CURRICULUM VITAE

ERIN L. HEINZEN, PHARM.D., PH.D.

1. PERSONAL INFORMATION

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2. EDUCATION

September 2005 – March 2009	Postdoctoral Fellow, Human Disease Genetics, Duke University, Durham, NC, Institute for Genome Sciences & Policy, Mentor: David B. Goldstein, Ph.D.
April 2004 – August 2005	Postdoctoral Fellow, Neuropharmacology, University of North Carolina, Chapel Hill, NC, School of Medicine, Curriculum in Toxicology, Mentor: Richard B. Mailman, Ph.D.
September 2001 – May 2004	Ph.D., Pharmaceutical Sciences, University of North Carolina, Chapel Hill, NC Thesis: *The role of nitric oxide as a mediator of morphine antinociceptive tolerance* Mentor: Gary M. Pollack, Ph.D.
August 1997 – August 2001	Pharm.D., University of North Carolina, Chapel Hill, NC
August 1995 – May 1997	Pre-pharmacy curriculum, University of North Carolina, Chapel Hill, NC

3. PROFESSIONAL EXPERIENCE – EMPLOYMENT HISTORY

POSITIONS

January 2020 – present	Associate Professor, Division of Pharmacotherapy and Experimental Therapeutics in the Eshelman School of Pharmacy and Department of Genetics in the School of Medicine, University of North Carolina at Chapel Hill, Chapel Hill, NC
May 2020 – present	Faculty, University of North Carolina Curriculum in Bioinformatics and Computational Biology in the School of Medicine, University of North Carolina at Chapel Hill, Chapel Hill, NC
May 2020 – present	Faculty, University of North Carolina Curriculum in Genetics and Molecular Biology in the School of Medicine, University of North Carolina at Chapel Hill, Chapel Hill, NC
March 2020 – present	Faculty, University of North Carolina Neuroscience Curriculum in the School of Medicine, University of North Carolina at Chapel Hill, Chapel Hill, NC
July 2019 – December 2019	Herbert Irving Assistant Professor of Pathology and Cell Biology, Institute for Genomic Medicine, Columbia University Medical Center, New York, NY
January 2015 – May 2019	Assistant Professor, Department of Pathology and Cell Biology, Institute for Genomic Medicine, Columbia University Medical Center, New York, NY
January 2015 – October 2018	Deputy Director of the Institute for Genomic Medicine, Columbia University Medical Center, New York, NY
June 2012 – December 2014	Faculty Director, Genome Analysis Facility, Center for Human Genome Variation, Duke University, Durham, NC
April 2009 – December 2014	Assistant Professor, Department of Medicine, Center for Human Genome Variation, Duke University Medical Center, Durham, NC

LICENSURE/CERTIFICATIONS

May 2015 – December 2020	Pharmacist licensure New York #060405
March 2015 – present	American Pharmaceutical Association (APhA) Pharmacy-Based Immunizer
February 2002 – present	Pharmacist licensure North Carolina #16251

4. HONORS

2019	Irving Institute for Clinical and Translational Research Scholar
2005	American Society for Pharmacology and Experimental Therapeutics Young Scientist Travel Award
2005	American Association of Pharmaceutical Scientists Postdoctoral Travel Award

2004 Division of Drug Delivery and Disposition Graduate Scholar Award
2003 American Association of Pharmaceutical Sciences Outstanding Graduate Student Research Award in Pharmacokinetics, Pharmacodynamics, and Drug Metabolism
2002 – 2004 American Foundation of Pharmaceutical Education Pre-doctoral Fellowship
2000 Miya Endowment Fund Academic Scholarship
1999 George T. Cornwell Academic Scholarship

5. BIBLIOGRAPHY AND PRODUCTS OF SCHOLARSHIP

PUBLISHED BOOKS AND BOOK CHAPTERS

1. Hunter SE, Jalazo E, Felton TR, **Heinzen EL**, Shiloh-Malawsky Y. *Epilepsy Genetics: Advancements in the Field and Impact on Clinical Practice.* Exon Publications; 2022. Chapter 3.
2. Poduri AH, George AL, **Heinzen EL**, Lowenstein DH. *How We Got to Where We're Going.* Elements in Genetics in Epilepsy. Cambridge University Press; 2021.
3. Ruzzo EK, Radtke RA, Goldstein DB, and **Heinzen EL**. *Practical Epilepsy: Genetics*. Demos Medical Publishing, New York City; 2016; 11-27.

PUBLISHED PEER-REVIEWED PUBLICATIONS

Original Research with Named Authorship

‡ indicates shared first or last author

1. Perucca P, Stanley K, Harris N, McIntosh AM, Asadi-Pooya AA, Mikati MA, Andrade DM, Dugan P, Depondt C, Choi H, **Heinzen EL**, Cavalleri GL, Buono RJ, Devinsky O, Sperling MR, Berkovic SF, Delanty N, Goldstein DB, O'Brien TJ; **EPIGEN Consortium**. *Rare genetic variation and outcome of surgery for mesial temporal lobe epilepsy.* Ann Neurol. 2022 Dec 19.
2. Oliver KL, Ellis CA, Scheffer IE, Ganesan S, Leu C, Sadleir LG, **Heinzen EL**, Mefford HC, Bass AJ, Curtis SW, Harris RV; **Epi4K Consortium,** Whiteman DC, Helbig I, Ottman R, Epstein MP, Bahlo M, Berkovic. *Common risk variants for epilepsy are enriched in families previously targeted for rare monogenic variant discovery.* EBioMedicine. 2022 May 27;81:104079.
3. Lai D, Gade M, Yang E, Koh HY, Lu J, Walley NM, Buckley AF, Sands TT, Akman CI, Mikati MA, McKhann GM, Goldman JE, Canoll P, Alexander AL, Park KL, Von Allmen GK, Rodziyevska O, Bhattacharjee MB, Lidov HGW, Vogel H, Grant GA, Porter BE, Poduri AH‡, Crino PB‡, **Heinzen EL**‡. *Somatic variants in diverse genes lead to a spectrum of focal cortical malformations.* Brain. 2022 Aug 27;145(8):2704-2720.
4. Campbell C, McCormack M, Patel S, Stapleton C, Bobbili D, Krause R, Depondt C, Sills GJ, Koeleman BP, Striano P, Zara F, Sander JW, Lerche H, Kunz WS, Stefansson K, Stefansson H, Doherty CP, **Heinzen EL**, Scheffer IE, Goldstein DB, O'Brien T, Cotter D, Berkovic SF; EpiPGX Consortium, Sisodiya SM, Delanty N, Cavalleri GL. *A pharmacogenomic assessment of psychiatric adverse drug reactions to levetiracetam.* Epilepsia. 2022 Jun;63(6):1563-1570.
5. Green TE, Motelow JE, Bennett MF, Ye Z, Bennett CA, Griffin NG, Damiano JA, Leventer RJ, Freeman JL, Harvey AS, Lockhart PJ, Sadleir LG, Boys A, Scheffer IE, Major H, Darbro BW, Bahlo M, Goldstein DB, Kerrigan JF, **Heinzen EL**, Berkovic SF, Hildebrand MS. *Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions.* Hum Mol Genet. 2022 Jul 21;31(14):2307-2316.
6. Alkelai A, Greenbaum L, Docherty AR, Shabalin AA, Povysil G, Malakar A, Hughes D, Delaney SL, Peabody EP, McNamara J, Gelfman S, Baugh EH, Zoghbi AW, Harms MB, Hwang HS, Grossman-Jonish A, Aggarwal V, **Heinzen EL**, Jobanputra V, Pulver AE, Lerer B, Goldstein DB. *The benefit of diagnostic whole genome sequencing in schizophrenia and other psychotic disorders.* Mol Psychiatry. 2022 Mar;27(3):1435-1447.
7. Uchitel J, Wallace K, Tran L, Abrahamsen T, Hunanyan A, Prange L, Jasien J, Caligiuri L, Pratt M, Rikard B, Fons C, De Grandis E, Vezyroglou A, **Heinzen EL**, Goldstein DB, Vavassori R, Papadopoulou MT, Cocco I, Moré R; Duke AHC Research Group; French AHC Consortium, Arzimanoglou A, Panagiotakaki E, Mikati MA. *Alternating hemiplegia of childhood: evolution over time and mouse model corroboration*. Brain Commun. 2021 Jun 4;3(3):fcab128.
8. Ernst ME, Baugh EH, Thomas A, Bier L, Lippa N, Stong N, Mulhern MS, Kushary S, Akman CI, **Heinzen EL**, Yeh R, Bi W, Hanchard NA, Burrage LC, Leduc MS, Chong JSC, Bend R, Lyons MJ, Lee JA, Suwannarat P,

Brilstra E, Simon M, Koopmans M, van Binsbergen E, Groepper D, Fleischer J, Nava C, Keren B, Mignot C, Mathieu S, Mancini GMS, Madan-Khetarpal S, Infante EM, Bluvstein J, Seeley A, Bachman K, Klee EW, Schultz-Rogers LE, Hasadsri L, Barnett S, Ellingson MS, Ferber MJ, Narayanan V, Ramsey K, Rauch A, Joset P, Steindl K, Sheehan T, Poduri A, Vasquez A, Ruivenkamp C, White SM, Pais L, Monaghan KG, Goldstein DB, Sands TT, Aggarwal V. *CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity*. Epilepsia. 2021 Jul;62(7):e103-e109.

