IBR STAFF PUBLICATIONS—2024

with 2023 Journal Impact Factors (JIFs) from 2023 Journal Citation Reports
(released June 2024), if available (downloaded from ResearchGate.net)

1. Alldred MJ, Pidikiti H, …, Wisniewski T, **Wegiel Jerzy**, Stutzmann GE,…, Mufson EJ, Ginsberg SD. Analysis of microisolated frontal cortex excitatory layer III and V pyramidal neurons reveals a neurodegenerative phenotype in individuals with Down syndrome. *Acta Neuropathol* 2024; 148(1):16. [DOI: 10.1007/s00401-024-02768-0]. *(2023 JIF: 6.2)*

2. Araldi GL, **Hwang YW,** Raghu G: Development and evaluation of ABI-171, a new fluoro-catechin derivative, for the treatment of idiopathic pulmonary fibrosis. *Int J Mol Sci* 2024; 25(21):11827. [DOI: 10.3390/ijms252111827]. *(2023 JIF: 4.9)*

3. **Basurto-Islas G, Tung YC, Dai CL, Iqbal K, Gong CX:** Diabetic ketoacidosis induces tau hyperphosphorylation in rat brain. *J Alzheimers Dis Rep* 2024; 8(1):615–626. [DOI: 10.3233/ADR-240040]

4. Boerwinkle AH, Wisch JK, …, **Krinsky-McHale SJ,** …, Lee JH, Ances BM; Alzheimer's Biomarker Consortium-Down Syndrome: The mediating role of plasma glial fibrillary acidic protein in amyloid and tau pathology in Down's syndrome. *Alzheimers Dement* 2024 Nov 13. [DOI: 10.1002/alz.14359]. Epub ahead of print. *(2023 JIF: 13.0)*

5. **Carter DC, Kierzkowska O, Sarino K, Guo L, Marchi E, Lyon GJ:** Ocular manifestations in a cohort of 43 patients with KBG syndrome. *Am J Med Genet A* 2024; 194(4):e63473. [DOI: 10.1002/ajmg.a.63473] *(2023 JIF: 1.7)*

6. Chen YA, Liu TY, Wen KY, Hsu CY, Sze CI, **Chang NS:** Zfra overrides WWOX in suppressing the progression of neurodegeneration. *Int J Mol Sci* 2024; 25(6):3507. [DOI: 10.3390/ijms25063507]. *(2023 JIF: 4.9)*

7. Chowdhury SF, Prout N, …, **Salafia CM,** …, Miller RK, O'Connor TG: PFAS alters placental arterial vasculature in term human placentae: a prospective pregnancy cohort study. *Placenta* 2024; 149:54–63. [DOI: 10.1016/j.placenta.2024.03.002] *(2023 JIF: 3.0)*

8. **Chu D**, Lei L, Gu S, **Liu F, Wu F:** Dual-specificity tyrosine phosphorylation-regulated kinase 1A promotes the inclusion of amyloid precursor protein exon 7. *Biochem Pharmacol* 2024; 224:116233. [DOI: 10.1016/j.bcp.2024.116233] *(2023 JIF: 5.3)*

9. Chu D, Yang X, Wang J, Zhou Y, Gu JH, …, **Liu F:** Tau truncation in the pathogenesis of Alzheimer's disease: a narrative review. *Neural Regen Res* 2024; 19(6):1221–1232.[DOI: 10.4103/1673-5374.385853]. *(2023 JIF: 5.91)*

10. Demirci GM, **Kittler PM, Phan HTT, Gordon AD, Flory MJ,** …, Tsai CL: Predicting mental and psychomotor delay in very pre-term infants using machine learning. *Pediatr Res* 2024; 95(3):668–678. [DOI: 10.1038/s41390-023-02713-z]. *(2023 JIF: 3.1)*

11. DiFilippo A, Jonaitis E, Makuch R, Gambetti B, Fleming V, …, **Krinsky-McHale S,** …, Christian B: Measurement of synaptic density in Down syndrome using PET imaging: a pilot study. *Sci Rep* 2024; 14(1):4676. [DOI: 10.1038/s41598-024-54669-7]. *(2023 JIF: 3.8)*

12. DiProspero N, Sathishkumar M, Janecek J, Smith A, …, **Krinsky-McHale S,** …, Yassa MA: Neurofilament light chain concentration mediates the association between regional medial temporal lobe structure and memory in adults with Down syndrome. *Alzheimers Dement (Amst)* 2024; 16(1):e12542. [DOI: 10.1002/dad2.12542] *(2023 JIF: 4.0)*

