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CARVER COLLEGE OF MEDICINE CURRICULUM VITAE

Alexander G Bassuk

(*Updated 2/26/2024*)

I. EDUCATIONAL AND PROFESSIONAL HISTORY

A. Education, Certification and Licensure

A. Higher Education

June, 1991	AB with Highest Honors (Fundamentals: Issues and Texts) - University of Chicago, Chicago, Illinois
November,1996	PhD (Biological Sciences Pathology) - University of Chicago, Chicago, Illinois Supervisor: Jeffrey Leiden, MD, PhD
June, 1999	MD - University of Chicago Pritzker School of Medicine, Chicago, Illinois

Post Graduate

1999 - 2001	Resident (Pediatrics) - Children's Memorial Hospital, Northwestern University Mentor: Sharon Unti, MD
2001 - 2004	Resident (Pediatric Neurology) - Children's Memorial Hospital, Northwestern University Mentor: Leon Epstein, MD and John Kessler, MD

Licensures

2009-2014	Illinois Medical License, D36-106050
2007 -	Iowa Permanent Medical License, 37386, issued 7/23/2007, renewed 11/09/2016, renewal 1/1/2019

Certifications

2005 -	Board Certified, Neurology with Special Qualification in Child Neurology, Certificate Number: 11467, certified 6/10/2005, recertified 2/9/2015, valid through 2025
2006	Board Certified, General Pediatrics, ABP ID#: 657180, Certificate Number: 85592, valid through 2026

B. Professional and Academic Positions

2004 - 2005	Instructor, Pediatrics and Neurology Northwestern University Supervisor- Thomas Green, MD

2005 - 2007	Assistant Professor, Neurology and Pediatrics Center for Genetic Medicine, Northwestern University Supervisor- Thomas Green, MD
2007 - 2011	Assistant Professor of Pediatrics, Division of Neurology, Graduate Program in Genetics, Graduate Program in Neuroscience, Graduate Program in Molecular and Cellular Biology, and the Medical Scientist Training Program University of Iowa, Iowa City, Iowa
2011 - 2016	Associate Professor of Pediatrics, Division of Neurology, Graduate Program in Genetics, Graduate Program in Neuroscience, Graduate Program in Molecular and Cellular Biology, and the Medical Scientist Training Program, Secondary appointment in Neurology University of Iowa, Iowa City, Iowa
2016 - Present	Stead Family Chair, Professor of Pediatrics, Division of Neurology, Graduate Program in Genetics, Graduate Program in Neuroscience, Graduate Program in Molecular and Cellular Biology, and the Medical Scientist Training Program, Secondary appointment in Neurology University of Iowa, Iowa City, Iowa
2018 - 2021	Director, Division of Pediatric Neurology (May 1, 2018) University of Iowa Hospitals & Clinics, Iowa City, Iowa
2021-present	Chair and DEO, Stead Family Department of Pediatrics Joel and Jay Stead Chair for Leadership in Children's Medicine Physician-in-Chief, University of Iowa Stead Family Children's Hospital

C. Honors, Awards, Recognitions, Outstanding Achievements

1987 - 1991	Dean's List - University of Chicago
1988	Early Acceptance Program - University of Chicago Pritzker School of Medicine
1988 - 1991	Richter Award for Undergraduate Research - University of Chicago
1990	Student Marshall - University of Chicago
1991	Phi Beta Kappa
1991 - 1999	NIH-MSTP Scholarship
1994	National Defense Science and Engineering Graduate Fellowship
1999	Leon E Ledbetter Prize for Most Outstanding Research Contribution by a Senior Medical Student
2012	Member of University of Chicago team for Boston Children's "CLARITY CHALLENGE" (team was second place finisher) - Annual Meeting of the American Society of Human Genetics (11/7/2012)

II. TEACHING

A. Teaching assignments

Classroom, Seminar, Teaching Laboratory

2003 - 2006	"Human Congenital CNS Malformations Board Review" Northwestern University Medical School, Yearly lecture Pathophysiology Students – 2nd year medical student
2003 - 2006	"Human Congenital CNS Malformations" Northwestern University Medical School Students – Neurology residents
Fall 2007	"New Investigator Panel", Fellow Core Curriculum Seminar (8/1/2007)
Fall 2007	"The Genetics of Human Neural Tube Defects" - Neurobiology of Disease lecture series University of Iowa (11/27/2007)
2007 - 2013	"Laboratory and Research Transition from Fellowship to Faculty" Fellows Core Curriculum Seminar
2007 – Present	Regular Teaching to University of Iowa rotating graduate and medical students
Spring 2008	"The Genetics of Human Neural Tube Defects", Genetics & Environment Seminar Series University of Iowa Interdisciplinary Graduate Program in Genetics (2/25/2008)
Summer 2008	"Recessive Epilepsies", SUMR MSTP program (7/18/2008)
Fall 2008	"New Epilepsy Genes", Molecular and Cellular Biology lecture series (8/28/2008)
Fall 2008	"Recessive Epilepsy Genes", Neuroscience lecture series (10/7/2008)
Fall 2008	"New Epilepsy Genes", Genetics Program lecture series (11/11/2008)
2008 - 2014	"Fellowship-Attending Transition", Pediatric Fellows Core Curriculum
Fall 2009	"Neural Tube Defects", Neurobiology of Disease lecture series, University of Iowa (9/17/2009)
Fall 2009	"Human Genetics-simple genetic traits", Genetic Analysis of Biological Systems course University of Iowa (11/12/2009)
2009 - Present	Neurobiology of Disease course
Fall 2010	"Epilepsy: Neurobiology", Neurobiology of Disease lecture series University of Iowa (11/19/2010)
Spring 2014	“Exome Sequencing in Neurology”, Developmental Neurobiology BIOL:4753; NCSI: 6184; MPB:5184: old number 2:184 (5/7/2014)
2016-2017	Responsible Conduct of Research Series Topic #7: Collaborative Research Including Collaborations, Pediatric Fellows Core Curriculum

2007-present	Continuing training of rotating graduate students, medical students, and residents
2015-present	Child Neurology attending M3 weekly Thursday presentation sessions, (rotating faculty)
2019	Responsible Conduct of Research Series Topic #7: Collaborative Research Including Collaborations with industry, Pediatric Fellows Core Curriculum, University of Iowa (2/13/19)
2019	Student Interest Group in Neurology (SIGN). Neurology Specialty Panel, University of Iowa (4/2/2019)
2020	University of Iowa Summer Health Professions Education Program at the University of Iowa Medicine Panel. (7/7/2020)
2020	MSTP Analyzing and Presenting Med Lit: Forest vs Trees: Asking good questions (7/20/20)
2021	Responsible Conduct of Research Series: Collaborative Research Including Collaborations with Industry, Fellows Core Curriculum 2/24/21

B. Student Supervision (* indicates chair of the committee)

Clinical Fellows

2009 - 2021	Dina Al-Zubeidi - Pediatric GI Fellow, Rohini Sing- Pediatric Heme-Once Fellow, Yezan Abderrhman-Pediatric PICU Fellow, member research oversight committee

Graduate Students

2009 - 2012	Levi Sowers - Molecular and Cellular Biology *(Graduated with PhD, currently Junior Investigator Award, University of Iowa VA)*
2009 – 2014	Lily Paekma - Genetics *(Graduated with PhD, currently Assistant Professor, University of Ghana)*
2012 – 2016	Allison Cox - Genetics *Awarded 2013 Genetics Training Program Grant, 2014 Bioinformatics Training Grant (Graduated with PhD, awarded* Ballard Seashore Dissertation Fellowship *currently Post-Doc, Yale)*
2017 – 2020	Lucy Evans - MSTP, Molecular Medicine *Awarded University of Iowa Graduate College Post-Comprehensive Research Fellowship and NIH F31 Award*
2018 - 2020	Lindsay Agostinelli – MSTP, Neuroscience *Awarded NIH Diversity Supplement*
2020 -present	Saul Rodriguez – MSTP, Neuroscience

Medical Students

2010	Amy Keller - Medical Student Research fellow awarded (*Summer)*
2010	Mark Fox – Mentor, Medical Student Research Fellow *Mark was awarded the Outstanding Presentation in the Discipline of Translational Neuroscience award for his research poster "Increased Hippocampal Neurogenesis May Contribute to Epilepsy" for the Medical Student Research Day Awards September 10, 2010*
2012	Emilie Dore – Mentor, Medical Student Research Fellow (*Summer), awarded 2012 AOA Student Research Fellowship*
2012	Tyler Mouw – Mentor, Medical Student Research Fellow (Summer)
2015	Madeline Cross (M3), Research Distinction track
2018	Fili Bogdamic – *Awarded Iowa Medical Student Research Program Fellowship*
2019	Saul Rodriguez – *Awarded Iowa Medical Student Research Program Fellowship, accepted to MSTP 2020*
2019	Saul Rodriguez – *Awarded Iowa Medical Student Research Program Fellowship, accepted to MSTP 2020*
2020	Ariel Roghair – *Awarded Iowa Medical Student Research Program Fellowship*
2020	Joseph Haight – *Awarded Iowa Medical Student Research Program Fellowship*
2021	Kyle Jackson – *Awarded Iowa Medical Student Research Program Fellowship; and awarded Excellence in Pediatric Basic Science Research*. “Evaluating the Disease-Modifying Effects of Naltrexone on the Development of Post-Traumatic Epilepsy using a Novel, Automated EEG Analysis Algorithm” Mentor: Alexander Bassuk, Pediatrics

Pediatric Neurology Trainees

2004 - 2005	Jason Umfleet - Trainee - Pediatric Neurology *Current Position: Private Practice, Grand Rapids, MI*
2004 - 2005	Jeremy Freeman - Trainee - Pediatric Neurology *Current Position: Consultant for the Monash Medical Center, Melbourne, Australia*
2004 - 2006	Robert Little - Trainee - Pediatric Neurology *Current Position: Private Practice, Phoenix Children's Hospital, Phoenix, AZ*
2004 - 2007	Charulata Venkatesan - Trainee - Pediatric Neurology *Current Position: Assistant Professor, Northwestern University Children's Memorial Hospital, Chicago, IL*

2004 - 2007	Jennifer Reuben - Trainee - Pediatric Neurology *Current Position: Assistant Professor, Northwestern University Children's Memorial Hospital, Chicago, IL*
2010 – 2013	Satsuki Matsumoto – Trainee – Pediatric Neurology *Current Position: Clinical Assistant Professor, University of Iowa, Iowa City, IA*
2011 – 2014	Himali Jayakody – Trainee – Pediatric Neurology *Current Position: Attending Child Neurologist, Tampa General Hospital and St Joseph's Hospital, Tampa, FL* *Adjunct Assistant Professor of Neurology, University of South Florida, Department of Neurology, Tampa, FL*
2014 – 2017	Dimah Saade – Trainee – Pediatric Neurology *Current Position: Clinical Fellow at the National Institute of Neurological Disorders and Stroke (NINDS), Bethesda, MD*
2014 – 2017	Megan Rohlf – Trainee – Pediatric Neurology *Current Position: Clinical Assistant Professor, University of Iowa, Iowa City, IA*
2015 – 2018	Ioanna Kouri – Trainee – Pediatric Neurology *Current Position: Pediatrician, Pediatric Neurologist, Sleep Medicine Specialist in private practice, Greece*
2017 – 2018	Hannah Klein – Trainee – Pediatric Neurology *Current Position: Pediatric Neurologist and Epileptologist, Boys Town Research Hospital, Omaha, NE*
2016 – 2019	Katie Lutz – Trainee – Pediatric Neurology *Current Position: Clinical Assistant Professor, University of Iowa, Iowa City, IA*

Post-doctoral Trainees

2014 – Present	Elizabeth Newell, MD. PICU Attending. CHRCA (K12) scholar (since 2015), K08 award (Bassuk, K08 mentor) in lab studying traumatic brain injury and inflammation
2014 - Present	Rohini Singh, MD. Hematology Oncology Fellow studying autism-oncology genetic and epidemiological overlap
2017 – Present	Marco Hefti, MD, CTSA KL2 award (since 2018), NIH K23 award, 2019 (Bassuk, Abel mentors), studying the role to Tau in neurodevelopment
2020 – Present	Shaunik Sharma, PhD, studying novel epilepsy treatments

Thesis Committee

Terry Yin - MCB, Welsh Lab, Mark Schulz - MCB, Davidson Lab, Farah Alul - Genetics, Murray Lab, Elizabeth Leslie - Genetics, Murray Lab, Xue Mei - Biology, Slusarksi Lab, Tian Yang - Biology, Fritzch Lab, Juan Santana - Biology, Manak Lab, Megan Kaiser - Neuroscience, Davidson Lab), David Ho (Genetics, Potash lab), Juan Santana (Biology, Manak lab), Allison Cox (Genetics, Bassuk lab), Sophie Gaynor (Genetics, Willour lab), Hannah Seberg (Genetics, Cornell lab), Amanda Benavides (Neuroscience, Nopoulos lab), Tricia Braun (Genetics, Potash lab), Hung-Lin Chen (Genetics, Kitamoto lab), Morgan Sturgeon (MCB, Cornell lab), Kelli Shaefer (Genetics, Mahajan lab), Sean Tompkins (Biochemistry, Taylor lab), Dylan Todd (Neuroscience, Bonthius lab), Leo Brueggeman (Genetics, Michaelson lab)

Undergraduate Students

2007-2008	Andrew Butler, former undergraduate student at University of Iowa *Now an Assistant Professor at The University of Wisconsin, Madison*
2013 - Present	Nicole Felderman - Freshman - research mentorship University of Iowa
2014 – Present	Brittany Todd, University of Iowa Undergraduate, awarded ICRU with Beth Newell, accepted to the University of Iowa MSTP Program (Fall 2019)
2017 – 2018	Alexis Finer, University of Iowa Undergraduate
2018 – 2021	Dustin Fykstra, University of Iowa Undergraduate (Received INI fellowship Summer 2020)
2018 – 2021	Prateek Raikwar, University of Iowa Undergraduate (Received ICRU Fellowship Summer 2020)
2020-Present	Noah Gilkes, University of Iowa Undergraduate (Received ICRU Fellowship Summer 2020)
2021-Present	Angela Wong, University of Iowa Undergraduate
2021-Present	David Keffala-Gerhard, University of Iowa Undergraduate (Received ICRU Fellowship Summer 2022)

C. Other Contributions to Institutional Programs

2009	Adeshwari Ramesh - Secondary Training Program (SSTP) (Summer)
2009 - Present	Oral Examination Panel *Neuroscience Graduate Program*
2011 - Present	Recruiter - University of Iowa Children's Hospital Research Center, Co-wrote grant renewal for 2012-2017 cycle
2012	Mentor - University of Iowa, Carver College of Medicine, Office of Cultural Affairs and Diversity Initiatives SNMA & LMSA (11/12/2012)
2018 – Present	Genetics Program, Faculty selection committee

2018 – 2020 Director – University of Iowa MSTP Admissions Committee

Institutional Conferences, Grand Rounds, Journal Clubs, Etc.

2007	"The Genetics of Human Neural Tube Defects" - Department of Pediatrics, University of Iowa Children's Hospital *Grand Rounds presentation* (1/26/2007)
2009	"Prickle Mutations in Epilepsy" - University of Iowa *Pediatric Research Day* (3/27/2009)
2010	"Flies, and zebrafish, and mice, and epileptics, oh my" - University of Iowa *Department of Pharmacology Faculty Seminar Series* (3/25/2010)
2010	"Flies, and zebrafish, and mice, and epileptics, oh my" - University of Iowa *Pediatric Grand Rounds* (4/2/2010)
2011	"Modeling epilepsy in Drosophila" - University of Iowa *Neurology Grand Rounds* (1/10/2011)
2013	"Exome Sequencing for Pediatricians" *Pediatric Grand Rounds* (11/8/2013)
2014	"Exome Sequencing in Pediatrics" - Next-Generation Sequencing Interest Group

III. SCHOLARSHIP/PROFESSIONAL PRODUCTIVITY

A. Publications or creative works (earliest to most recent)

Peer-reviewed papers

1. Wang CY, **Bassuk AG**, Boise LH, Thompson CB, Bravo R, Leiden JM. Activation of the granulocyte-macrophage colony-stimulating factor promoter in T cells requires cooperative binding of Elf-1 and AP-1 transcription factors., Mol. Cell Biol. 1994 Feb;14(2):1153-9. PMID: 8289796. PMCID: PMC358471.
   Role: Designed and performed or assisted in all experiments, co-wrote paper.
2. **Bassuk AG**, Leiden JM. A direct physical association between ETS and AP-1 transcription factors in normal human T cells., Immunity. 1995 Aug;3(2):223-37. PMID: 7648395.
   Role: Designed and performed all experiments, co-wrote paper.
3. **Bassuk AG**, Anandappa RT, Leiden JM. Physical interactions between Ets and NF-kappaB/NFAT proteins play an important role in their cooperative activation of the human immunodeficiency virus enhancer in T cells., J Virol. 1997 May;71(5):3563-73. PMID: 9094628, PMC191503.
   Role: Designed and performed all experiments, co-wrote paper.
4. **Bassuk AG**, Barton KP, Anandappa RT, Lu MM, Leiden JM. Expression pattern of the Ets-related transcription factor Elf-1., Mol. Med. 1998 June;4(6):392-401. PMID: 10780882. PMCID: PMC2230273.
   Role: Designed imaging analysis, wrote paper.
5. **Bassuk AG**, Burrowes DM, McRae W. Acute necrotizing encephalopathy of childhood with radiographic progression over 10 hours., Neurology. 2003 May

13;60(9):1552-3. PMID: 12743257.
Role: Designed imaging analysis, wrote paper.