9. Heron SE, Regan BM, Harris RV, Gardner AE, Coleman MJ, Bennett MF, Grinton BE, Helbig KL, Sperling MR, Haut S, Geller EB, Widdess-Walsh P, Pelekanos JT, Bahlo M, Petrovski S, **Heinzen EL**, Hildebrand MS, Corbett MA, Scheffer IE, Gécz J, Berkovic SF. *Association of SLC32A1 missense variants with genetic epilepsy with febrile seizures plus.* Neurology. 2021 May 4;96(18):e2251-e2260.
10. Alkelai A, Shohat S, Greenbaum L, Schechter T, Draiman B, Chitrit-Raveh E, Rienstein S, Dagaonkar N, Hughes D, Aggarwal VS, **Heinzen EL**, Shifman S, Goldstein DB, Kohn Y. *Expansion of the GRIA2 phenotypic representation: a novel de novo loss of function mutation in a case with childhood onset schizophrenia.* J Hum Genet. 2021 Mar;66(3):339-343.
11. Weng PL, Majmundar AJ, Khan K, Lim TY, Shril S, Jin G, Musgrove J, Wang M, Ahram DF, Aggarwal VS, Bier LE, **Heinzen EL**, Onuchic-Whitford AC, Mann N, Buerger F, Schneider R, Deutsch K, Kitzler TM, Klämbt V, Kolb A, Mao Y, Moufawad El Achkar C, Mitrotti A, Martino J, Beck BB, Altmüller J, Benz MR, Yano S, Mikati MA, Gunduz T, Cope H, Shashi V; Undiagnosed Diseases Network, Trachtman H, Bodria M, Caridi G, Pisani I, Fiaccadori E, AbuMaziad AS, Martinez-Agosto JA, Yadin O, Zuckerman J, Kim A; UCLA Clinical Genomics Center, John-Kroegel U, Tyndall AV, Parboosingh JS, Innes AM, Bierzynska A, Koziell AB, Muorah M, Saleem MA, Hoefele J, Riedhammer KM, Gharavi AG, Jobanputra V, Pierce-Hoffman E, Seaby EG, O'Donnell-Luria A, Rehm HL, Mane S, D'Agati VD, Pollak MR, Ghiggeri GM, Lifton RP, Goldstein DB, Davis EE, Hildebrandt F, Sanna-Cherchi S. *De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis.* Am J Hum Genet. 2021 Feb 4;108(2):357-367.
12. **Heinzen EL.** *Somatic variants in epilepsy - advancing gene discovery and disease mechanisms.* Curr Opin Genet Dev. 2020 Dec;65:1-7.
13. Choi H, Detyniecki K, Bazil C, Thornton S, Crosta P, Tolba H, Muneeb M, Hirsch LJ, **Heinzen EL**, Sen A, Depondt C, Perucca P, Heiman GA; **EPIGEN Consortium**. *Development and validation of a predictive model of drug-resistant genetic generalized epilepsy.* Neurology. 2020 Oct 13;95(15):e2150-e2160.
14. Miceli F, Carotenuto L, Barrese V, Soldovieri MV, **Heinzen EL**, Mandel AM, Lippa N, Bier L, Goldstein DB, Cooper EC, Cilio MR, Taglialatela M, Sands TT. *A novel Kv7.3 Variant in the voltage-sensing S 4 segment in a family with benign neonatal epilepsy: functional characterization and in vitro by β-hydroxybutyrate.* Front Physiol. 2020 Sep 4;11:1040.
15. Prange L, Pratt M, Herman K, Schiffmann R, Mueller DM, McLean M, Mendez MM, Walley N, **Heinzen EL**, Goldstein D, Shashi V, Hunanyan A, Pagadala V, Mikati MA. *D-DEMØ, a distinct phenotype caused by ATP1A3 mutations.* Neurol Genet. 2020 Aug 4;6(5):e466.
16. McCormack M, McGinty RN, Zhu X, Slattery L, **Heinzen EL**; **EPIGEN Consortium**, Costello DJ, Delanty N, Cavalleri GL. *De-novo mutations in patients with chronic ultra-refractory epilepsy with onset after age five years*. Eur J Med Genet. 2020 Jan;63(1):103625.
17. Sands TT, Miceli F, Lesca G, Beck AE, Sadleir LG, Arrington DK, Schönewolf-Greulich B, Moutton S, Lauritano A, Nappi P, Soldovieri MV, Scheffer IE, Mefford HC, Stong N, **Heinzen EL**, Goldstein DB, Perez AG, Kossoff EH, Stocco A, Sullivan JA, Shashi V, Gerard B, Francannet C, Bisgaard AM, Tümer Z, Willems M, Rivier F, Vitobello A, Thakkar K, Rajan DS, Barkovich AJ, Weckhuysen S, Cooper EC, Taglialatela M, Cilio MR. *Autism and developmental disability caused by KCNQ3 gain-of-function variants.* Ann Neurol. 2019 Aug;86(2):181-192.
18. Alkelai A, Greenbaum L, **Heinzen EL**, Baugh EH, Teitelbaum A, Zhu X, Strous RD, Tatarskyy P, Zai CC, Tiwari AK, Tampakeras M, Freeman N, Müller DJ, Voineskos AN, Lieberman JA, Delaney SL, Meltzer HY, Remington G, Kennedy JL, Pulver AE, Peabody EP, Levy DL, Lerer B. *New insights into tardive dyskinesia genetics: Implementation of whole-exome sequencing approach.* Prog Neuropsychopharmacol Biol Psychiatry. 2019 Aug 30;94:109659.

19. Cogné B, Ehresmann S, Beauregard-Lacroix E, Rousseau J, Besnard T, Garcia T, Petrovski S, Avni S, McWalter K, Blackburn PR, Sanders SJ, Uguen K, Harris J, Cohen JS, Blyth M, Lehman A, Berg J, Li MH, Kini U, Joss S, von der Lippe C, Gordon CT, Humberson JB, Robak L, Scott DA, Sutton VR, Skraban CM, Johnston JJ, Poduri A, Nordenskjöld M, Shashi V, Gerkes EH, Bongers EMHF, Gilissen C, Zarate YA, Kvarnung M, Lally KP, Kulch PA, Daniels B, Hernandez-Garcia A, Stong N, McGaughran J, Retterer K, Tveten K, Sullivan J, Geisheker MR, Stray-Pedersen A, Tarpinian JM, Klee EW, Sapp JC, Zyskind J, Holla ØL, Bedoukian E, Filippini F, Guimier A, Picard A, Busk ØL, Punetha J, Pfundt R, Lindstrand A, Nordgren A, Kalb F, Desai M, Ebanks AH, Jhangiani SN, Dewan T, Coban Akdemir ZH, Telegrafi A, Zackai EH, Begtrup A, Song X, Toutain A, Wentzensen IM, Odent S, Bonneau D, Latypova X, Deb W; CAUSES Study, Redon S, Bilan F, Legendre M, Troyer C, Whitlock K, Caluseriu O, Murphree MI, Pichurin PN, Agre K, Gavrilova R, Rinne T, Park M, Shain C, **Heinzen EL**, Xiao R, Amiel J, Lyonnet S, Isidor B, Biesecker LG, Lowenstein D, Posey JE, Denommé-Pichon AS; Deciphering Developmental Disorders study, Férec C, Yang XJ, Rosenfeld JA, Gilbert-Dussardier B, Audebert-Bellanger S, Redon R, Stessman HAF, Nellaker C, Yang Y, Lupski JR, Goldstein DB, Eichler EE, Bolduc F, Bézieau S, Küry S, Campeau PM. *Missense variants in the histone acetyltransferase complex component gene TRRAP cause autism and syndromic intellectual disability.* Am J Hum Genet. 2019 Mar 7;104(3):530-541.
20. Verbitsky M, Westland R, Perez A, Kiryluk K, Liu Q, Krithivasan P, Mitrotti A, Fasel DA, Batourina E, Sampson MG, Bodria M, Werth M, Kao C, Martino J, Capone VP, Vivante A, Shril S, Kil BH, Marasà M, Zhang JY, Na YJ, Lim TY, Ahram D, Weng PL, **Heinzen EL**, Carrea A, Piaggio G, Gesualdo L, Manca V, Masnata G, Gigante M, Cusi D, Izzi C, Scolari F, van Wijk JAE, Saraga M, Santoro D, Conti G, Zamboli P, White H, Drozdz D, Zachwieja K, Miklaszewska M, Tkaczyk M, Tomczyk D, Krakowska A, Sikora P, Jarmoliński T, Borszewska-Kornacka MK, Pawluch R, Szczepanska M, Adamczyk P, Mizerska-Wasiak M, Krzemien G, Szmigielska A, Zaniew M, Dobson MG, Darlow JM, Puri P, Barton DE, Furth SL, Warady BA, Gucev Z, Lozanovski VJ, Tasic V, Pisani I, Allegri L, Rodas LM, Campistol JM, Jeanpierre C, Alam S, Casale P, Wong CS, Lin F, Miranda DM, Oliveira EA, Simões-E-Silva AC, Barasch JM, Levy B, Wu N, Hildebrandt F, Ghiggeri GM, Latos-Bielenska A, Materna-Kiryluk A, Zhang F, Hakonarson H, Papaioannou VE, Mendelsohn CL, Gharavi AG, Sanna-Cherchi S. *The copy number variation landscape of congenital anomalies of the kidney and urinary tract.* Nat Genet. 2019 Jan;51(1):117-127.
21. Mulhern MS, Stumpel C, Stong N, Brunner HG, Bier L, Lippa N, Riviello J, Rouhl RPW, Kempers M, Pfundt R, Stegmann APA, Kukolich MK, Telegrafi A, Lehman A; CAUSES study, Lopez-Rangel E, Houcinat N, Barth M, den Hollander N, Hoffer MJ, Weckhuysen S; EuroEPINOMICS-RES-MAE working group, Roovers J, Djemie T, Barca D, Ceulemans B, Craiu D, Lemke JR, Korff C, Mefford HC, Meyers CT, Siegler Z, Hiatt SM, Cooper GM, Martina Bebin E, Snijders-Blok L, Veenstra-Knol HE, Baugh EH, Brilstra EH, Volker-Touw CML, van Binsbergen E, Revah-Politi A, Pereira E, McBrian D, Pacault M, Isidor B, Le Caignec C, Gilbert-Dussardier B, Bilan F, **Heinzen EL**, Goldstein DB, Stevens SJC, Sands TT. *NBEA: developmental disease gene with early generalized epilepsy phenotypes.* Ann Neurol. 2018 Nov; 84(5): 788–795
22. Assoum M, Lines MA, Elpeleg O, Darmency V, Whiting S, Edvardson S, Devinsky O, **Heinzen E**, Hernan RR, Antignac C, Deleuze JF, Des Portes V, Bertholet-Thomas A, Belot A, Geller E, Lemesle M, Duffourd Y, Thauvin-Robinet C, Thevenon J, Chung W, Lowenstein DH, Faivre L. *Further delineation of the clinical spectrum of de novo TRIM8 truncating mutations.* Am J Med Genet A. 2018 Nov;176(11):2470-2478.
23. Hemati P, Revah-Politi A, Bassan H, Petrovski S, Bilancia CG, Ramsey K, Griffin NG, Bier L, Cho MT, Rosello M, Lynch SA, Colombo S, Weber A, Haug M, **Heinzen EL**, Sands TT, Narayanan V, Primiano M, Aggarwal VS, Millan F, Sattler-Holtrop SG, Caro-Llopis A, Pillar N, Baker J, Freedman R, Kroes HY, Sacharow S, Stong N, Lapunzina P, Schneider MC, Mendelsohn NJ, Singleton A, Loik Ramey V, Wou K, Kuzminsky A, Monfort S, Weiss M, Doyle S, Iglesias A, Martinez F, Mckenzie F, Orellana C, van Gassen KLI, Palomares M, Bazak L, Lee A, Bircher A, Basel-Vanagaite L, Hafström M, Houge G; C4RCD Research Group; DDD study, Goldstein DB, Anyane-Yeboa K. *Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature.* Am J Med Genet A. 2018 Nov;176(11):2259-2275.
24. Helbig I, **Heinzen EL**, Mefford HC; *International League Against Epilepsy Genetics Commission. Genetic literacy series: Primer part 2-Paradigm shifts in epilepsy genetics*. Epilepsia. 2018 Jun;59(6):1138-1147.
25. Winawer MR, Griffin NG, Samanamud J, Baugh EH, Rathakrishnan D, Ramalingam S, Zagzag D, Schevon CA, Dugan P, Hegde M, Sheth SA, McKhann GM, Doyle WK, Grant GA, Porter BE, Mikati MA, Muh CR, Malone CD, Bergin AMR, Peters JM, McBrian DK, Pack AM, Akman CI, LaCoursiere CM, Keever KM,

Madsen JR, Yang E, Lidov HGW, Shain C, Allen AS, Canoll PD, Crino PB‡, Poduri AH‡, **Heinzen EL**‡. *Somatic SLC35A2 variants in the brain are associated with intractable neocortical epilepsy.* Ann Neurol. 2018 Jun;83(6):1133-1146.

