

University of Calgary
 Cumming School of Medicine
CURRICULUM VITAE

I. BIOGRAPHICAL DATA

Name	Ping-Yee Billie Au
Address	2016 38 Ave SW Calgary, Alberta T2T 2K4
Telephone	Office (403) 955-7246 Mobile (403) 969-3332
Present rank	Clinical Associate Professor
Department	Pediatrics/Genetics
Faculty	Medicine
Institution	Alberta Children's Hospital

II. ACADEMIC RECORD

Final degree	MD
Date completed	2009
Specialty	Medicine
Institution/City/Country	University of Toronto/Toronto/Canada
i. Undergraduate	BSc (Hons), Biochemistry, Queen's University, Kingston, Ontario.
ii. Special professional	RCPSC Residency Medical Genetics, University of Calgary, Calgary, Alberta.
iii. Graduate	PhD, Cellular and Molecular Biology, Dept. of Medical Biophysics, University of Toronto, Toronto, Ontario.
iv. Licensure, Certification, Boards	FRCP(C), Medical Genetics. FCCMG, Fellow, Canadian College of Medical Geneticists.

III. AWARDS AND DISTINCTIONS**Scholarships:**

2007–2008	Robert and Francine Ruggles Family Graduate Fellowship
2007–2009	Vision Science Fund Award
2000–2001	Queen's University Arts and Science Undergraduate Society Scholarship
1998	Heart and Stroke Foundation of Ontario, John D. Schultz Scholar Research Studentship
1996–1997	Queen's University Trillium Scholarship

Awards:

2025	Resident Teaching Award “Favorite Teacher of the Year”, Medical Genetics
2024	Cumming School of Medicine, University of Calgary PGME Teaching Award
2014	Best Resident Presentation Award (2 nd place). Rocky Mountain Basic Science Symposia.
2013	Canadian College of Medical Geneticists Linda Stevens Memorial Fund
2013	April PARA Resident of the Month Award

IV. ACADEMIC APPOINTMENTS

2025–present	Clinical Associate Professor, Medical Genetics, Cumming School of Medicine, University of Calgary.
2018–2025	Clinical Assistant Professor, Medical Genetics, Cumming School of Medicine, University of Calgary.
2015–present	Clinical Geneticist, Section of Clinical Genetics, Department of Pediatrics, Alberta Health Services.

V. EDUCATIONAL ACTIVITIES

Post Graduate Education / Medical Education

Jul. 2022–present	Program director, Suchowersky/Eagle Neurogenetics fellowship
Oct. 2021–present	RCPSC Genetics and Genomics Residency Competence Committee member
Mar. 2021–present	Supervisory committee member (Medical Science) for PhD candidate Emma Gillese.
Sep. 2019–present	RCPSC Genetics and Genomics Residency program Committee member
2019–present	Supervisory committee member (Medical Genetics) for PhD candidate Vladamir Avramovic.
Nov. 2024	MSc. Thesis Examiner for candidate Omoseni Idowu (Cardiovascular / Respiratory Sciences), University of Calgary.
Feb. 2024	Genetics core month lecture <u>VUS interpretation(when ACMG criteria fails you).</u>
2016–2024	Medical Genetics practice OSCE examiner.
Dec. 2023	Pediatric Psychiatry half day talk <u>Genetic neurodevelopmental syndrome overview.</u>

Jan. 2023	Genetics Core Month Lecture <u>Genetic Approach to Paroxysmal Movement Disorders.</u>
Jun. 2023	MSc. External Thesis Examiner for candidate Karen Kelm (Medical Sciences Pediatrics, University of Alberta).
2022–2023	Research Supervisor, Clinical Neurogenetics fellow Xiao-Ru Yang.
2020–2023	Gene panels and SNV interpretation 101 Lecture annually for first year residents.
Jan. 2022	Genetics Core Month Lecture <u>Overview of Genetic Therapies.</u>
2019–2021	Committee Member CCMG Molecular Genetics fellowship candidate Amanda Tyndall.
Feb. 2021	Genetics core month lecture <u>Genetics of Parkinsons Disease.</u>
2019–2020	Thesis proposal/Field of Study Examiner for PhD candidate (Neuroscience) Albert Isaacs.
Jan. 2020	Genetics Core Month Lecturer <u>Approach to Craniosynostosis.</u>
Sep. 2019	Pediatric Neurology practice OSCE examiner.
Jan. 2019	Genetics Core Month Lecturer <u>Genetic Approach to Epilepsy and Genetic Movement Disorder Overview.</u>
May 2018	CCMG Telegraf series <u>Approach to Genetics of Primary Dementias.</u>
2016–2018	Formal mentor for Pediatric Neurology Resident Momen Al-Momen.
2016–2017	Committee Member CCMG Molecular Genetics fellowship candidate Yanwei Xi.
2017	ACHRI Symposium poster judge.
Jan. 2016	Genetics Core Month Lecturer <u>Genetics of Dementia and Ribosomopathies.</u>
Mar. 2014	Speaker and participant <u>Genetics of Respiratory Chain Disorders.</u> Rocky Mountain Basic Science Symposia on Neurometabolics. Kananaskis, Alberta, Canada.
Feb. 2013	Pediatric Neurology Academic Half Day Guest Speaker <u>Approach to Inherited Leukodystrophy.</u> Alberta Children’s Hospital, University of Calgary.
2012–2013	Coordinator, Medical Genetics Wednesday Morning Resident Teaching. Department of Medical Genetics, Alberta Children’s Hospital, University of Calgary.
May 2010	Pediatrics Academic Half Day Speaker <u>Approach to Hypotonia.</u> Alberta Children’s Hospital, University of Calgary.

2009–2012 Speaker on various topics for Medical Genetics Core Teaching Month. Department of Medical Genetics, Alberta Children’s Hospital, University of Calgary.

Undergraduate Medical Education

2015–present UME Preceptor for Med440 AEBM Elective (5 students to date).

Jan. 2024 RIME Research Supervisor, Julia Zebak, Medical Student, University of Calgary.

2022–2023 AEBM Research supervisor, Kara Vanden Broek, Medical Student, University of Calgary.

2020–2023 UME “Clinical Genomics” Lecture for Course 6.

2018–2023 UME Course 6 Genetics Coordinator with Renee Perrier.

2011–2023 Small Group/Problem Based Learning preceptor for Course 6 (Integrated Clinical presentations related to Reproductive Medicine and Pediatrics Course, Medical Genetics module), Undergraduate Medical Education, University of Calgary.

2020–2021 Research supervisor, Aya Ebdalla, Medical Student, University of Calgary.

