

IBR STAFF PUBLICATIONS—2023

with 2022 Journal Impact Factors (JIFs) (released June 2023), if available

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2023 IBR STAFF PUBLICATIONS

1. Araldi GL, **Hwang YW**: Development of novel fluorinated polyphenols as selective inhibitors of DYRK1A/B kinase for treatment of neuroinflammatory diseases including Parkinson's disease. *Pharmaceuticals (Basel)* 2023; 16(3):443. [DOI: 10.3390/ph16030443]. (2022 JIF: 4.6)
2. Basheer N, Smolek T, Hassan I, **Liu F, Iqbal K, ...**, Novak P: Does modulation of tau hyperphosphorylation represent a reasonable therapeutic strategy for Alzheimer's disease? From preclinical studies to the clinical trials. *Mol Psychiatry* 2023; 28(6): 2197–2214. [DOI: 10.1038/s41380-023-02113-z]. (2022 JIF: 11)
3. Belbachir N, Wu Y, Shen M, ..., **Lyon GJ**, Ma N, Wu JC: Studying long QT syndrome caused by NAA10 genetic variants using patient-derived induced pluripotent stem cells. *Circulation* 2023; 148:1598–1601. [DOI: 10.1161/CIRCULATIONHA.122.061864] (2022 JIF: 37.8)
4. Boerwinkle AH, Gordon BA, Wisch J, Flores S, ...; Alzheimer's Biomarker Consortium-Down Syndrome, including **Krinsky-McHale S**; Dominantly Inherited Alzheimer Network: Comparison of amyloid burden in individuals with Down syndrome versus autosomal dominant Alzheimer's disease: a cross-sectional study. *Lancet Neurol* 2023; 22(1):55–65. [DOI: 10.1016/S1474-4422(22)00408-2]. (2022 JIF: 48)
5. Carey ME, Rando J, Melnyk S, James SJ, ..., **Salafia C, ...**, Lyall K: Examining associations between prenatal biomarkers of oxidative stress and ASD-related outcomes using quantile regression. *J Autism Dev Disord* 2023; 53:2975–2985. [DOI: 10.1007/s10803-022-05625-9]. (2022 JIF: 3.9)
6. **Carter DC, Kierzkowska O, Sarino K, Guo L, Marchi E, Lyon GJ**: Ocular manifestations in a cohort of 43 patients with KBG syndrome. *medRxiv* 2023; 2023.05.31.23290743. [DOI: 10.1101/2023.05.31.23290743]
7. Castellanos MR, Fanous E, ..., **Flory MJ, ...**, Dhar M, Starr A, Strange TJ: Expression patterns and clinical significance of estrogen receptor in non-small cell lung cancer. *Pathol Res Pract* 2023; 241:154298. [DOI: 10.1016/j.prp.2022.154298] (2022 JIF: 2.8)
8. **Chadman KK, Adayev T, ...**, **Dai C-L, Goodman JH, Meeker H, Dolzhanskaya N, Velinov M**: Efficient delivery of FMR1 across the blood brain barrier using AAVphp construct in adult FMR1 KO mice suggests the feasibility of gene therapy for fragile X syndrome. *Genes* 2023; 14:505. [DOI: 10.3390/genes14020505] (2022 JIF: 3.5)

