical and Metabolic Genetics Research Day March 17, 2017, Toronto, Canada.

April 2016 Desroches CL*, Bruun T*, Wang P, Marshall CR, **Mercimek-Mahmutoglu** S. Arginine-glycine amidinotransferase deficiency and functional characterization of missense variants in GATM. Clinical and Metabolic Genetics 5th Annual Research Day, April 1, 2016. (Poster Presentation). **Senior Responsible Author.** Research Technician Presentation.

April 2016 Teneiji AA*, Bruun T*, Sidky S, Cordeiro D, Cohn R, Raiman J, Siriwardena K, **Mercimek-Mahmutoglu S**. Phenotypic and genotypic spectrum of N-linked congenital disorders of glycosylation. Clinical and Metabolic Genetics 5th Annual Research Day, April 1, 2016. (Poster Presentation). **Senior Responsible Author.** Metabolic Fellow Presentation.

April 2016 **Senior Responsible Author.** Teneiji AA*, Bruun T*,Patel J*, Cordeiro D, **Mercimek-Mahmutoglu S**. Phenotypic spectrum of patients with pyridoxine dependent epilepsy caused by mutations in the ALDH7A in a single center. Clinical and Metabolic Genetics 5th Annual Research Day, April 1, 2016. (Oral Platform Presentation). Metabolic Fellow Presentation.

April 2016 **Senior Responsible Author.** DesRoches CL*, Struys E, **Mercimek-Mahmutoglu S**. Behavioral and biochemical features of aldh7a1 zebrafish for pyridoxine dependent epilepsy due to ALDH7A1 mutations. Clinical and Metabolic Genetics 5th Annual Research Day, April 1, 2016. (Oral Platform Presentation). Research Technician Presentation.

April 2016 **Senior Responsible Author.** Inbar-Feigenberg M*, Cameron JM, Clark JT, Feigenbaum A, Hewson S, Siriwardena K, Robinson BH, **Mercimek-Mahmutoglu S.** Phenotypic and biochemical features of pyruvate dehydrogenase complex deficiency: a retrospective cohort study at The Hospital for Sick Children. Clinical and Metabolic Genetics 5th Annual Research Day, April 1, 2016. (Poster Presentation). Metabolic Fellow Presentation.

April 2016 **Senior Responsible Author.** Bruun T*, DesRoches C*, Marshall C, **Mercimek-Mahmutoglu S.** Neonatal encephalopathy in term newborns: identification of underlying genetic defects using whole exome sequencing. Clinical and Metabolic Genetics 5th Annual Research Day, April 1, 2016. (Oral Platform Presentation). Research Student Presentation.

May 2015 **Senior Responsible Author**. Title: Carrier Frequency of Guanidinoacetate Methyltransferase Deficiency in the General Population by Functional

Characterization of Missense Variants in the GAMT Gene. Presenter Desroches CL. Authors: Desroches CL*, Patel J, Wang P, Minassian B, Marshall CR, Salomons GS, **Mercimek-Mahmutoglu S**. Clinical and Metabolic Genetics 4th Annual Research Day, Toronto, Ontario, Canada, May 29, 2015. Research Technician Presentation.

May 2014 **Collaborator.** Title: What the rheumatologist need to know about MSK manifestations of MPS. Rheumatology Research Rounds, The Hospital for Sick Children, Toronto, ON, Canada. Authors: **Mahmutoglu S,** Raiman J.

Mar 2014 **Senior Responsible Author.** Title: Underlying genetic etiologies in epilepsy patients: a retrospective large single center study. Clinical and Metabolic Genetics 3rd Annual Research Day, The Hospital for Sick Children, Toronto, Canada. Presenter Guerin A. Authors: Guerin A*, Imhof E*, Zak M, Mercimek-Mahmutoglu S. Trainee presentation.

Oct 2010 **Collaborator.** Title: Tone, Transporter and Treatment- making the connection. Advances in Pediatrics Department of Pediatrics, UBC, British Columbia Children's Hospital, Vancouver, BC, Canada. Authors: Veer D, Mickelson E, **Mahmutoglu S**, Roland E.

Jan 2010 **Speaker.** Title: Paroxysmal movement disorder caused by late onset non-ketotic hyperglycinemia. Medical Genetics-Biochemical Diseases Joint Rounds, British Columbia Children's Hospital, Vancouver, British Columbia, Canada.

Jan 2010 **Speaker.** Title: Mild GDD in a male patient: early diagnosis of cerebral creatine transporter deficiency. Medical Genetics-Biochemical Diseases Joint Rounds, British Columbia Children's Hospital, Vancouver, British Columbia, Canada.

Oct 2009 **Collaborator.** Title: Seizures: start pyridoxine think antiquitin. Advances in Pediatrics Department of Pediatrics, UBC, British Columbia Children's Hospital, Vancouver, BC, Canada. Authors: **Mahmutoglu S**, Connolly M, Stockler S.