2018–2020 UME Clerkship “Clinical Genetics” Lecture (every 2 months).

Mar. 2013 Guest TA for CMMB 413 Undergraduate Human Genetics Course, Session on Careers in Medical Genetics, Department of Biological Sciences, University of Calgary.

2008 Research Proposal Reviewer. Comprehensive Research Experience for Medical Students (CREMS) Program. Faculty of Medicine, University of Toronto. Toronto, Ontario.

2007 Mock OSCE examiner. Faculty of Medicine, University of Toronto, Clinical Skills Mentorship Program.

2004–2006 Undergraduate Thesis Project Student Supervisor. Department of Medical Biophysics, University of Toronto.

Continuing Medical Education

Grand Rounds:

Dec. 2024 Functional Genomics Lab. Medical Genetics Grand Rounds, University of Calgary, Alberta, Canada.

Dec. 2024 Genetic Neurodevelopmental Disorders Initiative. Presentation for the Department of Pediatrics Innovation Grant at Pediatric Grand Rounds,

University of Calgary, Alberta, Canada.

Nov. 2012 [It's Time to SKI: New developments in Shprintzen-Goldberg Syndrome.](#) Medical Genetics Grand Rounds, University of Calgary, Alberta, Canada.

Dec. 2009 [Congenital Myasthenic Syndromes.](#) Pediatric Neurology Grand Rounds, University of Calgary, Alberta, Canada.

Medical Genetics Journal Club:

Mar. 2024 [Misattributed Parentage Identified Through Diagnostic Exome Sequencing: Frequency of Detection and Reporting Practices.](#) J Genet Couns. 2022;31:631–640.

May 2023 [A systematic Review of Fragile X Associated Neuropsychiatric Disorders.](#) J Neuropsychiatry Clin Neurosci 35:2,110-120.

Oct. 2021 [Phase 1-2 Trial of Antisense Oligonucleotide Tofersen for SOD1 ALS.](#) NEJM July 2020. 283:109-119.

Nov. 2020 [Predicting Functional Effects of Missense Variants in Voltage-Gated Sodium and Calcium Channels.](#) Science Translational Medicine. August 2020.

Mar. 2017 [Assessment of Incidental Findings In 232 Whole-Exome Sequences from the Baylor-Hopkins Center for Mendelian Genomics.](#) Genetics in Medicine. October 2015.

Jan. 2015 [A SWI/SNF Related Autism Syndrome Caused by De Novo Mutations in ADNP.](#) Nature Genetics. April 2014.

Oct. 2012 [Rate of De Novo Mutations and the Importance of Father's Age to Disease Risk.](#) Nature. Aug 2012. 488:471-5.

Nov. 2011 [Mirror Extreme BMI Phenotypes Associated with Gene Dosage at the Chromosome 16p11.2 Locus.](#) Nature. Aug 2011. 478:97-102.

CME for Pediatrics and other subspecialties:

Apr. 2024 [Neurogenetics Approach to Ataxia.](#) Movement Disorder Rounds. Dept of Clinical Neurosciences, University of Calgary. In person.

Sep. 2023 [Canadian Position Statement for Genetic Testing in Development Disorders and Local Initiatives.](#) Co-presenter Dr. Pamela Veale. Community Pediatrics CME Rounds. Virtual.

Apr. 2023 [Canadian Position Statement for Genetic Testing in Development Disorders and Local Initiatives.](#) Co-presenter Dr. Pamela Veale. Owerko Neurodevelopmental Clinical Rounds.

Feb. 2020 [Neurogenetics Testing in Alberta.](#) Pediatric Neurology Rounds, Alberta

Children's Hospital.

- May 2019 Genetic Evaluation – How Does This Contribute to Pediatric Health Care? Southern Alberta Community Paediatric Update, Alberta Children's Hospital, Calgary, Alberta.
- Sep. 2016 Update on Common Syndromes. Southern Alberta Community Paediatric Update, Delta Lodge at Kananaskis, Alberta, Canada.
- Sep. 2016 Approach to Investigation of Neuro-Developmental Concerns (Including Autism, Developmental Delay, New Syndromes Etc.). Southern Alberta Community Paediatric Update, Delta Lodge at Kananaskis, Alberta, Canada.

CME for Genetic counselors:

- May 2013 It's Time to SKI: New Developments in Shprintzen-Goldberg Syndrome and What We've Learned About Tgfbeta Related Disorders. Annual Alberta Medical Genetics In-Service for Genetic Counselors, Calgary, Alberta, Canada.
- Apr. 2013 ATM Carriers and Breast Cancer Risk. Presentation to Cancer Genetics group, Department of Medical Genetics, University of Calgary.
- Apr. 2013 Li Fraumeni Syndrome. Presentation to Cancer Genetics, Department of Medical Genetics, University of Calgary.
- Jun. 2012 Fragile X Premutation. Presentation to General Genetic Counseling, Department of Medical Genetics, University of Calgary.
- Mar. 2012 BRCA1/2 Expedited Testing. Molecular Genetics Lab Rounds, Molecular Diagnostic Lab, Alberta Children's Hospital.

VI. ADMINISTRATIVE RESPONSIBILITIES

Departmental: Pediatrics/Genetics

- 2023–present Member, Alberta Provincial Working Group for Genetics Episode in Connect Care.
- 2020–present Committee Member, APL Discipline Council Biochemical Genetics Standing Working Group.
- Sep. 2019 Committee Member, Search and Selection for Clinical Genetics Physician.
- Feb. 2019 Committee Member, Search and Selection for Metabolics Physician.
- 2012–2013 Chief Resident, Medical Genetics Residency Program. University of Calgary.
- 2011 – 2013 Committee Member, Resident Training Committee. Medical Genetics Residency Program. University of Calgary.

Apr. 2013	Resident Representative, Internal Review of Pediatric Hematology/Oncology Residency Program.
2011 & 2012	Coordinator, Medical Genetics Core Month. Department of Medical Genetics, University of Calgary.

