CURRICULUM VITAE

Dr. Ping-Yee Billie Au

1. BIOGRAPHICAL DATA

Clinical Assistant Professor	Department of Medical Genetics, University of Calgary
Clinical Geneticist	Section of Clinical Genetics, Department of Pediatrics, Alberta Health Services
	28 Oki Drive, Alberta Children's Hospital, Calgary AB, T3B 6A8
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Research Institutes:	Alberta Children's Research Hospital Research Institute – Full Member
	Owerko Center – Full Member

2. EDUCATION

1996-2000	BSc (Hons), Biochemistry, Queen's University, Kingston, ON, Canada
2000-2002, 2007-2009	MD (as part of MD/PhD program), University of Toronto, Toronto, ON, Canada
2002-2007	PhD Cellular and Molecular Biology, Dept. of Medical Biophysics, University of Toronto
2009-2015	FRCPC (Medical Genetics and Genomics), University of Calgary, Calgary, AB, Canada
2016	FCCMG (Fellow, Canadian College of Medical Geneticists)

3. EMPLOYMENT HISTORY

2018 - present	Clinical Assistant Professor, Medical Genetics, Cumming School of Medicine, University of Calgary
2018 - present	Clinical Assistant Professor, Pediatrics, Cumming School of Medicine, University of Calgary
2015 - present	Clinical Geneticist, Section of Clinical Genetics, Department of Pediatrics, Alberta Children's Hospital

4. RECENT AWARDS

2025 Department of Medical Genetics Clinical Teaching award

2024 Cumming School of Medicine, University of Calgary PGME Teaching Award.

5. CURRENT RESEARCH SUPPORT

2026 – 2028 Co-Applicant, CIHR Knowledge Synthesis Grant. GeneXchange: Linking Genetic Discovery to Real World Impact. $98,450.

2025 – 2026 **Principal Applicant**, University of Calgary VPR Catalyst Grant. Identification of RNA binding targets affected by variants associated with HNRNPK-related neurodevelopmental disorder. $12,750.

2025 – 2026 **Principal Applicant**, Department of Pediatrics Innovation Grant. GEnetic Neurodevelopmental disorder Initiative (GENIe): a quality improvement project to improve care for families with Genetic neurodevelopmental disorders. $10,500.

2025 – 2030 Co-Investigator. Expanding the use of genomics to unravel rare diseases: Care4Rare EXPAND. Genome Canada, CPHI Pillar 1: Generating population-level genomic data. ~$20 million.

2025 – 2010 Co-Applicant. Mechanistic understanding of pathogenic autophagy and stem cell regulation in a neurodevelopmental disorder. CIHR Project Grant. $1,585,000.

2020 – ongoing **Principal Investigator.** HNRNPK-RNDD/Au-Kline Syndrome Natural History Study. ACHRI start up funds and HNRNP Family Foundation (~$10,000/year from HNRNP FF x 5 years).

2019 – ongoing **Principal Investigator**. Angelman Natural History Study. Funded by Boston Children's Hospital/Angelman Syndrome Foundation Canada. (~$10,000/year)

5. PUBLICATIONS (last 5 years)

(* denotes corresponding/senior author. h-index = 30)