May 2009 **Principal Author.** Plenary session. Title: A female with severe X-linked cerebral creatine transporter deficiency: successful treatment with l-arginine and glycine supplementation. Medical Genetics UBC & Seattle Exchange, Vanocuver, BC, Canada May 15, 2009. Presenter: **Mahmutoglu S.** Authors: Mahmutoglu S, Stockler S.

Jan 2009 **Collaborator.** Title: Saving the brain under attack. Advances in Pediatrics Department of Pediatrics, UBC, British Columbia Children's Hospital, Vancouver, BC, Canada. Authors: **Mahmutoglu S**, J. Hukin.

Sep 2008 **Principal Author.** Title: Lipopigments in non-neuronal cells and their diagnostic role in NCL. Neurometabolic Rounds, Department of Pediatrics, UBC, British Columbia Children's Hospital, Vancouver, BC, Canada. Presenter: **Mahmutoglu S.**

Mar 2008 **Principal Author.** Title: Is there a treatable cause of cerebral palsy?. Pediatric Neuroscience Research Rounds, Department of Pediatrics, UBC, British Columbia Children's Hospital, Vancouver BC, Canada. Presenter: **Mahmutoglu S.**

Oct 2006 **Collaborator.** Title: Adrenoleukodystrophy - windows of opportunity for diagnosis and treatment of a multifaceted disease. Advances in Pediatrics, Department of Pediatrics, British Columbia Children' Hospital, UBC, Vancouver, BC, Canada. Authors: **Mahmutoglu S**, Lillquist Y, Stockler S.

Invited Lectures and Presentations

July 2018	**Invited Speaker.** Title: Outcome of inherited Neurotransmitter Disorders. Metabolic Rounds, The Hospital for Sick Children, Toronto, Ontario Canada. July 6, 2018
June 2017	**Invited Presenter.** Title: Genetic epilepsies. Pediatric Epilepsy Fellowship Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA. June 12, 2017
May 2017	**Invited Presenter** Title: Metabolic epilepsy. Pediatric Epilepsy Fellowship Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA. May 8, 2017
April 2017	**Invited Presenter.** Title: How to diagnose rare genetic causes of epilepsy? Telegraf, Nationwide Telehealth for Genetic Residents and Fellows, CCMG organized rounds, Canada. April 21, 2017
April 2017	**Invited Presenter.** Title: Neonatal metabolic and vitamin responsive epilepsy. Pediatric Epilepsy Fellowship Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA. April 3, 2017
March 2017	**Invited speaker.** Title: Epilepsy Genetics Grand Rounds- How to diagnose rare genetic causes of epilepsy? Webcast presentation and youtube recording (https://youtu.be/Ckl_4FDBW7c). March 30, 2017
Aug 2016	**Invited Speaker.** Title: Creatine deficiency syndromes: phenotype, genotype and treatment outcomes. Metabolic Rounds, The Hospital for Sick Children, Toronto, Ontario Canada. August 5, 2016
Oct 2015	**Invited Speaker**. Title: Metabolic approach to movement disorder. Metabolic Rounds, The Hospital for Sick Children, Toronto, Ontario Canada. October 2, 2015
Jan 2015	**Invited Speaker**. Title: Diagnostic yield of genetic testing in epileptic encephalopathy in childhood. Neurology Grand Rounds, The Hospital for Sick Children, Toronto, Ontario Canada. January 8, 2015
Nov 2014	**Invited Speaker**. Title: Diagnostic yield of genetic testing in epileptic encephalopathy in childhood. Genetic Grand Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada, November 13, 2014.
Aug 2014	**Invited Speaker**. Title: Genetic white matter disorders. Metabolic Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada.
May 2014	**Invited speaker.** Title: What the rheumatologist needs to know about the MSK manifestations of MPS. Paediatric Rheumatology Academic Half-Day. University of Toronto, Toronto, Ontario, CA.
Feb 2014	**Invited Key Note Speaker**. Title: Guanidinoacetate methyltransferase deficiency: first steps to newborn screening for a treatable neurometabolic disease. International Rare Disease Day 2014, Toronto, Ontario, Canada, February 27, 2014.

May 2013 **Invited Speaker**, Title: Mimickers of HIE in term newborns: with focus on diagnosis and treatment. Neonatology Rounds. The Hospital for Sick Children, Toronto, Ontario, Canada, May 14, 2013.

Apr 2013 **Invited Speaker**, Title: Neonatal encephalopathy: identification of underlying causes by next generation sequencing. Neonatal Intensive Care Unit Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada, April 22, 2013.

Feb 2013 **Invited Speaker.** Title: Inherited metabolic disorders presenting with seizures in infants and children. Metabolic Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada.

Jul 2013 **Invited Speaker.** Title: Developmental delay and inherited metabolic disorders. Metabolic Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada.

May 2013 **Invited Speaker.** Title: Mimickers of HIE in term newborns: with focus on diagnosis and treatment. Neonatology Rounds. The Hospital for Sick Children, Toronto, Ontario, Canada.

Apr 2013 **Invited Speaker**. Title: Neonatal encephalopathy: identification of underlying causes by next generation sequencing. Neonatal Intensive Care Unit Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada.

Nov 2012 **Invited Speaker.** Title: Metabolic disorders presenting with seizures in childhood. DPLM Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada.