VII. PROFESSIONAL ACTIVITIES

Advisory Committees

2022–present	Member, HNRNP Family Foundation Scientific and Medical Advisory Board
2021–present	Member, Canadian Rett Syndrome Clinic Consortium
2021–present	Member, Angelman Syndrome Natural History Study Steering Committee
2020–present	Member, EpiSign Canada Advisory Committee
2020–2023	Member, CCMG Neurodevelopmental Working Group Committee
2020–2022	Member, Au-Kline Syndrome Medical Advisory Board

Professional and Society Memberships

2015–present	Fellow, Royal College of Physicians and Surgeons
2009–present	Member, College of Physicians and Surgeons of Alberta
2009–present	Member, Alberta Medical Association, Canadian Medical Association
2016–present	Fellow, Canadian College of Medical Geneticists
2015–present	Member, Alberta Children’s Hospital Research Institute

Professional Services

1. Peer Reviewer: American Journal of Human Genetics, American Journal of Medical Genetics, Clinical Genetics, Genetics in Medicine, Seizure: European Journal of Epilepsy, GeneReviews, Pediatric Neurology.
2. Abstract Reviewer: David Smith Workshop, Vancouver, BC, August 2024.
3. Judge: Trainee Oral Presentations for David Smith Workshop, Vancouver, BC, August 2024.

Professional Development - Workshops / Courses completed (selected)

1. David Smith Workshop on Malformations and Morphogenesis. (annual) 2013–2015 & 2018–2024.
2. American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting. 2009, 2011,

2012, 2018, 2022, 2024.

3. American Society of Human Genetics Meeting. 2012, 2016, 2019, 2020-2022.
4. International Congress of Parkinson's Disease and Movement Disorders. Vancouver BC. June 4-8, 2017.
5. Department Competence by Design Retreat, 2017.
6. Department of Pediatrics Mentorship Program, University of Calgary. 2016-2017.
7. 10th Annual Course Introductory Course on Skeletal Dysplasias. Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland. July 4-8, 2016.
8. Society for Inherited Metabolic Disorders North America Metabolic Academy (SIMD-NAMA), William F. Bolger Center, Potomac, Maryland. Sep. 11-17, 2011.

Event Organization

May 2023	Co-Organizer, HNRNP family meeting, Columbia University NYC.
Jul. 2020	Co-Organizer, Au-Kline Syndrome Patient meeting and mini research symposia. Virtual.
Jul. 2018	Co-Organizer, Au-Kline Syndrome Patient meeting. GBMC, Baltimore Maryland.
Jan. 2017	Organizer, Skeletal Dysplasia Symposium. University of Calgary.
Jul. 2012	Coordinator Angelman Syndrome Symposium. University of Calgary.

Internal Grant Review

2021	Trainee Small Grants Reviewer, Alberta Children's Hospital Research Institute.
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VIII. RESEARCH SUPPORT / ACTIVITIES

Active Research Support, as PI or co-PI

2025 – 2026	Principal Applicant. <u>Identification of RNA binding targets affected by variants associated with HNRNPK-related neurodevelopmental disorder</u> . VPR Catalyst Grant, \$12,750.
2025 – 2026	Principal Applicant. <u>GENetic Neurodevelopmental disorder Initiative (GENie): a quality improvement project to improve care for families with Genetic neurodevelopmental disorders</u> . Department of Pediatrics Innovation Grant. \$10,500. *Also received the audience choice award.
2024 – 2029	Co-Applicant, <u>Equity Diversity, Inclusion Sub-Platform</u> . \$19,999,843.63.

- 2023 – 2025 Co-Applicant (G. Yang, PI). USP15 in neurodevelopmental disease. RareKids-CAN: Pediatric Rare Disease Clinical Trials and Treatment Network Team Grant. CIHR. NPA: Thierry Lacaze-Masmonteil. Azrieli Foundation. \$100,000.
- 2020 – ongoing Principal Investigator. Au-Kline Syndrome Natural History Study. ACHRI start up funds and HNRNP Family Foundation.
- 2019 – ongoing Principal Investigator. Angelman Natural History Study. Funded by Boston Children’s Hospital/Angelman Syndrome Foundation Canada. Maximum \$53,000.

Completed Funding

- 2022 – 2023 Co- Investigator (G. Yang, Co-PI). Developing a mouse model for USP15, a gene implicated in a rare developmental disorder. CIHR/Genome Canada Rare Diseases Models & Mechanisms Catalyst Grant, Canadian Institute of Health Research. \$25,000.
- 2019 – 2023 Co-Principal Investigator (M. Tarailo-Graovac, PI). CombGene: Combined Genomics to Uncover Genetics of Rare Disease. Clinical Research Fund, ACHRI Start Up. \$210,000.
- 2022 – 2023 Co-Investigator (J. Jacobs-LeVan, Site-PI). Anavex2-73 RCT for Rett Syndrome. Sponsored by Anavex Life Sciences Corp.
- 2021 – 2022 Principal Investigator (B. Hallgrimson, S. Post, Co-PIs). Phenotype characterization of a mouse model for Au-Kline Syndrome. CIHR/Genome Canada Rare Diseases Models & Mechanisms Catalyst Grant. Canadian Institute of Health Research. \$25,000.
- 2019–2021 Co-Investigator (M. Leroux, W. T. Allison, L. Doucette, I.M. MacDonald, Co-PIs). CFAP20 in Retinitis Pigmentosa and Central Nervous System abnormalities. CIHR/Genome Canada Rare Diseases Models & Mechanisms Catalyst Grant. \$50,000.
- 2017–2021 Co-Investigator (J.S. Parboosingh, PI). Rapid Access to Pediatric Diagnoses (RAPiD) Kidomics. Alberta Children’s Hospital Foundation. \$177,000.
- 2019–2021 Principal Investigator. Characterization of rare variants in CACNA1C neurodevelopmental disorder. Rare Disease Foundation Microgrant. \$5000.
- 2016–2019 Principal Investigator. Phenotypic Characterization of Au-Kline Syndrome. Rare Disease Foundation Microgrant. \$3000.
- 2010–2013 Resident Grant (A.M. Innes, PI). Investigation of FBXO28, a candidate causative gene for a chromosome 1q41q42 microdeletion syndrome. ACHRI Small Grant Competition. \$1500.
- 2001–2006 Training Grant. TNF responsive genes in inflammation and carcinogenesis. Canadian Institutes of Health Research MD/PhD Studentship. \$114,000.

Other Research Activities (without direct funding support)

2023–ongoing	Principal Investigator (Pam Veale, Co-PI). <u>Clinical quality improvement and research for genetic neurodevelopmental disorders</u> . Genetic Neurodevelopmental Disorder Initiative (GENIe).
2021–2024	Calgary Site Investigator (B. Sedikovic, PI). <u>Beyond Genomics: Assessing the Improvement in Diagnosis of Rare Diseases using Clinical Epigenomics in Canada (EpiSign-CAN)</u> . Genome Canada Genomic Applications Partnership Program Grant.
2019–2023	Co-investigator (M. Chahine, PI). Funded in kind. <u>Functional studies on novel NaV variants</u> .
2019–2024	Co-Investigator (F. Bolduc, PI). <u>Double-Blind, Placebo-controlled trial of Metformin in Fragile X Syndrome</u> . Azrieli Foundation. Recruitment only from Calgary site.
2019–2020	Collaborator (Q. Long, PI). <u>Developing a Machine Learning Framework for Medical Genomics Projects with Small or Moderate Sample Sizes</u> . New Frontiers in Research Fund.
2019–2020	Collaborator (J. De Koning, PI). <u>An Open Platform for Rapid, Reproducible, Phenotype-Centric Variant Prioritization in Poorly Characterized Rare Genetic Disorders</u> . Genome Alberta Enabling Bioinformatic Solutions (EBS) Competition.