6. **Bassuk AG**, Burrowes DM, Velimirovic B, Grant J, Keating GF. A child with spinal cord AVM presenting with raised intracranial pressure., Neurology. 2003 May 27;60(10):1724-5. PMID: 12771285.
Role: Designed sequencing strategy, analyzed genetic data, designed imaging analysis, wrote paper.

7. **Bassuk AG**, Joshi A, Burton BK, Larsen MB, Burrowes DM, Stack C. Alexander disease with serial MRS and a new mutation in the glial fibrillary acidic protein gene., Neurology. 2003 Oct 14;61(7):1014-5. PMID: 14557587.
Role: Designed sequencing strategy, analyzed genetic data, designed imaging analysis, wrote paper.

8. **Bassuk AG**, Keating GF, Stumpf DA, Burrowes DM, Stack C. Systemic lymphoma mimicking acute disseminated encephalomyelitis., Pediatr Neurol. 2004 Feb;30(2):129-31. PMID: 14984907. doi: 10.1016/S0887-8994(03)00414-4.
Role: Designed imaging analysis, wrote paper.

9. **Bassuk AG**, McLone D, Bowman R, Kessler JA. Autosomal dominant occipital cephalocele., Neurology. 2004 May 25;62(10):1888-90. PMID:1 5159504.
Role: Identified syndrome, designed genetic analysis, wrote paper.

10. Benz LP, Swift FE, Graham FL, Enterline DS, Melvin EC, Hammock P, Gilbert JR, Speer MC, **Bassuk AG**, Kessler JA, George TM. TERC is not a major gene in human neural tube defects., Birth Defects Res A Clin Mol Teratol. 2004 August;70(8):531-3. PMID: 15329831. doi: 10.1002/bdra.20057.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

11. **Bassuk AG**, Craig D, Jalali A, Mukhopadhyay A, Kim F, Charrow J, Gulbu U, Epstein LG, Bowman R, McLone D, Yagi H, Matsuoka R, Stephan DA, Kessler JA. The genetics of tethered cord syndrome., Am J Med Genet. A. 2005 Feb 1;132A(4):450-3. PMID: 15558749. doi: 10.1002/ajmg.a.30439.
Role: Designed phenotype-genotype computer matrix, wrote paper.

12. Deak KL, Boyles AL, Etchevers HC, Melvin EC, Siegel DG, Graham FL, Slifer SH, Enterline DS, George TM, Vekemans M, McClay D, **Bassuk AG**, Kessler JA, Linney E, Gilbert JR, Speer MC. SNPs in the neural cell adhesion molecule 1 gene (NCAM1) may be associated with human neural tube defects., Hum Genet. 2005 July;117(2-3):133-42. Epub 2005 May 10. PMID: 15883837. PMCID: PMC3130962. doi: 10.1007/s00439-005-1299-7.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

13. Deak KL, Dickerson ME, Linney E, Enterline DS, George TM, Melvin EC, Graham FL, Siegel DG, Hammock P, Mehltretter L, **Bassuk AG**, Kessler JA, Gilbert JR, Speer MC. Analysis of ALDH1A2, CYP26A1, CYP26B1, CRABP1, and CRABP2 in human neural tube defects suggests a possible association with alleles in ALDH1A2., Birth Defects Res A Clin Mol Teratol. 2005 November;73(11):868-75. PMID: 16237707. doi: 10.1002/bdra.20183.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

14. Rampersaud E, **Bassuk AG**, Enterline DS, George TM, Siegel DG, Melvin EC, Aben J, Allen J, Aylsworth A, Brei T, Bodurtha J, Buran C, Floyd LE, Hammock P, Iskandar B, Ito J, Kessler JA, Lasarsky N, Mack P, Mackey J, McLone D,

Meeropol E, Mehltretter L, Mitchell LE, Oakes WJ, Nye JS, Powell C, Sawin K, Stevenson R, Walker M, West SG, Worley G, Gilbert JR, Speer MC. Whole genomewide linkage screen for neural tube defects reveals regions of interest on chromosomes 7 and 10., J Med Genet. 2005 Dec;42(12):940-6. Epub 2005 Apr 14. PMID: 15831595. PMCID: PMC1735960. doi: 10.1136/jmg.2005.031658.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

15. Sebold CD, Melvin EC, Siegel D, Mehltretter L, Enterline DS, Nye JS, Kessler J, **Bassuk A**, Speer MC, George TM. Recurrence risks for neural tube defects in siblings of patients with lipomyelomeningocele, Genet Med. 2005;7(1):64-7. PMID: 15654231. doi: 10.109701.gim.0000151158.09278.2b.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

16. Boyles AL, Billups AV, Deak KL, Siegel DG, Mehltretter L, Slifer SH, **Bassuk AG**, Kessler JA, Reed MC, Nijhout HF, George TM, Enterline DS, Gilbert JR, Speer MC. Neural tube defects and folate pathway genes: family-based association tests of gene-gene and gene-environment interactions., Environ Health Perspect. 2006 Oct;114(10):1547-52. PMID: 17035141. PMCID: PMC1626421.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

17. **Bassuk AG**, Mohile NA, Stack C. T-cell lymphoma presenting with neurologic features in immunocompetent children., Pediatr Neurol. 2006 Nov;35(5):314-7. PMID: 17074600. doi: 10.1016/j.pediatrneurol.2006.05.005.
Role: Organized and oversaw study, wrote paper.

18. **Bassuk AG**, Chen YZ, Batish SD, Nagan N, Opal P, Chance PF, Bennett CL. In cis autosomal dominant mutation of Senataxin associated with tremor/ataxia syndrome., Neurogenetics. 2007 Jan;8(1):45-9. Epub 2006 Nov 10. PMID: 17096168. doi: 10.1007/s10048-006-0067-8.
Role: Identified syndrome variant, designed genotyping strategy, co-wrote paper.

19. Lu W, Quintero-Rivera F, Fan Y, Alkuraya FS, Donovan DJ, Xi Q, Turbe-Doan A, Li QG, Campbell CG, Shanske AL, Sherr EH, Ahmad A, Peters R, Rilliet B, Parvex P, **Bassuk AG**, Harris DJ, Ferguson H, Kelly C, Walsh CA, Gronostajski RM, Devriendt K, Higgins A, Ligon AH, Quade BJ, Morton CC, Gusella JF, Maas RL. NFIA haploinsufficiency is associated with a CNS malformation syndrome and urinary tract defects., PLoS Genet. 2007 May 25;3(5):e80. PMID: 17530927. PMCID: PMC1877820. doi: 10.1371/journal.pgen.0030080.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

20. Donsante A, Tang J, Godwin SC, Holmes CS, Goldstein DS, **Bassuk A**, Kaler SG. Differences in ATP7A gene expression underlie intrafamilial variability in Menkes disease/occipital horn syndrome, J Med Genet. 2007 Aug;44(8):492-7. Epub 2007 May 11. PMID: 17496194. PMCID: PMC2597922. doi: 10.1136/jmg.2007.050013.
Role: Identified syndrome variant, designed genotyping strategy, co-wrote paper

21. Kan L, Jalali A, Zhao LR, Zhou X, McGuire T, Kazanis I, Episkopou V, **Bassuk AG**, Kessler JA. Dual function of Sox1 in telencephalic progenitor cells., Dev Biol. 2007 Oct;310(1):85-98. Epub 2007 Jul 27. PMID: 17719572. PMCID: PMC3437622. doi: 10.1016/j.ydbio.2007.07.026.
Role: Designed novel neurosphere assay variation, co-wrote paper

22. Epstein LG, Jalali A, Chary AN, Khan S, Ross J, Coppinger J, Carlson K, Charrow J, Burton B, Zimmerman D, Curran J, Kim F, Nguyen P, Burrowes D,

Angle B, Stack C, Shaffer L, Kessler JA, **Bassuk AG**. Neuroimaging findings in children with rare or novel de novo chromosomal anomalies., Birth Defects Res A Clin Mol Teratol. 2008 Apr;82(4):200-10. PMID: 18302267. doi: 10.1002/bdra.20443.
Role: Designed all analyses, oversaw all aspects of the project, wrote paper.

23. Jalali A, Aldinger KA, Chary A, McLone DG, Bowman RM, Le LC, Jardine P, Newbury-Ecob R, Mallick A, Jafari N, Russell EJ, Curran J, Nguyen P, Ouahchi K, Lee C, Dobyns WB, Millen KJ, Pina-Neto JM, Kessler JA, **Bassuk AG**. Linkage to chromosome 2q36.1 in autosomal dominant Dandy-Walker malformation with occipital cephalocele and evidence for genetic heterogeneity., Hum Genet. 2008 Apr;123(3):237-45. Epub 2008 Jan 19. PMID: 18204864. PMCID: PMC2822644. doi: 10.1007/s00439-008-0467-y.
Role: Designed all experiments, oversaw all aspects of the project, wrote paper.

24. **Bassuk AG**, Wallace RH, Buhr A, Buller AR, Afawi Z, Shimojo M, Miyata S, Chen S, Gonzalez-Alegre P, Griesbach HL, Wu S, Nashelsky M, Vladar EK, Antic D, Ferguson PJ, Cirak S, Voit T, Scott MP, Axelrod JD, Gurnett C, Daoud AS, Kivity S, Neufeld MY, Mazarib A, Straussberg R, Walid S, Korczyn AD, Slusarski DC, Berkovic SF, El-Shanti HI. A homozygous mutation in human PRICKLE1 causes an autosomal-recessive progressive myoclonus epilepsy-ataxia syndrome., Am J Hum Genet. 2008 Nov;83(5):572-81. Epub 2008 Oct 30. PMID: 18976727. PMCID: PMC2668041. doi: 10.1016/j.ajhg.2008.10.003.
Role: Designed all experiments, oversaw all aspects of the project, wrote paper.
Note: This paper was reviewed in two separate publications (A New, Progressive Myoclonic Epilepsy: Is It a Chronicle of the Noncanonical or a Failure to REST? Epilepsy Curr. 2009 May; 9(3): 82-84., and A prickly cause of progressive myoclonic epilepsy Clin Genet. 2009 Mar;75(3):225-6.)

25. Neilson DE, Adams MD, Orr CM, Schelling DK, Eiben RM, Kerr DS, Anderson J, **Bassuk AG**, Bye AM, Childs AM, Clarke A, Crow YJ, Di Rocco M, Dohna-Schwake C, Dueckers G, Fasano AE, Gika AD, Gionnis D, Gorman MP, Grattan-Smith PJ, Hackenberg A, Kuster A, Lentschig MG, Lopez-Laso E, Marco EJ, Mastroyianni S, Perrier J, Schmitt-Mechelke T, Servidei S, Skardoutsou A, Uldall P, van der Knaap MS, Goglin KC, Tefft DL, Aubin C, de Jager P, Hafler D, Warman ML. Infection-triggered familial or recurrent cases of acute necrotizing encephalopathy caused by mutations in a component of the nuclear pore, RANBP2., Am J Hum Genet. 2009 Jan;84(1):44-51. PMID: 19118815. PMCID: PMC2668029. doi: 10.1016/j.ajhg.2008.12.009.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

26. Kibar Z, Bosoi CM, Kooistra M, Salem S, Finnell RH, De Marco P, Merello E, **Bassuk AG**, Capra V, Gros P. Novel mutations in VANGL1 in neural tube defects., Hum Mutat. 2009 Jul;30(7):E706-15. PMID: 19319979. PMCID: PMC2885434. doi: 10.1002/humu.21026.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

27. **Bassuk AG**, Kibar Z. Genetic basis of neural tube defects., Semin Pediatr Neurol. 2009 Sep;16(3):101-10. PMID: 19778707. doi: 10.1016/j.spen.2009.06.001.

28. Aldinger KA, Lehmann OJ, Hudgins L, Chizhikov VV, **Bassuk AG**, Ades LC, Krantz ID, Dobyns WB, Millen KJ. FOXC1 is required for normal cerebellar development and is a major contributor to chromosome 6p25.3 Dandy-Walker

malformation., Nat Genet. 2009 Sep;41(9):1037-42. Epub 2009 Aug 9. PMID:19668217. PMCID: PMC2843139. doi: 10.1038/ng.422.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

29. Dibbens LM, Michelucci R, Gambardella A, Andermann F, Rubboli G, Bayly MA, Joensuu T, Vears DF, Franceschetti S, Canafoglia L, Wallace R, **Bassuk AG**, Power DA, Tassinari CA, Andermann E, Lehesjoki AE, Berkovic SF. SCARB2 mutations in progressive myoclonus epilepsy (PME) without renal failure., Ann Neurol. 2009 Oct;66(4):532-6. PMID: 19847901. doi: 10.1002/ana.21765.
Role: Contributed phenotype-genotype analysis, co-wrote paper

30. Mefford HC, Muhle H, Ostertag P, von Spiczak S, Buysse K, Baker C, Franke A, Malafosse A, Genton P, Thomas P, Gurnett CA, Schreiber S, **Bassuk AG**, Guipponi M, Stephani U, Helbig I, Eichler EE. Genome-wide copy number variation in epilepsy: novel susceptibility loci in idiopathic generalized and focal epilepsies., PLoS Genet. 2010 May 20;6(5):e1000962. PMID: 20502679. PMCID: PMC2873910. doi: 10.1371/journal.pgen.1000962.
Role: Contributed phenotype-genotype analysis, co-wrote paper.

31. Mahajan VB, Olney AH, Garrett P, Chary A, Dragan E, Lerner G, Murray J, **Bassuk AG**. Collagen XVIII mutation in Knobloch syndrome with acute lymphoblastic leukemia, Am J Med Genet A. 2010 Nov;152A(11):2875-9. PMID: 20799329. PMCID: PMC2965270.
Role: Designed all experiments, oversaw all aspects of the project, wrote paper.

32. Jalali A, **Bassuk AG**, Kan L, Israsena N, Mukhopadhyay A, McGuire T, Kessler JA. HeyL promotes neuronal differentiation of neural progenitor cells., J Neurosci Res. 2011 Mar;89(3):299-309. Epub 2011 Jan 5. PMID: 21259317. PMCID: PMC3079914. doi: 10.1002/jnr.22562.
Role: Co-first author, wrote paper.

33. Tao H, Manak JR, Sowers L, Mei X, Kiyonari H, Abe T, Dahdaleh NS, Yang T, Wu S, Chen S, Fox MH, Gurnett C, Montine T, Bird T, Shaffer LG, Rosenfeld JA, McConnell J, Madan-Khetarpal S, Berry-Kravis E, Griesbach H, Saneto RP, Scott MP, Antic D, Reed J, Boland R, Ehaideb SN, El-Shanti H, Mahajan VB, Ferguson PJ, Axelrod JD, Lehesjoki AE, Fritzsch B, Slusarski DC, Wemmie J, Ueno N, **Bassuk AG**. Mutations in prickle orthologs cause seizures in flies, mice, and humans, Am J Hum Genet. 2011 Feb 11;88(2):138-49. Epub 2011 Feb 3. PMID:21276947. PMCID: PMC3035715. doi: 10.1016/j.ajhg.2010.12.012.
Role: I am the senior and corresponding author. I designed all of the experiments and wrote the paper.