1. Bereshneh AH, …**Au PYB**, … Berger S, Bellen HJ. Rare heterozygous de novo variants in RAPGEF2 are associated with a neurodevelopmental disorder. Genet Med. 2026 Jan 16;28(4):101685. doi: 10.1016/j.gim.2026.101685. Epub ahead of print. PMID: 41556274.
2. 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; PMCID: PMC12883507.
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. Epub 2025 Jul 31. PMID: 40742146.
4. Asadollahi R, … **Au PYB**, … 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. Epub 2025 Oct 22. PMID: 41125872; PMCID: PMC12597829.
5. 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.
6. Erdogan EN, … **Au PYB,** Dobyns WB, Aldinger KA. Further Delineation of the AUTS2 HX Repeat Domain-Related Phenotype. Am J Med Genet A. 2025 May 3:e64093. doi: 10.1002/ajmg.a.64093. Epub ahead of print. PMID: 40317680.
7. Peron A, D'Arco F, Aldinger KA, Smith-Hicks C, Zweier C, Gradek GA, Bradbury K, Accogli A, Andersen EF, **Au PYB,** … 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.
8. 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.
9. Sabeh P, Dumas SA, Maios C, Daghar H, Korzeniowski M, Rousseau J, Lines M,…**Au PYB,**… Parker JA, Burnett BG, Campeau PM. Heterozygous UBR5 variants result in a neurodevelopmental syndrome with developmental delay, autism, and intellectual disability. Am J Hum Genet. 2025 Jan 2;112(1):75-86. doi: 10.1016/j.ajhg.2024.11.009. PMID: 39721588.
10. Gillesse E, Wade A, Parboosingh JS, **Au PYB,** Bernier FP; C4R Consortium; Lamont RE, Innes AM. Genome sequencing identifies biallelic variants in SCLT1 in a patient with syndromic nephronophthisis: Reflections on the SCLT1-related ciliopathy spectrum. Am J Med Genet A. 2024 Nov;194(11):e63789. doi: 10.1002/ajmg.a.63789. PMID: 38924217.
11. Shafiq T, … **Au PYB**…Bain J, Gillentine M et al. An expansion of the phenotype in individuals with SYNCRIP-Related Neurodevelopmental Disorder. November 2024. Rare. 2(9): 100052. 10.1016/j.rare.2024.100052. PMID: n/a.
12. Colijn MA, Vrijsen S, **Au PYB**, Abou El Asrar R, Houdou M, Van den Haute C, Sarna J, Montgomery G, Vangheluwe P. Kufor-Rakeb syndrome-associated pyschosis: a novel loss-of-function ATP13A2 variant and response to antipsychotic therapy. 2024 Oct;25(4):405-415. doi: 10.1007/s10048-024-00767-7. PMID: 39023817.
13. Smith CS, Riddell M, Baladalo L, **Au PYB***. Adults with Paternal UPD14 Causing Kagami-Ogata syndrome: Case Report and Review of the Literature. Am J Med Genet A. 2024 Sep;194(9):e63625. doi: 10.1002/ajmg.a.63625. PMID: 38741340.

14. Bhoj E, Layo-Carris D, Lubin E, …**Au B,** … Bryant L, Carere D. Expanded phenotypic spectrum of neurodevelopmental and neurodegenerative disorder Bryant-Li-Bhoj syndrome with 38 additional individuals. Eur J Hum Genet. 2024 Aug;32(8):928-937. doi: 10.1038/s41431-024-01610-1. Erratum in: Eur J Hum Genet. 2024 Aug;32(8):1032. doi: 10.1038/s41431-024-01659-y. PMID: 38678163.
15. Rinaldi B, Bayat A, Zachariassen LG, Sun JH, Ge YH, Zhao D,…**Au PYB**… Lesca G, Møller RS, Tümer Z, Musgaard M, Gerard B, Lemke JR, Shi YS, Kristensen AS. Gain-of-function and loss-of-function variants in GRIA3 lead to distinct neurodevelopmental phenotypes. Brain. 2024 May 3;147(5):1837-1855. doi: 10.1093/brain/awad403. PMID: 38038360.
16. Vanden Broek K, Ryu J-R, Perrier R, Tyndall AV, Childs SJ, **Au PYB***. SAM domain variants of EPHB4 associated with aberrant signalling are linked to lymphatic-related fetal hydrops and facial dysmorphology. Clin Genet. 2024 Apr;105(4):386-396. doi: 10.1111/cge.14467. Erratum in: Clin Genet. 2024 Nov;106(5):664. doi: 10.1111/cge.14597. PMID: 38151336.
17. Al-Kateb H, **Au PYB**, Berland S, Cogne B….Willing M, Shinawi M. CAMTA-1 related disorder: Phenotypic and molecular characterization of 26 new individuals and literature review. Clin Genet. 2024 Mar;105(3):294-301. doi: 10.1111/cge.14464. PMID: 38044714.
18. Pijpers JA, **Au PYB**, Weeke LC, Vein AA, Smit LS, Vilan A,…Appendino JP, Peeters-Scholte CMPCD. Early recognition of characteristic conventional and amplitude-integrated EEG patterns of seizures in SCN2A and KCNQ3-related epilepsy in neonates. eizure. 2023 Aug;110:212-219. doi: 10.1016/j.seizure.2023.06.016. PMID: 37429183.
19. Von Brauchitsch S, Haslinger D, Lindlar S, Thiele H, Bernsen N, Zahnert F, Reif PS, Balcik Y, **Au PYB**, Josephson C, Altmuller J, Strzelczyk A, Knake S, Rosenow F, Chiocchetti A, Klein KM. The phenotypic and genotypic spectrum of epilepsy and intellectual disability in adults: Implications for genetic testing. Epilepsia Open. 2023 Jun;8(2):497-508. doi: 10.1002/epi4.12719. PMID: 36896643.
20. Carter MT, Srour M, **Au PB**, Buhas D, Dyack S, Eaton A, Inbar-Feigenberg M, Howley H, Kawamura A, Lewis SME, McCready E, Nelson TN, Vallance H; Canadian College of Medical Geneticists. Genetic and metabolic investigations for neurodevelopmental disorders: position statement of the Canadian College of Medical Geneticists (CCMG). J Med Genet. 2023 Jun;60(6):523-532. doi: 10.1136/jmg-2022-108962. PMID: 36822643.
21. Salpietro V, Galassi Deforie V, Efthymiou S, … , **Au PYB**,… Houlden H, Männikkö R. De novo KCNA6 variants with attenuated KV 1.6 channel deactivation in patients with epilepsy. Epilepsia. 2023 Feb;64(2):443-455. doi: 10.1111/epi.17455. PMID: 36318112.
22. Li Q, Perera D, Cao C, He J, Bian J, Chen X, Azeem F, Howe A, **Au B**, Wu J, Yan J, Long Q. Interaction-integrated linear mixed model reveals 3D-genetic basis underlying Autism. Genomics. 2023 Mar;115(2):110575. doi: 10.1016/j.ygeno.2023.110575. PMID: 36758877.
23. Chrystal PW, Lambacher NJ, Doucette LP …. Allison TW*, Au **PYB***, MacDonald IM*, Arno G*, Leroux MR*. The inner junction protein CFAP20 functions in motile and non-motile cilia and is critical for vision. Nature Communications. Nat Commun. 2022 Nov 3;13(1):6595. doi: 10.1038/s41467-022-33820-w. PMID: 36329026.
24. AlAbdi L, Desbois M, Rusnac DV…**Au PYB**…Grill B, Alkuraya F. Loss of function variants in MYCBP2 cause neurobehavioural phenotypes and corpus collosum defects. Brain. 2023 Apr 19;146(4):1373-1387. doi: 10.1093/brain/awac364. PMID: 36200388.
25. Choufani S, McNiven V, Cytrynbaum C, … 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. 2022 Oct 6;109(10):1867-1884. doi: 10.1016/j.ajhg.2022.08.014. PMID: 36130591.
26. Plumereau Q, Ebdalla A, Poulin H, Appendino JP, Scantlebury M, **Au PYB**, Chahine M. De novo Y1460C missense variant in $Na_V1.1$ impedes the pore region and results in epileptic encephalopathy**.** Sci Rep. 2022 Oct 13;12(1):17182. doi: 10.1038/s41598-022-22208-x. PMID: 36229510.

