

Curriculum Vitae

Contact Details

Name: **Mag.rer.nat. Martin Werner Breuss, PhD**

Position: **Assistant Professor**
University of Colorado, School of Medicine, Department of Pediatrics, Section of Genetics and Metabolism

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Education

2009 **Molecular Biology (Diplomstudium)**, University of Vienna, Austria (Mag.rer.nat., Master's equivalent)

2013 **Molecular Biology (VBC PhD Program)**, University of Vienna, Austria | Institute of Molecular Pathology, Vienna, Austria

Positions

2013-2015 **Postdoctoral Researcher**, Institute of Molecular Pathology, Vienna, Austria; laboratory of Dr. David Keays

2015-2019 **Postdoctoral Researcher**, Department of Neurosciences, UC San Diego, Howard Hughes Medical Institute, Rady Children's Institute for Genomic Medicine, La Jolla, CA, USA; laboratory of Dr. Joseph Gleeson

2019-2020 **Project Scientist**, Department of Neurosciences, UC San Diego, Rady Children's Institute for Genomic Medicine, La Jolla, CA, USA; laboratory of Dr. Joseph Gleeson

2021- **Assistant Professor**, Department of Pediatrics, Section of Clinical Genetics and Metabolism, CU Anschutz Medical Campus, Aurora, CO, USA

Selected and Invited Talks

11/2016 USA EMBO fellows' meeting 2016, Boston, USA (selected for an oral presentation)

07/2017 Invited talk at the IMP Vienna, Austria (Host: Dr. David Keays)

10/2017 American Society of Human Genetics Meeting, Orlando, USA (selected for a plenary talk)

06/2018	EMBO fellows' meeting 2018, Heidelberg, Germany (selected for an oral presentation)
09/2018	Invited talk at San Diego State University, San Diego, USA (Host: Dr. Elizabeth Waters)
10/2018	Germ Cells Conference (CSHL) 2018, Cold Spring Harbor, USA (selected for a platform talk)
02/2020	Invited talk at King's College, London, Centre for Developmental Neurobiology (Host: Dr. Oscar Marin)
02/2020	Invited talk at the University of Colorado Anschutz Medical Campus (Host: Dr. Shawn McCandless)
03/2021	Internal invited talk (CU Anschutz) for the CSD graduate program (Host: Dr. Jeffrey Moore)
04/2021	Internal invited talk (CU Anschutz) for the HMGG (Host: Dr. Tamim Shaikh)
04/2021	Internal invited talk (CU Anschutz) for the Developmental Biology Section in the Department of Pediatrics (Host: Dr. Bruce Appel)
08/2021	Invited talk at Xuanwu Hospital, Capital Medical University, Beijing (Host: Dr. Guoliang Chai)
11/2021	Internal invited talk at the HMGG annual retreat (Host: HMGGP students)
07/2022	Invited talk at Boston Children's Hospital/Harvard Medical School Data Science Seminar Series (Host: Dr. Ann Poduri)
08/2022	Invited talk at Beijing Brain Conference – Big Data and Intelligent Medicine for Brain Diseases Symposium
10/2022	Invited talk at the 12 th European Congress of Andrology
11/2022	Invited talk at the National Center for Neurological Disorders, Huaxia Forum
02/2023	Invited talk at Case Western Reserve University Department of Genetics and Genome Sciences (Host: Dr. Ashleigh Schaffer)
04/2023	Invited talk at CRU326 'Male Germ Cells', University of Münster/University Hospital, Germany (Host: Dr. Nina Neuhaus)
10/2023	Internal invited talk at the HMGG annual retreat (Host: HMGGP students)
11/2023	Invited Session selected for the 2023 AHSG meeting (proposer and moderator)
12/2023	Invited talk at the Nanopore Sequencing Seminar, CU Anschutz (Host: Oxford Nanopore Technologies)
01/2024	Invited talk at Nebraska University Medical Center Genetics, Cell Biology and Anatomy Seminar Series (Host: Dr. Jared Graham)
04/2024	Internal invited talk (CU Anschutz) for the Basic & Translational Research Seminar Series (Host: Department of Pediatrics; Dr. Fred Suchy)
06/2024	European Society of Human Genetics Meeting 2024, Berlin, Germany (invited Educational Session talk)