IX. INVITED ADDRESSES

1. Au-Kline Syndrome, a 10-Year Journey. Invited speaker. University of Alberta Rare Disease Day, February 29, 2024.
2. EpiSignatures of the HNRNP-RNDDs: Focus on HNRNP. Invited speaker. HNRNP-Related Neurodevelopmental Disorders Scientific Symposia, May 4-6, 2023. Columbia University, New York.
3. Au-Kline Syndrome: One Decade Down, Forever to Go. Invited speaker. ACHRI Rare Disease Symposium, February 24, 2023.
4. Angelman Syndrome: Genetics 101. Canadian Angelman Syndrome Society (CASS) Moving Mountains Conference for families. Invited Speaker. Oct 13-15, 2022. Kelowna BC.
5. HNRNPK: Recent Insights from Human Methylation and Animal Studies. Invited speaker. ACMG Scientific Concurrent Session: The Role of HNRNP Gene Family in Neurodevelopmental Disorders. March 24, 2022.
6. Au-Kline Syndrome: Diagnostic Criteria and Phenotypic Spectrum. Invited speaker. HNRNP related disorders mini symposia. Virtual. June 3, 2022.

X. PUBLICATIONS/PRESENTATIONS

* denotes contributions as senior/corresponding author

Peer-Reviewed Publications:

1. Gillesse EH, Wan M, Ashtiani S, Suchowersky O, Parboosingh JS, Bernier FP, Lamont RE, Innes AM, **Au PYB**; Care4Rare Canada Consortium. Identification of an Additional Deep Intronic Splice Variant Prompts Critical Evaluation of SPG7 Inheritance. *Neurogenetics*. 2026 Feb 9;27(1):12. doi: 10.1007/s10048-026-00882-7. PMID: 41656397.
2. Das D, Lamothe SM, Duta NC, Koens CC, Hao Y, **Au PYB**, Kurata HT. Regulation of Kv2.1 Biogenesis and Gating by Candidate Disease-Linked Kv6.1 Variants. *J Biol Chem*. 2026 Jan;302(1):110943. doi: 10.1016/j.jbc.2025.110943. PMID: 41241096.
3. Alsayed A, Hakim Z, Merrikh D, Khanbabaei M, Kaur N, Hader W, Soule T, Ashtiani S, Polanco-Tovar G, Scantlebury M, Rahi H, Cao Y, Chan JA, Appendino JP, Pfeffer G, **Au PYB**, Klein KM. Identification of a Second-Hit Brain Somatic DEPDC5 Variant Supports Causality of a DEPDC5 Germline Variant of Uncertain Significance. Time for a Classification Update? *Am J Med Genet A*. 2025 Dec;197(12):e64204. doi: 10.1002/ajmg.a.64204. PMID: 40742146.
4. Asadollahi R, Ahmad A, Boonsawat P, Shahanoor Hinzen J, Lohse M, Bouazza-Arostegui B, Sun S, Utesch T, Sommer JD, Ilic D, Padmanarayana M, Fischermanns K, Ranjan M, Boll M, Ka C, Piton A, Mattioli F, Isidor B, Öunap K, Reinson K, Wojcik MH, Marshall CR, Mercimek-Andrews S, Matsumoto N, Miyake N, Stephan BO, Honjo RS, Bertola DR, Kim CA, Yusupov R, Mefford HC, Christodoulou J, Lee J, Heath O, Brown NJ, Baker N, Stark Z, Delatycki M, Lake NJ, Zeidler S, Zuurbier L, Maas SM, de Kruiff CC, Rajabi F, Rodan LH, Coury SA, Platzer K, Oppermann H, Abou Jamra R, Beblo S, Maxton C, Śmigiel R, Underhill H, Dubbs H, Rosen A, Helbig KL, Helbig I, Ruggiero SM, Fitzgerald MP, Kraemer D, Prada CE, Tenney J, Jayakar P, Redon S, Lefranc J, Uguen K, Race S, Efthymiou S, Maroofian R, Houlden H, Coppens S, Deconinck N, Ashokkumar B, Varalakshmi P, Gowda K VR, Eghbal F, Ghayoor Karimiani E, Heidari M, Neidhardt J, Owczarek-Lipska M, Korenke GC, Bamshad MJ, Campeau PM, Lehman A, Hendon LG, Wentzensen IM, Monaghan KG, Chen Y, Szuto A, Cohn RD, **Au PYB**, Hübner C, Boschann F, Manickam K, Koboldt DC, Rad A, Oprea G, Bachman KK, Seeley AH, Agolini E, Terracciano A, Carmelo P, Bupp C, Grysko B, Rein-Rothschild A, Ben Zeev B, Margolin A, Morrison J, Dagli A, Stolerman E, Louie RJ, Washington C, Stevens SJC, Heijligers M, Alkuraya FS, Lisfeld J, Neu A, Paoli Monteiro F, Santos Pessoa AL, Camelo-Filho AE, Kok F, Koeberl D, Riley K, Burglen L, Doummar D, Héron B, Mignot C, Keren B, Charles P, Nava C, Bernhard FP, Kühn AA, Thoms S, Morrie RD, Mekhoubad S, Green EM, Barmada SJ, Gitler AD, Jahn O, Rhee JS, Rosenmund C, Mitkovski M, Sticht H, Sun H, Le Gac G, Taschenberger H, Brose N, Dittman JS, Rauch A, Lipstein N. Pathogenic UNC13A Variants Cause a Neurodevelopmental Syndrome by Impairing Synaptic Function. *Nat Genet*. 2025 Nov;57(11):2691-2704. doi: 10.1038/s41588-025-02361-5. PMID: 41125872.
5. Erdogan EN, Cheng CV, Caraffi SG, Ivanovski I, Piatelli G, Errichiello E, Papavasiliou AS, Vasileiou G, Reis A, Prince B, Hickey SE, Koboldt DC, Schneider MC, Pormann J, Di Donato N, Leis T, Perry MS, Humberson J, Rotenberg J, Bakhtiari S, Magee H, Kheradmand S, Krueger MC, Swale A, Weber A, Landes C, Zuffardi O, Garavelli L, van Haeringen A, Ruivenkamp CAL, Pauly M, **Au PYB**, Dobyns WB, Aldinger KA. Further Delineation of the AUTS2 HX Repeat Domain-Related Phenotype. *Am J Med Genet A*. 2025 Sep;197(9):e64093. doi: 10.1002/ajmg.a.64093. PMID: 40317680.