9. Chen X, ..., **Dong X**, Liu J, ..., **Ju W**, ..., Cheng L, **Zhong N** and On behalf of the Chinese Consortium for Prenatal Ultrasound Screening of Congenital Heart Defects: Incidence, distribution, disease spectrum, and genetic deficits of congenital heart defects in China: implementation of prenatal ultrasound screening identified 18,171 affected fetuses from 2,452,249 pregnancies. *Cell & Bioscience* 2023; [doi.org/10.1186/s13578-023-01172-7]. (2022 JIF: 7.5)
10. Chu D, Yang X, Wang J, Zhou Y, Gu JH, Miao J, Wu F, **Liu F**: Tau truncation in the pathogenesis of Alzheimer's disease: a narrative review. *Neural Regen Res* [DOI: 10.4103/1673-5374.385853]. Epub 2023 Sep 22. (2022 JIF: 6.1)
11. Coyte RM, Darrah TH, ..., **Salafia CM**, **Shah R**, Love T, Miller RK: Comparison of trace element concentrations in paired formalin-fixed paraffin-embedded and frozen human placentae. *Placenta* 2023; 131:98–103. [DOI: 10.1016/j.placenta.2022.12.003] (2022 JIF: 3.8)
12. 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* 2023 Jul 27. [DOI: 10.1038/s41390-023-02713-z]. Epub ahead of print. (2022 JIF: 3.6)
13. **DiLiberto E**, **Phatarpekar S**, **Theodorakis K**, **Chadman KK**: Does the stranger mouse strain matter to female BTBR mice? *Behav Brain Res* 2023; 437:114132. [DOI: 10.1016/j.bbr.2022.114132] (2022 JIF: 2.7)
14. Dingemans AJM, Hinne M, Truijien KMG, ..., **Lyon GJ**, ..., Vissers LELM, de Vries BAB: PhenoScore quantifies phenotypic variation for rare genetic diseases by combining facial analysis with other clinical features using a machine-learning framework. *Nat Genet* 2023; 55(9):1598–1607. [DOI: 10.1038/s41588-023-01469-w]. (2022 JIF: 30.8)
15. Dong X, **Zhong N** (eds.): *New Perspectives on Asthma*. 2023. *IntechOpen* [DOI:10.5772/intechopen.101004]
16. Fleming V, Helsel BC, ..., **Krinsky-McHale S**, ..., Lee JH, Hartley SL; Alzheimer's Biomarker Consortium –Down Syndrome (ABC-DS) Consortium: Weight loss and Alzheimer's disease in Down syndrome. *J Alzheimers Dis* 2023; 91:1215–1227. [DOI: 10.3233/JAD-220865]. (2022 JIF: 4.0)
17. **Frackowiak J**, **Mazur-Kolecka B**: Intraneuronal accumulation of amyloid- β peptides as the pathomechanism linking autism and its co-morbidities: epilepsy and self-injurious behavior - the hypothesis. *Front Mol Neurosci* 2023; 16:1160967. [DOI: 10.3389/fnmol.2023.1160967]. (2022 JIF: 4.8)
18. Godler DE, **Brown WT**: Editorial for the Fragile X Syndrome Genetics Special Issue: May 2023. *Genes (Basel)*. 2023; 14:1148. [DOI: 10.3390/genes14061148]. (2022 JIF: 3.5)