06/2024	Internal invited talk (CU Anschutz) for the MOLB graduate program (Host: Dr. Rytis Prekeris)
10/2024	Internal invited talk at the MOLB annual retreat (Host: MOLB students)
11/2024	Internal invited talk at the CSD shop talk series (Host: CSD program)
02/2025	Keystone Symposium: Somatic Mosaicism in Human Development, Aging, and Diseases 2025 (selected for an oral presentation)
03/2025	Invited talk at the University of Colorado Colorado Springs Department of Biology and the Biology Graduate Program (Host: Dr. Jeremy Bono)

Teaching

2021-2023	Biomedical Sciences Program – Journal Club (first-year students), discussion lead (HMGGP representative)
2022-	Rigor and Reproducibility in Biomedical Research (Co-Director); BMSC 7812
2022	Responsible Conduct in Research (Co-Instructor for discussion groups); BMSC 7811
2022-	Genetics of Development, Disease, and Regeneration (Course Instructor); CSDV 7607 (bi-annually: 2022, 2024)
2023-	Foundations in Biomedical Sciences, Block III: Genetics (Block Instructor for Human Genetics); BMSC 7806
2024-	Foundations in Genetics (Instructor); BSBT 6075

Mentorship

Graduate program member in PhD programs: Human Medical Genetics and Genomics (HMGG), Cell Biology, Stem Cells, and Development (CSD), Molecular Biology (MOLB) program, the Biomedical Sciences Program (BSP), Medical Scientists Training Program (MSTP). Graduate program member in Master's programs: Biomedical Sciences and Biotechnology (BSBT), PATH-GDS. Research Mentor in the PATH-GREU undergraduate program. Attended the Maximizing Mentorship Training (2022; on campus), the Raising a Resilient Scientist training (2022-2023; through the NIH, remote), the Health Equity in Action Lab's (HEAL) Foundations in Equity Certificate Program (2023; on campus), the Implicit Bias Training (yearly since 2022; on campus), and the Bystander/Upstander Training (2024; on campus).

Postdoctoral Scholars

Breuss Lab

2022-2023 Dr. Jared Graham (Current: Postdoctoral Scholar, UN Medical Campus)

Other

2022-2023 Dr. Lisa Gabler (ASCINA Mentee; Current: Independent Postdoctoral Scholar/Principal Investigator, Austria)

Graduate Students

2023- Rose Doss (HMGG)

Rotation Students

2021/2022 Helen Gomez (HMGG)

2022/2023 Rose Doss, Abigail Cozart (HMGG)

2023/2024 Rachel Garstang (BSBT)

2024/2025 Nicole Costantino (CSD), Juan Avila Pagan (HMGG), Johannes Strauss (PATH-GDS), Emily Decurtis, Sierra Simmermann (MOLB)

Comprehensive Exam/Thesis Committee

2021-2024 Kyle Northington (CSD)

2022- Abigail Mumme-Monheit (CSD)

2022- Jason Wilson (HMGG)

2022- Lauren Dunn (HMGG)

2023- Harrison Wells (CSD; chair)

2024- Megan Kraus (HMGG)

2024- Abigail Cozart (HMGG; chair until 2025)

2024- Ashlyn Stahly (CSD)

Undergraduate Students

2022-2024 Josh Mejia (Boettcher Collaboration Grant, Student Worker)

2024 Ava Warren (Summer Child Health Research Internship)

High School Students

2023 Vinisha Tripathi (Summer Intern)

2023-2024 Meena Abdali (SRS Intern)

Before 2021

Graduate Students (UC San Diego: Dr. Shereen Ghosh); Master's Students (IMP Vienna: Dr. Jasmin Morandell, Dr. Andi Harley Hansen); Post-Bac (IMP Vienna: Kelvin Chan; UC San Diego: Thai Nguyen); Undergraduate Students (IMP Vienna: Freddy Vonberg; UC San Diego: An Nguyen)

Service

Internal

2021- Section of Genetics and Metabolism Chalk Talks Organizer

2021- Graduate Programs Interviewer (all active programs)

2021-2023 HMGG Retreat Committee Co-Chair

2022 NRSA Mock Study Section Reviewer

2022	Residency Programs Interviewer (Genetics, PSTP)
2022-	HMGG Steering Committee Member
2022-	BSP Admissions Committee Member
2022-2023	Vice Chair of Research (Pediatrics) Interview Committee Member
2022-2023	Faculty (Section of Genetics and Metabolism) Search Committee Member
2023	Mentored Scholarly Activity Projects Poster Judge
2023-2024	PIKE-PREP mock reviewer
2023-	HMGG Seminar Committee Chair
2023-	Advisory committee to the Vice Chair of Basic Research (Pediatrics) Member
2024	Focus group participant for the Access to Biobank Committee (ABC) Optimization Project