13. Dy-Hollins ME, Carr SJ, Essa A, …, **Lyon GJ,** …, Mathews CA, Scharf JM: The challenge of examining social determinants of health in people living with Tourette syndrome. *Pediatr Neurol* 2024; 155:55–61. [DOI: 10.1016/j.pediatrneurol.2024.02.008] 38608551. *(2023 JIF: 3.2)*

14. Fringuello A, Colbourn R, **Goodman J,** Michelson H, Ling D, Hrabetova S: Rapid volume pulsations of the extracellular space accompany epileptiform activity in trauma-injured neocortex and depend on the sodium-bicarbonate cotransporter NBCe1. *Epilepsy Res* 2024; 107337. 10.1016/j.eplepsyres.2024.107337. [DOI:10.1016/j.eplepsyres.2024.107337] *(2023 JIF: 2.0)*

15. **Goodman JH:** Closed-loop low-frequency stimulation: seizure reduction and more. *Epilepsy Currents* 2024;0(0). [DOI:10.1177/15357597241280683] *(2023 JIF: 5.8)*

16. **Goodman JH:** Hippocampal inhibitory neuron loss is epileptogenic but not likely a sole cause of mesial temporal lobe epilepsy (MTLE). *Epilepsy Currents* 2024;0(0). [DOI: 10.1177/15357597241303611] *(2023 JIF: 5.8)*

17. **Goodman JH:** It matters who starts the fire in mesial temporal lobe epilepsy. *Epilepsy Currents* 2024;0(0). [DOI:10.1177/15357597241238901] *(2023 JIF: 5.8)*

18. **Goodman JH:** Sometimes steroids get a bad rap. *Epilepsy Currents* 2024;0(0). [DOI:10.1177/15357597241254279] *(2023 JIF: 5.8)*

19. Hillerstrom H, Fisher R, Janicki MP, …, **Krinsky-McHale S,** …, Zaman SH, Zetterberg H: Adapting prescribing criteria for amyloid-targeted antibodies for adults with Down syndrome. *Alzheimers Dement* 2024; 20(5):3649–3656. [DOI: 10.1002/alz.13778] *(2023 JIF: 13.0)*

20. Iban-Arias R, Wang SH, Soares Dias Portela A, Yang EJ, Griggs E, Masieri S, **Hu W,** Chen LC, Pasinetti GM: Exposure to the World Trade Center particulate matter alters the gut-brain axis in early onset Alzheimer's disease mice. *J Alzheimers Dis* 2024; 100(s1):S305-S325. [DOI: 10.3233/JAD-240635]. *(2023 JIF: 3.4)*

21. **Iqbal K:** Tau and Alzheimer's disease: past, present and future. *Cytoskeleton (Hoboken).* 2024 Jan; 81(1):116–121. [DOI: 10.1002/cm.21822]. *(2023 JIF: 2.4)*

22. **Keane SP, Chadman KK, Gomez AR, Hu W:** Pros and cons of narrow- versus wide-compartment rotarod apparatus: an experimental study in mice. *Behav Brain Res* 2024 2024; 463:114901. [DOI: 10.1016/j.bbr.2024.114901]. *(2023 JIF: 2.6)*

23. Kim-McManus O, Gleeson JG, …, **Lyon GJ,** Schule R, Yu T: A framework for N-of-1 trials of individualized gene-targeted therapies for genetic diseases. *Nat Commun* 2024;15(1):9802. [DOI: 10.1038/s41467-024-54077-5]. *(2023 JIF: 14.7)*

24. **Kleyner R, Ung N, Arif M, Marchi E, Amble K, Gavin M, Madrid R, Lyon G:** *ITPR1*-associated spinocerebellar ataxia with craniofacial features-additional evidence for germline mosaicism. *Cold Spring Harb Mol Case Stud* 2024; 9(4):a006303. [DOI: 10.1101/mcs.a006303]. *(2023 JIF: 1.8)*

25. Lesmann H, Hustinx A, …, **Marchi E,** …, **Lyon GJ,** Krawitz PM, Hsieh TC: GestaltMatcher Database - A global reference for the facial phenotypic variability of rare human diseases. *medRxiv* [Preprint]. 2024 Mar 8:2023.06.06.23290887. [DOI: 10.1101/2023.06.06.23290887]

26. **Listwan TA, Krinsky-McHale SJ, Kovacs CM,** …, **Pang DI**, …, **Zigman WB,** Silverman W: Prodromal Alzheimer's disease can affect activities of daily living for adults with Down syndrome. *Alzheimers Dement (Amst)* 2024; 16(1):e12562. [DOI: 10.1002/dad2.12562] *(2023 JIF: 4.0)*

27. **London EB,** Zimmerman-Bier BL, **Yoo JH,** Gaffney JW. High-dose propranolol for severe and chronic aggression in autism spectrum disorder: a pilot, double-blind, placebo-controlled, randomized crossover study. *J Clin Psychopharmacol* 2024; 44(5):462–467. [DOI: 10.1097/JCP.0000000000001895]. *(2023 JIF: 2.9)*