26. **Heinzen EL**, O'Neill AC, Zhu X, Allen AS, Bahlo M, Chelly J, Chen MH, Dobyns WB, Freytag S, Guerrini R, Leventer RJ, Poduri A, Robertson SP, Walsh CA, Zhang M; **Epi4K Consortium; Epilepsy Phenome/Genome Project.** *De novo and inherited private variants in MAP1B in periventricular nodular heterotopia*. PLoS Genet. 2018 May 8;14(5):e1007281.
27. Zhu X, Padmanabhan R, Copeland B, Bridgers J, Ren Z, Kamalakaran S, O'Driscoll-Collins A, Berkovic SF, Scheffer IE, Poduri A, Mei D, Guerrini R, Lowenstein DH, Allen AS, **Heinzen EL**‡, Goldstein DB‡. *A case-control collapsing analysis identifies epilepsy genes implicated in trio sequencing studies focused on de novo mutations.* PLoS Genet. 2017 Nov 29;13(11):e1007104.
28. Myers CT‡, Stong N‡, Mountier EI, Helbig KL, Freytag S, Sullivan J, Zeev BB, Nissenkorn A, Tzadok M, Heimer G, Shinde DN, Rezazadej A, Regan BM, Oliver KL, Ernst ME, Lippa NC, Mulhern MS, Ren Z, Poduri A, Andrade DM, Bird LM, Bahlo M, Berkovic SF, Lowenstein DH, Scheffer IE, Sadleir LG, Goldstein DB, Mefford HC‡, **Heinzen EL**‡. *De novo mutations in PPP3CA cause severe neurodevelopmental disease with seizures*. Am J Hum Genet. 2017 Oct 5;101(4):516-524.
29. Griffin NG, Cronin KD, Walley NM, Hulette CM, Grant GA, Mikati MA, LaBreche HG, Rehder CW, Allen AS, Crino PB, **Heinzen EL.** *Somatic uniparental disomy of chromosome 16p in hemimegalencephaly.* Cold Spring Harb Mol Case Stud. 2017 Sep 1;3(5).
30. Gelfman S, Wang Q, McSweeney KM, Ren Z, La Carpia F, Halvorsen M, Schoch K, Ratzon F, **Heinzen EL**, Boland MJ, Petrovski S, Goldstein DB. *Annotating pathogenic non-coding variants in genic regions*. Nat Commun. 2017 Aug 9;8(1):236.
31. Depondt C, **Heinzen EL**, Goldstein DB. *Reply to Two patients with TNK2 mutations and late-onset infantile spasm*. Ann Neurol. 2017 Jan;81(1):161-162.
32. Hildebrand MS‡, Griffin NG‡, Damiano JA, Cops EJ, Burgess R, Ozturk E, Jones NC, Leventer RJ, Freeman JL, Harvey AS, Sadleir LG, Scheffer IE, Major H, Darbro BW, Allen AS, Goldstein DB, Kerrigan JF, Berkovic SF‡, **Heinzen EL**‡. *Mutations of the Sonic Hedgehog pathway underlie hypothalamic hamartoma with gelastic epilepsy.* Am J Hum Genet. 2016 Aug 4;99(2):423-9.
33. Helbig I, **Heinzen EL**, Mefford HC; *ILAE Genetics Commission. Primer Part 1-The building blocks of epilepsy genetics.* Epilepsia. 2016 Jun;57(6):861-8.
34. Petrovski S, Küry S, Myers CT, Anyane-Yeboa K, Cogné B, Bialer M, Xia F, Hemati P, Riviello J, Mehaffey M, Besnard T, Becraft E, Wadley A, Politi AR, Colombo S, Zhu X, Ren Z, Andrews I, Dudding-Byth T, Schneider AL, Wallace G; University of Washington Center for Mendelian Genomics., Rosen AB, Schelley S, Enns GM, Corre P, Dalton J, Mercier S, Latypova X, Schmitt S, Guzman E, Moore C, Bier L, **Heinzen EL**, Karachunski P, Shur N, Grebe T, Basinger A, Nguyen JM, Bézieau S, Wierenga K, Bernstein JA, Scheffer IE, Rosenfeld JA, Mefford HC, Isidor B, Goldstein DB. *Germline de novo mutations in GNB1 cause severe neurodevelopmental disability, hypotonia, and seizures.* Am J Hum Genet. 2016 May 5;98(5):1001-10.
35. Griffin NG, Wang Y, Hulette CM, Halvorsen M, Cronin KD, Walley NM, Haglund MM, Radtke RA, Skene JH, Sinha SR, **Heinzen EL**. *Differential gene expression in dentate granule cells in mesial temporal lobe epilepsy with and without hippocampal sclerosis.* Epilepsia. 2016 Mar;57(3):376-85.
36. Panagiotakaki E, De Grandis E, Stagnaro M, **Heinzen EL**, Fons C, Sisodiya S, de Vries B, Goubau C, Weckhuysen S, Kemlink D, Scheffer I, Lesca G, Rabilloud M, Klich A, Ramirez-Camacho A, Ulate-Campos A, Campistol J, Giannotta M, Moutard ML, Doummar D, Hubsch-Bonneaud C, Jaffer F, Cross H, Gurrieri F, Tiziano D, Nevsimalova S, Nicole S, Neville B, van den Maagdenberg AM, Mikati M, Goldstein DB, Vavassori R, Arzimanoglou A; Italian IBAHC Consortium.; French AHC Consortium.; International AHC Consortium. *Clinical profile of patients with ATP1A3 mutations in alternating hemiplegia of childhood-a study of 155 patients*. Orphanet J Rare Dis. 2015 Sep 26;10:123.
37. **Heinzen EL**, Neale BM, Traynelis SF, Allen AS, Goldstein DB. *The genetics of neuropsychiatric diseases: looking in and beyond the exome.* Annu Rev Neurosci. 2015 Jul 8;38:47-68.
38. Dhindsa RS, Bradrick SS, Yao X, **Heinzen EL**, Petrovski S, Krueger BJ, Johnson MR, Frankel WN, Petrou S, Boumil RM, Goldstein DB. *Epileptic encephalopathy-causing mutations in DNM1 impair synaptic vesicle endocytosis*. Neurol Genet. 2015 Apr 17;1(1):e4.

39. Li M, Jazayeri D, Corry B, McSweeney KM, **Heinzen EL**, Goldstein DB, Petrou S. *A functional correlate of severity in alternating hemiplegia of childhood.* Neurobiol Dis. 2015 May;77:88-93.
40. Jiang Y, Satten GA, Han Y, Epstein MP, **Heinzen EL**, Goldstein DB, Allen AS. *Utilizing population controls in rare-variant case-parent association tests.* Am J Hum Genet. 2014 Jun 5;94(6):845-53.
41. **Heinzen EL**, Arzimanoglou A, Brashear A, Clapcote SJ, Gurrieri F, Goldstein DB, Jóhannesson SH, Mikati MA, Neville B, Nicole S, Ozelius LJ, Poulsen H, Schyns T, Kathleen J Sweadner KJ, van den Maagdenberg A, Vilsen BV, ATP1A3 Working Group. *ATP1A3 mutations in distinct neurological disorders*. Lancet Neurol. 2014 May;13(5):503-14.
42. Sánchez Fernández I, Abend NS, Agadi S, An S, Arya R, Carpenter JL, Chapman KE, Gaillard WD, Glauser TA, Goldstein DB, Goldstein JL, Goodkin HP, Hahn CD, **Heinzen EL**, Mikati MA, Peariso K, Pestian JP, Ream M, Riviello JJ Jr, Tasker RC, Williams K, Loddenkemper T; **Pediatric Status Epilepticus Research Group (pSERG).** *Gaps and opportunities in refractory status epilepticus research in children: A multi-center approach by the Pediatric Status Epilepticus Research Group (pSERG).* Seizure. 2014 Feb;23(2):87-97.
43. Poduri A, **Heinzen EL**, Chitsazzadeh V, Lasorsa FM, Elhosary PC, LaCoursiere CM, Martin E, Yuskaitis CJ, Hill RS, Atabay KD, Barry B, Partlow JN, Bashiri FA, Zeidan RM, Elmalik SA, Kabiraj MM, Kothare S, Stödberg T, McTague A, Kurian MA, Scheffer IE, Barkovich AJ, Palmieri F, Salih MA, Walsh CA. *SLC25A22 is a novel gene for migrating partial seizures in infancy.* Ann Neurol. 2013 Dec;74(6):873-82.
44. Hitomi Y, **Heinzen EL**, Donatello S, Dahl HH, Damiano JA, McMahon JM, Berkovic SF, Scheffer IE, Legros B, Rai M, Weckhuysen S, Suls A, De Jonghe P, Pandolfo M, Goldstein DB, Van Bogaert P, Depondt C. *Mutations in TNK2 in severe autosomal recessive infantile-onset epilepsy*. Ann Neurol. 2013 Sep;74(3):496-501
45. Petrovski S, Wang Q, **Heinzen EL**, Allen AS, Goldstein DB. *Genic intolerance to functional variation and the interpretation of personal genomes*. PLoS Genet. 2013 Aug;9(8):e1003709.
46. Manzini MC, Tambunan DE, Hill RS, Yu TW, Maynard TM, **Heinzen EL**, Shianna KV, Stevens CR, Partlow JN, Barry BJ, Rodriguez J, Gupta VA, Al-Qudah AK, Eyaid WM, Friedman JM, Salih MA, Clark R, Moroni I, Mora M, Beggs AH, Gabriel SB, Walsh CA. *Exome sequencing and functional validation in zebrafish identify GTDC2 mutations as a cause of Walker-Warburg Syndrome.* Am J Hum Genet. 2012;91(3):541-7.
47. Qianqian Zhu Q, Ge D, **Heinzen EL**, Dickson SP, Urban TJ, Zhu M, Maia JM, He M, Zhao Q, Shianna KV, Goldstein DB. *Prioritizing genetic variants for causality on the basis of preferential linkage disequilibrium.* Am J Hum Genet. 2012 Sep 7;91(3):422-34.
48. Zhu M., Need AC, Han Y, Ge D, Maia JM, Zhu Q, **Heinzen EL**, Cirulli ET, Pelak K, He M, Ruzzo EK, Gumbs C, Singh A, Feng S, Shianna KV, Goldstein DB. *Using ERDS to infer copy-number variants in high-coverage genomes.* Am J Hum Genet. 2012 Sep 7;91(3):408-21.
49. Need AC, McEvoy J, Gennarelli M, **Heinzen EL**, Ge D, Maia JM, Shianna KV, He M, Cirulli E, Gumbs C, Zhao Q, Campbell CR, Hong L, Rosenquist P, Putkonen A, Hallikainen T, Repo-Tiihonen E, Tiihonen J, Levy D, Meltzer H, Goldstein DB. *Exome sequencing followed by large-scale genotyping suggests a limited role for moderately rare risk factors of strong effect in schizophrenia*. Am J Hum Genet. 2012 Aug 10;91(2):303-12.
50. **Heinzen EL**, Depondt C, Cavalleri GL, Ruzzo EK, Walley NM, Need AC, Ge D, He M, Cirulli ET, Zhao Q, Cronin KD, Gumbs CE, Campbell CR, Hong LK, Maia JM, Shianna KV, McCormack M, O'Conner GD, Radtke RA, Mikati MA, Gallentine WB, Husain AM, Sinha SR, Chinthapalli K, Puranam RS, McNamara JO, Ottman R, Sisodiya SM, Norman Delanty, Goldstein DB *Exome sequencing of idiopathic generalized epilepsy patients.* Am J Hum Genet. 2012 Aug 10;91(2):293-302.
51. **Heinzen EL**,‡ Swoboda KJ,‡ Hitomi Y,‡ Gurrieri F, Nicole S, de Vries B, Tiziano FD, Fontaine B, Walley NM, Heavin S, Panagiotakaki E, European AHC Genetics Consortium, **I.B.AHC Consortium**, ENRAH for SME consortium, Fiori F, Abiusi E, Di Pietro L, Sweney MT, Newcomb TM, Viollet L, Huff C, Jorde L, Reyna SP, Murphy KJ, Shianna KV, Gumbs CE, Little L, Silver K, Ptáček LJ, Haan J, Ferrari MD, Bye AM, Herkes GK, Whitelaw CM, Webb D, Lynch BJ, Uldall P, King MD, Scheffer IE, Neri G, Arzimanoglou A, van den Maagdenberg AMJM, Sisodiya SM, Mikati MA, Goldstein DB. *ATP1A3 de novo mutations in alternating hemiplegia of childhood*. Nat Genet. 2012 Jul 29;44(9):1030-4**.**
52. Poduri A, Evrony GD, Cai X, Elhosary PC, Hills L, Beroukhim R, Lehtinen MK, **Heinzen EL**, Hill A, Hill RS, Barry BJ, Bourgeois BFD, Riviello JJ, Barkovich AJ, Black PM, Ligon KL, Walsh CA. *Somatic activation of AKT3 causes hemispheric developmental brain malformations*. Neuron. 2012 Apr 12;74(1):41-8.