Funding Agencies

2020	Mega-grants (Ministry of Science and Higher Education of the Russian Federation) External Reviewer
2023	NIH Study Section (DEV2) Early Career Reviewer

External Review

2021	KU Leuven (faculty candidate review; application retracted before review) External Reviewer
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Societies

2018-2022	ASHG Meeting Platform Moderator (2018, 2019, 2022)
2021	ASHG Meeting Abstract Reviewer (2021)
2024	ASHG Career Development Committee Member
2025	ASHG Career Development Committee Incoming Chair
2025	ASHG Career Development Committee Training Excellence Spotlight Subgroup Member

Journals

<i>Ad hoc</i> reviewer:	Nature, eLife, American Journal of Human Genetics, Development, Genome Research, Trends in Genetics, American Journal of Psychiatry, PLOS Biology, PLOS Genetics, BMC Biology, BMC Pediatrics, Scientific Reports, Journal of Molecular Diagnostics, Heliyon, European Journal of Medical Genetics, Neurology: Genetics, Journal of Pediatric Neurology, Progress in Neurobiology, Epilepsy Research, Journal of Molecular Modeling, Journal of Movement Disorders, Communications Biology, Frontiers in Genetics, Frontiers in Neuroscience, Frontiers in Pediatrics, Frontiers in Reproductive Health
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Outreach

2020	JRNL club, recorded presentation to disseminate Breuss et al. Nature Medicine 2020
2021	Presentation on 'Genotypes and Phenotypes in Humans' to 7 th -grade students at the South Middle School in Nemo, South Dakota; JRNL club, recorded presentation to disseminate Yang*, Breuss*, et al. Cell 2021
2022	Presented at the Denver Public Schools CareerSpark program to 5 th -grade students at the Valdez Elementary School in Denver, Colorado
2023	Interviewed and quoted as an independent expert for the news article "Your cells don't have the genome you were born with. Project aims to chart impact, of new mutations.", published in Science on August 15 th .
2024	Interview with Project Bridge Colorado (Anschutz Medical Campus) for 'Meet the Scientist'

Memberships

2017-	American Society of Human Genetics
2018-	European Society of Human Genetics
2024-	NHGRI Technology Development Coordinating Center

Scholarships and Awards

2004	Excellence Scholarship from the University of Vienna
2015-2017	EMBO Long Term Fellowship (ALTF 174-2015), EMBO
2018-2019	Erwin Schrödinger Fellowship (J4197-B30), FWF Austria
2020	ASCINA Young Scientist Award
2022	Boettcher Investigator (see also Funding), Boettcher Foundation
2024	Translational Research Scholar (see also Funding), CU SOM

Funding

Past

2022/05-2023/04	CCTSI Pilot Grant Award TM-T-22-117, "Targeted long-read sequencing for parental haplotype determination of diagnostic variants" (PI: Breuss, Co-I: James); \$20,000.00.
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Current

- 2022/06-2025/05** Webb-Waring Biomedical Research Grant, “Elucidation of the features of germ cell mosaicism and its impact on human health” (PI Breuss); \$235,000.000.
- 2024/07-2029/04** NIH/NHGRI Katz R01 1R01HG013472, “Genome-wide assessment of transcriptional state history”(PI Breuss); \$1,250,000 (direct costs)
- 2024/07-** Translational Research Scholar Program, “The Mosaicism Landscape of Aging and Disease in the Colorado Biobank” (PI Breuss); \$75,000/year.

Patents

1. PCT ref. no. SD2017-181-2PCT: Assessing risk of *de novo* mutations in males (filed by UC San Diego; abandoned)

Publications

Google Scholar page:

<https://scholar.google.com/citations?user=3vW6Gf4AAAAJ&hl=en>

NCBI Bibliography:

<https://www.ncbi.nlm.nih.gov/myncbi/1beNt9tECboQt/bibliography/public/>

*shared first author; # co-corresponding authors; Breuss lab trainees/staff

Peer-reviewed original research

1. Braun A, **Breuss M**, Salzer MC, Flint J, Cowan NJ, Keays DA. Tuba8 is expressed at low levels in the developing mouse and human brain. *Am J Hum Genet.* 2010;86(5):819-22; author reply 22-3. Epub 2010/05/15. doi: 10.1016/j.ajhg.2010.03.019. PubMed PMID: 20466094; PMCID: PMC2869021.
2. Edwards A, Treiber CD, **Breuss M**, Pidsley R, Huang GJ, Cleak J, Oliver PL, Flint J, Keays DA. Cytoarchitectural disruption of the superior colliculus and an enlarged acoustic startle response in the Tuba1a mutant mouse. *Neuroscience.* 2011;195(4):191-200. Epub 2011/08/31. doi: 10.1016/j.neuroscience.2011.08.035. PubMed PMID: 21875651; PMCID: PMC3188702.
3. **Breuss M***, Heng JI*, Poirier K, Tian G, Jaglin XH, Qu Z, Braun A, Gstrein T, Ngo L, Haas M, Bahi-Buisson N, Moutard ML, Passemard S, Verloes A, Gressens P, Xie Y, Robson KJ, Rani DS, Thangaraj K, Clausen T, Chelly J, Cowan NJ, Keays DA. Mutations in the beta-tubulin gene TUBB5 cause microcephaly with structural brain abnormalities. *Cell Rep.* 2012;2(6):1554-62. Epub 2012/12/19. doi: 10.1016/j.celrep.2012.11.017. PubMed PMID: 23246003; PMCID: PMC3595605.
4. Treiber CD, Salzer MC, Riegler J, Edelman N, Sugar C, **Breuss M**, Pichler P, Cadiou H, Saunders M, Lythgoe M, Shaw J, Keays DA. Clusters of iron-rich cells in the upper beak of pigeons are macrophages not magnetosensitive neurons. *Nature.* 2012;484(7394):367-70. Epub 2012/04/13. doi: 10.1038/nature11046. PubMed PMID: 22495303.
5. Treiber CD, Salzer M, **Breuss M**, Ushakova L, Lauwers M, Edelman N, Keays DA. High resolution anatomical mapping confirms the absence of a magnetic sense system in the rostral