6. Choufani S, McNiven V, Cytrynbaum C, Jangjoo M, Adam MP, Bjornsson HT, Harris J, Dymant DA, Graham GE, Nezarati MM, Aul RB, Castiglioni C, Breckpot J, Devriendt K, Stewart H, Banos-Pinero B, Mehta S, Sandford R, Dunn C, Mathevet R, van Maldergem L, Piard J, Brischoux-Boucher E, Vitobello A, Faivre L, Bournez M, Tran-Mau F, Maystadt I, Fernández-Jaén A, Alvarez S, García-Prieto ID, Alkuraya FS, Alsaif HS, Rahbeeni Z, El-Akouri K, Al-Mureikhi M, Spillmann RC, Shashi V, Sanchez-Lara PA, Graham JM Jr, Roberts A, Chorin O, Evrony GD, Kraatari-Tiri M, Dudding-Byth T, Richardson A, Hunt D, Hamilton L, Dyack S, Mendelsohn BA, Rodríguez N, Sánchez-Martínez R, Tenorio-Castaño J, Nevado J, Lapunzina P, Tirado P, Carminho Amaro Rodrigues MT, Quteineh L, Innes AM, Kline AD, **Au PYB**, Weksberg R. An HNRNPK-Specific DNA Methylation Signature Makes Sense of Missense Variants and Expands the Phenotypic Spectrum of Au-Kline Syndrome. *Am J Hum Genet*. 2025 Aug 7;112(8):1979. doi: 10.1016/j.ajhg.2025.07.001. Erratum for: *Am J Hum Genet*. 2022 Oct 6;109(10):1867-1884. doi: 10.1016/j.ajhg.2022.08.014. PMID: 40780051.
7. Hodgson AKO, Baxandall L, Aiyedun D, Li A, **Au PYB**, Bain JM, Gillentine MA, Goel H, Kline AD, Ricupero CL, Sánchez-Carpintero R, Seward EP, Sidlow R, Wilson SA, Balasubramanian M. Expanding the Phenotypic Spectrum of HNRNPU-Related Disorder, Documenting the First Familial Presentation and Comprehensive Review. *Am J Med Genet A*. 2025 Jun;197(6):e64013. doi: 10.1002/ajmg.a.64013. PMID: 39976380.
8. Peron A, D'Arco F, Aldinger KA, Smith-Hicks C, Zweier C, Gradek GA, Bradbury K, Accogli A, Andersen EF, **Au PYB**, Battini R, Belefond D, Bird LM, Bouman A, Bruel AL, Busk ØL, Campeau PM, Capra V, Carlston C, Carmichael J, Chassevent A, Clayton-Smith J, Bamshad MJ, Earl DL, Faivre L, Philippe C, Ferreira P, Graul-Neumann L, Green MJ, Haffner D, Haldipur P, Hanna S, Houge G, Jones WD, Kraus C, Kristiansen BE, Lespinasse J, Low KJ, Lynch SA, Maia S, Mao R, Kalinauskiene R, Melder C, McDonald K, Montgomery T, Morleo M, Motter C, Openshaw AS, Palumbos JC, Parikh AS, Perilla-Young Y, Powell CM, Person R, Desai M, Piard J, Pfundt R, Scala M, Serey-Gaut M, Shears D, Slavotinek A, Suri M, Turner C, Tvrdik T, Weiss K, Wentzensen IM, Zollino M, Hsieh TC; C4RCD Research Group; Telethon Undiagnosed Disease Program (TUDP); University of Washington Center for Mendelian Genomics (UW-CMG); de Vries BBA, Guillemot F, Dobyns WB, Viskochil D, Dias C. BCL11A Intellectual Developmental Disorder: Defining the Clinical Spectrum and Genotype-Phenotype Correlations. *Eur J Hum Genet*. 2025 Mar;33(3):312-324. doi: 10.1038/s41431-024-01701-z. PMID: 39448799.
9. Sjøstrøm E, Bruel AL, Philippe C, Delanne J, Faivre L, Menke LA, **Au PYB**, Cormick JJ, Moosa S, Bayat A. Exploring the Cognitive and Behavioral Aspects of Shprintzen-Goldberg Syndrome; a Novel Cohort and Literature Review. *Clin Genet*. 2025 Mar;107(3):328-334. doi: 10.1111/cge.14646. PMID: 39600231.
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Other Publications / Book Chapters:

1. **Au PYB***, McNiven V, Phillips L, Innes AM, Kline AD. Au-Kline Syndrome. 2019 Apr 18 [updated 2024 Feb 1]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2024.
2. **Au PYB**, Eaton AE, Dyment D. Chapter 23- Genetic Mechanisms of Neurodevelopmental Disease. In: Gallagher, Bulteau, Cohen and Michaud, editors. Handbook of Clinical Neurology. Elsevier, New York, New York. Volume 173, 2020, Pages 307-32.
3. **Au PYB**. Trial and Error in HIV Microbicide Research: In University of Toronto Medical Journal. 2007 May; 84:140-41.
4. **Au PYB**. Editor. Evidence Based Medicine For Primary Care And Core In: Sagar Dugani and Danica Lam, Eds. The Toronto Notes 2009. Toronto, ON: University of Toronto Medical Society; 2009.
5. **Au PYB**, Cohen M, Correia J, Eds. Essentials of Infectious Disease In: Butalia S, Ko HH, Lam C, and Tan J, Eds. Essentials of Clinical Examination Handbook. 4th Ed. Toronto, ON: University of Toronto Medical Society; 2002:265-78.