34. Kibar Z, Salem S, Bosoi CM, Pauwels E, De Marco P, Merello E, **Bassuk AG**, Capra V, Gros P. Contribution of VANGL2 mutations to isolated neural tube defects., Clin Genet. 2011 Jul;80(1):76-82. Epub 2010 Jul 22. PMID: 20738329. PMCID: PMC3000889. doi: 10.1111/j.1399-0004.2010.01515.x.
Role: Contributed phenotype-genotype analysis, genotyped controls.

35. Bosoi CM, Capra V, Allache R, Trinh VQ, De Marco P, Merello E, Drapeau P, **Bassuk AG**, Kibar Z. Identification and characterization of novel rare mutations in the planar cell polarity gene PRICKLE1 in human neural tube defects., Hum Mutat. 2011 Dec;32(12):1371-5. Epub 2011 Sep 23. PMID: 21901791. PMCID: PMC3217084. doi: 10.1002/humu.21589.
Role: Contributed phenotype-genotype analysis, genotyped controls.

36. Mahajan VB, Skeie JM, **Bassuk AG**, Fingert JH, Braun TA, Daggett HT, Folk JC, Sheffield VC, Stone EM. Calpain-5 mutations cause autoimmune uveitis, retinal neovascularization, and photoreceptor degeneration., PLoS Genet. 2012;8(10):e1003001. Epub 2012 Oct 4. PMID: 23055945. PMCID: PMC3464205. doi: 10.1371/journal.pgen.1003001.
Role: Performed exome sequencing. contributed phenotype-genotype analysis, genotyped controls.

37. Rowell HA, **Bassuk AG**, Mahajan VB. Monozygotic twins with CAPN5 autosomal dominant neovascular inflammatory vitreoretinopathy., Clin. Ophthalmol. 2012;6:2037-44. Epub 2012 Dec 6. PMID: 23271883. PMCID: PMC3526908. doi: 10.2147/OPTH.S40086.
Role: Contributed phenotype-genotype analysis, genotyped controls.

38. Tao H, Inoue K, Kiyonari H, **Bassuk AG**, Axelrod JD, Sasaki H, Aizawa S, Ueno N. Nuclear localization of Prickle2 is required to establish cell polarity during early mouse embryogenesis., Dev Biol. 2012 Apr 15;364(2):138-48. Epub 2012 Feb 4. PMID: 22333836. PMCID: PMC3299875. doi: 10.1016/j.ydbio.2012.01.025.
Role: Contributed phenotype-genotype analysis.

39. Safra N, **Bassuk AG**, Ferguson PJ, Aguilar M, Coulson RL, Thomas N, Hitchens PL, Dickinson PJ, Vernau KM, Wolf ZT, Bannasch DL. Genome-wide association mapping in dogs enables identification of the homeobox gene, NKX2-8, as a genetic component of neural tube defects in humans., PLoS Genet. 2013 Jan;9(7):e1003646. Epub 2013 Jul 18. PMID: 23874236. PMCID: PMC3715436. doi: 10.1371/journal.pgen.1003646.
Role: I designed all of the human genetics experiments and co-wrote the paper.

40. **Bassuk AG**, Muthuswamy LB, Boland R, Smith TL, Hulstrand AM, Northrup H, Hakeman M, Dierdorff JM, Yung CK, Long A, Brouillette RB, Au KS, Gurnett C, Houston DW, Cornell RA, Manak JR. Copy number variation analysis implicates the cell polarity gene glypican 5 as a human spina bifida candidate gene., Hum Mol Genet. 2013 Mar 15;22(6):1097-111. Epub 2012 Dec 7. PMID: 23223018. PMCID: PMC3578410. doi: 10.1093/hmg/dds515.
Role: I designed all the experiments and co-wrote the paper.

41. Mei X, Wu S, **Bassuk AG**, Slusarski DC. Mechanisms of prickle1a function in zebrafish epilepsy and retinal neurogenesis., Dis Model Mech. 2013 May;6(3):679-88. Epub 2013 Jan 11. PMID: 23324328. PMCID: PMC3634651. doi: 10.1242/dmm.010793.
Role: I designed all of the experiments and co-wrote the paper.

42. **Bassuk AG**, Geraghty E, Wu S, Mullen SA, Berkovic SF, Scheffer IE, Mefford HC. Deletions of 16p11.2 and 19p13.2 in a family with intellectual disability and generalized epilepsy., Am J Med Genet A. 2013 Jul;161A(7):1722-5. Epub 2013 May 17. PMID: 23686817. PMCID: PMC4169108. doi: 10.1002/ajmg.a.35946.
Role: I designed all of the experiments and co-wrote the paper.

43. Cherepanova NS, Leslie E, Ferguson PJ, Bamshad MJ, **Bassuk AG**. Presence of epilepsy-associated variants in large exome databases., J Neurogenet. 2013 Jun;27(1-2):1-4. Epub 2013 Mar 25. PMID: 23527921. PMCID: PMC3672316. doi: 10.3109/01677063.2013.772176.
Role: I designed all of the experiments and co-wrote the paper.

44. Robinson A, Partridge D, Malhas A, De Castro SC, Gustavsson P, Thompson DN, Vaux DJ, Copp AJ, Stanier P, **Bassuk AG**, Greene ND. Is LMNB1 a susceptibility gene for neural tube defects in humans?, Birth Defects Res A Clin Mol Teratol. 2013 Jun;97(6):398-402. Epub 2013 Jun 3. PMID: 23733478. PMCID: PMC3738925. doi: 10.1002/bdra.23141.
Role: I helped designed all of the experiments and co-wrote the paper.

45. Darbro BW, Mahajan VB, Gakhar L, Skeie JM, Campbell E, Wu S, Bing X, Millen KJ, Dobyns WB, Kessler JA, Jalali A, Cremer J, Segre A, Manak JR, Aldinger KA, Suzuki S, Natsume N, Ono M, Hai HD, le Viet T, Loddo S, Valente EM, Bernardini L, Ghonge N, Ferguson PJ, **Bassuk AG**. Mutations in extracellular matrix genes NID1 and LAMC1 cause autosomal dominant Dandy-Walker malformation and occipital cephaloceles., Hum Mutat. 2013 Aug;34(8):1075-9. Epub 2013 May 28. PMID: 23674478. PMCID: PMC3714376. doi: 10.1002/humu.22351.
Role: I designed all of the experiments and co-wrote the paper.

46. Sowers LP, Mouw TJ, Ferguson PJ, Wemmie JA, Mohapatra DP, **Bassuk AG**. The non-canonical Wnt ligand Wnt5a rescues morphological deficits in Prickle2-deficient hippocampal neurons., Mol Psychiatry. 2013 Oct;18(10):1049. PMID: 24056908. doi: 10.1038/mp.2013.119.
Role: I designed all of the experiments and co-wrote the paper.

47. Sowers LP, Loo L, Wu Y, Campbell E, Ulrich JD, Wu S, Paemka L, Wassink T, Meyer K, Bing X, El-Shanti H, Usachev YM, Ueno N, Manak JR, Shepherd AJ, Ferguson PJ, Darbro BW, Richerson GB, Mohapatra DP, Wemmie JA, **Bassuk AG**. Disruption of the non-canonical Wnt gene PRICKLE2 leads to autism-like behaviors with evidence for hippocampal synaptic dysfunction., Mol Psychiatry. 2013 Oct;18(10):1077-89. Epub 2013 May 28. PMID: 23711981. PMCID: PMC4163749. doi: 10.1038/mp.2013.71.
Role: I designed all of the experiments and co-wrote the paper.

48. Reiff A, **Bassuk AG**, Church JA, Campbell E, Bing X, Ferguson PJ. Exome sequencing reveals RAG1 mutations in a child with autoimmunity and sterile chronic multifocal osteomyelitis evolving into disseminated granulomatous disease, J Clin Immunol. 2013 Nov;33(8):1289-92. PMID: 24122031. PMCID: PMC3873094. doi: 10.1007/s10875-013-9953-7.
Role: I co-designed all of the genetics experiments and co-wrote the paper.

49. Yang T, **Bassuk AG**, Fritzsch B. Prickle1 stunts limb growth through alteration of cell polarity and gene expression., Dev Dyn. 2013 Nov;242(11):1293-306. Epub 2013 Sep 6. PMID: 23913870. PMCID: PMC3985166. doi: 10.1002/dvdy.24025.
Role: I co-designed all of the genetics experiments and co-wrote the paper.

50. Paemka L, Mahajan VB, Skeie JM, Sowers LP, Ehaideb SN, Gonzalez-Alegre P, Sasaoka T, Tao H, Miyagi A, Ueno N, Takao K, Miyakawa T, Wu S, Darbro BW, Ferguson PJ, Pieper AA, Britt JK, Wemmie JA, Rudd DS, Wassink T, El-Shanti H, Mefford HC, Carvill GL, Manak JR, **Bassuk AG**. PRICKLE1 interaction with SYNAPSIN I reveals a role in autism spectrum disorders., PloS One. 2013 Dec 3;8(12):e80737. eCollection 2013. PMID: 24312498. PMCID: PMC3849077. doi: 10.1371/journal.pone.0080737.
Role: I co-designed all of the genetics experiments and co-wrote the paper.

51. Cassel SL, Janczy JR, Bing X, Wilson SP, Olivier AK, Otero JE, Iwakura Y, Shayakhmetov DM, **Bassuk AG**, Abu-Amer Y, Brogden KA, Burns TL, Sutterwala FS, Ferguson PJ. Inflammasome-independent IL-1β mediates autoinflammatory disease in Pstpip2-deficient mice., Proc Natl Acad Sci U S A. 2014 Jan 21;111(3):1072-7. Epub 2014 Jan 6. PMID: 24395802. PMCID: PMC3903222. doi: 10.1073/pnas.1318685111.
Role: I co-designed all of the genetics experiments and co-wrote the paper.

52. Yang T, Jia Z, Bryant-Pike W, Chandrasekhar A, Murray JC, Fritzsch B, **Bassuk AG**. Analysis of PRICKLE1 in human cleft palate and mouse development demonstrates rare and common variants involved in human malformations., Mol Genet Genomic Med. 2014 Mar;2(2):138-51. Epub 2013 Dec 17. PMID: 24689077. PMCID: PMC3960056. doi: 10.1002/mgg3.53.
Role: Designed all analyses, oversaw all aspects of the project, wrote paper.

53. Sowers LP, Yin T, Mahajan VB, **Bassuk AG**. Defective Motile Cilia in Prickle2-Deficient Mice, J Neurogenet. 2014 Mar-Jun;28(1-2):146–152. Epub 2014 Apr 7. doi: 10.3109/01677063.2014.885966.
Role: Co-designed all analyses, co-wrote paper.

54. **Bassuk AG**, Sujirakul T, Tsang SH, Mahajan VB. A novel RPGR mutation masquerading as Stargardt disease., Br J Pphthalmol. 2014 May;98(5):709-11. Epub 2014 Jan 31. PMID: 24489377. PMCID: PMC4170590. doi: 10.1136/bjophthalmol-2013-304822.
Role: I co-designed all of the genetics experiments and co-wrote the paper.
See: http://now.uiowa.edu/2014/02/taking-long-view.

55. Wert KJ, Skeie JM, **Bassuk AG**, Olivier AK, Tsang SH, Mahajan VB. Functional validation of a human CAPN5 exome variant by lentiviral transduction into mouse retina., Hum Mol Genet. 2014 May 15;23(10):2665-77. Epub 2013 Dec 30. PMID: 24381307. PMCID: PMC3990166. doi: 10.1093/hmg/ddt661.
Role: I co-designed all of the genetics experiments and co-wrote the paper.

56. Ehaideb SN, Iyengar A, Ueda A, Iacobucci GJ, Cranston C, **Bassuk AG**, Gubb D, Axelrod JD, Gunawardena S, Wu CF, Manak JR. Prickle modulates microtubule polarity and axonal transport to ameliorate seizures in flies., Proc Natl Acad Sci U S A. 2014 Jul 29;111(30):11187-92. Epub 2014 Jul 14. PMID: 25024231. PMCID: PMC4121842. doi: 10.1073/pnas.1403357111.
Role: Assisted with writing and design.

57. Mei X, Westfall TA, Zhang Q, Sheffield VC, **Bassuk AG**, Slusarski DC. Functional characterization of Prickle2 and BBS7 identify overlapping phenotypes yet distinct mechanisms., Dev Biol. 2014 Aug 15;392(2):245-55. Epub 2014 June 2. PMID: 24938409. PMCID: PMC4114335. doi: 10.1016/j.ydbio.2014.05.020.
Role: Co-designed all analyses, co-wrote paper.

58. Yang T, **Bassuk AG**, Stricker S, Fritzsch B. Prickle1 is necessary for the caudal migration of murine facial branchiomotor neurons., Cell Tissue Res. 2014 Sep;357(3):549-61. Epub 2014 June 15. PMID: 24927917. PMCID: PMC4149827. doi: 10.1007/s00441-014-1925-6.
Role: Co-designed all analyses, co-wrote paper.

59. Yin TC, Britt JK, De Jesús-Cortés H, Lu Y, Genova RM, Khan MZ, Voorhees JR, Shao J, Katzman AC, Huntington PJ, Wassink C, McDaniel L, Newell EA, Dutca

LM, Naidoo J, Cui H, **Bassuk AG**, Harper MM, McKnight SL, Ready JM, Pieper AA. P7C3 neuroprotective chemicals block axonal degeneration and preserve function after traumatic brain injury., Cell Rep. 2014 Sep 25;8(6):1731-40. Epub 2014 Sep 15. PMID: 25220467. PMCID: PMC4206693. doi: 10.1016/j.celrep.2014.08.030.
Role: Assisted with writing and design.

60. Gonzalez-Alegre P, Di Paola J, Wang K, Fabbro S, Yu H-C, Shaikh TH, Darbro BW, **Bassuk AG**. Evaluating Familial Essential Tremor with Novel Genetic Approaches: Is it a Genotyping or Phenotyping Issue?, Tremor Other Hyperkinet Mov (N Y). 2014 Oct 20;4:258. eCollection 2014. PMID: 25374765. PMCID: PMC4219111. doi: 10.7916/D8FB51G3.
Role: Designed all analyses, oversaw all aspects of the project, wrote paper.

61. Paemka L, Mahajan VB, Ehaideb SN, Skeie JM, Tan MC, Wu S, Cox AJ, Sowers LP, Gecz J, Jolly L, Ferguson PJ, Darbro B, Schneider A, Scheffer IE, Carvill GL, Mefford HC, El-Shanti H, Wood SA, Manak JR, **Bassuk AG**. Seizures are regulated by ubiquitin-specific peptidase 9 X-linked (USP9X), a de-ubiquitinase., PLoS Genet. 2015 Mar 12;11(3):e1005022. eCollection 2015 Mar. PMID: 25763846. PMCID: PMC4357451. doi: 10.1371/journal.pgen.1005022.
Role: Designed all analyses, oversaw all aspects of the project, wrote paper.

62. **Bassuk AG**, Yeh S, Wu S, Martin DF, Tsang SH, Gakhar L, Mahajan VB. Structural modeling of a novel CAPN5 mutation that causes uveitis and neovascular retinal detachment., PLoS One. 2015 Apr 9;10(4):e0122352. eCollection 2015. PMID: 25856303. PMCID: PMC4391918. doi: 10.1371/journal.pone.0122352.
Role: Designed all analyses, oversaw all aspects of the project, wrote paper.