Aug 2012 **Invited Speaker**, Title: Inborn errors of metabolic disorders presenting with seizures in neonates and infants. Metabolic Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada.

Feb 2012 **Invited Speaker.** Inborn errors of metabolic disorders presenting with epilepsy. Neurology Grand Rounds, The Hospital for Sick Children, Toronto, Ontario, Canada.

Jan 2011 **Invited Speaker.** Title: GLUT1 deficiency. Molecular Genetics Laboratory Rounds, British Columbia Children's Hospital, Vancouver, British Columbia, Canada.

Mar 2010 **Invited Speaker.** Title: Two cases with GAMT deficiency in BC; treatment and monitoring. Biochemical Diseases Clinic Academic Rounds, British Columbia Children's Hospital, Vancouver, British Columbia, Canada.

Nov 2009 **Invited Speaker.** Title: A female patient with SLC6A8 deficiency. Biochemical Diseases Clinic Academic Rounds, British Columbia Children's Hospital, Vancouver, British Columbia, Canada.

Other Presentations- Abstracts- Poster Presentations

August 2019 **Senior Responsible Author.** Poster presentation. Urine Creatine Panel for the Diagnosis of Creatine Deficiency Disorders and its Diagnostic Yield. Authors: Shalini Bahl*, Lauren MacNeil, Andreas Schulze, **Saadet Mercimek-Andrews**. Trainee presentation. SSURE Research Day, August 2019, Toronto, Canada.

March 2019 **Senior Responsible Author.** Poster Presentation. Title: Phenotypic and genotypic spectrum of mitochondrial aminoacyl-tRNA synthetase deficiencies

from a single clinic. Authors: Al Balushi A*, Matviychuk D, Jobling R, Blaser S, Mercimek-Andrews S. Clinical and Metabolic Divisional Research Day. Trainee presentation.

March 2019 **Senior Responsible Author.** Poster Presentation. Title: High diagnostic yield of direct Sanger sequencing in the diagnosis of neuronal ceroid lipofuscinoses. Authors: Jilani A*, Matviychuk D, Blaser S, Dyack S, Mathieu J, Prasad N, Prasad C, Kyriakopoulou L, Mercimek-Andrews S. Clinical and Metabolic Divisional Research Day. Trainee presentation.

May 2019 **Senior Responsible Author.** Poster Presentation. Title: Genetics landscape of pediatric neurometabolic leukodystrophies. Authors: Damseh N*, Inbar-Feigenberg M, Blaser S, Mercimek-Andrews S. Clinical and Metabolic Divisional Research Day. Trainee presentation.

August 2018 **Senior Responsible Author.** Poster presentation. Diagnostic Yield of Neuronal Ceroid Lipofuscinoses Molecular Genetic Tests: Phenotype and Genotype of Neuronal Ceroid Lipofuscinoses. Authors: Jilani A*, Matviychuk D*, Blaser S, Dyack S, Mathieu J, Prasad N, Prasad C, Kyriakopoulou L, **Mercimek-Andrews** S. Trainee presentation. SSURE Research Day, August 2018, Toronto, Canada.

May 2017 **Senior Responsible Author**. Characterization of aldh7a1 knock-out zebrafish using CRISPR-Cas9 technology. Genetics & Genome Biology Program Annual Retreat, May 3, 2017, Toronto, Canada. Presenter Bullivant G*. Authors: Cao F, Fernandez M, Jia ZP, Wen XY, Dowling JJ, Salomons G, **Mercimek-Mahmutoglu S**. Trainee presentation.

Nov 2012 **Senior Responsible Author**. Title: A retrospective study to evaluate underlying etiology in patients with intractable epilepsy at The Hospital for Sick Children. SickKids Research Retreat Day, Toronto, Ontario, Canada, November 2012. Authors: Chan J*, Zak M, Minassian B, **Mercimek-Mahmutoglu S**. Research student presentation.

Nov 2012 **Senior Responsible Author**. Title: A Retrospective series of patients with myoclonic epilepsy: The First Step Towards Improved Diagnostics for a Challenging Group of Disorders. Research Retreat SickKids Hospital, Toronto, Ontario, Canada, November 2012. Authors: Rodan L*, Martyn F, Zak M, Chan J, Minassian B, **Mercimek-Mahmutoglu S**. Trainee presentation.

Aug 2012 **Senior Responsible Author**. Title: Retrospective review of patients with myoclonic epilepsy: First steps to determine aetiology using next generation exome/genome sequencing. The Hospital for Sick Children Summer Research Program, Toronto, Ontario, Canada, August 2012, Authors: Martyn F*, **Mahmutoglu S**. Research student presentation.

Mar 2012 **Senior Responsible Author**. Title: X-linked adrenoleukodystrophy with atypical neuroimaging features: case report. Clinical and Metabolic Genetics 1st Annual Research Day, March 2, 2012, The Hospital for Sick Children, Toronto, Ontario, Canada. Authors: Tran C*, Brandsema J, Widjaja E, Donner E, Raiman J, Blaser S, **Mercimek-Mahmutoglu S**. Trainee presentation.