53. McCormack M, Urban TJ, Shianna KV, Walley N, Pandolfo M, Depondt C, Chaila E, O'Conner GD, Kasperavičiūtė D, Radtke RA, **Heinzen EL**, Sisodiya SM, Delanty N, Cavalleri GL. *Genome-wide mapping for clinically relevant predictors of lamotrigine- and phenytoin-induced hypersensitivity reactions.* Pharmacogenomics. 2012 Mar;13(4):399-405.
54. McCormack M, Alfirevic A, Bourgeois S, Farrell JJ, Kasperavičiūtė D, Carrington M, Sills GJ, Marson T, Jia X, de Bakker PI, Chinthapalli K, Molokhia M, Johnson MR, O'Connor GD, Chaila E, Alhusaini S, Shianna KV, Radtke RA, **Heinzen EL**, Walley N, Pandolfo M, Pichler W, Park BK, Depondt C, Sisodiya SM, Goldstein DB, Deloukas P, Delanty N, Cavalleri GL, Pirmohamed M. *HLA-A*3101 and carbamazepine-induced hypersensitivity reactions in Europeans.* N Engl J Med. 2011 Mar 24;364(12):1134-43.
55. Cirulli ET, **Heinzen EL**, Dietrich FS, Shianna KV, Singh A, Maia JM, Goedert JJ, Goldstein DB. *A whole-genome analysis of premature termination codons.* Genomics. 2011 Nov;98(5):337-42.
56. Catarino CB, Kasperavičiūtė D, Thom M, Cavalleri GL, Martinian L, **Heinzen EL**, Dorn T, Grunwald T, Chaila E, Depondt C, Krämer G, Delanty N, Goldstein DB, Sisodiya SM. *Genomic microdeletions associated with epilepsy: not a contraindication to resective surgery*. Epilepsia. 2011 Aug;52(8):1388-92.
57. Ge D, Ruzzo EK, Shianna KV, He M, Pelak K, **Heinzen EL**, Need AC, Cirulli ET, Maia JM, Dickson SP, Zhu M, Singh A, Allen AS, Goldstein DB. *SVA: Software for annotating and visualizing sequenced human genomes.* Bioinformatics 2011 Jul 15;27(14):1998-2000.
58. Zhu Q, Ge D, Maia JM, Zhu M, Petrovski S, Dickson SP, **Heinzen EL**, Shianna KV, Goldstein DB. A Genome-wide *Comparison of the functional properties of rare and common genetic variants in humans.* Am J Hum Genet. 2011 Apr 8;88(4):458-68.
59. Pelak K, Shianna KV, Ge D, Maia JM, Zhu M, Smith JP, Cirulli ET, Fellay J, Dickson SP, Gumbs CE, **Heinzen EL**, Need AC, Ruzzo EK, Singh A, Campbell CR, Hong LK, Lornsen KA, McKenzie AM, Sobreira NL, Hoover-Fong JE, Milner JD, Ottman R, Haynes BF, Goedert JJ, Goldstein DB. *The characterization of twenty sequenced human genomes*. PLoS Genet. 2010 Sep 9;6(9). pii: e1001111.
60. Kasperaviciūte D, Catarino CB, **Heinzen EL**, Depondt C, Cavalleri GL, Caboclo LO, Tate SK, Jamnadas-Khoda J, Chinthapalli K, Clayton LM, Shianna KV, Radtke RA, Mikati MA, Gallentine WB, Husain AM, Alhusaini S, Leppert D, Middleton LT, Gibson RA, Johnson MR, Matthews PM, Hosford D, Heuser K, Amos L, Ortega M, Zumsteg D, Wieser HG, Steinhoff BJ, Krämer G, Hansen J, Dorn T, Kantanen AM, Gjerstad L, Peuralinna T, Hernandez DG, Eriksson KJ, Kälviäinen RK, Doherty CP, Wood NW, Pandolfo M, Duncan JS, Sander JW, Delanty N, Goldstein DB, Sisodiya SM. *Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study.* Brain. 2010 Jul;133(Pt 7):2136-47.
61. Beskow LM, Linney KN, Radtke RA, **Heinzen EL**, Goldstein DB. *Ethical challenges in genotype-driven research recruitment.* Genome Res. 2010 Jun;20(6):705-9.
62. **Heinzen EL**‡, Radtke RA‡, Urban TJ‡, Cavalleri GL, Depondt C, Need AC, Walley NM, Nicoletti P, Ge D, Catarino CB, Duncan JS, Kasperaviciūte D, Tate SK, Caboclo LO, Sander JW, Clayton L, Linney KN, Shianna KV, Gumbs CE, Smith J, Cronin KD, Maia JM, Doherty CP, Pandolfo M, Leppert D, Middleton LT, Gibson RA, Johnson MR, Matthews PM, Hosford D, Kälviäinen R, Eriksson K, Kantanen AM, Dorn T, Hansen J, Krämer G, Steinhoff BJ, Wieser HG, Zumsteg D, Ortega M, Wood NW, Huxley-Jones J, Mikati M, Gallentine WB, Husain AM, Buckley PG, Stallings RL, Podgoreanu MV, Delanty N, Sisodiya SM, Goldstein DB. *Rare deletions at 16p13.11 predispose to a diverse spectrum of sporadic epilepsy syndromes*. Am J Hum Genet. 2010 May 14;86(5):707-18.
63. Rotger M, Dang K, Fellay J, **Heinzen EL**, Feng S, Descombes P, Shianna KV, Ge D, Günthard HF, Goldstein DB, Telenti A. *Genome-wide mRNA expression correlates of viral control in CD4+ T-cells from HIV-1-Infected Individuals*. PLoS Pathogens 2010 Feb 26;6(2):e1000781
64. Cirulli ET, Singh A, Shianna KV, Ge D, Smith JP, Maia JM, **Heinzen EL**, Goedert JJ, Goldstein DB; Center for HIV/AIDS Vaccine Immunology (CHAVI). *Screening the human exome: a comparison of whole genome and whole transcriptome sequencing.* Genome Biol. 2010;11(5):R57.
65. **Heinzen EL**, Need AC, Hayden KM, Chiba-Falek O, Roses AD, Strittmatter WJ, Burke JR, Hulette CM, Welsh-Bohmer KA, Goldstein DB. *Genome-wide scan of copy number variation in late-onset Alzheimer's disease*. J Alzheimers Dis. 2010;19(1):69-77.
66. Need AC, Attix DK, McEvoy JM, Cirulli ET, Linney KL, Hunt P, Ge D, **Heinzen EL**, Maia JM, Shianna KV, Weale ME, Cherkas LF, Clement G, Spector TD, Gibson G, Goldstein DB. *A genome-wide study of common*

SNPs and CNVs in cognitive performance in the CANTAB. Hum Mol Genet. 2009; 18(23):4650-61.Ge D, Fellay J, Thompson AJ, Simon JS, Shianna KV, Urban TJ, **Heinzen EL**, Qiu P, Bertelsen AH, Muir AJ, Sulkowski M, McHutchison JG, Goldstein DB. *Genetic variation in IL28B predicts hepatitis C treatment-induced viral clearance.* Nature. 2009; 461(7262):399-401. Need AC, Ge D, Weale ME, Maia J, Feng S, **Heinzen EL**, Shianna KV, Yoon W, Kasperaviciūte D, Gennarelli M, Strittmatter WJ, Bonvicini C, Rossi G, Jayathilake K, Cola PA, McEvoy JP, Keefe RS, Fisher EM, St Jean PL, Giegling I, Hartmann AM, Möller HJ, Ruppert A, Fraser G, Crombie C, Middleton LT, St Clair D, Roses AD, Muglia P, Francks C, Rujescu D, Meltzer HY, Goldstein DB. *A genome-wide investigation of SNPs and CNVs in schizophrenia.* PLoS Genet. 2009; 5(2):e1000373.**Heinzen EL**‡, Ge D‡, Cronin KD, Maia JM, Shianna KV, Gabriel WN, Welsh-Bohmer KA, Hulette CM, Denny TN, Goldstein DB. *Tissue-specific genetic control of splicing: Implications for the study of complex traits.* PLoS Biol 2008; 6(12): e1000001.