Peer Reviewed Abstracts/Presentations:

1. Gillentine MA, Hansen A, ... **Au PYB***. Understanding the Autism Phenotype Among Individuals with HNRNP-Related Neurodevelopmental Disorders. International Society for Autism Research, Seattle April 30-May 3, 2025. Accepted

2. Gillentine MA, Hansen A, ... **Au PYB***. HNRNP-Related Neurodevelopmental Disorders: Expanding Knowledge of Known Conditions and Prompting Novel Studies into Candidate Gene. American Society of Human Genetics Meeting, Denver Nov 5-9, 2024.
3. **Au PYB***, Malaney P, Rodriguez A, Kline AD, Post S. Ribosome Dysfunction in Au-Kline Syndrome. Oral Platform Presentation. David Smith Workshop Aug 16-21, 2024, Vancouver, BC, Canada.
4. Prince B, Al Khatib I, Shutt T, **Au PYB***. Apparent Missense Variant Disrupting a Regulatory Element in a Patient with an AUTS2 Phenotype. Poster Presentation. David Smith Workshop Aug 16-21, 2024. Vancouver, BC, Canada.
5. Bradbrook S, **Au PYB**, Innes AM. A Novel Form of Osteopetrosis Associated with Bell's Palsy in the Hutterite Brethren. Poster Presentation. David Smith Workshop Aug 16-21, 2024. Vancouver, BC, Canada.
6. Kowalzyk Z, Bermudez M, Porrmann J, DiDonato N, Hübner C, Dahl A, **Au PYB**, Phillips L, Choufani S, Weksberg R, William D, Schröck E, Jahn A. Deep-Intronic De Novo Variant in HNRNPK as Likely Cause for Au-Kline Syndrome. Poster Presentation. European Human Genetics Conference. Berlin, Germany. June 1-4, 2024.
7. Friedman B, Potter SN... **Au PYB**... Tan W-H, Angelman Syndrome Natural History Consortium. Early Manifestations Prompting Diagnosis of Angelman Syndrome. Gatlinburg Conference on Research and Theory in Intellectual and Developmental Disabilities. April 2024. Kansas City, MO. USA.
8. Tan WH, Sathwani A, **Au PYB**... Bird LM. Angelman Syndrome Natural History Study – How Has it Benefited the Angelman Community Over the Last 17 Years? Foundation for Angelman Syndrome Therapeutics (FAST) Summit. Miami, FL. USA. Nov 9-11, 2023.
9. Burns K, Moebius Syndrome Research Consortium, Ramond F, Wei XC, Yang G, **Au PYB***. USP15 is a Novel Candidate Gene for a Neurodevelopmental Syndrome Associated with Cranial Nerve Dysfunction and Dysmorphic Features. Oral Platform Presentation. David Smith Workshop Aug 25-30, 2023. Southbridge, MA. USA.
10. Kline AD, **Au PYB**, Balasubramanian M, Bain JM, Ricupero CL, Ferguson M, Phillips L, Gillentine MA. HNRNP Related Neurodevelopmental Disorders: A Diverse Yet Overlapping Gene Family. Oral Platform Presentation. 44th Annual David Smith Workshop. Aug 25-30, 2023. Southbridge, MA, USA.
11. Smith CS, Thomas MA, Dymont DA, **Au PYB***. Clinical And Molecular Characterization Of 3 Adult Patients with Infantile Onset Spinocerebellar Ataxia Type 5. Poster Presentation. 43rd Annual David Smith Workshop. August 19 – 24, 2022, Norfolk, VA. USA.
12. Tan, W-H, Anderson KN, Sathwani A, **Au PYB**, Bacino CA, Berry-Kravis E, Boelman C, Carson RP, DeRamus M, Duis JB, Murias K, Ochoa-Lubinoff C, Peters SU, Sell E, Skinner SA, Talboy A, Wheeler AC, Wink LK, Gwaltney A, Bird LM. The Natural History of Angelman Syndrome: Sixteen Years And 450 Individuals Later. Oral Platform Presentation. American College of Medical Genetics Meeting March 15-17, 2023, Salt Lake City, UT. USA.
13. **Au PYB**, Chrystal PW, Lambacher NJ, Doucette LP, Allison TW, MacDonald IM, Arno G, Leroux

- MR. The Inner Junction Protein CFAP20 Functions in Motile and Non-Motile Cilia and Is Critical for Vision. Oral Platform Presentation. American Society of Human Genetics Annual Meeting Oct 25-29, 2022. Los Angeles CA. USA.
14. Choufani S, McNiven V, Cytrynbaum C, ... Kline AD, **Au PYB***, Weksberg R*. Au-Kline Syndrome Phenotypic Expansion: Genome Wide DNA Methylation Signature Makes Sense of Missense. Oral Platform Presentation. David Smith Workshop Aug 20-23, 2022. Norfolk, VA. USA.
 15. Smith CS, Thomas MA, Dyment DA, **Au PYB**. Characterization Of 3 Adult Patients with Infantile Onset Spinocerebellar Ataxia Type 5 and Recurrent Variants at Codon 255. Poster Presentation. David Smith Workshop Aug 20-23, 2022. Norfolk, VA. USA.
 16. Pijpers JA, **Au PYB**, Weeke L, Vein AA, Smit L, Vilan A, Jacobs E, de Vries LS, Steggerda SJ, Peeters CMPCD*, Appendino JP*. Early Recognition of a Unique Amplitude-Integrated EEG Pattern and Clinical Semiology of Neonatal Seizures Caused by SCN2A And KCNQ3 Mutations. Poster Presentation. American Epilepsy Society Meeting, Dec 3-7, 2021. Virtual.
 17. Yang XR, Ginjupalli VK, Theriault O, Poulin H, Appendino JP, **Au PYB***, Chahine M*. SCN2A-Related Epilepsy of Infancy with Migrating Focal Seizures: Report of a Variant with Apparent Gain and Loss of Function Effects. Canadian League Against Epilepsy Meeting, Sept 24-26, 2021. Virtual.
 18. **Au PYB**, Tyndall AV, Phillips E, Gurram Venkata SKR, Leijser LM, Walker L, Bernier FP, Parboosingh HS, Lamont E, Innes AM. Novel C-Terminal EPHB4 Variants in Two Individuals with Lymphatic Related Fetal Hydrops Spectrum Disorder. Poster Presentation. David Smith Workshop Sept 10-15, 2021. Stevenson WA. USA.
 19. Cherian C, Appendino JP, Ashtiani S, Federico P, Molnar C, Kerr M, Khan A, **Au PYB**, Klein KM. The Phenotypic Spectrum of KCNT1: A New Family with Variable Epilepsy Syndromes and Severity. Poster presentation. 34th International Epilepsy Congress. Aug 28-Sept 1, 2021. Virtual meeting.
 20. **Au PYB***, Choufani S, McNiven V... Kline AD, Weksberg R. Au-Kline Syndrome: Genome Wide DNA Methylation Signature Makes Sense of Missense. Poster presentation. American Society of Human Genetics, Oct 27-30. 2021. Virtual Meeting. Selected as Reviewer's Choice Award, top 10% of abstracts.
 21. Al-Kateb H, Frank M, Willing M, Lichtarge O, Katsonis P, **Au PYB**, Shinawi M. Nonsense and Missense Variants in CAMTA1 Gene are Associated with Developmental Delay/Intellectual Disability, Gait Abnormalities, and Hypotonia. Poster presentation. American Society of Human Genetics, Oct 15-19, 2019. Houston, TX. USA.
 22. **Au PYB**, Tago J, Novak J, MacDonald I. CFAP20 (Bug22) is a Candidate Gene for Autosomal Recessive Syndromic Retinitis Pigmentosa. Poster presentation. David Smith Workshop Aug 24-29, 2019. Snowbird Resort, UT. USA.
 23. Tan TY, Dong X, Costain G, Rodan LH, Lynch S, Gupta A, Dowling JJ, Walker S, Chung WK, **Au PYB**, Griffith C, Ben Zeev B, Moutton S, Devinsky O, Schaefer GB, Mantovan JF, Torti E, Pais L, FBXW7 Collaborative Group, Christodoulou. De Novo Variants in FBXW7 Associated with a Variable Neurodevelopmental and Congenital Anomaly Phenotype. Poster presentation. David Smith Workshop Aug 24-29, 2019. Snowbird Resort, UT. USA.
 24. Suchowersky O, Ashtiani S, **Au PYB**, McLeod S, Rouleau G, Gan-Or Z. Hereditary Spastic