Jun 2011 **Senior Responsible Author**. Title: Two different clinical phenotype in two siblings with 3-methylglutaconic aciduria type I. Student Research Forum, June 16, 2011, Vancouver, British Columbia, Canada. Authors: Tucker T*, Casey B, **Mercimek-Mahmutoglu S**. Trainee presentation.

Jun 2010 **Senior Responsible Author**. Title: Juvenile neuronal lipofuscinosis in a patient with compound heterozygous CLN3 mutations: a 9 year follow-up. 2010 Student Research Forum, June 17, 2010, Vancouver, British Columbia, Canada. Authors: Al-Thihli K*, Carey Matsuba, Roland E, **Mercimek-Mahmutoglu S**. Trainee presentation.

Jun 2006 **Principal Author**. Title: GAMT deficiency: features, treatment, and outcome in a Newly recognized cerebral creatine deficiency syndrome. University of British Columbia C&W, Student Research Forum, June 18, 2006, Vancouver, British Columbia, Canada. Authors: **Mercimek-Mahmutoglu S**, Stockler-Ipsiroglu S.

H. Teaching and Design

Please see the Teaching and Educational Report for full details.

1. Innovations and Development in Teaching and Education.

1.1. Biochemical Genetics and Clinical Metabolic Genetics Fellowship Programs: I am the Biochemical Genetics Fellowship and Clinical Metabolic Genetics Program director. Biochemical Genetics Fellowship program is a Canadian College of Medical Geneticist (CCMG) accredited program. The program re-accreditation application process was completed in 2014 and I prepared application document for re-accreditation as part of Medical Genetics Training Program. CCMG Accreditations Committee reviewed the document and arranged site visit in 2015 with no specific concerns, challenges or questions for the Biochemical Genetics Fellowship program.

I developed Biochemical Genetics Fellowship Program Objectives. I developed elective program for Biochemical Genetics and Clinical Metabolic Genetics Fellows.

I am co-developer of the Metabolic Fellowship Teaching rounds, which is a weekly teaching round to review topics for inherited metabolic disorders as well as Journal club.

1.2. Teaching Curriculum Development

- 1.2.1. Medical Genetics Residency Program Metabolic Genetics Curriculum Coordinator. I developed 3-year curriculum and arranged lecturer for Current Topics in Medical Genetics, Academic Half-Day, Medical Genetics Residency Program.
- 1.2.2. Medical Genetics Residency Program Epidemiology and Critical Appraisal Curriculum Coordinator. I developed 3-year curriculum and arranged lecturer for Current Topics in Medical Genetics, Academic Half-Day, Medical Genetics Residency Program.
- 1.2.3. Paediatric Neurology Training Program Teaching Curriculum Developer. I developed 2-year curriculum and arranged lecturer for Paediatric Neurology Teaching Rounds.
- 1.2.4. Paediatric Residency Program PeRLS Academic Half Day Curriculum Co-Developer together with Dr. Kronick.

1.3. I developed a systematic case based teaching style for known metabolic genetic diagnosis (e.g. PDE, GLUT1 deficiency, CDS, PA, MMA, MSUD, diagnosis, clinical features, genetic defect, pathways, biomarkers, acute and chronic treatment) or approach to the major symptoms related to biochemical and metabolic genetics including liver disease, cholestasis, hypotonia, acidosis, seizures, hypoglycaemia, developmental delay, encephalopathy, lysosomal storage disorders. The trainees include PGY2-5 Medical Genetics Residents, Pediatric Residents, Neurology Residents, Metabolic and Clinical Genetics Fellows, National Elective Residents (e.g. McGill, Ottawa), National Elective Medical Students, IMG Observers. I provide key references to read which are discussed 2-3 hours/week as formal teaching.

2. Summary of Teaching and Education

Undergraduate

May 2018 **Station Examiner**, Clinical Skills 2 Final Objectively Structured Clinical Examination, Women's College Hospital Education Office, MD Program, Faculty of Medicine, UFT, Undergraduate 2nd year medical students, Toronto, Ontario, CA. May 28, 2018 (3hrs)

I supervised an OSCE station, asked questions and assessed 20 students for the station.

April 4, 2017 **Section Leader.** MGY470, Medical genetics ethics, Molecular Genetics Undergraduate Courses, University of Toronto, Canada.

Mar 3, 2014 **Station Examiner**, Integrated Objectively Structured Clinical Examination, University of Toronto/Centre for the Evaluation of Health Professionals Educated Abroad (CEHPEA), Department of Paediatrics, Undergraduate 3rd year medical students, Toronto, Ontario, CA.

I supervised an OSCE station, asked questions and assessed 12 students for the station.

Aug 27, 2013 **Station Examiner**, Integrated Objectively Structured Clinical Examination, University of Toronto/Centre for the Evaluation of Health Professionals Educated Abroad (CEHPEA), Department of Paediatrics, Undergraduate 3rd year medical students, Toronto, Ontario, CA.

I supervised an OSCE station, asked questions and assessed 12 students for the station.