70. Ervin JF, **Heinzen EL**, Goldstein DB, Szymanski MH, Burke JR, Welsh-Bohmer KA, and Hulette CM. *Human brain tissue may be retrieved up to 30 hours post mortem with little effect on RNA integrity.* J Neuropathol Exp Neurol 2007;66:1093-1099.
71. **Heinzen EL**, Yoon W, Weale ME, Wood NW, Sen A, Burke JR, Welsh-Bohmer KA, Hulette CM, Sisodiya SM, Goldstein DB. *Alternative ion channel splicing in mesial temporal lobe epilepsy and Alzheimer's disease*. Genome Bio 2007; 8: R32.
72. **Heinzen EL**‡, Yoon W‡, Tate SK, Sen A, Wood NW, Sisodiya SM, Goldstein DB. *NOVA2 interacts with a cis-acting polymorphism to influence the proportions of pharmacologically relevant splice variants.* Am J Hum Genet. 2007; 80: 876-883.
73. Naderer OJ, Dupuis RE, **Heinzen EL**, Wiwattanawongsa K, Johnson MW, Smith PC. *Influence of antibiotics on the disposition of mychophenolic acid in healthy volunteers.* J Clin Pharmacol 2005; 45: 219-226.
74. **Heinzen EL**, Booth RG, Pollack GM. *Neuronal nitric oxide modulates morphine antinociceptive tolerance by enhancing constitutive activity of the μ-opioid receptor.* Biochem Pharmacol 2005; 69: 679-688.
75. **Heinzen EL** and Pollack GM. *Pharmacodynamics of morphine-induced neuronal nitric oxide production and antinociceptive tolerance development.* Brain Res 2004; 1023:175-184.
76. **Heinzen EL** and Pollack GM. *The development of morphine antinociceptive tolerance in nitric oxide synthase-deficient mice.* Biochem Pharmacol 2003; 67, 735-741.
77. **Heinzen EL** and Pollack GM. *Pharmacokinetics and pharmacodynamics of L-arginine in rats: A model of stimulated neuronal nitric oxide production*. Brain Res 2003; 989: 67-75.
78. **Heinzen EL** and Pollack GM. *The use of an electrochemical nitric oxide sensor to detect neuronal nitric oxide in conscious, unrestrained rats.* Pharmacol and Toxicol Methods 2002; 3: 139-146.
79. Wiwattanawongsa K, **Heinzen EL**, Kemp DC, Dupuis RE, Smith PC. *Determination of mycophenolic acid and its phenol glucuronide metabolite in human plasma and urine by high-performance liquid chromatography.* J Chromatogr B 2001; 763: 35-45.

Original Research From Collaborative Networks

*** Specific contributions in collaborative work indicated using Contributor Roles Taxonomy (CRediT) (https://credit.niso.org/) for publications with a significant role

80. Koko M, Motelow JE, Stanley KE, Bobbili DR, Dhindsa RS, May P; Canadian Epilepsy Network; **Epi4K Consortium**; Epilepsy Phenome/Genome Project; EpiPGX Consortium; EuroEPINOMICS-CoGIE Consortium. *Association of ultra-rare coding variants with genetic generalized epilepsy: A case-control whole exome sequencing study.* Epilepsia. 2022 Mar;63(3):723-735.
81. Koko M, Krause R, Sander T, Bobbili DR, Nothnagel M, May P, Lerche H; **Epi25 Collaborative**. *Distinct gene-set burden patterns underlie common generalized and focal epilepsies*. EBioMedicine. 2021 Oct;72:103588.
82. Stevelink R, Luykx JJ, Lin BD, Leu C, Lal D, Smith AW, Schijven D, Carpay JA, Rademaker K, Rodrigues Baldez RA, Devinsky O, Braun KPJ, Jansen FE, Smit DJA, Koeleman BPC; **International League Against Epilepsy Consortium on Complex Epilepsies; Epi25 Collaborative.** *Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations.* Epilepsia. 2021 Jul;62(7):1518-1527.

83. **Epi25 Collaborative.** *Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals.* Am J Hum Genet. 2021 Jun 3;108(6):965-982.

84. Epilepsy Phenome/Genome Project, **Epi4K Consortium.** *Diverse genetic causes of polymicrogyria with epilepsy*. Epilepsia. 2021 Apr;62(4):973-983.

    ***CRediT: Conceptualization, Formal Analysis, Funding acquisition, Project administration, Writing – original draft

85. Schijven D, Stevelink R, McCormack M, van Rheenen W, Luykx JJ, Koeleman BPC, Veldink JH; Project MinE ALS GWAS Consortium; **International League Against Epilepsy Consortium on Complex Epilepsies**. *Analysis of shared common genetic risk between amyotrophic lateral sclerosis and epilepsy.* Neurobiol Aging. 2020 Aug;92:153.e1-153.e5.

86. Niestroj LM, Perez-Palma E, Howrigan DP, Zhou Y, Cheng F, Saarentaus E, Nürnberg P, Stevelink R, Daly MJ, Palotie A, Lal D; **Epi25 Collaborative**. *Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17,458 subjects.* Brain. 2020 Jul 1;143(7):2106-2118.

    ***CRediT: Supervision, Writing – review & editing

87. Grasby KL, Jahanshad N, Painter JN, Colodro-Conde L, Bralten J, Hibar DP, Lind PA, Pizzagalli F, Ching CRK, McMahon MAB, Shatokhina N, Zsembik LCP, Thomopoulos SI, Zhu AH, Strike LT, Agartz I, Alhusaini S, Almeida MAA, Alnæs D, Amlien IK, Andersson M, Ard T, Armstrong NJ, Ashley-Koch A, Atkins JR, Bernard M, Brouwer RM, Buimer EEL, Bülow R, Bürger C, Cannon DM, Chakravarty M, Chen Q, Cheung JW, Couvy-Duchesne B, Dale AM, Dalvie S, de Araujo TK, de Zubicaray GI, de Zwarte SMC, den Braber A, Doan NT, Dohm K, Ehrlich S, Engelbrecht HR, Erk S, Fan CC, Fedko IO, Foley SF, Ford JM, Fukunaga M, Garrett ME, Ge T, Giddaluru S, Goldman AL, Green MJ, Groenewold NA, Grotegerd D, Gurholt TP, Gutman BA, Hansell NK, Harris MA, Harrison MB, Haswell CC, Hauser M, Herms S, Heslenfeld DJ, Ho NF, Hoehn D, Hoffmann P, Holleran L, Hoogman M, Hottenga JJ, Ikeda M, Janowitz D, Jansen IE, Jia T, Jockwitz C, Kanai R, Karama S, Kasperaviciute D, Kaufmann T, Kelly S, Kikuchi M, Klein M, Knapp M, Knodt AR, Krämer B, Lam M, Lancaster TM, Lee PH, Lett TA, Lewis LB, Lopes-Cendes I, Luciano M, Macciardi F, Marquand AF, Mathias SR, Melzer TR, Milaneschi Y, Mirza-Schreiber N, Moreira JCV, Mühleisen TW, Müller-Myhsok B, Najt P, Nakahara S, Nho K, Olde Loohuis LM, Orfanos DP, Pearson JF, Pitcher TL, Pütz B, Quidé Y, Ragothaman A, Rashid FM, Reay WR, Redlich R, Reinbold CS, Repple J, Richard G, Riedel BC, Risacher SL, Rocha CS, Mota NR, Salminen L, Saremi A, Saykin AJ, Schlag F, Schmaal L, Schofield PR, Secolin R, Shapland CY, Shen L, Shin J, Shumskaya E, Sønderby IE, Sprooten E, Tansey KE, Teumer A, Thalamuthu A, Tordesillas-Gutiérrez D, Turner JA, Uhlmann A, Vallerga CL, van der Meer D, van Donkelaar MMJ, van Eijk L, van Erp TGM, van Haren NEM, van Rooij D, van Tol MJ, Veldink JH, Verhoef E, Walton E, Wang M, Wang Y, Wardlaw JM, Wen W, Westlye LT, Whelan CD, Witt SH, Wittfeld K, Wolf C, Wolfers T, Wu JQ, Yasuda CL, Zaremba D, Zhang Z, Zwiers MP, Artiges E, Assareh AA, Ayesa-Arriola R, Belger A, Brandt CL, Brown GG, Cichon S, Curran JE, Davies GE, Degenhardt F, Dennis MF, Dietsche B, Djurovic S, Doherty CP, Espiritu R, Garijo D, Gil Y, Gowland PA, Green RC, Häusler AN, Heindel W, Ho BC, Hoffmann WU, Holsboer F, Homuth G, Hosten N, Jack CR Jr, Jang M, Jansen A, Kimbrel NA, Kolskår K, Koops S, Krug A, Lim KO, Luykx JJ, Mathalon DH, Mather KA, Mattay VS, Matthews S, Mayoral Van Son J, McEwen SC, Melle I, Morris DW, Mueller BA, Nauck M, Nordvik JE, Nöthen MM, O'Leary DS, Opel N, Martinot MP, Pike GB, Preda A, Quinlan EB, Rasser PE, Ratnakar V, Reppermund S, Steen VM, Tooney PA, Torres FR, Veltman DJ, Voyvodic JT, Whelan R, White T, Yamamori H, Adams HHH, Bis JC, Debette S, Decarli C, Fornage M, Gudnason V, Hofer E, Ikram MA, Launer L, Longstreth WT, Lopez OL, Mazoyer B, Mosley TH, Roshchupkin GV, Satizabal CL, Schmidt R, Seshadri S, Yang Q; Alzheimer's Disease Neuroimaging Initiative; CHARGE Consortium; **EPIGEN Consortium**; IMAGEN Consortium; SYS Consortium; Parkinson's Progression Markers Initiative, Alvim MKM, Ames D, Anderson TJ, Andreassen OA, Arias-Vasquez A, Bastin ME, Baune BT, Beckham JC, Blangero J, Boomsma DI, Brodaty H, Brunner HG, Buckner RL, Buitelaar JK, Bustillo JR, Cahn W, Cairns MJ, Calhoun V, Carr VJ, Caseras X, Caspers S, Cavalleri GL, Cendes F, Corvin A, Crespo-Facorro B, Dalrymple-Alford JC, Dannlowski U, de Geus EJC, Deary IJ, Delanty N, Depondt C, Desrivières S, Donohoe G, Espeseth T, Fernández G, Fisher SE, Flor H, Forstner AJ, Francks C, Franke B, Glahn DC, Gollub RL, Grabe HJ, Gruber O, Håberg AK, Hariri AR, Hartman CA, Hashimoto R, Heinz A, Henskens FA, Hillegers MHJ, Hoekstra PJ, Holmes AJ, Hong LE, Hopkins WD, Hulshoff Pol HE, Jernigan TL, Jönsson EG, Kahn RS, Kennedy MA, Kircher TTJ, Kochunov P, Kwok JBJ, Le Hellard S, Loughland CM, Martin NG, Martinot JL, McDonald C, McMahon KL, Meyer-Lindenberg A, Michie PT, Morey RA, Mowry B, Nyberg L, Oosterlaan J, Ophoff RA, Pantelis C, Paus T, Pausova Z, Penninx BWJH, Polderman TJC, Posthuma D, Rietschel M,