63. Wert KJ, **Bassuk AG**, Wu WH, Gakhar L, Coglan D, Mahajan M, Wu S, Yang J, Lin CS, Tsang SH, Mahajan VB. CAPN5 mutation in hereditary uveitis: the R243L mutation increases calpain catalytic activity and triggers intraocular inflammation in a mouse model., Hum Mol Genet. 2015 Aug 15;24(16):4584-98. Epub 2015 May 20. PMID: 25994508. PMCID: PMC4512628. doi: 10.1093/hmg/ddv189.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

64. **Bassuk AG**, Sherr EH. A de novo mutation in PRICKLE1 in fetal agenesis of the corpus callosum and polymicrogyria, J Neurogenet. 2015;29(4):174-7. Epub 2016 Jan 4. PMID: 26727662. PMCID: PMC4813514. doi: 10.3109/01677063.2015.1088847.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

65. **Bassuk AG**, Zheng A, Li Y, Tsang SH, Mahajan VB. Precision Medicine: Genetic Repair of Retinitis Pigmentosa in Patient-Derived Stem Cells, Sci Rep. 2016 Jan 27;6:19969. PMID: 26814166. PMCID: PMC4728485. doi: 10.1038/srep19969.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

66. Darbro BW, Singh R, Zimmerman MB, Mahajan VB, **Bassuk AG**. Autism Linked to Increased Oncogene Mutations but Decreased Cancer Rate, PloS One. 2016 Mar 2;11(3):e0149041. eCollection 2016. PMID: 26934580. PMCID:

PMC4774916. doi: 10.1371/journal.pone.0149041.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote pape

67. Mahajan VB, **Bassuk AG**. Response to Sandford et al.: PRICKLE2 Variants in Epilepsy: A Call for Precision Medicine, Am J Hum Genet. 2016 March 3;98(3):590–591. PMID: 26942292. PMCID: PMC4800048. doi: 10.1016/j.ajhg.2016.02.002.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

68. Velez G, Roybal CN, Colgan D, Tsang SH, **Bassuk AG**, Mahajan VB. Precision Medicine: Personalized Proteomics for the Diagnosis and Treatment of Idiopathic Inflammatory Disease, JAMA Ophthalmol. 2016 Apr;134(4):444-8. PMID: 26848019. PMCID: PMC4833518. doi: 10.1001/jamaophthalmol.2015.5934.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

69. Wert KJ, Mahajan VB, Zhang L, Yan Y, Li Y, Tosi J, Hsu CW, Nagasaki T, Janisch KM, Grant MB, Mahajan M, **Bassuk AG**, Tsang SH. Neuroretinal hypoxic signaling in a new preclinical murine model for proliferative diabetic retinopathy., Signal Transduct Target Ther. 2016;1. Epub Apr 22. PMID: 27195131. PMCID: PMC4868361. doi: 10.1038/sigtrans.2016.5.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

70. Schaefer KA, Toral MA, Velez G, Cox AJ, Baker SA, Borcherding NC, Colgan DF, Bondada V, Mashburn CB, Yu C-G, Geddes JW, Tsang SH, **Bassuk AG**, Mahajan VB. Calpain-5 Expression in the Retina Localizes to Photoreceptor Synapses., Invest Ophthalmol Vis Sci. 2016 May 1;57(6):2509–2521. PMID: 27152965. PMCID: PMC4868102. doi: 10.1167/iovs.15-18680.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

71. Cham A, Bansal M, Banda HK, Kwon Y, Tlucek PS, **Bassuk AG**, Tsang SH, Sobol WM, Folk JC, Yeh S, Mahajan VB. Secondary glaucoma in CAPN5-associated neovascular inflammatory vitreoretinopathy, Clin Ophthalmol. 2016 Jun 27;10:1187–1197. eCollection 2016. PMID: 27390515. PMCID: PMC4930228. doi: 10.2147/OPTH.S103324.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

72. Moshfegh Y, Velez G, Li Y, **Bassuk AG**, Mahajan VB, Tsang SH. BESTROPHIN1 mutations cause defective chloride conductance in patient stem cell-derived RPE., Hum Mol Genet. 2016 Jul 1;25(13):2672-2680. Epub 2016 May 18. PMID: 27193166. PMCID: PMC5181636. doi: 10.1093/hmg/ddw126.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

73. Ehaideb SN, Wignall EA, Kasuya J, Evans WH, Iyengar A, Koerselman HL, Lilienthal AJ, **Bassuk AG**, Kitamoto T, Manak JR. Mutation of orthologous prickle genes causes a similar epilepsy syndrome in flies and humans., Ann Clin Transl Neurol. 2016 Aug 3;3(9):695-707. eCollection 2016 Sep. PMID: 27648459. PMCID: PMC5018582. doi: 10.1002/acn3.334.

Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

74. Wu W-H, Tsai Y-T, Justus S, Lee T-T, Zhang L, Lin C-S, **Bassuk AG**, Mahajan VB, Tsang SH. CRISPR Repair Reveals Causative Mutation in a Preclinical Model of Retinitis Pigmentosa, Mol Ther. 2016 Aug;24(8):1388-94. Epub 2016 May 20. PMID: 27203441. PMCID: PMC5023380. doi: 10.1038/mt.2016.107.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

75. Yang J, **Bassuk AG**, Merl-Pham J, Hsu CW, Colgan DF, Li X, Au KS, Zhang L, Smemo S, Justus S, Nagahama Y, Grossbach AJ, Howard, 3rd MA, Kawasaki H, Feldstein NA, Dobyns WB, Northrup H, Hauck SM, Ueffing M, Mahajan VB, Tsang SH. Catenin delta-1 (CTNND1) phosphorylation controls the mesenchymal to epithelial transition in astrocytic tumors., Hum Mol Genet. 2016 Oct 1;25(19):4201-4210. Epub 2016 Aug 11. PMID: 27516388. PMCID: PMC5291196. doi: 10.1093/hmg/ddw253.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

76. Zhang L, Justus S, Xu Y, Pluchenik T, Hsu CW, Yang J, Duong JK, Lin CS, Jia Y, **Bassuk AG**, Mahajan VB, Tsang SH. Reprogramming towards anabolism impedes degeneration in a preclinical model of retinitis pigmentosa., Hum Mol Genet. 2016 Oct 1;25(19):4244-4255. Epub 2016 Aug 11. PMID: 27516389. PMCID: PMC5291198. doi: 10.1093/hmg/ddw256.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

77. Wang Y, Williams J, Rattner A, Wu S, **Bassuk AG**, Goffinet AM, Nathans J. Patterning of papillae on the mouse tongue: A system for the quantitative assessment of planar cell polarity signaling., Dev Biol. 2016 Nov 15;419(2):298-310. Epub 2016 Sep 6. PMID: 27612405. doi: 10.1016/j.ydbio.2016.09.004.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

78. Gakhar L, **Bassuk AG**, Velez G, Khan S, Yang J, Tsang SH, Mahajan VB. Small-angle X-ray scattering of calpain-5 reveals a highly open conformation among calpains., J Struct Biol. 2016 Dec;196(3):309-318. Epub 2016 Jul 27. PMID: 27474374. PMCID: PMC5118095. doi: 10.1016/j.jsb.2016.07.017.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

79. Kambouris M, Thevenon J, Soldatos A, Cox A, Stephen J, Ben-Omran T, Al-Sarraj Y, Boulos H, Bone W, Mullikin JC, Masurel-Paulet A, St-Onge J, Dufford Y, Chantegret C, Thauvin-Robinet C, Al-Alami J, Faivre L, Riviere JB, Gahl WA, **Bassuk AG**, Malicdan MC, El-Shanti H. Biallelic *SCN10A* mutations in neuromuscular disease and epileptic encephalopathy., Ann Clin Transl Neurol. 2016 Dec 20;4(1):26-35. eCollection 2017 Jan. PMID: 28078312. PMCID: PMC5221474. doi: 10.1002/acn3.372.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

80. T Toral MA, Velez G, Boudreault K, Schaefer KA, Xu Y, Saffra N, **Bassuk AG**, Tsang SH, Mahajan VB. Structural modeling of a novel SLC38A8 mutation that

causes foveal hypoplasia., Mol Genet Genomic Med. 2017 Feb 26;5(3):202-209. eCollection 2017 May. PMID: 28546991. PMCID: PMC5441399. doi: 10.1002/mgg3.266.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

81. Cox AJ, Darbro BW, Laxer RM, Velez G, Bing X, Finer AL, Erives A, Mahajan VB, **Bassuk AG***, Ferguson PJ. Recessive coding and regulatory mutations in FBLIM1 underlie the pathogenesis of chronic recurrent multifocal osteomyelitis (CRMO)., PLoS One. 2017 Mar 16;12(3):e0169687. eCollection 2017. PMID: 28301468. PMCID: PMC5354242. doi: 10.1371/journal.pone.0169687.
*Co-corresponding author
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

82. Velez G, Roybal CN, Binkley E, **Bassuk AG**, Tsang SH, Mahajan VB. Proteomic Analysis of Elevated Intraocular Pressure with Retinal Detachment., Am J Ophthalmol Case Rep. 2017 Apr;5:107-110. Epub 2017 Jan 3. PMID: 28825049. PMCID: PMC5560621. doi: 10.1016/j.ajoc.2016.12.023.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

83. Schaefer KA, Wu WH, Colgan DF, Tsang SH, **Bassuk AG**, Mahajan VB. Unexpected mutations after CRISPR-Cas9 editing in vivo., Nat Methods. 2017 May 30;14(6):547-548. PMID: 28557981. PMCID: PMC5796662. doi: 10.1038/nmeth.4293.
Role: Retracted Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

84. Cox AJ, Darbro BW, Laxer RM, Velez G, Bing X, Finer AL, Erives A, Mahajan VB, **Bassuk AG**, Ferguson PJ. Correction: Recessive coding and regulatory mutations in FBLIM1 underlie the pathogenesis of chronic recurrent multifocal osteomyelitis (CRMO)., PLoS One. 2017 Jul 7;12(7):e0181222. eCollection 2017. PMID: 28686717. PMCID: PMC5501673. doi: 10.1371/journal.pone.0181222.
*Co-corresponding author
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

85. Yang T, Kersigo J, Wu S, Fritzsch B, **Bassuk AG**. Prickle1 regulates neurite outgrowth of apical spiral ganglion neurons but not hair cell polarity in the murine cochlea., PLoS One. 2017 Aug 24;12(8):e0183773. eCollection 2017. PMID: 28837644. PMCID: PMC5570324. doi: 10.1371/journal.pone.0183773.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

86. Cabral T, Toral MA, Velez G, DiCarlo JE, Gore AM, Mahajan M, Tsang SH, **Bassuk AG**, Mahajan VB. Dissection of Human Retina and RPE-Choroid for Proteomic Analysis., J Vis Exp. 2017 Nov 12;(129). PMID: 29155757. doi: 10.3791/56203.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

87. Schaefer K, Mahajan M, Gore A, Tsang SH, **Bassuk AG**, Mahajan VB. Calpain-5 gene expression in the mouse eye and brain., BMC Res Notes. 2017 Nov 21;10(1):602. PMID: 29157313. PMCID: PMC5697233. doi: 10.1186/s13104-017-2927-8.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

88. Velez G, Tsang SH, Tsai YT, Hsu CW, Gore A, Abdelhakim AH, Mahajan M, Silverman RH, Sparrow JR, **Bassuk AG**, Mahajan VB. Gene Therapy Restores Mfrp and Corrects Axial Eye Length., Sci Rep. 2017 Nov 23;7(1):16151. PMID: 29170418. PMCID: PMC5701072. doi: 10.1038/s41598-017-16275-8.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

89. Velez G, **Bassuk AG**, Colgan D, Tsang SH, Mahajan VB. Therapeutic drug repositioning using personalized proteomics of liquid biopsies., JCI Insight. 2017 Dec 21;2(24). pii: 97818. [Epub ahead of print]. PMID: 29263305. PMCID: PMC5752263. doi: 10.1172/jci.insight.97818.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

90. Wu WH, Tsai YT, Justus S, Cho GY, Sengillo JD, Xu Y, Cabral T, Lin CS, **Bassuk AG**, Mahajan VB, Tsang SH. CRISPR Repair Reveals Causative Mutation in a Preclinical Model of Retinitis Pigmentosa: A Brief Methodology., Methods Mol Biol. 2018;1715:191-205. PMID: 29188514. doi: 10.1007/978-1-4939-7522-8_13.

91. Roybal CN, Velez G, Toral MA, Tsang SH, **Bassuk AG**, Mahajan VB. Personalized Proteomics in Proliferative Vitreoretinopathy Implicate Hematopoietic Cell Recruitment and mTOR as a Therapeutic Target., Am J Ophthalmol. 2018 Feb;186:152-163. Epub 2017 Dec 13. PMID: 29246578. PMCID: PMC5805631. doi: 10.1016/j.ajo.2017.11.025.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

92. Apatoff MBL, Sengillo JD, White EC, Bakhoum MF, Bassuk AG, Mahajan VB, Tsang SH. Autologous stem cell therapy for inherited and acquired retinal disease. Regen Med. 2018 Jan;13(1):89-96. doi: 10.2217/rme-2017-0089. Epub 2018 Jan 23. PMID: 29360008; PMCID: PMC6123878.

93. Velez G, Machlab DA, Tang PH, Sun Y, Tsang SH, **Bassuk AG**, Mahajan VB. Proteomic analysis of the human retina reveals region-specific susceptibilities to metabolic- and oxidative stress-related diseases., PLoS One. 2018 Feb 21;13(2):e0193250. eCollection 2018. PMID: 29466423. PMCID: PMC5821407. doi: 10.1371/journal.pone.0193250.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

94. Velez G, **Bassuk AG**, Schaefer KA, Brooks B, Gakhar L, Mahajan M, Kahn P, Tsang SH, Ferguson PJ, Mahajan VB. A novel de novo CAPN5 mutation in a patient with inflammatory vitreoretinopathy, hearing loss, and developmental delay., Cold Spring Harb Mol Case Stud. 2018 Feb 22. pii: mcs.a002519. [Epub ahead of print]. PMID: 29472286. doi: 10.1101/mcs.a002519.

Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

95. Evans LP, Newell EA, Mahajan M, Tsang SH, Ferguson PJ, Mahoney J, Hue CD, Vogel EW 3rd, Morrison B 3rd, Arancio O, Nichols R, **Bassuk AG**, Mahajan VB. Acute vitreoretinal trauma and inflammation after traumatic brain injury in mice., Ann Clin Transl Neurol. 2018 Feb 26;5(3):240-251. eCollection 2018 Mar. PMID: 29560370. PMCID: PMC5846452. doi: 10.1002/acn3.523.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

96. Jauregui R, Cho GY, Takahashi VKL, Takiuti JT, **Bassuk AG**, Mahajan VB, Tsang SH. Caring for Hereditary Childhood Retinal Blindness., Asia Pac J Ophthalmol (Phila). 2018 Mar 14. [Epub ahead of print]. PMID: 29536675. doi: 10.22608/APO.201851.
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

97. Newell EA, Todd BP, Mahoney J, Pieper AA, Ferguson PJ, Bassuk AG. Combined Blockade of Interleukin-1α and -1β Signaling Protects Mice from Cognitive Dysfunction after Traumatic Brain Injury. eNeuro. 2018 Apr 13;5(2):ENEURO.0385-17.2018. doi: 10.1523/ENEURO.0385-17.2018. PMID: 29662944; PMCID: PMC5898697.

98. Lin MK, Yang J, Hsu CW, Gore A, **Bassuk AG**, Brown LM, Colligan R, Sengillo JD, Mahajan VB, Tsang SH. HTRA1, an age-related macular degeneration protease, processes extracellular matrix proteins EFEMP1 and TSP1., Aging Cell. 2018 May 5:e12710. . [Epub ahead of print]. PMID: 29730901. doi: 10.1111/acel.12710
Role: Co-designed all analyses, oversaw all aspects of the project, co-wrote paper.

99. Todd BP, **Bassuk AG**. A de novo mutation in PRICKLE1 associated with myoclonic epilepsy and autism spectrum disorder., J Neurogenet. 2018 May 23:1-3. PMID: 29790814. [Epub ahead of print]. doi: 10.1080/01677063.2018.1473862.
Role: Co-wrote the paper.