- upper beak of pigeons. *Commun Integr Biol.* 2013;6(4):e24859. Epub 2013/08/14. doi: 10.4161/cib.24859. PubMed PMID: 23940826; PMCID: PMC3738016.
6. Ngo L, Haas M, Qu Z, Li SS, Zenker J, Teng KS, Gunnersen JM, **Breuss M**, Habgood M, Keays DA, Heng JI. TUBB5 and its disease-associated mutations influence the terminal differentiation and dendritic spine densities of cerebral cortical neurons. *Hum Mol Genet.* 2014;23(19):5147-58. Epub 2014/05/17. doi: 10.1093/hmg/ddu238. PubMed PMID: 24833723.
7. **Breuss M***, Morandell J*, Nimpf S, Gstrein T, Lauwers M, Hochstoeger T, Braun A, Chan K, Sanchez Guajardo ER, Zhang L, Suplata M, Heinze KG, Elsayad K, Keays DA#. The Expression of Tubb2b Undergoes a Developmental Transition in Murine Cortical Neurons. *J Comp Neurol.* 2015;523(15):2161-86. Epub 2015/06/25. doi: 10.1002/cne.23836. PubMed PMID: 26105993.
8. Isrie M, **Breuss M**, Tian G, Hansen AH, Cristofoli F, Morandell J, Kupchinsky ZA, Sifrim A, Rodriguez-Rodriguez CM, Dapena EP, Doonanco K, Leonard N, Tinsa F, Moortgat S, Ulucan H, Koparir E, Karaca E, Katsanis N, Marton V, Vermeesch JR, Davis EE, Cowan NJ, Keays DA, Van Esch H. Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. *Am J Hum Genet.* 2015;97(6):790-800. Epub 2015/12/08. doi: 10.1016/j.ajhg.2015.10.014. PubMed PMID: 26637975; PMCID: PMC4678434.
9. **Breuss M**, Fritz T, Gstrein T, Chan K, Ushakova L, Yu N, Vonberg FW, Werner B, Elling U, Keays DA. Mutations in the murine homologue of TUBB5 cause microcephaly by perturbing cell cycle progression and inducing p53-associated apoptosis. *Development.* 2016;143(7):1126-33. Epub 2016/02/24. doi: 10.1242/dev.131516. PubMed PMID: 26903504.
10. **Breuss MW**, Sultan T, James KN, Rosti RO, Scott E, Musaev D, Furia B, Reis A, Sticht H, Al-Owain M, Alkuraya FS, Reuter MS, Abou Jamra R, Trotta CR, Gleeson JG. Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. *Am J Hum Genet.* 2016;99(1):228-35. Epub 2016/07/09. doi: 10.1016/j.ajhg.2016.05.023. PubMed PMID: 27392077; PMCID: PMC5005448.
11. **Breuss MW***, Hansen AH*, Landler L, Keays DA#. Brain-specific knockin of the pathogenic Tubb5 E401K allele causes defects in motor coordination and prepulse inhibition. *Behav Brain Res.* 2017;323:47-55. Epub 2017/01/29. doi: 10.1016/j.bbr.2017.01.029. PubMed PMID: 28130172.
12. **Breuss MW**, Nguyen T, Srivatsan A, Leca I, Tian G, Fritz T, Hansen AH, Musaev D, McEvoy-Venneri J, James KN, Rosti RO, Scott E, Tan U, Kolodner RD, Cowan NJ, Keays DA, Gleeson JG. Uner Tan syndrome caused by a homozygous TUBB2B mutation affecting microtubule stability. *Hum Mol Genet.* 2017;26(2):258-69. Epub 2016/12/26. doi: 10.1093/hmg/ddw383. PubMed PMID: 28013290; PMCID: PMC6075555.
13. **Breuss MW**, Nguyen A, Song Q, Nguyen T, Stanley V, James KN, Musaev D, Chai G, Wirth SA, Anzenberg P, George RD, Johansen A, Ali S, Zia-Ur-Rehman M, Sultan T, Zaki MS, Gleeson JG. Mutations in LNPK, Encoding the Endoplasmic Reticulum Junction Stabilizer Lunapark, Cause a Recessive Neurodevelopmental Syndrome. *Am J Hum Genet.* 2018;103(2):296-304. Epub 2018/07/24. doi: 10.1016/j.ajhg.2018.06.011. PubMed PMID: 30032983; PMCID: PMC6080764.
14. Gstrein T, Edwards A, Pristoupilova A, Leca I, **Breuss M**, Pilat-Carotta S, Hansen AH, Tripathy R, Traunbauer AK, Hochstoeger T, Rosoklija G, Repic M, Landler L, Stranecky V, Durnberger G, Keane TM, Zuber J, Adams DJ, Flint J, Honzik T, Gut M, Beltran S, Mechtler K, Sherr E, Kmoch S, Gut I, Keays DA. Mutations in Vps15 perturb neuronal migration in mice and are