28. Luchsinger JA, **Pang D, Krinsky-McHale SJ,** …, Lee JH, Silverman W, **Zigman WB:** Obesity, diabetes and their metabolic correlates in middle-aged adults with Down syndrome. *J Intellect Disabil Res* 2024; 68(3):212–222. [DOI: 10.1111/jir.13103]. *(2023 JIF: 2.1)*

29. **Lyon GJ, Longo J, Garcia A, Inusa F, Marchi E, Shi D,** …, **Nashat MA, Bolton D:** Evaluating possible maternal effect lethality and genetic background effects in *Naa10* knockout mice. *PLoS One* 2024; 19(5):e0301328. [DOI: 10.1371/journal.pone.0301328]

30. **Makwana R, Christ C, Marchi E, Harpell R, Lyon GJ:** Longitudinal adaptive behavioral outcomes in Ogden syndrome by seizure status and therapeutic intervention. *Am J Med Genet A* 2024; 194(9):e63651. [DOI: 10.1002/ajmg.a.63651] *(2023 JIF: 2.0)*

31. **Makwana R, Christ C, Marchi E, Harpell R, Lyon GJ:** A natural history of NAA15-related neurodevelopmental disorder through adolescence. *medRxiv* [Preprint]. 2024 Jul 26:2024.04.20.24306120. [DOI: 10.1101/2024.04.20.24306120]

32. Mottolese N, Loi M, Trazzi S, …, **Iqbal K,** Medici G, Ciani E: Effects of a ciliary neurotrophic factor (CNTF) small-molecule peptide mimetic in an in vitro and in vivo model of CDKL5 deficiency disorder. *J Neurodev Disord* 2024; 16(1):65. [DOI: 10.1186/s11689-024-09583-4] *(2023 JIF: 4.1)*

33. **Patel R, Makwana R, Christ C, Marchi E, Ung N, Harpell R,** …, **Lyon GJ,** Whitehead MT: Neuroanatomical features of NAA10- and NAA15-related neurodevelopmental syndromes. *medRxiv* [Preprint]. 2024 Jun 25:2024.06.24.24309433. [DOI: 10.1101/2024.06.24.24309433].

34. **Patel R, Park AY, Marchi E,** Gropman AL, Whitehead MT, **Lyon GJ:** Ophthalmic manifestations of NAA10-related and NAA15-related neurodevelopmental syndrome: analysis of cortical visual impairment and refractive errors. *Am J Med Genet A* 2024 Dec;194(12):e63821. [DOI: 10.1002/ajmg.a.63821]. *(2023 JIF: 2.0)*

35. Rodrigues B, …**, Glicksman A, Tortora N, Nolin SL,** Nogueira AJA, Jorge P: FMR1 allelic complexity in premutation carriers provides no evidence for a correlation with age at amenorrhea. *Reprod Biol Endocrinol* 2024; 22(1):71. [DOI: 10.1186/s12958-024-01227-5]. *(2023 JIF: 4.2)*

36. **Salafia CM,** Rukat C, Dygulska B, Miller RK, Misra DP: Placental chronic inflammatory histopathology and fetal growth in a cohort with universal placental examination. *Placenta* 2024; 154:193–200. [DOI: 10.1016/j.placenta.2024.06.020]. *(2023 JIF: 3.0)*

37. **Sarino KP, Guo L, Yi E, Park J, Kierzkowska O, Carter D, Marchi E, Lyon GJ:** Assessment of adaptive functioning and the impact of seizures in KBG syndrome. *Am J Med Genet A*. 2024 Oct 4:e63896. [DOI: 10.1002/ajmg.a.63896]. Epub ahead of print. *(2023 JIF: 1.7)*

38. Schworer EK, Handen BL, …, **Krinsky-McHale SJ,** … Hartley SL; Alzheimer Biomarker Consortium-Down Syndrome: Cognitive and functional performance and plasma biomarkers of early Alzheimer's disease in Down syndrome. *Alzheimers Dement (Amst)* 2024; 16(2):e12582. [DOI: 10.1002/dad2.12582]. *(2023 JIF: 4.0)*

39. **Shah RG, Salafia CM, Girardi T, Rukat C**, …, Miller RK; program collaborators for Environmental influences on Child Health Outcomes: Maternal affective symptoms

and sleep quality have sex-specific associations with placental topography. *J Affect Disord* 2024; 360:62–70. [DOI: 10.1016/j.jad.2024.05.108]. *(2023 JIF: 4.9)*