100. Liu KY, Sengillo JD, Velez G, Jauregui R, Sakai LY, Maumenee IH, **Bassuk AG**, Mahajan VB, Tsang SH. Missense mutation in SLIT2 associated with congenital myopia, anisometropia, connective tissue abnormalities, and obesity., Orphanet J Rare Dis. 2018 Aug 15;13(1):138.
doi: 10.1186/s13023-018-0885-4.
PMID: 30111362
Role: Co-wrote the paper

101. Wert KJ, Velez G, Cross MR, Wagner BA, Teoh-Fitzgerald ML, Buettner GR, McAnany JJ, Olivier A, Tsang SH, Harper MM, Domann FE, **Bassuk AG**, Mahajan VB. Extracellular superoxide dismutase (SOD3) regulates oxidative stress at the vitreoretinal interface., Free Radic Biol Med. 2018 Aug 20;124:408-419.
doi: 10.1016/j.freeradbiomed.2018.06.024. Epub 2018 Jun 22.
PMID: 29940351
Role: Co-wrote the paper

102. Jauregui R, Park KS, **Bassuk AG**, Mahajan VB, Tsang SH. Deferoxamine-induced electronegative ERG responses. Doc Ophthalmol. 2018 Aug;137(1):15-23. doi: 10.1007/s10633-018-9640-3. Epub 2018 May 16. PMID: 29770904.
Role: Co-wrote the paper

103. Velez G, Tang PH, Cabral T, Cho GY, Machlab DA, Tsang SH, **Bassuk AG**, Mahajan VB. Personalized Proteomics for Precision Health: Identifying Biomarkers of Vitreoretinal Disease., Transl Vis Sci Technol. 2018 Sep 26;7(5):12. doi: 10.1167/tvst.7.5.12. eCollection 2018 Sep.
PMID: 30271679
Role: Co-wrote the paper

104. Tang PH, Kinnick TR, Folk JC, Mahajan M, **Bassuk AG**, Tsang SH, Mahajan VB. Progression of scotopic single-flash electroretinography in the stages of capn5 vitreoretinopathy., Retin Cases Brief Rep. 2018 Oct 8. doi: 10.1097/ICB.0000000000000828. [Epub ahead of print]
PMID: 30300311
Role: Co-wrote the paper

105. Machlab DA, Velez G, **Bassuk AG**, Mahajan VB. ProSave: an application for restoring quantitative data to manipulated subsets of protein lists., Source Code Biol Med. 2018 Nov 12;13:3. doi: 10.1186/s13029-018-0070-0. eCollection 2018.
PMID: 30459825
Role: Co-wrote the paper

106. Brueggeman L, Sturgeon ML, Martin RM, Grossbach AJ, Nagahama Y, Zhang A, Howard MA 3rd, Kawasaki H, Wu S, Cornell RA, Michaelson JJ, **Bassuk AG.** Drug repositioning in epilepsy reveals novel antiseizure candidates., Ann Clin Transl Neurol. 2018 Dec 11;6(2):295-309. doi: 10.1002/acn3.703. eCollection 2019 Feb.
PMID: 30847362
Role: Co-wrote the paper

107. Tang PH, Velez G, Tsang SH, **Bassuk AG**, Mahajan VB. VCAN Canonical Splice Site Mutation is Associated With Vitreoretinal Degeneration and Disrupts an MMP Proteolytic Site., Invest Ophthalmol Vis Sci. 2019 Jan 2;60(1):282-293. doi: 10.1167/iovs.18-25624.
PMID: 30657523
Role: Co-wrote the paper

108. Kousa YA, Zhu H, Fakhouri WD, Lei Y, Kinoshita A, Roushangar RR, Patel NK, Agopian AJ, Yang W, Leslie EJ, Busch TD, Mansour TA, Li X, Smith AL, Li EB, Sharma DB, Williams TJ, Chai Y, Amendt BA, Liao EC, Mitchell LE, **Bassuk AG**, Gregory S, Ashley-Koch A, Shaw GM, Finnell RH, Schutte BC. The TFAP2A-IRF6-GRHL3 genetic pathway is conserved in neurulation., Hum Mol Genet. 2019 Jan 25. doi: 10.1093/hmg/ddz010. [Epub ahead of print]
PMID: 30689861
Role: Co-wrote the paper

109. Tang PH, Jauregui R, Tsang SH, **Bassuk AG**, Mahajan VB. Optical Coherence Tomography Angiography of RPGR-Associated Retinitis Pigmentosa Suggests Foveal Avascular Zone is a Biomarker for Vision Loss., Ophthalmic Surg Lasers Imaging Retina. 2019 Feb 1;50(2):e44-e48. doi: 10.3928/23258160-20190129-18.

PMID: 30768229
Role: Co-wrote the paper

110. Sancho-Pelluz J, Cui X, Lee W, Tsai YT, Wu WH, Justus S, Washington I, Hsu CW, Park KS, Koch S, Velez G, **Bassuk AG**, Mahajan VB, Lin CS, Tsang SH., Mechanisms of neurodegeneration in a preclinical autosomal dominant retinitis pigmentosa knock-in model with a RhoD190N mutation., Cell Mol Life Sci. 2019 Apr 11. doi: 10.1007/s00018-019-03090-9. [Epub ahead of print]
PMID: 30976840
Role: Co-wrote the paper

111. Wert KJ, Bakall B, **Bassuk AG**, Tsang SH, Mahajan VB. Insights into Retinal Development Using Live Imaging in Female Carriers of Choroideremia. Ophthalmic Surg Lasers Imaging Retina. 2019 May 1;50(5):e158-e162. doi:10.3928/23258160-20190503-15. PubMed PMID: 31100169.
Role: Co-wrote the paper

112. Velez G, Yang J, Li AS, Tsang SH, **Bassuk AG**, Mahajan VB. Proteomic insight into the pathogenesis of CAPN5-vitreoretinopathy. Sci Rep. 2019 May 20;9(1):7608. doi: 10.1038/s41598-019-44031-7. PubMed PMID: 31110225; PubMed Central PMCID: PMC6527583.
Role: Co-wrote the paper

113. Abe K, Cox A, Takamatsu N, Velez G, Laxer RM, Tse SML, Mahajan VB, **Bassuk AG**, Fuchs H, Ferguson PJ, Hrabe de Angelis M. Gain-of-function mutations in a member of the Src family kinases cause autoinflammatory bone disease in mice and humans. Proc Natl Acad Sci U S A. 2019 Jun 11;116(24):11872-11877. doi:10.1073/pnas.1819825116. Epub 2019 May 28. PubMed PMID: 31138708; PubMed Central PMCID: PMC6575637.
Role: Co-wrote the paper

114. Cox AJ, Grady F, Velez G, Mahajan VB, Ferguson PJ, Kitchen A, Darbro BW, **Bassuk AG**. In trans variant calling reveals enrichment for compound heterozygous variants in genes involved in neuronal development and growth. Genet Res (Camb). 2019 Jun 13;101:e8. doi: 10.1017/S0016672319000065. PubMed PMID: 31190668.
Role: Co-wrote the paper

115. Wert KJ, Koch SF, Velez G, Hsu CW, Mahajan M, **Bassuk AG**, Tsang SH, Mahajan VB. CAPN5 genetic inactivation phenotype supports therapeutic inhibition trials. Hum Mutat. 2019 Aug 12. doi: 10.1002/humu.23894. [Epub ahead of print]. DOI: 10.1002/humu.23894. PMID: 31403230.
Role: Co-wrote the paper

116. Zhang L, Cui X, Han Y, Park KS, Gao X, Zhang X, Yuan Z, Hu Y, Hsu CW, Li X, **Bassuk AG**, Mahajan VB, Wang NK, Tsang SH. Hypoxic drive caused type 3 neovascularization in a preclinical model of exudative age-related macular degeneration. Hum Mol Genet. 2019 Oct 15;28(20):3475-3485. doi: 10.1093/hmg/ddz159. PubMed PMID: 31518400.
Role: Co-wrote the paper

117. Harper MM, Woll AW, Evans LP, Delcau M, Akurathi A, Hedberg-Buenz A, Soukup DA, Boehme N, Hefti MM, Dutca LM, Anderson MG, **Bassuk AG**. Blast Preconditioning Protects Retinal Ganglion Cells and Reveals Targets for Prevention of Neurodegeneration Following Blast-Mediated Traumatic Brian

Injury. Invest Ophthalmol Vis Sci. 2019 Oct 1;60(13):4159-4170. doi: 10.1167/iovs.19-27565. PubMed PMID: 31598627; PubMed Central PMCID: PMC6785841.
Role: Co-wrote the paper

118. Oh JK, Lima de Carvalho JR, Sun YJ, Ragi S, Yang J, Levi SR, Ryu J, **Bassuk AG**, Mahajan VB, Tsang SH. Novel mutations in the 3-box motif of the BACK domain of KLHL7 associated with nonsyndromic autosomal dominant retinitis pigmentosa. Orphanet J Rare Dis. 2019 Dec 19;14(1):295. doi: 10.1186/s13023-019-1275-2. PubMed PMID: 31856884; PubMed Central PMCID: PMC6924004.
Role: Co-wrote the paper

119. Velez G, Sun YJ, Khan S, Yang J, Herrmann J, Chemudupati T, MacLaren RE, Gakhar L, Wakatsuki S, **Bassuk AG**, Mahajan VB. Structural Insights into the Unique Activation Mechanisms of a Non-classical Calpain and Its Disease-Causing Variants. Cell Rep. 2020 Jan 21;30(3):881-892.e5. doi: 10.1016/j.celrep.2019.12.077. PubMed PMID: 31968260.
Role: Co-wrote the paper

120. Wert KJ, Velez G, Kanchustambham VL, Shankar V, Evans LP, Sengillo JD, Zare RN, **Bassuk AG**, Tsang SH, Mahajan VB. Metabolite therapy guided by liquid biopsy proteomics delays retinal neurodegeneration. EBioMedicine. 2020 Feb 3;52:102636. doi: 10.1016/j.ebiom.2020.102636. [Epub ahead of print] PMID: 32028070
Role: Co-wrote the paper

121. Cho A, Lima de Carvalho JR, Tanaka AJ, Jauregui R, Levi SR, **Bassuk AG**, Mahajan VB, Tsang SH. Fundoscopy-directed genetic testing to re-evaluate negative whole exome sequencing results. Orphanet J Rare Dis. 2020 Jan 30;15(1):32. doi: 10.1186/s13023-020-1312-1. PMID: 32000842
Role: Co-wrote the paper

122. Newell EA, Todd BP, Luo Z, Evans LP, Ferguson PJ, Bassuk AG. A Mouse Model for Juvenile, Lateral Fluid Percussion Brain Injury Reveals Sex-Dependent Differences in Neuroinflammation and Functional Recovery. J Neurotrauma. 2020 February 15; 37(4). doi: 10.1089/neu.2019.6675. [Epub ahead of print] PubMed PMID: 31621484.
Role: Co-wrote the paper

123. Harper MM, Rudd D, Meyer KJ, Kanthasamy AG, Anantharam V, Pieper AA, Vázquez-Rosa E, Shin MK, Chaubey K, Koh Y, Evans LP, **Bassuk AG**, Anderson MG, Dutca L, Kudva IT, John M. Identification of chronic brain protein changes and protein targets of serum auto-antibodies after blast-mediated traumatic brain injury. Heliyon. 2020 Feb 17;6(2):e03374. doi: 10.1016/j.heliyon.2020.e03374. eCollection 2020 Feb. PubMed PMID: 32099918; PubMed Central PMCID: PMC7029173.

124. Tang PH, Chemudupati T, Wert KJ, Folk JC, Mahajan M, Tsang SH, **Bassuk AG**, Mahajan VB. Phenotypic variance in Calpain-5 retinal degeneration. Am J Ophthalmol Case Rep. 2020 Feb 24;18:100627. doi: 10.1016/j.ajoc.2020.100627. eCollection 2020 Jun. PubMed PMID: 32274441; PubMed Central PMCID: PMC7132063.
Role: Co-wrote the paper

125. Sepah YJ, Velez G, Tang PH, Yang J, Chemudupati T, Li AS, Nguyen QD, **Bassuk AG**, Mahajan VB. Proteomic analysis of intermediate uveitis suggests myeloid cell recruitment and implicates IL-23 as a therapeutic target. Am J Ophthalmol Case Rep. 2020 Mar 6;18:100646. doi: 10.1016/j.ajoc.2020.100646. eCollection 2020 Jun. PubMed PMID: 32274442; PubMed Central PMCID: PMC7132169.
Role: Co-wrote the paper

126. Evans LP, Woll AW, Wu S, Todd BP, Hehr N, Hedberg-Buenz A, Anderson MG, Newell EA, Ferguson PJ, Mahajan VB, Harper MM, **Bassuk AG**. Modulation of Post-Traumatic Immune Response Using the IL-1 Receptor Antagonist Anakinra for Improved Visual Outcomes. J Neurotrauma. 2020 Jun 15;37(12):1463-1480. doi: 10.1089/neu.2019.6725. Epub 2020 Mar 16. PubMed PMID: 32056479; PubMed Central PMCID: PMC7249480.
Role: Co-wrote the paper

127. Hildebrand JM, Kauppi M, Majewski IJ, Liu Z, Cox AJ, Miyake S, Petrie EJ, Silk MA, Li Z, Tanzer MC, Brumatti G, Young SN, Hall C, Garnish SE, Corbin J, Stutz MD, Di Rago L, Gangatirkar P, Josefsson EC, Rigbye K, Anderton H, Rickard JA, Tripaydonis A, Sheridan J, Scerri TS, Jackson VE, Czabotar PE, Zhang JG, Varghese L, Allison CC, Pellegrini M, Tannahill GM, Hatchell EC, Willson TA, Stockwell D, de Graaf CA, Collinge J, Hilton A, Silke N, Spall SK, Chau D, Athanasopoulos V, Metcalf D, Laxer RM, **Bassuk AG**, Darbro BW, Fiatarone Singh MA, Vlahovich N, Hughes D, Kozlovskaia M, Ascher DB, Warnatz K, Venhoff N, Thiel J, Biben C, Blum S, Reveille J, Hildebrand MS, Vinuesa CG, McCombe P, Brown MA, Kile BT, McLean C, Bahlo M, Masters SL, Nakano H, Ferguson PJ, Murphy JM, Alexander WS, Silke J. A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. Nat Commun. 2020 Jun 19;11(1):3150. doi:10.1038/s41467-020-16819-z. PubMed PMID: 32561755.
Role: Co-wrote the paper

128. Agostinelli LJ, Mix MR, Hefti MM, Scammell TE, **Bassuk AG**., Input-output connections of LJA5 prodynorphin neurons. J Comp Neurol. 2020 Jun 30. doi: 10.1002/cne.24974. Online ahead of print. PMID: 32602558.
Role: Co-wrote paper

129. Li AS, Velez G, Darbro B, Toral MA, Yang J, Tsang SH, Ferguson PJ, Folk JC, **Bassuk AG**, Mahajan VB. Whole-Exome Sequencing of Patients with Posterior Segment Uveitis. Am J Ophthalmol. 2020 Jul 21:S0002-9394(20)30378-0. doi: 10.1016/j.ajo.2020.07.021. Online ahead of print.PMID: 32707200.
Role: Co-wrote the paper

130. Evans LP, Boehme N, Wu S, Burghardt EL, Akurathi A, Todd BP, Newell EA, Ferguson PJ, Mahajan VB, Dutca LM, Harper MM, **Bassuk AG**. Sex Does Not Influence Visual Outcomes After Blast-Mediated Traumatic Brain Injury but IL-1 Pathway Mutations Confer Partial Rescue. Invest Ophthalmol Vis Sci. 2020 Oct 1;61(12):7. doi: 10.1167/iovs.61.12.7.PMID: 33030508.
Role: Co-wrote the paper

131. Agostinelli LJ, **Bassuk AG**. Novel inhibitory brainstem neurons with selective projections to spinal lamina I reduces both pain and itch. J Comp Neurol. 2020 Nov 27. doi: 10.1002/cne.25076 Epub ahead of print. PMID: 33247430.
Role: Co-wrote the paper

132. Sun YJ, Velez G, Parsons DE, Li K, Ortiz ME, Sharma S, McCray PB Jr, **Bassuk AG**, Mahajan VB. Structure-based phylogeny identifies Avoralstat as a TMPRSS2 inhibitor that prevents SARS-CoV-2 infection in mice. J Clin Invest. 2021 Apr 12:147973. doi: 10.1172/JCI147973. Epub ahead of print. PMID: 33844653.
Role: Co-wrote the paper, co-corresponding author

133. Burke EA, Sturgeon M, Zastrow DB, Fernandez L, Prybol C, Marwaha S, Frothingham EP, Ward PA, Eng CM, Fresard L, Montgomery SB, Enns GM, Fisher PG, Wolfe LA, Harding B, Carrington B, Bishop K, Sood R, Huang Y, Elkahloun A, Toro C, **Bassuk AG**, Wheeler MT, Markello TC, Gahl WA, Malicdan MCV. Compound heterozygous KCTD7 variants in progressive myoclonus epilepsy. J Neurogenet. 2021 May 10:1-10. doi: 10.1080/01677063.2021.1892095. Epub ahead of print. PMID: 33970744.
Role: Co-wrote the paper

134. Todd BP, Chimenti MS, Luo Z, Ferguson PJ, **Bassuk AG**, Newell EA. Traumatic brain injury results in unique microglial and astrocyte transcriptomes enriched for type I interferon response. J Neuroinflammation. 2021 Jul 5;18(1):151. doi: 10.1186/s12974-021-02197-w. PMID: 34225752; PMCID: PMC8259035.
Role: Co-corresponding author

135. Bayat A, Iqbal S, Borredy K, Amiel J, Zweier C, Barcia G, Kraus C, Weyhreter H, **Bassuk AG**, Chopra M, Rubboli G, Møller RS. PRICKLE2 revisited-further evidence implicating PRICKLE2 in neurodevelopmental disorders. Eur J Hum Genet. 2021 Jun 7. doi: 10.1038/s41431-021-00912-y. Epub ahead of print. PMID: 34092786.
Role: Co-author

136. Sturgeon ML, Langton R, Sharma S, Cornell RA, Glykys J, **Bassuk AG**. The opioid antagonist naltrexone decreases seizure-like activity in genetic and chemically induced epilepsy models. *Epilepsia Open*. 2021 Sep;6(3):528-538. doi: 10.1002/epi4.12512. Epub 2021 Jun 9. PMID: 34664432; PMCID: PMC8408599.
Role: Co-author, co-wrote the paper

    This paper was selected as the **Basic Science Epilepsia One Prize** for 2022.