- Paraplegia Misdiagnosed as Cerebral Palsy: Review of Pediatric Hereditary Spastic Paraplegia Cases in an Alberta Cohort. Poster presentation. XXIV World Congress on Parkinson's Disease and Related Disorders. Montreal, QC. Canada. 2019 June.
25. **Au PYB**, Goedhart C, Ferguson M, Breckpot J, Devriendt K, Wierenga K, Fanning E, Grange DK, Graham GE, Dymont DA, Galarreta C, Jones MC, Kini U, Stewart H, Mehta S, Castiglioni C, Nezarati MM, Aul RB, Rosenfeld JA, Hanchard N, Burrage L, Weiss K, Care for Rare Canada Consortium, Parboosingh JS, Kline AD, Innes AM. Phenotypic Spectrum of Au-Kline Syndrome: 12 New Cases and Review of the Literature. Oral platform presentation at the David Smith Workshop Aug 24-29, 2018. Banff, Alberta. Canada.
 26. Chang C, **Au PYB**. Expanded Phenotype of the Blepharocheilodontic Syndrome in a Mother and Daughter with CTNND1 Mutation. Poster presentation. American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting in Seattle, WA. USA. April 3-5, 2019.
 27. Ganapathi M, Padgett L, Yamada K, **Au PYB**, Tagoe J, McDonald M, Karłowicz D, Devinsky O, Willaert R, Person R, Deng L, Leduc C, Mirmira R, Mastracci T, Chung W. Recessive Rare Variants in Deoxyhypusine Synthase (DHPS) An Enzyme Involved in Synthesis of Hypusine, Cause A Novel Condition Including Global Developmental Delay and Seizures. **Oral platform presentation** at the American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting in Charlotte, North Carolina, April 10-14, 2018.
 28. Chang CA, Lemiski N, **Au PYB**. A Novel Frameshift Mutation Causing NOG-Symphalangism Spectrum Disorder (NOG-SSD). Poster presentation. European Society of Human Genetics Meeting in Copenhagen, Denmark, Europe. May 27-30, 2017.
 29. **Au PYB**, Goedhart C, Kerr M, Ferguson M, Breckpot J, Devriendt K, Wierenga KJ, Fanning E, Kini U, Stewart H, Lange L, Pagnamenta A, Taylor J, Keays D, Grange DK, Dugan SL, Care for Rare Canada Consortium, Parboosingh JS, Kline AD, Innes AM. Further Delineation of Au-Kline Syndrome: A Craniosynostosis and Kabuki-Like Syndrome Due to Loss of Function Mutations and Deletions of HNRNPK. Poster presentation. American Society of Human Genetics in Vancouver BC, Canada. October 18-22, 2016.
 30. Chang C, Argiropoulos B, Innes M, **Au PYB***. A New Case of 5q31 Microdeletion Syndrome and Putative Causative Genes. Presented at the 40th Annual Scientific Conference of the CCMG in Edmonton, AB, Canada. June 19-21, 2016.
 31. **Au PYB**, Chernos JE, Thomas MA. Review of the Recurrent 8q13.2q13.3 Branchio-Oto-Renal Related Microdeletion, and Report of an Additional Case with Associated Distal Arthrogyrosis. Poster presentation. American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting in Tampa, Florida, USA. March 8-12, 2016.
 32. **Au PYB**, You J, Caluseriu O, Schwartzenruber J, Majewski J, Bernier FP, Ferguson M, Care for Rare Consortium, Valle D, Parboosingh JS, Sorbreira N, Innes AM, and Kline AD. De Novo Loss of Function Mutations in Ribonucleoprotein HNRNPK Cause a Novel Malformation Syndrome Characterized by Unique Facial Dysmorphisms, Skeletal and Connective Tissue Abnormalities, and Intellectual Disability. Oral platform presentation. 36th Annual David W. Smith Workshop on Malformations and Morphogenesis, Aug 14-19, 2015, St. Michaels, MD, USA.
 33. **Au PYB**, Smith CS, Lamont RE, Racher HE, Parboosingh J, Bernier F, Innes AM. De Novo Truncating Mutation in the Chromatin Remodeler CHD8 in a Patient with Autism Macrocephaly and Overgrowth. Oral platform presentation. 35th Annual David W. Smith Workshop on Malformations

and Morphogenesis, July 25-30, 2014. Madison, Wisconsin, USA.