Mar 4, 2013 Station **Examiner**, Integrated Objectively Structured Clinical Examination, University of Toronto/Centre for the Evaluation of Health Professionals Educated Abroad (CEHPEA), Department of Paediatrics, Undergraduate 3rd year medical students, Toronto, Ontario, CA.

I supervised an OSCE station, asked questions and assessed 12 students for the station.

Oct 24, 2012 **Seminar Leader**, MSB-LABS Genetics Seminars: Mechanisms, Manifestations & Management of Disease (MMMD) - Weeks 1-9, University of Toronto, Department of Paediatrics, Undergraduate Medical School, Toronto, Ontario, CA.

I led the seminar for a group of approximately 10 students for genetic cases as an interactive session for 3 hours.

Oct 23, 2011 **Seminar Leader**, MSB-LABS Genetics Seminars: Mechanisms, Manifestations & Management of Disease (MMMD) - Weeks 1-9, University of Toronto, Department of Paediatrics, Undergraduate, Toronto, Ontario, CA.

I led the seminar for a group of approximately 10 students for genetic cases as an interactive session for 3 hours.

Graduate

December 2018 **Lecturer.** Title: Title: Childhood epilepsy: from molecular defect to phenotype burden. Concepts of Clinical Genetics Course (MMG-1226Y), MSc Genetic

	Counseling Students. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, Canada.
March 2018	**Lecturer.** Title: Title: Childhood epilepsy: from molecular defect to phenotype burden. Concepts of Clinical Genetics Course (MMG-1226Y), MSc Genetic Counseling Students. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, Canada.
Nov 2017	**Lecturer.** Title: Leukodystrophy, peroxisomal disorders, disorders of copper transport. Concepts in Clinical Genetics (MMG-1226Y). Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
March 2017	**Lecturer.** Title: Title: Childhood epilepsy: from molecular defect to phenotype burden. Concepts of Clinical Genetics Course (MMG-1226Y), MSc Genetic Counseling Students. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, Canada.
Oct 2016	**Lecturer.** Title: Leukodystrophy, peroxisomal disorders, disorders of copper transport. Concepts in Clinical Genetics (MMG-1226Y). Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Mar 2016	**Lecturer.** Title: Title: Childhood epilepsy: from molecular defect to phenotype burden. Concepts of Clinical Genetics Course (MMG-1226Y), MSc Genetic Counseling Students. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, Canada.
Oct 2015	**Lecturer.** Title: Leukodystrophy, peroxisomal disorders, disorders of copper transport. Concepts in Clinical Genetics (MMG-1226Y). Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Jan 2015	**Lecturer.** Title: Title: Childhood epilepsy: from molecular defect to phenotype burden. Concepts of Clinical Genetics Course (MMG-1226Y), MSc Genetic Counseling Students. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, Canada.
Dec 2014	**Lecturer.** Title: Inborn errors of amino acid catabolism organic acidopathies. Master's in Genetic Counselling, Concepts of Clinical Genetics Course (MMG-1226Y). University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Oct 2014	**Lecturer.** Title: Leukodystrophy, peroxisomal disorders, disorders of copper transport. Concepts in Clinical Genetics (MMG-1226Y). Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Feb 2014	**Lecturer.** Title: Childhood epilepsy: from molecular defect to phenotype burden. Master's in Genetic Counselling, Concepts of Clinical Genetics Course (MMG-1226Y). University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.

Oct 2013	**Lecturer.** Title: Leukodystrophy, peroxisomal disorders, disorders of copper transport. Concepts in Clinical Genetics (MMG-1226Y). Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Jan 2013	**Lecturer.** Title: Childhood epilepsy: from molecular defect to phenotype burden. Master's in Genetic Counselling, Concepts of Clinical Genetics Course (MMG-1226Y). Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Jan 2012	**Lecturer.** Title: Childhood epilepsy: from molecular defect to phenotype burden. Concepts of Clinical Genetics Course (MMG-1226Y), MSc Genetic Counseling Students. Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.

Postgraduate

Apr 2018	**Station Examiner,** April 20, 2018. Objectively Structured Clinical Examination, Pediatric Residency Training Program, University of Toronto, Toronto, Ontario, CA
Jun 2017	**Lecturer,** June 12, 2017. Title: Genetic epilepsies. Pediatric Epilepsy Fellowship Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA.
May 2017	**Lecturer,** May 8, 2017. Title: Metabolic epilepsy. Pediatric Epilepsy Fellowship Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA.
Apr 2017	**Station Examiner,** April 28, 2017. Objectively Structured Clinical Examination, Pediatric Residency Training Program, University of Toronto, Toronto, Ontario, CA
Apr 2017	**Lecturer,** April 21, 2017. Title: How to diagnose rare genetic causes of epilepsy? Telegraf, Nationwide Telehealth for Genetic Residents and Fellows, CCMG organized rounds, Canada.
Apr 2017	**Lecturer,** April 3, 2017. Title: Neonatal metabolic and vitamin responsive epilepsy. Pediatric Epilepsy Fellowship Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA.
Sep 2016	**Lecturer.** Title: GLUT1 and Pyridoxine Dependent Epilepsy. Pediatric Neurology Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA.
Apr 2016	**Station Examiner,** Objectively Structured Clinical Examination, Medical Genetics Training Program, University of Toronto, Toronto, Ontario, CA
May-Jun 2016	**Written and OSCE examiner**, Biochemical Genetics Fellowship Training CCMG exam preparation, University of Toronto, Toronto, Ontario, CA
Feb 2016	**Lecturer.** Title: Metabolic Genetics Pediatrics Royal College Exam Preparation. Pediatric Residency Training. University of Toronto/The Hospital for Sick Children, Department of Pediatrics. Toronto, Ontario, CA.