27. Pavinato L, Vedove AD….**Au PYB**…Brusco A. CAPRIN1 haploinsufficiency causes a neurodevelopmental disorder with language impairment, ADHD and ASD. Brain. 2023 Feb 13;146(2):534-548. doi: 10.1093/brain/awac278. PMID: 35979925.
28. Yang X-R, Ginjupalli VKM, 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. J Neurophysiol. 2022 May 1;127(5):1388-1397
29. 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. J. Neurol. 2022 Apr;269(4):2162-2171
30. Johannesen KM, Liu Y, Koko M, …. **Au PYB**…Lerche H, Moller RS. Genotype-phenotype correlations in *SCN8A*-related disorders reveal prognostic and therapeutic implications. Brain. 2021 August 25. https://doi.org/10.1093/brain/awab321
31. Stephenson SEM, Costain G, Blok LER… **Au PYB**… Lockhart PJ, Christodoulu J, Yan TY. Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a novel neurodevelopmental syndrome. Am J Hum Genet. 2022 Apr 7;109(4):601-617
32. Maroilley T, Wright NAM, Diao C, MacLaren L, Pfeffer G, Sarna JR, **Au PYB***, Tarailo-Graovac M*. Case Report: Biallelic Loss of Function ATM due to Pathogenic Synonymous and novel deep intronic variant c.1803-270T>G identified by genome sequencing in child with Ataxia Telangiectasia. Frontiers in Genetics. 2022 Jan 25. doi: 10.3389/fgene.2022.815210
33. Johannesen KM, Iqbal S…. Klein KM**, Au PYB**, Smyth K….Moller RK, Gardella E. Structural Mapping of GABRB3 variants reveals genotype-phenotype correlations. Genet Med. 2022. 24(3):681-693
34. Kloth K, Lozic B, Tagoe J, Hoffer MJV, Van der Ven A, Thiele H, Altmuller J, Kubisch C, **Au PYB**, Denecke J, Bijlsma EK, Lessel D. ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss of function variants. Neurogenetics. 2021 Oct; 22(4):263-269.
35. Suchowersky O, Ashtiani S, **Au PYB**, McLeod S, Estiar MA, Gan-Or Z, Rouleau G. Hereditary spastic paraplegia initially diagnosed as cerebral palsy. Clinical Parkinsonism and Related Disorders. 2021 Nov. https://doi.org/10.1016/j.prdoa.2021.100114
36. Rodan LH, Spillman RC, Kurata HT, …. **Au PYB**, Shashi V. Phenotypic expansion of *CACNA1C*-associated disorders to include isolated neurological manifestations. Genetics in Medicine. 2021. Oct;23(10):1922-1932
37. Parenti I, Lehalle D…. **Au PYB**….Depienne C, Mignot C. Missense and truncating variants in CHD5 lead to neurodevelopmental disorders with intellectual disability, autism and epilepsy. Human Genetics. 2021 Jul; 140 (7):1109-1120.
38. Sheppard SE, Campbell IM… **Au PYB**….Quintero-Rivera F. Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner Syndrome. Am J Med Genet. 2021 June; 185(6):1649-1665.
39. Hanly C, Shah H, **Au PYB**, Murias K. Description of neurodevelopmental phenotypes associated with 10 genetic neurodevelopmental disorders: A scoping review. Clin Genet. 2021 Mar;99(3):335-346
40. **Au PYB**, Eaton AE, Dyment D. Au PYB, Eaton A, Dyment DA. 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.
41. Dyment DA, O'Donnell-Luria A, Agrawal PB, Coban Akdemir Z, Aleck KA, Antaki D, Al Sharhan H, **Au PB**,…Innes AM. Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. Am J Med Genet A. 2021 Jan;185(1):119-133.
42. **Au PYB***, Innes AM, Kline AD. *Au-Kline Syndrome*. 2019 Apr 18, [updated 2024 Feb 1]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016. Available from: https://www.ncbi.nlm.nih.gov/books/NBK540283/