Roffman JL, Rowland LM, Sachdev PS, Sämann PG, Schall U, Schumann G, Scott RJ, Sim K, Sisodiya SM, Smoller JW, Sommer IE, St Pourcain B, Stein DJ, Toga AW, Trollor JN, Van der Wee NJA, van 't Ent D, Völzke H, Walter H, Weber B, Weinberger DR, Wright MJ, Zhou J, Stein JL, Thompson PM, Medland SE; *Enhancing NeuroImaging Genetics through Meta-Analysis Consortium (ENIGMA)—Genetics working group. The genetic architecture of the human cerebral cortex.* Science. 2020 Mar 20;367(6484):eaay6690.

88. **Epi4K Consortium.** *Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies.* Epilepsia. 2019 Nov;60(11):2194-2203.
89. Stevelink R, Pangilinan F, Jansen FE, Braun KPJ; **International League Against Epilepsy Consortium on Complex Epilepsies**, Molloy AM, Brody LC, Koeleman BPC. *Assessing the genetic association between vitamin B6 metabolism and genetic generalized epilepsy.* Mol Genet Metab Rep. 2019 Oct 11;21:100518.
90. **Epi25 Collaborative.** *Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals.* Am J Hum Genet. 2019 Aug 1;105(2):267-282.
91. Lerche H, Berkovic SF, Lowenstein DH; EuroEPINOMICS-CoGIE Consortium; EpiPGX Consortium; **Epi4K Consortium**/Epilepsy Phenome/Genome Project. Response to *Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy.* N Engl J Med. 2019 Apr 18;380(16):e24.
92. **Epilepsy Genetics Initiative.** *The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield.* Epilepsia. 2019 Apr 5.

    ***CRediT: Conceptualization, Funding acquisition, Supervision, Writing – review & editing
93. **International League Against Epilepsy Consortium on Complex Epilepsies.** *Genome-wide mega-analysis identifies 16 loci and highlights diverse biological mechanisms in the common epilepsies.* Nat Commun. 2018 Dec 10;9(1):5269.
94. **Brainstorm Consortium.** *Analysis of shared heritability in common disorders of the brain.* Science. 2018 Jun 22;360(6395).
95. McCormack M, Gui H, Ingason A, Speed D, Wright GEB, Zhang EJ, Secolin R, Yasuda C, Kwok M, Wolking S, Becker F, Rau S, Avbersek A, Heggeli K, Leu C, Depondt C, Sills GJ, Marson AG, Auce P, Brodie MJ, Francis B, Johnson MR, Koeleman BPC, Striano P, Coppola A, Zara F, Kunz WS, Sander JW, Lerche H, Klein KM, Weckhuysen S, Krenn M, Gudmundsson LJ, Stefánsson K, Krause R, Shear N, Ross CJD, Delanty N; **EPIGEN Consortium**;, Pirmohamed M, Carleton BC; Canadian Pharmacogenomics Network for Drug Safety, Cendes F, Lopes-Cendes I, Liao WP, O'Brien TJ, Sisodiya SM; EpiPGX Consortium;, Cherny S, Kwan P, Baum L; International League Against Epilepsy Consortium on Complex Epilepsies;, Cavalleri GL. *Genetic variation in CFH predicts phenytoin-induced maculopapular exanthema in European-descent patients.* Neurology. 2018 Jan 23;90(4):e332-e341.
96. **Epilepsy Genetic Initiative.** *De novo variants in the alternative exon 5 of SCN8A cause epileptic encephalopathy.* Genet Med. 2018 Feb;20(2):275-281.

    ***CRediT: Conceptualization, Formal Analysis, Funding acquisition, Project administration, Writing – original draft
97. **Epi4K Consortium**; EuroEPINOMICS-RES Consortium; Epilepsy Phenome Genome Project. *Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data.* Eur J Hum Genet. 2017 Jun;25(7):894-899.
98. **Epi4K Consortium**; Epilepsy Phenome/Genome Project. *Ultra-rare genetic variation in common epilepsies: a case-control sequencing study.* Lancet Neurol. 2017 Feb;16(2):135-143.

    ***CRediT: Conceptualization, Formal Analysis, Funding acquisition, Supervision, Writing – review & editing
99. **Epi4K Consortium**. *De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies.* Am J Hum Genet. 2016 Aug 4;99(2):287-98.
100. **EpiPM Consortium.** *A roadmap for precision medicine in the epilepsies.* Lancet Neurol. 2015 Dec;14(12):1219-28.
101. Epilepsy Phenome/Genome Project, **Epi4K Consortium**. *Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy.* Ann Neurol. 2015 Aug;78(2):323-8.

102. EuroEPINOMICS-RES Consortium; Epilepsy Phenome/Genome Project; **Epi4K Consortium**. *De Novo Mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies.* Am J Hum Genet. 2014 Oct 2;95(4):360-70.

    ***CRediT: Conceptualization, Formal Analysis, Funding acquisition, Project administration, Writing – original draft

103. **International League Against Epilepsy Consortium on Complex Epilepsies**. *Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies.* Lancet Neurol. 2014 Sep;13(9):893-903.

104. **Epi4K Consortium** and Epilepsy Phenome/Genome Project. *De novo mutations in epileptic encephalopathies.* Nature. 2013 Sep 12;501(7466):217-21.

    ***CRediT: Conceptualization, Formal Analysis, Funding acquisition, Project administration, Writing – review & editing

105. **Epi4K Consortium.** *Epi4K: Gene discovery in 4,000 genome.* Epilepsia. 2012 Aug;53(8):1457-1467.

    ***CRediT: Conceptualization, Funding acquisition, Project administration, Writing – review & editing

Abstracts

1. Lai D, Sosicka P, Ressler AK, Freeze HH, Boland MJ, **Heinzen EL**. *A loss of function mutation in SLC35A2 generates asynchronous, hypoactive neural networks due to glutamatergic/GABAergic imbalance.* American Epilepsy Society Annual Meeting, 2022.
2. Gade M, Lai D, **Heinzen EL**. *Determining single-cell transcriptomic signatures in mosaic epileptogenic human brain tissue.* American Epilepsy Society Annual Meeting, 2022.
3. Gade M, Lai D, **Heinzen EL**. *Leveraging mosaic epileptogenic human brain tissue to study cell-type specific transcriptional changes associated with a pathogenic PIK3CA variant.* American Society for Human Genetics Annual Meeting, 2022.
4. Lai D, Sosicka P, Ressler AK, Freeze HH, Boland MJ, **Heinzen EL**. *Loss of function mutations in SLC35A2 disrupts the glutamatergic/GABAergic balance resulting in asynchronous, hypoactive neural networks.* Society for Neuroscience Annual Meeting, 2022.
5. **The International League Against Epilepsy Consortium on Complex Epilepsies, Epi25 Collaborative**, *Meta-analysis in 64,000 individuals reveals novel epilepsy-related GWAS signals*, American Epilepsy Society Annual Meeting, 2021.
6. Oliver KL, Ellis CA, Scheffer IE, Ganesan S, Leu, C, Sadleir L, **Heinzen EL**, Epilepsy Phenome/Genome Project, Epi4K Consortium, Whiteman DC, Ottman RO, Epstein MP, Bahlo M, Berkovic SF. *Common risk variants for epilepsy are enriched in families previously targeted for rare "monogenic" variant discovery*, American Epilepsy Society Annual Meeting, 2021.
7. Lai D, Williams D, Sosicka P, Ressler AK, Freeze HH, Boland MJ, **Heinzen EL**. *Establishing a human iPSC-derived neuron model of SLC35A2-associated epilepsy.* Society for Neuroscience Annual Meeting 2021.
8. Lu, J. De Araujo Martins Moreno C, Harms MB, **Heinzen EL**. *LUSTR: A powerful and user-friendly tool to call germline and somatic short tandem repeat variants from short read next-generation sequencing data.* American Society Human Genetics Annual Meeting, 2021.
9. Lai D, Yang E, Grant G, Porter B, Mikati M, Walley N, Canoll P, Lu J, Von Allmen G, Bhattacharjee M, Crino P, Poduri A, **Heinzen EL**. *Brain-tissue localized somatic variants in epilepsy genes in pediatric cortical brain malformations. American* Epilepsy Society Annual Meeting, 2020.

    *selected for a podium presentation

10. Lai D, Yang E, Grant G, Porter B, Mikati M, Walley N, Canoll P, Lu J, Von Allmen G, Bhattacharjee M, Crino P, Poduri A, **Heinzen EL**. *Somatic variants in known epilepsy genes are found in pediatric cortical brain malformations.* American Society Human Genetics Annual Meeting, 2020.

    *selected for a podium presentation

11. Lu, J. De Araujo Martins Moreno C, Harms MB, **Heinzen EL**. *LUSTR - a new pipeline for somatic short tandem repeat variant calling.* American Society Human Genetics Annual Meeting, 2020.
12. Alkelai A, Greenbaum L, Docherty AR, Shabalin AA, Hughes D, Povysil G, Delaney SL, Peabody EP, McNamara J, Gelfman S, Baugh EH, Zoghbi AW, Aggarwal V, **Heinzen EL**, Lerer B, Pulver AE, Levy DL,

Goldstein DB. *Clinical value of diagnostic whole genome sequencing in schizophrenia and psychotic disorders.* World Congress of Psychiatric Genetics 2020.