Jan 2016	**Lecturer.** Title: Developmental delay and developmental regression and approach to inherited metabolic disorders. Paediatric Residents Academic Half-Day Teaching, PeRLS. University of Toronto/The Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Dec 2015	**Lecturer.** Title: Mimickers of hypoxic-ischemic encephalopathy. Neonatal Neurology Training Program Teaching Rounds. Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
May 2015	**Lecturer.** Title: Metabolic approaches to movement disorder. Pediatric Neurology Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA.
Apr 2015	**Lecturer.** Title: Epilepsy and inborn errors of metabolism. Pediatric Neurology Residency Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA.
Apr 2015	**Lecturer.** Title: Metabolic disorders involving white matter. Pediatric Neurology Residency Training Program Teaching Rounds. University of Toronto/The Hospital for Sick Children, Division of Neurology. Toronto, Ontario, CA.
Jan 2015	**Lecturer.** Title: Leukoencephalopathy & leukodystrophy. Current Topics in Medical Genetics, Medical Genetics Residency Program. University of Toronto/The Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
May 2014	**Lecturer.** Title: Genetics disorders of white matter. Adult Neurology Residency Training Program Teaching Rounds. University of Toronto. Toronto Western Hospital, Toronto, Ontario, CA.
Feb 2014	**Lecturer.** Title: Metabolic Approach to Seizures. Current Topics in Medical Genetics, Academic Half-Day, Medical Genetics Residency Program. University of Toronto/The Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Feb 2014	**Co-Lecturer.** Title: Approaches to Common Metabolic Consults on the NICU and the challenges of Newborn Screening. Neonatal-Perinatal Fellows Academic Half Day. University of Toronto/The Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Oct 2013	**Lecturer.** Title: Genetics disorders of white matter-II. Pediatric Neurology Residency Training Program Teaching Rounds. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Sep 2013	**Lecturer.** Title: Genetics disorders of white matter-I. Pediatric Neurology Residency Training Program Teaching Rounds. The Hospital for Sick Children, Department of Paediatrics, Toronto, Ontario, CA.
Apr 2013	**Lecturer.** Title: Developmental delay and inherited metabolic disorders. Paediatric Residents Academic Half-Day Teaching, PeRLS. University of Toronto/The Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.
Jan 2013	**Lecturer.** Title: Hyperammonemia: metabolic approach. Current Topics in Medical Genetics, Academic Half-Day, Medical Genetics Residency Program.

University of Toronto/The Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.

Jan 2013 **Lecturer.** Title: Metabolic approach to dysmorphic features other than lysosomal storage disorders. Current Topics in Medical Genetics, Academic Half-Day, Medical Genetics Residency Program. University of Toronto/The Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.

Oct 2012 Co-**Lecturer.** Title: Hyperammonema and Acute Encephalopathy. Pediatric Residents Academic Half-Day Teaching, PeRLS. Lecturer. University of Toronto/Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.

Jan 2012 **Lecturer.** Title: Glycogen storage disorders and approach to hypoglycemia. Current Topics in Medical Genetics. Lecturer. University of Toronto/The Hospital for Sick Children, Department of Paediatrics. Toronto, Ontario, CA.

Other Health Professional

Registered Metabolic Nurse, Metabolic Nurse Practitioner, Metabolic Dietician and Genetic Counsellor:

- Case based teaching for inherited metabolic disorders during outpatient clinics or inpatient service.

I. Research Supervision

1. Multilevel Education

Undergraduate Education

2019 **Primary Supervisor**. Shalini Bahl. Current Position: Research student, University of Toronto. Urine creatine panel and its diagnostic yield.

2017-Present **Primary Supervisor**. Yulia Kungurova. Current Position: Research Student, University of Toronto. Aldh7a1 zebrafish project.

2016-2017 **Primary Supervisor**. Garrett Bullivant. Current Position: Research Student, University of Toronto. Aldh7a1 zebrafish, case report, cobalamin retrospective review study.

2016 **Primary Supervisor**. Ece Otazca. Position: Research Student, University of Montreal. Alexander retrospective review study.

2015-2016 **Primary Supervisor**. Yannay Khaikin. Current Position: Research Student, University of Toronto. STXBP1 related epileptic encephalopathy: literature review, book chapter.

2015-Present **Primary Supervisor**, Theodora Bruun. Current Position: Research Student, University of Toronto. *Neonatal epileptic encephalopathy: identification of underlying metabolic epilepsies using next generation exome sequencing*. Awards: 2015 Starbucks Studentship Award, Poster award-SSURE Symposium Day.