137. Toral, M.A., Charlesworth, C.T., Ng, B, Chemudupati T, Homma S, Nakauchi H, **Bassuk AG**, Porteus MH, Mahajan VB. Investigation of Cas9 antibodies in the human eye. *Nat Commun* 13**,** 1053 (2022). https://doi.org/10.1038/s41467-022-28674-1.

    Role: Co-corresponding author

138. Langton RL, Sharma S, Tiarks GC, **Bassuk AG, Glykys J**. Lacosamide decreases neonatal seizures without increasing apoptosis. *Epilepsia*. 2022 Sep 28. doi: 10.1111/epi.17423. Epub ahead of print. PMID: 36168798.

    Role: Co-author

139. Agostinelli LJ, Seaman SC, Saper CB, Fykstra D, Hefti MM, Koscik TR, Dlouhy BJ, **Bassuk AG**. Human Brainstem and Cerebellum Atlas- Chemoarchitecture and Cytoarchitecture Paired to MRI. *Journal of Neuroscience* 28 November 2022, JN-RM-0587-22; DOI: https://doi.org/10.1523/JNEUROSCI.0587-22.2022.

Role: Senior author

140. Nukala KM, Lilienthal AJ, Lye SH, **Bassuk AG**, Chtarbanova S, Manak JR. Downregulation of oxidative stress-mediated glial innate immune response suppresses seizures in a fly epilepsy model. Cell Rep. 2023 Jan 14;42(1):112004. doi: 10.1016/j.celrep.2023.112004. Epub ahead of print. PMID: 36641750.

    Role: Co-author

141. Jackson KJ, Sharma S, Tiarks G, Rodriguez S, **Bassuk AG**. (2023). Fast Detection and Quantification of Interictal Spikes and Seizures in a Rodent Model of Epilepsy Using an Automated Algorithm. *Bio-protocol* 13(6): e4632. DOI: 10.21769/BioProtoc.4632.

    Role: Co-author

142. Todd BP, Luo Z, Gilkes N, Chimenti MS, Peterson Z, Mix M, Harty JT, Nickl-Jockschat T, Ferguson PJ, **Bassuk AG**, Newell EA. Selective neuroimmune modulation by type I interferon drives neuropathology and neurologic dysfunction following traumatic brain injury. *bioRxiv* [Preprint]. 2023 Jun 7:2023.06.06.543774. doi: 10.1101/2023.06.06.543774. PMID: 37333385; PMCID: PMC10274693.

    Role: Co-author

143. Wolf J, Chemudupati T, Kumar A, Rasmussen DK, Wai KM, Chang RT, Montague AA, Tang PH, **Bassuk AG**, Dufour A, Mruthrunjaya P, Mahajan VB. Biobanking of Human Aqueous and Vitreous Liquid Biopsies for Molecular Analyses. *J Vis Exp*. 2023 Sep 11;(199). doi: 10.3791/65804. PMID: 37747194.

    Role: Co-author

144. Wolf J, Rasmussen DK, Sun YJ, Vu JT, Wang E, Espinosa C, Bigini F, Chang RT, Montague AA, Tang PH, Mruthyunjaya P, Aghaeepour N, Dufour A, **Bassuk AG**, Mahajan VB. Liquid-biopsy proteomics combined with AI identifies cellular drivers of eye aging and disease in vivo. *Cell*. 2023 Oct 6:S0092-8674(23)01033-4. doi: 10.1016/j.cell.2023.09.012. Epub ahead of print. PMID: 37863056.

    Role: Co-author

145. Wolf J, Franco JA, Yip R, Dabaja MZ, Velez G, Liu F, **Bassuk AG**, Mruthyunjaya P, Dufour A, Mahajan VB. Liquid Biopsy Proteomics in Ophthalmology. *J Proteome Res*. 2024 Jan 3. doi: 10.1021/acs.jproteome.3c00756. Epub ahead of print. PMID: 38171013.

    Role: Co-author

146. Charras A, Hofmann SR, Cox A, Schulze F, Russ S, Northey S, Liu X, Fang Y, Haldenby S, Hartmann H, **Bassuk AG**, Carvalho A, Sposito F, Grinstein L, Rösen-Wolff A, Meyer-Bahlburg A, Beresford MW, Lainka E, Föll D, Wittkowski H, Girschick HJ, Morbach H, Uebe S, Hüffmeier U, Ferguson PJ, Hedrich CM. P2RX7 gene variants associate with altered inflammasome assembly and reduced pyroptosis in chronic nonbacterial osteomyelitis (CNO). J Autoimmun. 2024 Feb 22;144:103183. doi: 10.1016/j.jaut.2024.103183. Epub ahead of print. PMID: 38401466.

Role: Co-author

Chapters

1. **Bassuk AG**. Ataxia. Just The Facts in Pediatrics. 1st Edition. Thomas, G., Granklin, W., Tanz, R. McGraw-Hill Professional. 2005.
2. **Bassuk AG**. Child neurology [section editor]. Comprehensive Board Review in Neurology. 1st and 2nd Editions. Borsody, M. Thieme Medical Publishers. 2007, 2012.
3. McMahon K, Paciorkowski AR, Walters-Sen LC, Milunsky JM, **Bassuk AG**, Darbro BW, Diaz J, Dobyns WB, Gropman A. Neurogenetics in the Genome Era. Swaiman's Pediatric Neurology: Principles and Practice. 6th edition. Swaiman KF, Ashwal S, Ferriero DM, Schor NF, Finkel RS, Gropman AL, Pearl PL, Shevell MI. Elsevier Inc. 2018.
4. **Darbro B, Bassuk, AG.** Microarray analysis in Pediatric Neurology. (Chapter) Swaiman's Pediatric Neurology: Principles and Practice. 6th edition. Swaiman KF, Ashwal S, Ferriero DM, Schor NF, Finkel RS, Gropman AL, Pearl PL, Shevell MI. Elsevier Inc. 2018

Reviews

1. **Bassuk AG**, Leiden JM. The role of Ets transcription factors in the development and function of the mammalian immune system., Adv Immunol. 1997;64:65-104. PMID:9100980.
   Role: Designed and performed all experiments, co-wrote paper. Note: Cover Photo.
2. Fox MH, **Bassuk AG**. PRICKLE1-Related Progressive Myoclonus Epilepsy with Ataxia, In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [internet]. University of Washington, Seattle. 1993-2018. 2009 Sep 8 [updated 2014 Apr 10] PMID: 20301774.
   Role: co-wrote invited review
3. Cho GY, Abdulla Y, Sengillo JD, Justus S, Schaefer KA, **Bassuk AG**, Tsang SH, Mahajan VB. CRISPR-mediated Ophthalmic Genome Surgery., Curr Ophthalmol Rep. 2017 Sep;5(3):199-206. Epub 2017 Jun 15. PMID: 28966884. PMCID: PMC5613978. doi: 10.1007/s40135-017-0144-1.
4. Cho GY, Schaefer KA, **Bassuk AG**, Tsang SH, Mahajan VB. CRISPR Genome surgery in the retina in light of off-targeting., Retina. 2018 May 7. [Epub ahead of print]. PMID: 29746416. doi: 10.1097/IAE.0000000000002197.
5. CRISPR Base Editing in Induced Pluripotent Stem Cells. Chang YJ, Xu CL, Cui X, **Bassuk AG**, Mahajan VB, Tsai YT, Tsang SH. Methods Mol Biol. 2019 Jun 28. doi: 10.1007/7651_2019_243. [Epub ahead of print] PMID: 31250381.
6. Bonkowsky JL, deVeber G, Kosofsky BE; **Child Neurology Society Research Committee**. Pediatric Neurology Research in the Twenty-First Century: Status, Challenges, and Future Directions Post-COVID-19. Pediatr Neurol. 2020 Dec;113:2-12. doi: 10.1016/j.pediatrneurol.2020.08.012. Epub 2020 Aug 26. PMID: 32979654.

7. Shaunik Sharma, Grant Tiarks, Joseph Haight and **Alexander G. Bassuk***. Neuropathophysiological Mechanisms and Treatment Strategies for Post-traumatic Epilepsy. Front. Mol. Neurosci., 23 February 2021 | ttps://doi.org/10.3389/fnmol.2021.612073.

8. Parsons DE, Lee SH, Sun YJ, Velez G, **Bassuk AG**, Smith M, Mahajan VB. Peptidomimetics Therapeutics for Retinal Disease. Biomolecules. 2021 Feb 24;11(3):339. doi: 10.3390/biom11030339. PMID: 33668179.

9. Evans Lucy P., Roghair Ariel M., Gilkes Noah J., **Bassuk Alexander G**. Visual Outcomes in Experimental Rodent Models of Blast-Mediated Traumatic Brain Injury. Front. Mol. Neurosci. April 15, 2021;14:58. doi: 10.3389/fnmol.2021.659576
Role: Co-wrote the paper

10. **Bassuk AG**. Gene therapy for Rett syndrome. Genes Brain Behav. 2021 May 30:e12754. doi: 10.1111/gbb.12754. Epub ahead of print. PMID: 34053173.
Role: Author

11. Brumback AC, Wilson RB, Augustine EF, Bass NE, **Bassuk AG**, Cejas DM, Shellhaas RA, Strober JB, Tilton AC, Pearl PL. Introducing the Child Neurology Society Leadership, Diversity, Equity, and Inclusion Task Force (LDEI). Ann Neurol. 2021 Jul 21. doi: 10.1002/ana.26176. Epub ahead of print. PMID: 34288089.
Role: Co-author

12. Parsons DE, Lee SH, Sun YJ, Velez G, **Bassuk AG**, Smith M, Mahajan VB. Peptidomimetics Therapeutics for Retinal Disease. Biomolecules. 2021 Feb 24;11(3):339. doi: 10.3390/biom11030339. PMID: 33668179 Free PMC article. Review.

13. Shellhaas RA, deVeber G, Bonkowsky JL; Child Neurology Society Research Committee. Gene-Targeted Therapies in Pediatric Neurology: Challenges and Opportunities in Diagnosis and Delivery. *Pediatr Neurol*. 2021 Sep 25;125:53-57. doi: 10.1016/j.pediatrneurol.2021.09.011. Online ahead of print. PMID: 34628144. Review.

14. Wolf J, Franco JA, Yip R, Dabaja MZ, Velez G, Liu F, **Bassuk AG**, Mruthyunjaya P, Dufour A, Mahajan VB. Liquid Biopsy Proteomics in Ophthalmology. J Proteome Res. 2024 Jan 3. doi: 10.1021/acs.jproteome.3c00756. Epub ahead of print. PMID: 38171013.

Selected Abstracts and Presentations

1. **Bassuk, A.G.**, Brenner, M., Burrowes, D.M., Joshi, A., Burton, B.K., DeLeon, G.A., Goldman, J., Larsen,M.B., Messing, M., and Stack, C. Fulminant Alexander disease with a novel mutation in glial fibrillary acidic protein gene. Presented to the 32nd annual meeting of the Child Neurology Society, Miami, Florida. 2003.

2. **Bassuk A.G.**, Burrowes, D.M., Keating, G.F., Ritacco, D.R., Bowman, R., Burton, B.K., Kessler, J. A. Variable Occipital Cephalocele with Autosomal Dominant Inheritance: A Three Generation Pedigree. Presented to the 32nd annual meeting of the Child Neurology Society, Miami, Florida. 2003.

3. **Bassuk, A.G.** and Kessler J.A. From Head to Tail, From High-tech to Low-tech: Evaluating Human Neural Tube Defects Using a Spectrum of Techniques. Presented to the 3rd annual NICHD Structural Birth Defects Conference, St. Louis, Missouri. 2004.
4. **Bassuk, A.G.**, Mohile, N., and Stack C.A. T-Cell Lymphoma Presenting with Neurological Features in Immunocompetent Children. Presented to the 57th Annual American Academy of Neurology Meeting, Miami, Florida. 2005.
5. **Bassuk, A.G.**, Jalali, A., Chary, A., Millen, K., Dobyns, W.B., and Kessler, J.A. Autosomal Dominant Dandy-Walker Malformation Maps to 2q36.1. Presented at the 34th Annual meeting of the Child Neurology Society, Los Angeles, California. 2005.
6. **Bassuk, A.G.**, Bejerano, G., Haussler, D., Chary, A., and Kessler J.A. Ultraconserved Element Mutations in Human Spina Bifida. Presented at the 4th annual NICHD Structural Birth Defects Meeting, Washington, D.C., 2005.
7. **Bassuk, A.G.**, The Genetics of Human Neural Tube Defects. Presented at the Symposium on Spina Bifida: Current Strategies, Chicago, Illinois, 2006.
8. Xhou, X., Kan, L., Jalali, A., Mcguire, T., Kessler, J.A., Zhao, L., Kazanis, V., Episkopou, V., and **Bassuk, A.G.** SOX1 Promotes Neuronal Lineage Commitment by Telencephalic Progenitor Cells. Presented to The Society for Neuroscience, Atlanta, Georgia, 2006.
9. Donsante, A., Tang, J., Goldstein, D.S., Holmes, C.S., **Bassuk, A.G.**, and Kaler, S.G. Biochemical and Molecular Characterization of Pronounced Intrafamilial Clinical Variation in Two Unrelated Families with Missense Mutations in Atp7a. Presented to the 5th International Copper Meeting: Copper and Related Metals in Biology, Sardinia, Italy, 2006.
10. Coppinger, J., **Bassuk, A.G.**, Mendelsohn, N., Ballif, B., Theisen, A., Bejjani, B., Shaffer, L. Characterization of deletion 4p16.3 by array CGH: unexpected inheritance, clinical phenotypes and counseling implications. Presented at the American College of Medical Genetics Meeting, 2007.
11. Shaffer, Lisa Bejjani, B., Torchia, B., Aylsworth, A., Curtis, M., Saitta, S., Shaikh, T., McDonald, M., Parisi, M., Tsai, Atkin, J., **Bassuk, A.G.**, Ballif, B. Identification and delineation of chromosomal deletions associated with the holoprosencephaly spectrum. Presented at the American College of Medical Genetics Meeting, 2007.
12. Wallace, R.H., **Bassuk, A.G.**, Mangelsdorf, M.E., Berkovic, S.F. PRICKLE1 MUTATIONS ARE ASSOCIATED WITH PROGRESSIVE MYOCLONIC EPILEPSY. Presented at the 32nd Human Genetics 6, 2008.
13. Kooistra, M., Fortin, P., Salem, S., De Marco, P., Merello E., **Bassuk, A.G.**, Capra, V., Gros, P., Kibar, Z. Identification of VANGL1 Variants Associated with Neural Tube Defects. Presented at the Advanced perspective in Neural Tube Defects 2nd Meeting September 27-28, 2008 Villa Quartara, Badia della Castagna, Genoa.
14. **Bassuk, A.G.**, Wallace, R.H., Buhr, A., Buller, A.R., Afawi, Z., Shimojo, M., Miyata, S., Chen, S., Gonzalez-Alegre, P., Griesbach, H.L., Wu, S., Nashelsky, M., Vladar, E.K., Antic, D., Ferguson, P.J., Cirak, S., Voit, T., Scott, M.P.,

Axelrod, J.D., Gurnett, C., Daoud, A.S., Kivity, S., Neufeld, M.Y., Mazarib, A., Straussberg, R., Walid, S., Korczyn, A.D., Slusarski, D.C., Berkovic, S.F., El-Shanti, H.I. Identification of the Gene for Myoclonic Epilepsy-Ataxia Syndrome. Presented at the 37th Annual Meeting of the Child Neurology Society, October, 2008.