13. Lai D, Sosicka P, Ressler AK, Boland MJ, Freeze HH, **Heinzen EL**. *Establishing a human ipsc-derived neuronal model of SLC35A2 epilepsy to study disease mechanisms.* American Epilepsy Society Annual Meeting, 2019.
14. Lu, J. De Araujo Martins Moreno C, Harms MB, **Heinzen EL**. *A new short tandem repeat realignment pipeline allowing simple customized query designed for high accuracy, reliability, and compatibility.* Genome Informatics Cold Spring Harbor Laboratories Meeting, 2019.
15. Lu, J. De Araujo Martins Moreno C, Harms MB, **Heinzen EL**. *A new pipeline designed for reliable and accurate short tandem repeat realignment.* American Society Human Genetics Annual Meeting, 2019.
16. Lai D, Sosicka P, Ressler AK, Boland MJ, Freeze HH, **Heinzen EL**. *Characterizing the mechanisms of SLC35A2 epilepsy in human iPSC-derived neurons.* American Society Human Genetics Annual Meeting, 2019.
17. **CSNK2B Working Group**. *CSNK2B: A novel cause of neurodevelopmental disease and epilepsy.* American College of Medical Genetics Annual Clinical Genetics Meeting, 2019.
18. Green, T, Ye Z, Damiano JA, Burgess R, Griffin NG, Bennett MF, Bahlo M, Scheffer IE, Leventer RJ, Freeman JL, Harvey SA, Goldstein DB, Kerrigan J, **Heinzen EL**, Hildebrand MS. *Two-hit genetic model provides evidence that sporadic hypothalamic hamartoma is a ciliopathy.* American Epilepsy Society Annual Meeting, 2019.
19. Perucca P, Stanley KE, McIntosh A, Asadi-Pooya AA, Mikati MA, Andrade DM, Dugan P, Depondt C, Choi H, **Heinzen EL**, Cavalleri G, Buono R, Devinsky O, Sperling MR, Goldstein D. *Seizure outcome after surgery for mesial temporal lobe epilepsy and ultra-rare genetic variation: An international, multicenter, case-control, whole-exome sequencing study.* American Epilepsy Society Annual Meeting, 2019.
20. Griffin NG, Winawer M, Crino P, Poduri A, **Heinzen EL**. *Post-zygotically acquired brain-tissue specific mutations in SLC35A2 lead to intractable neocortical epilepsy.* Gordon Research Conference, Mechanisms of Epilepsy and Neuronal Synchronization, 2018.
21. **Heinzen EL**, Allen AS, Cannoll P, Crino PB, Dugan P, Grant GA, Griffin NG, Hedge M, Lidov HGW, Malone CD, McKhann GM, Muh CR, Poduri AH, Schevon CA, Sheth SA, Winawer MR, Yang E, Zagzag D. *Somatic loss-of-function SLC35A2 mutations in refractory neocortical epilepsy.* Human Genetics in New York City Bi-Annual Meeting, 2018.
22. Goldstein D, **Heinzen EL**, Wapner R, Anyane-Yeboa K, Aggarwal V, Stong N, Bier L, Lippa N, Revah-Politi A, Ernst M, Mulhern M, Giordano J, Bialer M, Nichter C, Iglesias A, Alter A. *Characterization of SCN2A: A case series.* American College of Medical Genetics Annual Clinical Genetics Meeting, 2018.
23. Alkelai A, Greenbaum L, **Heinzen EL**, Goldstein D, Lerer B. *Potential Role of Rare Variants in the Genetics of Tardive Dyskinesia.* American Society Human Genetics Annual Meeting, 2018.
24. Lai D, Sosicka P, Ressler AK, Freeze H, Boland M, **Heinzen EL**. *Establishing a human ipsc-derived neuronal model of SLC35A2 epilepsy to study disease mechanisms.* American Epilepsy Society Annual Meeting, 2019.
25. **Heinzen EL**, Clark J, Vimla A, Amengual Gual M, Arya R, Carpenter JL, Chapman KE, Gaillard WD, Gaínza-Lein M, Glauser TA, Goldstein JL, Jackson M, Kapur K, McDonough TL, Mikati MA, Peariso K, Sánchez Fernández I, Stanley KE, Tasker RC, Tchapyjnikov D, Vasquez A, Wainwright MS, Wilfong A, Williams K, Goldstein D, Loddenkemper T. *Exome sequencing of a pediatric refractory status epilepticus cohort (the pSERG cohort).* American Epilepsy Society Annual Meeting, 2018
26. Winawer MR, Griffin NG, Samanamud J, Baugh E, Rathakrishnan D, Ramalingam S, Spagnolo Allende A, Zagzag D, Schevon CA, Dugan P, Hedge M, Sheth S, McKhann GM, Doyle WK, Grant GA, Porter BE, Mikati MA, Muh CR, Malone C, Bergin AM, Peters JM, McBrian DK, Pack AM, Akman CI, LaCoursiere C, Keever KM, Madsen JR, Yang E, Lidov HGW, Shain C, Allen AS, Canoll P, Crino PB, Poduri Annapurna, **Heinzen EL.** *Somatic mutation in SLC35A2 leads to focal epilepsy.* American Epilepsy Society Annual Meeting, 2018.
27. Song S, Dhindsa R, Williams D, **Heinzen EL**, Goldstein D, Boland M. *Modeling an epilepsy-associated cortical malformation Using hiPSC-derived organoids.* Society for Neuroscience Annual Meeting, 2018.
28. Poduri A, Winawer MR, Crino PB, **Heinzen EL**. *Somatic mutation in SLC35A2 leads to focal epilepsy.* Society for Neuroscience Annual Meeting, 2018.

29. McCormack M, Heavin S, Doherty CP, Costello D, Zhu X, **Heinzen EL**, Goldstein DB, Delanty N, Cavalleri GL. *An exploration of de novo mutations underpinning chronic refractory epilepsy.* Irish Institute for Clinical Neurosciences Meeting, 2017.
30. **Heinzen EL**, Allen AS, Berkovic SF, Dobyns W, Freytag S, Goldstein DB, Leventer R, Lowenstein DH, O'Neill AC, Poduri A, Robertson SP, Walsh C, Zhu X. *De novo and inherited variants in MAP1B in periventricular nodular heterotopia.* Gordon Research Conference, Human Genetics and Genomics, 2017
31. Mulhern M, Lippa L, Ernst M, Bazil C, Rivello J, Stong N, **Heinzen EL**, Goldstein D. *Review of Genetic testing in a cohort of neurology patients with genetic diagnoses made through research whole exome Sequencing.* American College of Medical Genetics Annual Clinical Genetics Meeting, 2017.
32. Lippa N, **Heinzen EL,** Bier L, Mulhern M, Ernst M, Stong N, Bazil C, Sands T, Harms M, Alcalay R, Gutierrez-Contreras J, Kuo SH, Kreisl W, Goldman J, Aggarwal V, Goldstein DB. *Whole exome sequencing in a cohort of adult and pediatric patients with neurologic disease.* American College of Medical Genetics Annual Clinical Genetics Meeting, 2017.
33. Griffin NG, Garcia-Tarodo S, Von Allmen G, Grant GA, Porter B, Mikati MA, Walley NM, Muh CR, LaCoursiere CM, Keever K, Shain C, Yang E, Crino PB, Poduri A, **Heinzen EL**. *Identification of somatic mutations in malformations of cortical development.* American Society Human Genetics Annual Meeting, 2017.
34. Sands TT, Miceli F, Lesca G, Beck A, Cimino M, **Heinzen EL**, Goldstein DB, Lowenstein DB, Weckhuysen S, Cooper EC, Tagliatela M, Cilio MR. *Autism with benzodiazepine-responsive electrical status epilepticus in sleep (ESES) caused by KCNQ3 gain-of-function variants*. American Epilepsy Society Annual Meeting, 2017.
35. **Heinzen, EL**. *De novo and inherited variants in MAP1B in periventricular nodular heterotopia.* American Epilepsy Society Annual Meeting, 2017.

    *selected for a podium presentation
36. Griffin NG, Garcia-Tarodo S, Von Allmen G, Grant GA, Porter B, Mikati MA, Walley NM, Muh CR, LaCoursiere CM, Keever K, Shain C, Yang E, Crino PB, Poduri A, **Heinzen EL**. *Identification of somatic mutations in malformations of cortical development.* American Epilepsy Society Annual Meeting, 2017.

    *selected for a podium presentation
37. Ernst, ME, **Heinzen EL**, Bier L, Lippa N, Mulhern M, Strong N, Bazil C, Sands T, Aggarwal V, Goldstein DB. *Experiences of whole exome sequencing in a broad adult and pediatric epilepsy cohort*. American Epilepsy Society Annual Meeting, 2017.

    *selected for a podium presentation
38. McSweeney K, **Heinzen EL**, Boland M, Goldstein D. *Mutations causing alternating hemiplegia of childhood disrupt normal neural network activity patterns.* Society for Neuroscience Annual Meeting, 2017.
39. Winawer M, Chen D, Misiewicz S, Samanamud j, Cannol P, **Heinzen EL**, Zagzag D, Wilson M, Scheyon C, Sheth S, McKhann G, Werner D, DeRisi J, Dugan P, Crino P. *Molecular abnormalities in non-lesional focal epilepsy.* American Epilepsy Society Annual Meeting, 2016.
40. Froukh T, Zhu X, Shashi V, Goldstein D, **Heinzen EL**. *Chromosomal translocation, cnv deletion and missense mutations associated with intellectual disability in consanguineous families from Jordan.* American Society Human Genetics Annual Meeting, 2016.
41. Hildebrand MS, Griffin NG, Damiano J, Cops EJ, Burgess R, Darbro B, Ozturk E, Jones N, Leventer R, Freeman JL, Harvey AS, Scheffer I, Goldstein D, Kerrigan J, Berkovic S, **Heinzen EL**. *Mutations of the sonic Hedgehog pathway underlie hypothalamic hamartoma and gelastic epilespy*. American Epilepsy Society Annual Meeting, 2015.
42. Goldstein, D, **Heinzen EL**, Lowenstein D, Berkovic S, Dixon-Salazar T, Milder J, White HS, Devinsky O, Dlugos D, Gallentine W, Mikati M, Poduri A, Scheffer I, Sullivan J. *Epilepsy Genetics Initiative (EGI).* American Epilepsy Society Annual Meeting, 2014.
43. Griffin N, Wang Y, Hong L, Hulette C, Haglund M, Radtke R, Skene P, Sinha S, **Heinzen EL**. *Gene Expression profiling in dentate granule cells of mesial temporal lobe epilepsy patients with and without hippocampal sclerosis.* American Epilepsy Society Annual Meeting, 2014.
44. **Heinzen EL**, Han Y, Poduri A, Epi4K and EPGP Investigators. *Somatic mutations in malformations of cortical development.* Gordon Research Conference, Human Genetics and Genomics, 2013.