19. Gordon LB, **Brown WT**, Collins FS: Hutchinson-Gilford Progeria Syndrome. 2003 Dec 12 [updated 2023 Oct 19]. 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; 1993–2023. [PMID: 20301300]
20. Hartley SL, Fleming V, Schworer EK, ..., **Krinsky-McHale S**, ..., Christian B; Alzheimer Biomarker Consortium-Down Syndrome: Timing of Alzheimer's disease by intellectual disability level in Down syndrome. *J Alzheimers Dis* 2023; 95:213–225. [DOI: 10.3233/JAD-230200]. (2022 JIF: 4.0)
21. **He XY, Dobkin C**, Brown WT, **Yang SY**: Infantile neurodegeneration results from mutants of 17 β -hydroxysteroid dehydrogenase type 10 rather than A β -binding alcohol dehydrogenase. *Int J Mol Sci* 2023; 24(10):8487. [DOI: 10.3390/ijms24108487]. (2022 JIF: 5.6)
22. **He XY, Frackowiak J, Dobkin C, Brown WT, Yang SY**: Involvement of type 10 17 β -hydroxysteroid dehydrogenase in the pathogenesis of infantile neurodegeneration and Alzheimer's disease. *Int J Mol Sci* 2023; 24(24):17604. [DOI: 10.3390/ijms242417604]. (2022 JIF: 5.6)
23. Im E, Jiang Y, ..., **Wegiel J**, Lee JH, Nixon RA: Lysosomal dysfunction in Down syndrome and Alzheimer mouse models is caused by v-ATPase inhibition by Tyr⁶⁸²-phosphorylated APP β CTF. *Sci Adv* 2023; 9(30):eadg1925. Epub 2023 Jul 26. [DOI: 10.1126/sciadv.adg1925]. (2022 JIF: 13.6)
24. **Iqbal K**: Tau and Alzheimer's disease: past, present and future. *Cytoskeleton (Hoboken)*. 2023 Dec 21. [DOI: 10.1002/cm.21822]. Epub ahead of print. (2022 JIF: 2.9)
25. Jain P, ...; Psychiatric Genomics Consortium Tourette Syndrome Working Group (PGC-TS), including **Lyon GL**; EMTICS collaborative group; Dietrich A; TS-EUROTRAIN Network; ..., Paschou P: Polygenic risk score-based phenome-wide association study identifies novel associations for Tourette syndrome. *Transl Psychiatry* 2023; 13:69. [DOI: 10.1038/s41398-023-02341-5] (2022 JIF: 6.8)
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27. **Kierzkowska O, Sarino K, Carter D, Guo L, Marchi E, ..., Lyon GJ**: Documentation and prevalence of prenatal and neonatal outcomes in a cohort of individuals with KBG syndrome. *Am J Med Genet A* 2023; 191(9):2364–2375. [DOI: 10.1002/ajmg.a.63311]. (2022 JIF: 3.1)
28. **Kleyner R, Ung N, ..., 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* 2023 Oct 11:mcs.a006303. [DOI: 10.1101/mcs.a006303]. Online ahead of print. (2022 JIF: 0.93)

29. Lesmann H, **Lyon GJ**, Caro P, Abdelrazek IM, ..., **Marchi E**, ..., Hsieh TC: GestaltMatcher Database—a FAIR database for medical imaging data of rare disorders. *medRxiv* [Preprint]. 2023 Jun 10:2023.06.06.23290887. [DOI: 10.1101/2023.06.06.23290887].
30. **Li L, Miao J, Jiang Y, Dai CL, Iqbal K, Liu F, Chu D**: Passive immunization inhibits tau phosphorylation and improves recognition learning and memory in 3xTg-AD mice. *Exp Neurol* 2023; 362:114337. [DOI: 10.1016/j.expneurol.2023.114337] (2022 JIF: 5.3)
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32. Luchsinger JA, **Pang D, Krinsky-McHale SJ**, Schupf N, ..., Silverman W, **Zigman WB**: Obesity, diabetes and their metabolic correlates in middle-aged adults with Down syndrome. *J Intellect Disabil Res* 2023 Oct 29. [DOI: 10.1111/jir.13103]. Online ahead of print. (2022 JIF: 3.6)
33. **Lyon GJ, Longo J, Garcia A, Inusa F, Marchi E, Shi D, ..., Nashat M, Bolton D**: Evaluating possible maternal effect lethality of Naa10 knockout mice, and modulation of phenotypes for embryonic and neonatal lethality by genetic background and environment. *bioRxiv* [Preprint]. 2023 Apr 27:2023.04.27.538618. [DOI: 10.1101/2023.04.27.538618].
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35. Mejia-Bautista A, Michelson HB, Sanjana A, Famuyiwa O, **Goodman JH**, Ling DSF: 4-AP challenge reveals that early intervention with brivaracetam prevents posttraumatic epileptogenesis in rats. *Epilepsy Res* 2023; 196:107217. [DOI: 10.1016/j.eplepsyres.2023.107217]. (2022 JIF: 2.2)
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39. **Shah R, Salafia C, Girardi T, Persaud P**, Brunner J, ..., Miller RK: Placental growth and development analyzed through 2D and 3D fractals. [Internet]. In: Ouadfeul S-A (ed.), *Fractal Analysis —Applications and Updates*. IntechOpen; 2023. Available from: <http://dx.doi.org/10.5772/intechopen.1001929>
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