15. **Bassuk, A.G.**, Mank, J.R. International Spina Bifida Genomics Consortium to detect copy number variants. Presented at the 6th International NTD Conference, Burlington, VT, September 12-15, 2009.
16. Manak, J.R., Sowers, L., Tao, H., Ueno, N., Axelrod, J.D., **Bassuk, A.G.** Mutations in Prickle homologues cause seizures in flies, mice and humans. Presented at the 2010 Drosophila Research Conference, Washington DC, April 7-11, 2010.
17. Alasti, F., Brophy, P., Darbro, B., Dierdorff, J., Nishimura, C., Cobb, B., Clarke, J., Hakeman, M., **Bassuk, A.G.**, Smith, R.J.H., Manak, J.R. Genome-wide copy number variation analysis of a branchio-oto-renal syndrome cohort identifies a recombination hotspot associated with an EYA1 deletion and novel candidate genomic regions. Presented at the American Society for Human Genetics Conference, San Francisco, November 6-10, 2012.
18. Allache, R., Capra, V., Wang, M.Q., Bosoi, C.M., Drapeau, P., **Bassuk, A.G.**, Kibar, Z. Role of the PRICKLE genes in neural tube defects in humans. Presented at the American Society for Human Genetics Conference, San Francisco, November 6-10, 2012.
19. Mahajan, V.B., Skeie, J.M., **Bassuk, A.G.**, Fingert, J.H., Braun, T.A., Daggett, H.T., Folk, J. C., Sheffield, V.C., Stone, E.M. Calpain-5 causes autoimmune uveitis, retinal neovascularization and photoreceptor degeneration. Presented at the American Society for Human Genetics Conference, San Francisco, November 6-10, 2012.
20. Paemka, L., Mahajan, V.B., Skeie, J.M., Sowers, L.P., Ehaideb, S.N., Gonzalez-Alegre, P., Sasaoka, T., Tao, H., Miyagi, A., Ueno, N., Wu, S., Darbro, B.W., Ferguson, P.J., Pieper, A.A., Britt, J.K., Wemmie, J.A., Rudd, D.S., Wassink, T., El-Shanti, H., Mefford, H.C., Carvill, G.L., Manak, J.R., **Bassuk, A.G.** PRICKLE1 interaction with SYNAPSIN I reveals a role in Autism Spectrum Disorder. Presented at the American Society for Human Genetics Conference, October 22-26, 2013.
21. Darbro, B.W., Mahajan, V.B., Gakhar, L., Skeie, J.M., Campbell, E., Wu, S., Bing, X., Millen, K.J., Dobyns, W.B., Kessler, J.A., Jalali, A., Cremer, J., Segre, A., Manak, J.R., Aldinger, K.A., Suzuki, S., Natsume, N., Ono, M., Hai, H.D., Viet le, T., Loddo, S., Valente, E.M., Bernardini, L., Ghonge, N., Ferguson, P.J., **Bassuk, A.G.** Mutations in extracellular matrix genes NID1 and LAMC1 cause autosomal dominant Dandy-Walker malformation and occipital cephaloceles. Presented at the American Society for Human Genetics Conference, October 22-26, 2013.
22. Mei, X., Wu, S., **Bassuk, A.G.**, Slusarski, D. C. Mechanisms of prickle in zebrafish epilepsy and retinal neurogenesis. Presented at the Society for Neuroscience Conference, November 9, 2013.

23. Sowers, L.P., Loo, L., Wu, Y., Wassikn, T., Ferguson, P.J., Wemmie, J.A., Richerson, G., Mohapatra, D.P., **Bassuk, A.G.** Hippocampal synaptic abnormalities and autism like behaviors associated with disruption of the non-canonical Wnt gene Prickle2. Presented at the Society for Neuroscience Conference, November 13, 2013.

24. Kousa, Y.A., Zhu, H., Fakhouri, W., Lei, Y., Kinoshita, A., Roushangar, R.R., Leslie, E.J., Busch, T.D., Williams, T.J., Chai, Y., Amendt, B.A., Murray, J.C., Shaw, G.M., **Bassuk, A.G.**, Ashley-Koch, A., Gregory, S., Finnell, R.H., Schutte, B. C. A conserved role for IRF6 in neurulation. Presented at the American Society for Human Genetics Conference, October 18-22, 2014.

25. Sturgeon, M., Malicdan, M.C., **Bassuk, A.G.**, Cornell, R., Validating putative epilepsy-causing genes in zebrafish; Abstract #35. Presented at the 2015 Midwest Zebrafish Meeting, June 11, 2015, St. Louis, Missouri.

26. Cox, A.J., Darbro, B.W., Finer, A.L., Bing, X., **Bassuk, A.G.**, Ferguson, P.J. Recessive coding and regulatory mutations in FBLIM1 cause chronic recurrent multifocal osteomyelitis (CRMO).

    a. Presented at American Society of Human Genetics (ASHG) meeting, October 2016, Vancouver, BC.

    b. Presented at the University of Iowa Health Sciences Research Week, April 2016, Awarded poster prize by the Institute for Clinical and Translational Science

27. Nessler, A.J., Han, S., Kim, Y., Wu, S., Sowers, L.P., Antic, D., Axelrod, J.D., Narayanan, N.S., **Bassuk, A.G.**, Parker, K.L. (November 2016). Timing impairments and cerebellar abnormalities in Prickle2 mice: Implications for autism. Presented at the at the Society for Neuroscience, November 2016, San Diego, CA.

28. Cox, A.J., Darbro, B.W., Bing, X., **Bassuk, A.G.**, Ferguson, P.J. Whole exome analysis of individuals and families with chronic recurrent multifocal osteomyelitis (CRMO).

    a. Presented at the University of Iowa Genetics Retreat, October 2013, October 2014, November 2015. Awarded 2nd place for poster presentations, November 2015.

    b. Presented at the World Congress on Inflammation, Boston, MA, August 2015.

    c. Presented at the University of Iowa Pediatrics Research Day, April 2015.

    d. Presented at the Great Lakes Bioinformatics Conference, Cincinnati, OH, May 2014.

    e. Presented at the University of Iowa Health Sciences Research Week, April 2014

29. Fyn-tau interaction in experimental models and human epilepsy | M. Putra; G. Lee; M. Hefti; **A. Bassuk**; T. Thippeswamy, American Epilepsy Society 2020 Meeting, December 5, 2020.

30. Franco J, Toral M Young JS, Schaefer K, Smits M, Wert K, Coglan D, Wu S, Majajan M, **Bassuk AG**, Mahajan VB. Iroquois-3 is a novel target of CAPN5

proteolytic processing and contributes to inflammatory eye disease OR CAPN5 regulates inflammatory cytokines through proteolytic processing of a transcription factor Iroquois-3 in inflammatory eye disease. Presented at the 2024 annual meeting of the Association for Research in Vision and Ophthalmology, May 5-9, 2024 in Seattle, WA.

31. Wolf J, Rasmussen DK, Sun YJ, Mruthyunjaya P, Dufour A, **Bassuk AG**, Mahajan VB. High-resolution proteomic profiling of aqueous humor liquid biopsies from patients with uveitis. Presented at the 2024 annual meeting of the Association for Research in Vision and Ophthalmology, May 5-9, 2024 in Seattle, WA.

Ph.D. Thesis

1. **Bassuk AG**. Protein-protein interactions and development expression of Ets transcription factors: ELF-1 as a model protein, The University of Chicago. 1996.

Intellectual Property (e.g. Patents, Copyrights)

1. Mahajan, V. B. 16340657, "Methods and Composition for Treating Genetic Eye Diseases."

B. Areas of Research Interest

- Genetics and mechanisms underlying human epilepsy and autism.
- Molecular mechanism of bone inflammation.
- Molecular mechanisms of retinal disease.
- Molecular mechanisms of traumatic brain injury.
- Neurodevelopmental genetics.

C. Grants Received

Active

Title: “1R01EY031952-01” - Proteomic Biomarkers of Intraocular Infection
Source of Grant: NIH
Principal Investigator: Alexander Bassuk, Polly J Ferguson, Vinit B Mahajan
Amount: $410,002
Period of Funding: 9/30/2020 -7/31/2024
Role: PI: 10% effort.
Goals: To identify factors involved in retinal infection.

Title: “1R01EY030151-01A1” - Inflammatory gene transcription in the retina
Source of Grant: NIH
Principal Investigator: Alexander Bassuk, Vinit B Mahajan
Amount: $407,791
Period of Funding: 1/1/2020 -12/31/2024
Role: PI: 10% effort.
Goals: To identify transcription factors involved in retinal pathology.

Title: “5KL2TR002536-02” - The University of Iowa Clinical and Translational Science Award
Source of Grant: NIH
Principal Investigator: Alexander Bassuk
Amount: $675,002
Period of Funding: 4/13/2018 -2/23/2023
Role: PI: 20% effort.
Goals: To train clinical scientists.

Title: “K12 HD27748-11”
Source of Grant: NIH/NICHD - Child Health Career Development Award
Principal Investigator: Alexander G. Bassuk (as of 9/1/21)
Amount: $300,000/year
Period of Funding: 7/1/2011 -11 /30/2022
Role: Recruiter (since 7/11): 6% effort.
Goals: The goal is to train young investigators in molecular genetics and molecular biology or use in pediatric research.

Title: Novel circuits and mechanisms of descending pain modular
Source of Grant/Sponsor: NIH - R01 NS-127428
PI(s): Yurily Usachev, Contact PI; Alex Bassuk, MPI
Amount: $2,752,407
Period of funding: 12/15/22-11/30/27
Dr. Bassuk’s role: MPI
Goals of the grant: To explore circuits of pain

Inactive

Title: Genetic and Immunologic etiology of chronic recurrent multifocal osteomyelitis (CRMO)
Source of Grant: 2R01AR059703-06A1
Principal Investigator: MPI, Bassuk, Ferguson
Amount:
Period of Funding: 9/28/2015-8/31/2020
Role: MPI: 10% effort

Title: Mechanistic Studies on Regenerative Medicine Approaches to Childhood Blindness
Source of Grant: 5R01EY026682
Principal Investigator: MPI, Bassuk, Mahajan, Tsang
Amount:
Period of Funding: 4/12016-3/31/2020
Role: MPI, Contact PI: 10% effort

Title: Structure and Function of Caplain-5.
Source of Grant: 1R01EY024665
Principal Investigator: Mahajan
Amount:
Period of Funding: 12/1/14-11/30/19
Role: Co-Investigator: 10% effort

Title: Mechanism-based therapies for photoreceptor degeneration.
Source of Grant: 1R01EY025225-01
Principal Investigator: Mahajan
Amount:
Period of Funding: 5/1/2015-4/30/2019
Role: Co-investigator: 10% effort

Title: Evaluating GWAS AMD Candidate Loci by Gene Editing in Human iPS Cells
Source of Grant: 1R21AG050437-01
Principal Investigator: MPI, Bassuk, Mahajan, Tsang
Amount:
Period of Funding: 4/15/2015-3/31/2017
Role: MPI: 10% effort

Title: A new dietary therapy for refractory epilepsy: its potential efficacy and underlying mechanisms
Source of Grant: University of Iowa ICTS IX
Principal Investigator: MPI, Bassuk, Kitamoto, Wu, Richerson
Amount:
Period of Funding: 2016-2016

Title: The Role of IL-1 in Traumatic Brain Injury Pathogenesis
Source of Grant: CCOM Carver Medical Research Initiative
Principal Investigator: Bassuk
Amount: $30,000
Period of Funding: 2/15-2/17
Role: PI

Title: The Prickle1 gene as a genetic factor for autism-like behaviors and epilepsy
Source of Grant: 1R21MH107782-01, GRANT11759824
Principal Investigator: Bassuk
Amount:
Period of Funding: October 27, 2014
Role: PI

Title: Evaluating GWAS AMD Candidate Loci by Gene Editing in Human iPS Cells
Source of Grant: 1R21AG050437-01, GRANT11728294
Principal Investigator: Bassuk
Amount:
Period of Funding: November 3, 2014
Role: PI
Goals:

Title: Gene Silencing and Gene Editing in Phototransduction
Source of Grant: 1R01EY024698-01A1, GRANT11802501
Principal Investigator: Bassuk
Amount:
Period of Funding: December 10, 2014
Role: PI

Title: Using Genetic Screens in Model Systems to Identify New Anti-Seizure Targets
Source of Grant: AN:3774979, GRANT11806527
Principal Investigator: Bassuk
Amount:
Period of Funding: December 18, 2014
Role: PI
Goals:

Title: *"The Genetics of Human Neural Tube Defects"*
Source of Grant: Northwestern Memorial Faculty Foundation Young Investigator Award
Principal Investigator:
Amount: $25,000
Period of Funding: 2005

Title: Genetics of Epilepsy
Source of Grant: Care for Kids Foundation
Principal Investigator:
Amount: $35,000
Period of Funding: March 2008

Title: Role of Herp3 in BMP Mediated Neurogenesis
Source of Grant: NINDS Mentored Clinical Scientist Development Award -- KO8 NS48174-01A1
Principal Investigator:
Amount: $157,100
Period of Funding: 2004 - 2009

Title: A novel microarray based technique for mapping human disease causing mutations
Source of Grant: Carver Trust Collaborative Pilot Grant
Principal Investigator: Bassuk, Alex (Co-Principal), Ferguson, Polly (Co-Principal), Manak, John (Co-Principal)
Amount: $50,000
Period of Funding: February 1, 2009 - January 31, 2010

Title: Exome Sequencing in Consagnuious Epilepsy Pedigrees
Source of Grant: Iowa Institute of Human Genetics
Principal Investigator: Bassuk
Amount: $50,000
Period of Funding: January 1, 2013 - December 31, 2013
Role: PI

Title: Evaluating PRICKLE mutations in human epilepsy and animal models
Source of Grant: NIH 1R01 NS064159-01A1
Principal Investigator: Bassuk
Amount: $1,861,848
Period of Funding: 2009 - 2014
Role: PI

Title: Genetic and Immunological Etiology of Chronic Recurrent Multifocal Osteomyelitis
Source of Grant: NIH 1R01AR059703-01A1
Principal Investigator: Ferguson, Polly (Principal Investigator)
Period of Funding: 2010 - 2015
Role: Bassuk, A (Co-Principal): 5% effort

Title: WNT modulation as a treatment for autism spectrum disorders
Source of Grant: NIH 1R21MH100086-01, NIMH R21
Principal Investigator: Bassuk
Amount: $553,704
Period of Funding: 2013 - 2015
Role: PI

Title: Modeling human epilepsy in zebrafish
Source of Grant: 1R21GM110302-01
Principal Investigator: Bassuk, A (Multi-PI), Slusarski (Multi-PI)
Amount: $250,000
Period of Funding: September 2014 - August 2016
Role: Multi-PI

Title: Gene Identification in Autosomal Recessive Familial Epilepsy
Source of Grant: Qatar National Research Fund
Principal Investigator: El-Shanti, Hatem
Amount: $3,000,000
Period of Funding: February 2, 2014 - December 2016
Role: Co-I: 15% effort

Title: Child Health Career Development Award
Source of Grant: K12 HD27748-11
Principal Investigator: Hirsch, Raphael
Amount: $1,800,000
Period of Funding: November 2012 - July 2017
Role: 20% effort

Title: Structure and Function of Calpain-5
Source of Grant: 1R01EY024665
Principal Investigator: Mahajan
Amount:
Period of Funding: December 2014 - November 2019
Role: Co-I: 10% effort

Title: NINDS R01 supplement Collaborative Activities to Promote Translational Research (CAPTR) NOT-NS-10-008: Translating PRICKLE-epilepsy discoveries to therapies using a fruit fly model
Source of Grant: 3R01NS064159-02S1
Principal Investigator: Bassuk
Amount: $75,000
Period of Funding:

Role: PI

Title: NIH R01 Exploring Novel Epilepsy Pathways
Source of Grant: 5R01NS098590-05
Principal Investigator: Bassuk
Amount:
Period of Funding: 2015-2022
Role: PI: 20% effort

D. Presentations

2003

- 32nd Annual Meeting of the Child Neurology Society. *Variable Occipital Cephalocele with Autosomal Dominant Inheritance: A Three Generation Pedigree.* Bassuk, AG, Burrowes, DM, Keating, GF, Ritacco, DR, Bowman, R, Burton, BK, Kessler, JA.

2005

- 57th Annual American Academy of Neurology Meeting. *T-Cell Lymphoma Presenting with Neurological Features in Immunocompetent Children.* Bassuk, AG, Mohile, N, Stack, CA.