- associated with neurodevelopmental disease in humans. *Nat Neurosci.* 2018;21(2):207-17. Epub 2018/01/10. doi: 10.1038/s41593-017-0053-5. PubMed PMID: 29311744; PMCID: PMC5897053.
15. Schaffer AE, **Breuss MW**, Caglayan AO, Al-Sanaa N, Al-Abdulwahed HY, Kaymakcalan H, Yilmaz C, Zaki MS, Rosti RO, Copeland B, Baek ST, Musaev D, Scott EC, Ben-Omran T, Kariminejad A, Kayserili H, Mojahedi F, Kara M, Cai N, Silhavy JL, Elsharif S, Fenercioglu E, Barshop BA, Kara B, Wang R, Stanley V, James KN, Nachnani R, Kalur A, Megahed H, Incecik F, Danda S, Alanay Y, Fageih E, Melikishvili G, Mansour L, Miller I, Sukhudyann B, Chelly J, Dobyns WB, Bilguvar K, Jamra RA, Gunel M, Gleeson JG. Biallelic loss of human CTNNA2, encoding alphaN-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. *Nat Genet.* 2018;50(8):1093-101. Epub 2018/07/18. doi: 10.1038/s41588-018-0166-0. PubMed PMID: 30013181; PMCID: PMC6072555.
 16. Tripathy R, Leca I, van Dijk T, Weiss J, van Bon BW, Sergaki MC, Gstrein T, **Breuss M**, Tian G, Bahi-Buisson N, Paciorkowski AR, Pagnamenta AT, Wenninger-Weinzierl A, Martinez-Reza MF, Landler L, Lise S, Taylor JC, Terrone G, Vitiello G, Del Giudice E, Brunetti-Pierri N, D'Amico A, Raymond A, Voisin N, Bernstein JA, Farrelly E, Kini U, Leonard TA, Valence S, Burglen L, Armstrong L, Hiatt SM, Cooper GM, Aldinger KA, Dobyns WB, Mirzaa G, Pierson TM, Baas F, Chelly J, Cowan NJ, Keays DA. Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. *Neuron.* 2018;100(6):1354-68 e5. Epub 2018/11/20. doi: 10.1016/j.neuron.2018.10.044. PubMed PMID: 30449657; PMCID: PMC6436622.
 17. **Breuss MW**, Antaki D, George RD, Kleiber M, James KN, Ball LL, Hong O, Mitra I, Yang X, Wirth SA, Gu J, Garcia CAB, Gujral M, Brandler WM, Musaev D, Nguyen A, McEvoy-Venneri J, Knox R, Sticca E, Botello MCC, Uribe Fenner J, Perez MC, Arranz M, Moffitt AB, Wang Z, Hervas A, Devinsky O, Gymrek M, Sebat J, Gleeson JG. Autism risk in offspring can be assessed through quantification of male sperm mosaicism. *Nat Med.* 2020;26(1):143-50. Epub 2019/12/25. doi: 10.1038/s41591-019-0711-0. PubMed PMID: 31873310; PMCID: PMC7032648.
 18. **Breuss MW***, Mamerto A*, Renner T*, Waters ER. The Evolution of the Mammalian ABCA6-like Genes: Analysis of Phylogenetic, Expression, and Population Genetic Data Reveals Complex Evolutionary Histories. *Genome Biol Evol.* 2020;12(11):2093-106. Epub 2020/09/03. doi: 10.1093/gbe/evaa179. PubMed PMID: 32877505; PMCID: PMC7674697.
 19. Garcia CAB, Carvalho SCS, Yang X, Ball LL, George RD, James KN, Stanley V, **Breuss MW**, Thome U, Santos MV, Saggiaro FP, Neder Serafini L, Silva WA, Jr., Gleeson JG, Machado HR. mTOR pathway somatic variants and the molecular pathogenesis of hemimegalencephaly. *Epilepsia Open.* 2020;5(1):97-106. Epub 2020/03/07. doi: 10.1002/epi4.12377. PubMed PMID: 32140648; PMCID: PMC7049797.
 20. Ghosh SG, Wang L, **Breuss MW**, Green JD, Stanley V, Yang X, Ross D, Traynor BJ, Alhashem AM, Azam M, Selim L, Bastaki L, Elbastawisy HI, Temtamy S, Zaki M, Gleeson JG. Recurrent homozygous damaging mutation in TMX2, encoding a protein disulfide isomerase, in four families with microlissencephaly. *J Med Genet.* 2020;57(4):274-82. Epub 2019/10/07. doi: 10.1136/jmedgenet-2019-106409. PubMed PMID: 31586943; PMCID: PMC7405652.
 21. Chai G, Webb A, Li C, Antaki D, Lee S, **Breuss MW**, Lang N, Stanley V, Anzenberg P, Yang X, Marshall T, Gaffney P, Wierenga KJ, Chung BH, Tsang MH, Pais LS, Lovgren AK, VanNoy GE, Rehm HL, Mirzaa G, Leon E, Diaz J, Neumann A, Kalverda AP, Manfield IW, Parry DA, Logan CV, Johnson CA, Bonthron DT, Valleley EMA, Issa MY, Abdel-Ghafar SF, Abdel-Hamid MS, Jennings P, Zaki MS, Sheridan E, Gleeson JG. Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly.