34. **Au PYB**, Smith CS, Lamont RE, Racher HE, Parboosingh J, Bernier F, Innes AM. A De Novo Truncating Mutation in the Chromatin Remodeler CHD8 in a Patient with Autism Macrocephaly and Overgrowth. Poster presentation. CCMG 38th Annual Scientific Conference, Vancouver, BC, Canada. November 2014.
35. **Au PYB**, Innes AM, Racher HE, Duff HJ, Gerull B, Khan A, Bernier F, Sinasac D, Snyder F. Dilated Cardiomyopathy Ataxia Syndrome: Approaches to Elucidating Underlying Biological Mechanisms and Update on the Natural History. Oral platform presentation. 34th Annual David W. Smith Workshop on Malformations and Morphogenesis. August 9-14, 2013, Mont Tremblant, Quebec, Canada.
36. **Au PYB**, Perrier R, Puffenberger EG, Hume S, Anderson R, Lafay-Cousin L, Strother D, McInnes B, Parboosingh J, Bernier F. Poster presentation. A Novel Bi-Allelic MSH2 Mutation Associated with Constitutional Mismatch Repair Deficiency Syndrome, and Review of the Clinical Phenotype (Abstract 1052F Cancer Genetics). 62nd Annual Meeting of American Society of Human Genetics, November 2012, San Francisco, CA. USA.
37. **Au PYB**, Argiropoulos B, Parboosingh J, Innes AM. FBXO28, a Candidate Causative Gene for a Chromosome 1q41q42 Microdeletion Syndrome Associated with Intellectual Disability and Seizures. Poster presentation. American College of Medical Genetics Conference, March 2012, Charlotte, NC. USA.
38. **Au PYB**, Dymant DA, Chernos JE, Thomas MA. Deletion 18q21.2q21.31 in a Case of Pitt-Hopkins Syndrome Presenting with Obstructive Hydrocephalus Due to an Arachnoid Cyst. Poster presentation. American College of Medical Genetics Conference, March 2011, Vancouver BC, Canada.
39. **Au PYB**, Kato M, Dupuis L, Mendoza-Londono R. Distinctive Clinical Phenotype in Patients with Distal Trisomy 5q and Distal Monosomy 13q Due to Paternal Balanced Translocations. Poster presentation. American College of Medical Genetics Conference, March 2009, Tampa, FL. USA.
40. **Au PYB**, Lin WJ, Katz D, Moemeni B, Mirtsos C, Yeh WC. Characterization of TRIM47: A Potential TNF Inducible Regulator of P53. Poster presentation. Canadian Society of Clinical Investigators Conference. September 2006. Ottawa, ON. Canada.
41. **Au PYB**, Chau H, Moemeni B, Martin N, Yeh WC. PDGF-B Provides a Key Switch Between Cell Death and Survival Induced By TNF. Poster presentation. NF- κ B: 20 Years on the Road from Biochemistry to Pathology, Keystone Symposia. March 23-28, 2006. Banff AB, Canada.
42. **Au PYB**, Chau H, Moemeni B, Martin N, Yeh WC. PDGF-B Provides a Key Switch Between Cell Death and Survival Induced By TNF. Oral presentation. Canadian Society of Clinical Investigators Conference. Sep. 2005. Vancouver, BC, Canada.
43. **Au PYB**, Martin N, Yeh WC. PDGF-B as a Potential Survival Effector Mediated by TNF α Through NF- κ B. Poster presentation. IRCM Sixth Conference on Signaling in Normal and Cancer Cells. March 2004. Banff, AB. Canada.
44. **Au PYB**, Suzuki S, Suzuki N, Lye E, Martin N, Yeh WC. Microarray Screening for Survival Genes Inducible by TNF α . Poster presentation. TNF Superfamily Conference 2002: 9th International Congress on TNF Related Cytokines. October 2002. San Diego, CA. USA.

Non- peer reviewed Presentations / Abstracts

1. **Au PYB**, Kline AD, Ferguson M, Phillips L, Plecas-Maricic S, Hallgrimmson B, Weksberg R, Choufani S, Malaney P, Post S. HNRNPK – Works in Progress. Oral presentation. Scientific networking session. HNRNP Family Foundation Meeting. July 28-30, Issaquah, WA, USA.
2. Ashtiani S, **Au PYB**, Gan-Or Z, Rouleau G, Suchowersky O. Hereditary Spastic Paraplegia Misdiagnosed as Cerebral Palsy: Preliminary Review of Pediatric Hereditary Spastic Paraplegia Cases in an Alberta Cohort. Poster presentation. Owerko Centre Conference. Trajectories and Translation in Neurodevelopmental Disorders. June 7, 2018.
3. **Au PYB**, Perrier R, Puffenberger EG, Hume S, Anderson R, Lafay-Cousin L, Strother D, McInnes B, Parboosingh J, Bernier F. A Novel Bi-Allelic MSH2 Mutation Associated with Constitutional Mismatch Repair Deficiency Syndrome, And Review of the Clinical Phenotype. Poster presentation. Alberta Genome Instability and Aging Conference, September 2013, Calgary, Alberta.

XI. CLINICAL and CLINICAL QI ACTIVITIES (selected)

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| 2023–present | Co-Lead with Pam Veal for GENie (Genetic Neurodevelopmental Disorder Initiative) clinical pathway. |
| 2018–present | Clinic lead. Genetic Epilepsy Clinic (GEC), interdisciplinary clinic with Dr JP Appendino (pediatric epileptologist) and Dr Karl Martin Klein (adult epileptologist) and Setareh Ashtiani (genetic counselor). |
| 2017–present | Clinic lead. Neurogenetics / Movement Disorder interdisciplinary clinic (with Dr. J. Sarna, movement disorder neurologist). |
| 2016–2020 | Epilepsy Genetics and Metabolic Evaluation mini task force, a quality improvement initiative to develop a diagnostic pathway and expedited genetic testing for epileptic encephalopathy patients. |
| 2015–2020 | Clinical follow-up of Calgary patients who were enrolled in the Genetics of Hereditary Spastic Paraplegia project (O. Suchowersky, PI). |
| 2016 | Development of genetics referral criteria for primary early onset and familial dementias. |

XII. PATIENT ADVOCACY/PATIENT KNOWLEDGE TRANSLATION

1. HNRNPK Natural History Update. Invited speaker. HNRNP Family Foundation Meeting. July 28-30, 2024. Issaquah, WA, USA.
2. Methylation Patterns of the HNRNP-RNDDs. HNRNP-Related Neurodevelopmental Disorders Family Meeting, May 4-6, 2023. Columbia University, NY. USA.
3. Fragile X 101. Invited speaker. Fragile X family presentation. Virtual. March 2021.
4. Intro to Genetics for Fragile X. Invited speaker. Fragile X family presentation on FMRP-tat, with Dr Ray Turner, Virtual. July 2020.

5. Updates on Au-Kline Syndrome. Speaker and coordinator. Au-Kline Syndrome Patient/Family Meeting. Virtual. July 2020.
6. Overview of Au-Kline Syndrome. Speaker and coordinator. Inaugural Au-Kline Syndrome Patient/Family Meeting, Baltimore MA. USA. July 2018.
7. Genetics 101: Angelman Syndrome. Invited speaker. Canadian Angelman Syndrome Society. Kananaskis, Alberta, Canada. July 2012.