2013 **Primary Supervisor**, Elizabeth Imhof. Current Position: MSc. Student, University of Calgary. *Is low creatine kinase a nonspecific-screening marker for creatine deficiency syndromes?* Completed 2013.

2012	**Primary Supervisor**, Fredrick Martyn. Current Position: Medical Student, Boston University. *Retrospective review of patients with myoclonic epilepsy: First steps to determine aetiology using next generation exome/genome sequencing.* Awards: 2012 Starbucks Studentship Award. Completed 2012
2012	**Primary Supervisor**, Jonathan Chan. Current Position: Graduate Student. *Myoclonic Epilepsy: generating a database to identify treatable causes*. Completed 2012.
2012-2013	**Primary Supervisor**, Priscilla Tang. Current Position: MSc candidate, University of Toronto. *Intractable epilepsy: retrospective review of all patients with intractable epilepsy at The Hospital or Sick Children to generate a database*. Completed 2013.
2012-2013	**Primary Supervisor**, Nellie Allam, Current Position: Unknown. *Prospective inherited disorders of neurotransmitter analysis for the diagnosis of treatable inherited disorders of neurotransmitter metabolism.* Completed 2013.
2012-2013	**Primary Supervisor**, Carmen Fang. Current Position: Unknown. *Global Developmental Delay (GDD)*. Completed 2013.
2012-2013	**Primary Supervisor**, Anum Karim. Current Position: Client Services, Pharma Communications Group Inc. *Retrospective Review of Cerbrospinal Fluid Neurotransmitter Analysis for the Diagnosis of Treatable Inherited Disorder Neurotransmitter Metabolism*. Completed 2013.
2012-2014	**Primary Supervisor**, Amrita Machado. Current Position: Coordinator, Appletree Medical Group. *Is low creatine kinase a nonspecific-screening marker for creatine deficiency syndromes?* Completed 2014.
2012-2014	**Primary Supervisor**, Sarah Sidky. Current Position: Medical Student, University of Limerick. *Retrospective Review of Cerbrospinal Fluid Neurotransmitter Analysis for the Diagnosis of Treatable Inherited Disorder Neurotransmitter Metabolism*.
2012-2015	**Primary Supervisor**, Jaina Patel. Current Position: Medical Student, Ireland. *Retrospective review of the patients with X-linked adrenoleukodystrophy at The Hospital for Sick Children.*

Graduate MD

2018-2019	**Primary Supervisor**. Abdulhakim Jilani. Current Position: Research student, University of Toronto. High yield of direct sequencing in neuronal ceroid lipofuscinosis.
2014- 2016	**Primary Supervisor,** Sarah Sidky. Current Position: Medical Student, University of Limerick. Retrospective registry of patients with creatine transporter deficiency using Research Electronic Data Capture (REDCap) software for evaluation of long-term treatment outcome of patients.
2015-2016	**Primary Supervisor**, Anath Lionel, Current Position: Medical Student, University of Toronto, MED23 caused epileptic encephalopathy: case report and review of the literature.
2015-2016	**Primary Supervisor**, Jaina Patel. Current Position: Medical School, University of

Postgraduate MD

2018-2019	**Primary Supervisor**. Gregory Costain. Current Position: Metabolic Genetics Resident, University of Toronto. Epilepsy and inherited metabolic disorders and clinical diagnostic yield of next generation sequencing in epilepsy.
2018-2019	**Primary Supervisor**. Graeme Nimmo. Current Position: Metabolic Genetics Fellow. University of Toronto. Genotypes and phenotypes of GLUT1 deficiency.
2018-2019	**Primary Supervisor**. Nadirah Damseh. Current Position: Metabolic Genetics Fellow, University of Toronto. Metabolic leukodystrophies: review of patients diagnosed in our center.
2018-2019	**Primary Supervisor**. Aaisha Al Balushi. Current Position: Metabolic Genetics Fellow, University of Toronto. Genotypes and phenotypes of tRNA defects.
2017-2018	**Primary Supervisor**. Bushra Afroz. Current Position: Metabolic Genetics Fellow, University of Toronto. Various Case reports.
2017-2018	**Primary Supervisor**. Danielle Veenma. Current Position: Clinical Genetics Fellow, University of Toronto. Case report for DNAJC12 patients.
2017-2018	**Primary Supervisor**. Fady Shmouni. Current Position: Metabolic Genetics Fellow, University of Toronto. Case report for CD320 patients.
2016-2017	**Primary Supervisor**. Graeme Nimmo. Current Position: Metabolic Genetics Fellow, University of Toronto. Case report for riboflavin transporter deficiency.
2016-2017	**Primary Supervisor**. Lizbeth Mellin. Current Position: Metabolic Genetics Fellow, University of Toronto. Cobalamin retrospective review study.
2015-2017	**Primary Supervisor**. Rebecca Barmherzig. Current Position: Pediatric Neurology Resident, University of Toronto. Salla disease case report.
2015-2017	**Primary Supervisor,** Saleh Al Banyan, Current Position: Medical Genetics Residency Program PGY2, University of Toronto. BCPA31 caused intellectual disability: case report and review of the literature.
2015-2016	**Primary Supervisor**, Dr. Michal Inbar-Feigenberg. Current Position: Metabolic Physician, The Hospital for Sick Children. *Phenotypic and biochemical features of pyruvate dehydrogenase complex deficiency: a retrospective cohort study at The Hospital for Sick Children/Retrospective review of all patients diagnosed with pyruvate dehydrogenase complex deficiency at The Hospital for Sick Children*
2014-2016	**Primary Supervisor,** Dr. Amal Al-Teneiji. Position: CCMG Biochemical Genetics Fellow, The Hospital for Sick Children. *Whole exome sequencing data analysis for mitochondrial disorders/Case report preparation/ Retrospective review of patients with pyridoxine dependent epilepsy diagnosed at The Hospital for Sick Children/ Retrospective review of patients with congenital disorders of glycosylation diagnosed at The Hospital for Sick Children*
2013	**Primary Supervisor**, Dr. Rebekah Jobling. Current Position: Resident, University of Toronto. *Low cerebrospinal fluid catecolamine, serotonin and 5-methyltetrahydrofolate levels in a patient with infantile onset CBLG deficiency*