2006

- 5th International Copper Meeting: Copper and Related Metals in Biology. *Biochemical and Molecular Characterization of Pronounced Intrafamilial Clinical Variation in Two Unrelated Families with Missense Mutations in Atp7a.* Dosante, A, Tang, J, Goldstein, DS, Holmes, CS, Bassuk, AG, Kaler, SG.
- The Society for Neuroscience. *SOX1 Promotes Neuronal Lineage Commitment by Telencephalic Progenitor Cells.* Xhou, X, Kan, L, Jalali, A, Mcguire, T, Kessler, JA, Zhao, L, Kazanis, V, Episkopou, V, Bassuk, AG.
- Symposium on Spina Bifida: Current Strategies. *The Genetics of Human Neural Tube Defects.* Bassuk, AG.

2008

- American Academy of Neurology Scientific Program. *"Introduction to Types of Genetic Testing".* Bassuk, A. (April 18, 2008).
- 37th Annual Meeting of the Child Neurology Society. *IDENTIFICATION OF THE GENE FOR MYOCLONIC EPILEPSY-ATAXIA SYNDROME.* Bassuk, AG, Wallace, RH, Buhr, A, Buller, AR, Afawi, Z, Shimojo, M, Miyata, S, Chen, S, Gonzalez-Alegre, P, Griesbach, HL, Wu, S, Nashelsky, M, Vladar, EK, Antic, D, Ferguson, PJ, Cirak, S, Voit, T, Scott, MP, Axelrod, JD, Gurnett, C, Daoud, AS, Kivity, S, Neufeld, MY, Mazarib, A, Straussberg, R, Walid, S, Korczyn, AD, Slusarski, DC, Berkovic, SF, El-Shanti, HI. (October 2008)
- 32nd Human Genetics 6. *Prickle 1 Mutations are Associated with Progressive Myoclonic Epilepsy.* Wallace, RH, Bassuk, A, Mangelsdorf, ME, Berkovic, SF.

2009

- 6th International NTD Conference. *International Spina Bifida Genomics Consortium to detect copy number variants.* Bassuk, AG, Mank, JR. (September 15, 2009)

2010

- August 30 National Institute of Neurological Disorder and Stroke Genetics of Epilepsy Workshop. Bassuk, A. (August 30, 2010).

2012

- November 10 American Society for Human Genetics Conference. *Calpain-5 causes autoimmune uveitis, retinal neovascularization and photoreceptor degeneration*. (November 10, 2012).

2014

- ASHG 2014 Meeting. *Moderator, "Genomics of Autism".* Bassuk, A.

Conference Presentation

2003

- 32nd annual meeting of the Child Neurology Society. *Fulminant Alexander disease with a novel mutation in glial fibrillary acidic protein gene.* Bassuk, AG, Brenner, M, Burrowes, DM, Joshi, A, Burton, BK, DeLeon, GA, Goldman, J, Larsen, MB, Messing, M, Stack, C.

2004

- 3rd Annual NICHD Structural Birth Defects Conference. *From Head to Tail, From High-tech to Low-tech: Evaluating Human Neural Tube Defects Using a Spectrum of Techniques.* Bassuk, AG, Kessler, JA.

2005

- 34th Annual Meeting of the Child Neurology Society. *Autosomal Dominant Dandy-Walker Malformation Maps to 2q36.1.* Bassuk, AG, Jalali, A, Chary, A, Millen, K, Dobyns, WB, Kessler, JA.
- 4th Annual NICHD Structural Birth Defects Meeting. *Ultraconserved Element Mutations in Human Spina Bifida.* Bassuk, AG, Bejerano, G, Haussler, D, Chary, A, Kessler, JA.

2007

- American College of Medical Genetics Meeting. *Characterization of deletion 4p16.3 by array CGH: unexpected inheritance, clinical phenotypes and counseling implications.* Coppinger, J, Bassuk, A, Mendelsohn, N, Ballif, B, Theisen, A, Bejjani, B, Shaffer, L.
- American College of Medical Genetics Meeting. *Identification and delineation of chromosomal deletions associated with the holoprosencephaly spectrum.* Shaffer, L, Bejjani, B, Torchia, B, Aylsworth, A, Curtis, M, Saitta, S, Shaikh, T, McDonald, M, Parisi, M, Tsai, M, Atkin, J, Bassuk, A, Ballif, B.

2008

- Advanced Perspective in Neural Tube Defects 2nd Meeting. *Identification of Vangl1 Variants Associated with Neural Tube Defects.* Bassuk, A, Capra, V, Gros, P, Kibar, Z. (September 28, 2008).

2009

- 6th International NTD Conference. *"International Spina Bifida Genomics Consortium to detect copy number variants".* Bassuk, A. (September 12, 2009).

2010

- 2010 Drosophila Research Conference. *Mutations in Prickle homologues cause seizures in flies, mice and humans.* Manak, JR, Sowers, L, Tao, H, Ueno, N, Axelrod, JD, Bassuk, AG. (April 11, 2010).
- Invited to the National Institute of Neurological Disorder and Stroke Genetics of Epilepsy Workshop, August 30-September 1, 2010, San Diego, CA.

2012

- American Society for Human Genetics Conference. *Genome-wide copy number variation analysis of a branchio-oto-renal syndrome cohort identifies a recombination hotspot associated with an EYA1 deletion and novel candidate genomic regions.* Alasti, F, Brophy, P, Darbro, B, Dierdorff, J, Nishimura, C, Cobb, B, Clarke, J, Hakeman, M, Bassuk, A, Smith, R, Manak, JR. (November 10, 2012).
- American Society for Human Genetics Conference. *Role of the PRICKLE genes in neural tube defects in humans.* Allache, R, Capra, V, Wang, MQ, Bosoi, CM, Drapeau, P, Bassuk, AG, Kibar, Z. (November 10, 2012).

2013

- American Society for Human Genetics Conference. *Mutations in extracellular matrix genes NID1 and LAMC1 cause autosomal dominant Dandy-Walker malformation and occipital cephaloceles.* Mahajan, Darbro, B, Gakhar, L, Skeie, J, Campbell, E, Wu, S, Bing, X, Millen, K, Dobyns, W, Kessler, J, Jalali, A, Cremer, J, Segre, A, Manak, J, Aldinger, K, Suzuki, S, Natsumi, N, Ono, M, Dai Hai, H, Thi Viet, L, Loddo, S, Valente, E, Bernardini, L, Ghonge, N, Ferguson, P, Bassuk, A. (October 26, 2013).
- American Society for Human Genetics Conference. *PRICKLE1 interaction with SYNAPSIN I reveals a role in Autism Spectrum Disorder.* Paemka, Mahajan, VB, Skeie, JM, Sowers, LP, Ehaideb, SN, Gonzalez-Alegre, P, Sasaoka, T, Tao, H, Miyagi, A, Ueno, N, Wu, S, Darbro, BW, Ferguson, PJ, Pieper, AA, Britt, JK, Wemmie, JA, Rudd, DS, Wassink, T, El-Shanti, H, Mefford, HC, Carvill, GL, Manak, JR, Bassuk, AG. (October 26, 2013).
- Society for Neuroscience Conference. *Mechanisms of prickle in zebrafish epilepsy and retinal neurogenesis.* Mei, X, Wu, S, Bassuk, AG, Slusarski, DC. (November 9, 2013).

- Society for Neuroscience Conference. *Hippocampal synaptic abnormalities and autism like behaviors associated with disruption of the non-canonical Wnt gene Prickle2.* Sowers, LP, Loo, L, Wu, Y, Wassink, T, Ferguson, PJ, Wemmie, JA, Richerson, G, Mohapatra, DP, Bassuk, AG. (November 13, 2013).

Invited Lecture

2005

- The University of Illinois Urbana-Champagne Institute for Genomic Biology. *"The Genetics of Human Neural Tube Defects".* Bassuk, A. (October 14, 2005).

2007

- Grand Rounds, University of Wisconsin, Neurology. *"Clinical Neurogenetics and Human Neural Tube Malformations".* Bassuk, A. (January 27, 2007).

2008

- American Academy of Neurology Scientific Program. "*Introduction to Types of Genetic Testing"*. Bassuk, A. (April 18, 2008
- Presented to Dr Matt Scott and Jeff Axelrod laboratories, Stanford University. *"Prickle genes and Epilepsy".* Bassuk, A. (May 27, 2008).
- IMNA: Management of Adult and Pediatric Epilepsy. *"Forever in Blue Genes? Progress and Promise of Genetics in Epilepsy".* Bassuk, A. (September 12, 2008).

2009

- American Academy of Neurology Scientific Program. *"Genetics Testing: The Future is Now".* Bassuk, A. (April 2009).
- 6th International NTD Conference. "*International Spina Bifida Genomics Consortium to detect copy number variants*". Bassuk, A. (September 12, 2009).

2010

- Grand Rounds, University of California San Diego, Neurology. Bassuk, A. (January 28, 2010).
- University of Chicago Pediatrics. *"Flies, and zebrafish, and mice, and epilepsy oh my".* Bassuk, A. (May 28, 2010).
- NIH Workshop. *"Coming Together on Epilepsy Genetics".* Bassuk, A. Jackson Laboratory, Bar Harbour, ME. (October 9, 2010).

2014

- ASHG 2014 Meeting. "*Genomics of Autism*", Moderator. San Diego. CA.

2015

- CHLA/USC Grand Rounds. "*Adventures of Neurogenetics*". Los Angeles, CA. (September 12, 2015)
- Scientific and Clinical Foundation for Precision Medicine in Epilepsy. "*Precision Epilepsy in Multiple Species*". Banbury Center, Cold Spring

Harbor Laboratory. The Banbury Center is the small conference center at Cold Spring Harbor holding meetings for groups of between 20 and 30 scientists. Participation is by invitation only. More information: http://www.cshl.edu/bandury. (November 1-4, 2015).

2016

- Pediatric Grand Rounds, Rady Children's Hospital, "Adventures in Neurogenetics" UCSD. (February, 2016).
- University of Arizona MARC (Minority Access to Research Careers) Program. "*My MSTP Adventure*". Guest Mentor & Speaker. (November 7, 2016)
- Neurosurgery Grand Rounds, University of Iowa. "*Adventures in Neurogenetics*". (November 11, 2016).

2017

- Weizmann Institute of Science Molecular Neuroscience Forum (MNF), "Exploring epilepsy mechanisms across evolution: Seizing drosophila, zebras, mice, and humans". (May 16, 2017).

2018

- Rochester University, Department of Pediatrics "Adventures in Neurogenetics". (June 5, 2020).

2020

- Kaiser Permanente Los Angeles Neurology Grand Rounds. November 10, 2020

2021

- Columbia University, Pediatric Epilepsy. February 26, 2021.

2023

- Child Neurologist Career Development Program (CNCDP) K12, Annual Meeting. "You have to be of use (if you can be of use)". October 1-3, 2023.

IV. SERVICE

A. Memberships in Professional Organizations

American Academy of Neurology

American Academy of Pediatrics

American Epilepsy Society

American Neurological Association

American Pediatric Society

American Society of Human Genetics

Association of Medical School Pediatric Department Chairs (AMSPDC)

Child Neurology Society

Society for Neuroscience

Society for Pediatric Research (SPR), elected member

B. Professional Service

2009 - 2010	Residency Selection Committee, Pediatrics
2009 – Present	Physician Scientist Training Program Interviewer
2010 – Present	MSTP Admission Committee
2011 - 2015	Permanent Study Section Member, Genetics of Human (GHD) July 2001-June 2015
2011 – 2018	Pediatric Research Day Poster Judge
2012	Medical Student Research Day Poster Judge
2014	University of Iowa SUMR program Mentor (Summer)
2014	MCB representative for MSTP interview weekend (Winter)
2014	University of Iowa CCOM, 5-year academic review panel – Urology
2014 - 2015	University of Iowa Pediatric Intensive Care Unit Director Search Committee Head
2014 – 2016	University of Iowa Neuroscience Director Search Committee Member
2014 – 2016	University of Iowa Pediatric Infectious Diseases Director Search Committee Member
2014 – Present	University of Iowa CCOM Bridge Funding Review Committee
2014 – 2017	University of Iowa eHealth and eNovation Center (under Vice-chair Dr. Patrick Brophy), leading research efforts
2016 – 2017	Pediatric GI Division Director Search Committee
2016 – 2021	Pediatric Pulmonary Division Director Search Committee
2018 – 2019	Pediatric Nephrology Division Director Search Committee
2018 – 2019	Pediatric Genetics Division Director Search Committee
2018 – 2020	Psychiatry DEO Search Committee
2018 – 2020	Admissions Committee Director, MSTP
2018	Ad Hoc CNCDP K12 National Advisory Council
2019 – Present	CNCDP K12 National Advisory Council – Member
2019 – Present	Child Neurology Society (CNS) Scientific Program Committee
2019 – Present	University of Iowa KL2 Director (as of July 1, 2019)
2020 – Present	Iowa Hawk-IDDRC Executive Committee
2021 – Present	Clinical Systems Committee, University of Iowa Hospitals and Clinics
2021 – Present	Medical Council, University of Iowa Carver College of Medicine
2021 – Present	Enterprise Committee, University of Iowa Carver College of Medicine
2021 – Present	Enterprise Committee, Finance Subcommittee, University of Iowa Carver College of Medicine
2021 – Present	Institute for Clinical and Translational Science Executive Committee
2022 – Present	Association of Medical School Pediatric Department Chairs (AMSPDC), Pediatric Scientist Development Program (PSDP), Selection Subcommittee. Three-year term July 2022 to June 2025.

2022 – Present	Association of Medical School Pediatric Department Chairs (AMSPDC), Research Committee. Two-year term, 2022-2024.
2023 – Present	University of Iowa Carver College of Medicine Vice President for Medical Affairs Search Committee Member
2023	Panel Member, "Starting Your Faculty Position/Research Group", Association of Medical School Pediatric Department Chairs (AMSPDC) Annual Meeting 2023, Clearwater, FL, March 2, 2023.
2023	University of Iowa Carver College of Medicine Internal Medicine DEO Search Co-Chair
2023	Departmental Consulting Group, Department of Dermatology, on the consideration of Dr. Ali Jabbari for promotion to Professor of Dermatology.
2024	Testimony before the Committee on Energy and Commerce, Subcommittee on Health, US House of Representatives on "Legislative Proposals to Support Patients with Rare Diseases". Invited by the Honorable Representative Mariannette Miller-Meeks of Iowa, 2/29/2024.

Ad hoc reviewer

2009	NIH Study Section, Genetics of Human Disease (GHD) (October 8-9)
2013	NIH Special Emphasis Panel (SEP), Epilepsy Genetics Review Panel (April 1)
2013	English Medical Research Council (MRC)
2015	NIH "Developing Disease Modifying or Prevention Therapies for Epilepsies Center Without Walls (CWOW)", Epilepsy Genetics Review Panel (March 3)
2016	NIH "Developing Disease Modifying or Prevention Therapies for Epilepsies Center Without Walls (CWOW)", Epilepsy Genetics Review Panel, mail-on reviewer (June)
2017	NIH DBD (Developmental Brain Disorders) Study Section, phone reviewer (February 10)
2017	NIH DBD (Developmental Brain Disorders) Study Section (June 1-2)
2018	NIH Gabriella Miller Kids First Study Section, phone reviewer (May 10)
2019	NIH Rare Disease Clinical Network (RDCRN) Review Meeting Feb 11-15, 2019 (Phone reviewer)
2019	Peer Review Medical Research Program (PRMRP) for the Department of Defense Congressionally Directed Medical Research Programs (CDMRP)
2020	NIH SBIR ETTN-A(15) Study Section (July 21-22, 2020)
2021	NINDS P01 review panel ZNS1 SRB K(28), 3/16/21-3/19/21 Mail reviewer

Manuscript Reviewer (Number within the last 5 years)

American Journal of Medical Genetics (3)
Birth Defects Research Part A (12)
Pediatric Neurology (2)

Journal of Neurogenetics (5)
Neurology (3)
Translational Neurology (2)
PLOS Genetics (6)
Molecular Psychiatry (2)
PLoS One (6)
Human Molecular Genetics (5)
Annals of Clinical and Translational Neurology (6)
American Journal of Human Genetics (2)
Seizure (1)
JCI (1)
JCI Insights (2)

C. Clinical assignments since last promotion (if applicable)

Inpatient

Inpatient service, consults and night call for Pediatric Neurology,

- 2010-2013 -- 14-15 weeks per year,
- 2013-2014 -- 4 weeks
- 2015 – 2 weeks
- 2016 – 1.5 weeks
- 2017 – 2 weeks
- 2018-present 2 weeks

Outpatient

Outpatient-Pediatric Neurology Outpatient Clinic

- 2007-2014 -- Approximately ½ day per week
- 2015-2016 – Varies with division needs
- 2017 – ½ day per week
- 2018-Present Varies with division needs