6. PRESENTATIONS

Selected Invited Presentations

1. Updates on the Natural History study for HNRNPK-RNDD. Invited speaker. HNRNP Family Foundation Patient and Family Meeting. July 27-30, 2025. Ponte Vedra Florida.
2. HNRNPK Natural History update. Invited speaker. HNRNP Family Foundation Meeting. July 28-30, 2024. Issaquah, WA, USA.
3. Au-Kline Syndrome, a 10-year journey. Invited speaker. University of Alberta Rare Disease Day, February 29, 2024.
4. Episignatures of the HNRNP-RNDDs: Focus on HNRNP. Invited speaker. HNRNP-Related Neurodevelopmental Disorders Scientific Symposia, May 4-6, 2023. Columbia University, New York.
5. Canadian position statement for genetic testing in development disorders and local initiatives. Invited speaker. April 2023. Owerko Neurodevelopmental Clinical Rounds.
6. "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
7. "Au-Kline Syndrome: diagnostic criteria and phenotypic spectrum." Invited speaker. HNRNP related disorders Mini Symposia. Columbia University. Virtual. June 3, 2022.

Selected Conference Presentations

1. **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.
2. 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.
3. **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.
4. 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.
5. **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**.

7. SELECTED OTHER CONTRIBUTIONS

Education Leadership:

June 2025 – May 2026: Co-interim program director RCPSC Genetics and Genomics Residency Program
2023 – 2025: RCPSC Genetics and Genomics Residency Competence Committee Chair

Selected Committees:

2022– current Clinical lead, Functional Genomics Lab, University of Calgary
2020– current HNRNP Family Foundation medical and scientific advisory board
2020– 2022 CCMG Neurodevelopmental working group committee

8. PERSONAL STATEMENT

I am a medical geneticist and clinician investigator with a previous PhD in cell signaling biology. My clinical and research interests focus on neurodevelopmental and neurogenetic disorders. I have contributed to discovery and characterization of over 12 novel genetic diseases, most notably HNRNPK as the causative gene for Au-Kline syndrome. I am an active investigator within the national genomics network Care4Rare. I co-run the Functional

Genomics program in Calgary, which leverages cell based assays to address genes and variants of uncertain significance. My research focuses on understanding how genetic variants drive molecular and cellular dysfunction, how this dysfunction influences clinical phenotypes and outcomes, and how these insights can be leveraged to develop therapies and improve patient management.