2013-2016 **Primary Supervisor**, Dr. Christel Tran. Current Position: Metabolic Geneticist, Centre Hospitalier Universitaire Vaudois CHUV. *Retrospective review of the patients with X-linked adrenoleukodystrophy at The Hospital for Sick Children*. Completed 2015.

2013-2014 **Primary Supervisor**, Dr. Andrea Guerin. Current Position: Assistant Professor and Medical Geneticist, Hotel Dieu Hospital. *Atypical clinical response and normal CSF neurotransmitters in a patient with PNPO deficiency (case report)*. Completed 2014.

2012-2013 **Primary Supervisor**, Dr. Anne Roscher. Current Position: Doctor, Medical University of Vienna. *Retrospective review of all patients with glycogen storage disease type VI and IX at the Hospital for Sick Children,* Completed 2013.

2012-2015 **Primary Supervisor**, Dr. Enas Nasr. Current Position: Clinical Biochemical Geneticist, Dubai, United Arab Emirates. *Is low creatine kinase a nonspecific-screening marker for creatine deficiency syndromes?* Completed 2015.

2011 **Primary Supervisor**, Dr. Tracy Tucker. Current Position: Clinical Scientist, British Columbia Cancer Agency.*3-methylglutaconic aciduria type I caused a novel intragenic deletion, case report.* Completed 2011.

2011 **Primary Supervisor**, Dr. Joseph Ting. Current Position: Neonatologist, Children's and Women's Hospitals of British Columbia. *Blueberry muffin rash and thrombocytopenia in a newborn with mucolipidosis type II (I-cell disease) masquerading as congenital infections.* Completed 2011

2011-2012 **Primary Supervisor**, Dr. Lance Rodan. Current Position: Clinical Fellow, Children's Hospital Boston. *A Retrospective series of patients with myoclonic epilepsy: The First Step Towards Improved Diagnostics for a Challenging Group of Disorders.* Completed 2012.

2010-2011 **Primary Supervisor**, Dr. Mary Dunbar. Current Position: Paediatric Resident, University of British Columbia. *A new GAMT patient and its long term treatment outcome (case report).* Completed 2011.

2010-2011 **Primary Supervisor**, Dr. Karen Neiderhoffer. Current Position: Medical Doctor, University of British Columbia. Two novel compound heterozygous mutations in the Twinkle Helicase (C100rf2) gene causing early onset mitochondrial encephalomyopathy. Completed 2011.

2009-2011 **Primary Supervisor**, Dr. Khalid Al-Thihli. Current Position: Acting Head of the Genetic Department/Consultant Clinical and Biochemical Geneticist, Oman Hospital and Health Care. *Juvenile neuronal lipofuscinosis in a patient with compound heterozygous CLN3 mutations: a 9 year follow-up.* Completed 2011.

2009-2011 **Primary Supervisor**, Catharine Brunel-Guitton. Current Position: Doctor, Saint-Justine University Hospital, Health and Social Services Agency Laurentians. *Late onset nonketotic hyperglycinemia caused by a novel homozygous missense mutation in the GLDC gene.* Completed 2011.

Other

2015-2017 **Primary Supervisor**. Stacy Hewson. Current Position: Genetic Counselor, Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto. Prevalence of genetic diagnoses and glucose transporter 1

	deficiency in patients with drug resistant epilepsy on the ketogenic diet. Variable expressivity of a likely pathogenic variant in KCNQ2 in a three-generation pedigree presenting with intellectual disability with childhood onset seizures.
2018-2019	**Primary Supervisor**. Diana Matviychuk. Current Position: Genetic Counselor, Clinical and Metabolic Genetics, Department of Pediatrics, University of Toronto. Various publications and poster presentations in medical genetics and metabolic meetings.

J. Creative Professional Activities

Please see Creative Professional Activities Dossier for full details