- Neuron. 2021;109(2):241-56 e9. Epub 2020/11/22. doi: 10.1016/j.neuron.2020.10.035. PubMed PMID: 33220177; PMCID: PMC8800389.
22. Ghosh SG*, **Breuss MW***#, Schlachetzki Z, Chai G, Ross D, Stanley V, Sonmez FM, Topaloglu H, Zaki MS, Hosny H, Gad S, Gleeson JG#. Biallelic hypomorphic mutations in HEATR5B, encoding HEAT repeat-containing protein 5B, in a neurological syndrome with pontocerebellar hypoplasia. *Eur J Hum Genet.* 2021;29(6):957-64. Epub 2021/04/08. doi: 10.1038/s41431-021-00832-x. PubMed PMID: 33824466; PMCID: PMC8187379.
23. Ghosh SG, Scala M, Beetz C, Helman G, Stanley V, Yang X, **Breuss MW**, Mazaheri N, Selim L, Hadipour F, Pais L, Stutterd CA, Karageorgou V, Begtrup A, Crunk A, Juusola J, Willaert R, Flore LA, Kennelly K, Spencer C, Brown M, Trapane P, Hurst ACE, Lane Rutledge S, Goodloe DH, McDonald MT, Shashi V, Schoch K, Undiagnosed Diseases N, Tomoum H, Zaitoun R, Hadipour Z, Galehdari H, Pagnamenta AT, Mojarrad M, Sedaghat A, Dias P, Quintas S, Eslahi A, Shariati G, Bauer P, Simons C, Houlden H, Issa MY, Zaki MS, Maroofian R, Gleeson JG. A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. *Eur J Hum Genet.* 2021;29(2):271-9. Epub 2020/09/10. doi: 10.1038/s41431-020-00717-5. PubMed PMID: 32901138; PMCID: PMC7868361 declare no conflict of interest.
24. Yang X*, **Breuss MW***, Xu X, Antaki D, James KN, Stanley V, Ball LL, George RD, Wirth SA, Cao B, Nguyen A, McEvoy-Venneri J, Chai G, Nahas S, Van Der Kraan L, Ding Y, Sebat J, Gleeson JG. Developmental and temporal characteristics of clonal sperm mosaicism. *Cell.* 2021;184(18):4772-83 e15. Epub 2021/08/14. doi: 10.1016/j.cell.2021.07.024. PubMed PMID: 34388390; PMCID: PMC8496133.
25. **Breuss MW***, Yang X*, Schlachetzki JCM*, Antaki D*, Lana AJ, Xu X, Chung C, Chai G, Stanley V, Song Q, Newmeyer TF, Nguyen A, O'Brien S, Hoeksema MA, Cao B, Nott A, McEvoy-Venneri J, Pasillas MP, Barton ST, Copeland BR, Nahas S, Van Der Kraan L, Ding Y, Network NBSM, Glass CK, Gleeson JG. Somatic mosaicism reveals clonal distributions of neocortical development. *Nature.* 2022;604(7907):689-96. Epub 2022/04/22. doi: 10.1038/s41586-022-04602-7. PubMed PMID: 35444276; PMCID: PMC9436791.
26. **Breuss MW***#, Yang X*, Stanley V, McEvoy-Venneri J, Xu X, Morales AJ, Gleeson JG#. Unbiased mosaic variant assessment in sperm: a cohort study to test predictability of transmission. *Elife.* 2022;11. Epub 2022/07/06. doi: 10.7554/eLife.78459. PubMed PMID: 35787314; PMCID: PMC9255958.
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1. **Breuss MW***, Yang X*, Gleeson JG. Developmental and temporal characteristics of clonal sperm mosaicism. American Society of Human Genetics (ASHG) Meeting 2021. Poster presentation.
2. **Breuss MW***, Yang X*, Stanley V, McEvoy-Venneri J, Xu X, Morales AJ, Gleeson JG. Sperm mosaicism predicts transmission of de novo mutations to human blastocysts. American Society of Human Genetics (ASHG) Meeting 2022. Poster presentation (selected as Reviewer's Choice, denoting the top 10% of scored abstracts).

3. Graham JH, Pitsch JW, **Breuss MW**. Development of a region specific enrichment and long read sequencing strategy to phase de novo mutations in human genetic disease. American Society of Human Genetics (ASHG) Meeting 2023. Poster presentation (selected as Reviewer's Choice, denoting the top 10% of scored abstracts).
4. Doss RM, Wirth SA, Pitsch JW, Gropman AL, **Breuss MW**. Identification of novel pathogenic variants in the neurodevelopmental gene *LNPK*. American Society of Human Genetics (ASHG) Meeting 2024. Poster presentation.
5. Pitsch JW, Wirth SA, Xiaoxu Yang, **Breuss MW**. A novel bioinformatic tool for large-scale primer design and specificity analysis. American Society of Human Genetics (ASHG) Meeting 2024. Poster presentation.
6. **Breuss MW**[#], Wirth SA, Pitsch JW, Doss RM, Graham J, Yang X, Gleeson JG. The origin, impact, and utility of mosaicism analysis in sperm. Keystone Symposium: Somatic Mosaicism in Human Development, Aging, and Diseases 2025. Poster presentation and selected talk (see also *Invited and Selected Talks*).