40. Song Y, **Dai CL,** Shinohara M, **Chyn Tung Y,** …, **Gong CX,** Lovell JF: A pentavalent peptide vaccine elicits Aβ and tau antibodies with prophylactic activity in an Alzheimer's disease mouse model. *Brain Behav Immun* 2024; 122:185–201. [DOI: 10.1016/j.bbi.2024.08.028]. *(2023 JIF: 8.8)*

41. Tan D, Liu Y, Luo H, …, **Zhong N** (co-corresponding author), Zhang H, Xiong H: A novel mouse model for LAMA2-related muscular dystrophy: analysis of molecular pathogenesis and clinical phenotype. *eLife* 2024; 13:RP94288 https://doi.org/10.7554/eLife.94288.2. *(2023 JIF: 6.4)*

42. Tsetsos F, …; PGC TS Working Group; TSAICG, **including Lyon G**; …, Hoekstra PJ, Paschou P: Genome-wide association study points to novel locus for Gilles de la Tourette syndrome. *Biol Psychiatry* 2024; 96(2):114–124. [DOI: 10.1016/j.biopsych.2023.01.023]. *(2023 JIF: 4.0)*

43. Varkey H, **Phan H, Kittler P, Gordon A**, Torres EB: Infants on the move: bibliometric analyses of observational vs. digital means of screening infant development. *Front Integr Neurosci* 2024; 17:1251252. [DOI: 10.3389/fnint.2023.1251252] *(2023 JIF: 2.6)*

44. **Wegiel Jarek, Chadman K, London E, Wisniewski T, Wegiel Jerzy:** Contribution of the serotonergic system to developmental brain abnormalities in autism spectrum disorder. *Autism Res* 2024; 17(7):1300–1321. [DOI: 10.1002/aur.3123]. *(2023 JIF: 5.3)*

45. Wesely J, Rusielewicz T, …, **Makwana R, Marchi E,** …, Moroziewicz D; NYSCF Global Stem Cell Array® Team; …, **Lyon GJ:** A repository of Ogden syndrome patient derived iPSC lines and isogenic pairs by X-chromosome screening and genome-editing. *bioRxiv* [Preprint]. 2024 Sep 28:2024.09.28.615067. [DOI: 10.1101/2024.09.28.615067]

46. Wisch JK, McKay NS, …, **Krinsky-McHale SJ,** …, Ances BM; Alzheimer's Biomarker Consortium-Down syndrome; Dominantly Inherited Alzheimer Network: Comparison of tau spread in people with Down syndrome versus autosomal-dominant Alzheimer's disease: a cross-sectional study. *Lancet Neurol* 2024 May; 23(5):500–510. [DOI: 10.1016/S1474-4422(24)00084-X]. *(2023 JIF: 46.5)*

47. Wu D, Yang J, Liu C, …, **Marchi E,** …, **Lyon GJ,** …, Wang K: GestaltMML: enhancing rare genetic disease diagnosis through multimodal machine learning combining facial images and clinical texts. *ArXiv* [Preprint]. 2024 Apr 22:arXiv:2312.15320v2. PMID: 38711434; PMCID: PMC11071539.

48. Xicota L, Cosentino S, …, **Krinsky-McHale S,** Handen BL, …, Barral S; Long-Life Family Study (LLFS); Alzheimer's Disease Genetic Consortium (ADGC); Alzheimer's Biomarkers Consortium-Down Syndrome (ABC-DS): Whole genome-wide sequence analysis of long-lived families (Long-Life Family Study) identifies MTUS2 gene

associated with late-onset Alzheimer's disease. *Alzheimers Dement* 2024; 20(4):2670–2679. [DOI: https://doi.org/10.1002/alz.13718]. *(2023 JIF: 13.0)*

49. Xicota L, Dang LT, Lee A, **Krinsky-McHale S, Pang D,** Melilli L, O'Bryant S, Henson RL, Laymon C, Lai F, Rosas HD, Ances B, Lott I, Hom C, Christian B, Hartley S, Zaman S, Head E, Mapstone M, Jin Z, Silverman W, Schupf N, Handen B, Lee JH; Alzheimer's Biomarker Consortium – Down Syndrome (ABC-DS): The effects of mosaicism on biological and clinical markers of Alzheimer's disease in adults with Down syndrome. *EBioMedicine* 2024; 110:105433. [DOI: 10.1016/j.ebiom.2024.105433]. *(2023 JIF: 9.7)*

50. Zeng X, Mai F, Li Z, Yuan T, Zhang J, Zhang Y, **Zhong N:** Newborn screening for congenital hearing impairment: a 17-year experience for improving quality of healthcare. *J Community Med Public Health* 2024; 8:400. [DOI: https://doi.org/10.29011/2577-2228.100400]

Last updated 2/26/2025