45. **Heinzen EL**, Poduri A, Epi4K and EPGP Investigators. *Trio sequencing in malformations of cortical development.* American Society Human Genetics Annual Meeting, 2013.
46. Hildebrand, M, Petrovski S, Damiano J, Regan B, Scheffer I, **Heinzen EL**, Goldstein D, Berkovic S. *Unravelling the genetics of the common epilepsies using discordant monozygotic twins*. American Epilepsy Society Annual Meeting, 2013.
47. Poduri A, Evrony GD, Cai X, Elhosary PC, Beroukhim R, Lehtinen MK, Hills LB, **Heinzen EL**, Hill A, Hill RS, Barry BJ, Bourgeois BF, Riviello JJ, Vining E, Carson B, Barovich AJ, Black PM, Ligon KL, Walsh CA. *Somatic and germline mutations in AKT3 cause epileptic cortical malformations*. American Epilepsy Society Annual Meeting, 2012.
48. Ruzzo EK, **Heinzen EL**, Wedel R, Shianna KV, Scheffer E, Berkovic SF, Ottman R, Goldstein DB. *Investigating the genetic etiology of familial epilepsies using next generation sequencing.* American Society Human Genetics Annual Meeting, 2012.

    * selected for a podium presentation
49. Poduri A, Evrony GD, Cai X, Elhosary PC, Beroukhim R, Lehtinen MK, Hills LB, **Heinzen EL**, Hill A, Hill RS, Barry BJ, Bourgeois BFD, Riviello JJ, Barkovich AJ, Black PM, Madsen J, Ligon KL, Walsh CA. *Mutations in AKT3 lead to hemimegalencephaly*. American Society Human Genetics Annual Meeting, 2012.
50. **Heinzen EL**, Ruzzo E, Depondt C, Cavalleri G, Radtke R, Shianna K, Ge D, Catarino C, O'Conner G, Sisodiya S, Delanty N, Goldstein D, EPIGEN Consortium. *Small-scale exome-sequencing study followed by large-scale follow up to detect variants that increase the risk of idiopathic generalized epilepsy.* American Society Human Genetics Annual Meeting, 2012.

    * selected for a podium presentation
51. Ruzzo EK, **Heinzen EL**, Wedel R, Shianna KV, Ge D, Ottman R, Goldstein DB. *Interpreting Familial Whole-Genome sequencing data for the identification of genetic variants influencing epilepsy susceptibility.* American Society Human Genetics Annual Meeting, 2012.
52. Ruzzo, E, **Heinzen EL**, Poduri A, Wedel R, Ottman R, Goldstein D. *Whole-Genome Sequencing in Multiplex Epilepsy Families: An approach to identify rare susceptibility variants.* American Epilepsy Society Annual Meeting, 2010.
53. **Heinzen EL**, Cavalleri G, McCormack M, Alhusaini S, O'Connor G, Radtke R, Depondt C, Sisodiya S, Delanty N, Goldstein D. *Next-generation sequencing of refractory juvenile myclonic rpilepsy patients.* American Epilepsy Society Annual Meeting, 2010.
54. Urban T, Walley N, Nicoletti P, **Heinzen EL**, Ge D, Shianna K, Radtke R, Goldstein D. *NRXN1 Variants are Associated with behavioral difficulty in response to levetiracetam.* American Epilepsy Society Annual Meeting, 2009.
55. Mailman RB, **Heinzen EL**, Huang X. *The role of D1 dopamine receptor activation in Parkinson's disease: Insight from apomorphine and other clinically used dopamine Agonists.* International Congress of Parkinson's Disease and Movement Disorders, 2006.
56. **Heinzen EL**, Southerland SB, Neitzel KL, Mailman RB. *Differential activation of G Proteins at the dopamine D1 receptor mediates behavioral tolerance to agonists in rats*. American Society for Pharmacology and Experimental Therapeutics Annual Meeting, 2005.
57. **Heinzen EL**, Booth RG, Pollack GM. *Morphine-induced neuronal nitric oxide (NO) production modulates constitutive activity.* American Association of Pharmaceutical Scientists Annual Meeting, 2004.
58. **Heinzen EL**, Pollack GM. *The role of nitric oxide as a mediator of morphine antinociceptive tolerance.* American Association of Pharmaceutical Scientists Annual Meeting, 2003.
59. **Heinzen EL**, Pollack GM. *Pharmacokinetics and pharmacodynamics of L-Arginine in rats: A model of stimulated nitric oxide synthesis.* American Association of Pharmaceutical Scientists Annual Meeting, 2002.
60. Wiwattanasongsa K, **Heinzen EL**, Smith PC. *Evidence for glucuronidation of mycophenolic acid by intestinal UGT in the Guinea pig.* Annual Meeting of the International Society for the Study of Xenobiotics, 1999.
61. Wiwattanasongsa K, **Heinzen EL**, Dupuis RE, Smith PC. *Influence of antibiotics on the disposition of mycophenolic acid in the guinea pig.* American Association of Pharmaceutical Scientists Annual Meeting, 1997.

IN PRESS/SUBMITTED MANUSCRIPTS

1. Lu J, Toro C, Adams DR, Undiagnosed Disease Network, Lee WP, Leung YY, Vardarajan B, **Heinzen EL**. *LUSTR: a new customizable tool for calling genome-wide germline and somatic short tandem repeat variants.* BMC Genomics (submitted for publication).
2. Panagiotakaki E, Tiziano FD, Mikati MA, Vijfhuizen LS, Nicole S, Leisca G, Harder AVE, Goldstein DB, Walley NM, Arzamangouliou A, Gurrieri F, Koenderink J, Thompson CH, George AL Jr. van den Maagdenberg A.M.J.M., **Heinzen EL**. *Exome sequencing of ATP1A3-negative alternating hemiplegia of childhood reveals mutations in additional genes.* Eur J Hum Genet (submitted for publication).
3. Miller KE, Rivaldi A, Shinagawa N, Navarro J, Sran S, Westfall J, Miller AR, Roberts R, Hernandez Gonzalez ME, Akkari Y, Supinger R, Hester M, Marhabaie M, Gade M, Rodziyevska O, Bhattacharjee MB, Von Allmen GK, Yang E, Lidov HGW, Goldman JE, Thomas D, Boué D, Ostendorf AP, Mardis ER, Poduri A, Daniel Koboldt D, **Heinzen EL**‡, Bedrosian TA‡. Post-zygotic rescue of meiotic errors causes brain mosaicism and focal epilepsy. Nature (submitted for publication).
4. **International League Against Epilepsy Consortium on Complex Epilepsies.** *Genome-wide meta-analysis of over 29,000 people with epilepsy reveals 26 loci and subtype-specific genetic architecture.* Nature Genetics (submitted for publication).
5. **Epi25 Collaborative.** *Shared and distinct ultra-rare genetic risk for different types of epilepsies: A whole-exome sequencing study of 54,423 individuals across multiple genetic ancestries.* Nature Genetics (submitted for publication).

PRESENTATIONS

Invited Speaker

Regional

2022 Invited Speaker, *The expanding role of ultra-rare genetic variants in the epilepsies*, Genetics Colloquium, UNC Department of Genetics, Chapel Hill, NC.

2022 Invited Speaker, *The case of the hidden genetics of epilepsy*, UNC Department of Genetics Annual Retreat, Wilmington, NC.

2022 Invited Speaker, *Using a CRISPR-screen to identify regulators of NFIX expression*, Malan Syndrome Family Foundation Meeting, Chapel Hill, NC.

2022 Invited Speaker, *Illuminating novel pathways in epilepsy through the study of somatic variants*, Current Topics in Medical and Human Genetics, UNC School of Medicine, Chapel Hill, NC.

2021 Invited Speaker, *The impact of recent gene discovery on treatment selection in epilepsy*, Mini-Symposium: Pharmacogenomics and Decision Support in the EHR, UNC School of Medicine, Chapel Hill, NC.

2021 Invited Speaker, *Identification and functional characterization of somatic variants in pediatric epilepsies*, Children's Research Institute Seminar Series, UNC School of Medicine, Department of Pediatrics, Chapel Hill, NC.

2020 Invited Speaker, *Identification and genomic characterization of somatic variants in epilepsy*, Bioinformatics and Computational Biology Colloquium Series, UNC Department of Genetics, Chapel Hill, NC.

2020 Invited Speaker, *Somatic variants in epilepsy*, Genetics Colloquium, UNC Department of Genetics, Chapel Hill, NC.

2020 Invited Speaker, *Somatic variants in epilepsy*, Catalyst Research Group Meeting, Eshelman School of Pharmacy, Chapel Hill, NC.

2020 Invited Speaker, *Discovery and functional characterization of SLC35A2 epilepsy*, UNC Neuroscience Seminar Series, Chapel Hill, NC.

2019 Invited Speaker, *Somatic variants further expand the phenotypic spectrum associated with epilepsy and brain malformation genes*, Neurogenetics Seminar Series, Columbia University Medical Center, New York, NY.

2019 Invited Speaker, *Identification and functional characterization of somatic variants in epilepsy*, Department of Neurology, Columbia University Medical Center, New York, NY.

2018 Invited Speaker, *Exome sequencing of a pediatric status epilepticus cohort*, Pediatric status epilepticus research meeting, Boston Children's Hospital, Boston, MA.

2017 Invited Speaker, *Elucidating the genetic basis of malformations of cortical development*, Med into Grad Symposium, Columbia University, New York, NY.

2017 Invited Speaker, *The expanding de novo paradigm in severe sporadic epilepsies*, Langone Comprehensive Epilepsy Center, New York University, New York, NY.

2013 Invited Speaker, *The role of post-zygotic mutations in malformations of cortical development*, Human Genetics Evening. Duke University School of Medicine, Durham, NC.

2012 Invited Speaker, *Next Generation Sequencing in Complex Neuropsychiatric Disease*, The Harriet and John Wooten Laboratory for Alzheimer's and Neurodegenerative Diseases Research, Brody School of Medicine, East Carolina University, Greenville, NC.

2008 Invited Speaker, *Genetic Regulation of Splicing & Expression in Human Tissue*, Affymetrix Integrated Genomics Seminar Series, Durham, NC.

National

2023 Invited Speaker, *Genetics of childhood generalized epilepsies*, Southern Epilepsy and EEG Society Meeting, Atlanta, GA.

2023 Invited Speaker, *Somatic genetics of epilepsy - a novel lens into disease mechanisms.* Seizure Focus Seminar Series, Northwestern University, Chicago, IL (virtual).

2022 Invited Speaker, *Illuminating novel pathways in epilepsy and other neurodevelopmental disorders through the study of somatic variants*, Pediatric Grand Rounds, McGovern Medical School, University of Texas – Houston, Houston, TX (virtual).

2022 Invited Speaker, *Landscape of somatic variants in pediatric intractable neocortical epilepsy.* Texas Pediatric Neuroscience Network Conference 2022, Epilepsy Foundation Texas, Houston, TX.

2022 Invited Speaker, *iPSC-derived neuronal models of SLC35A2 epilepsy and beyond*, Ultragenyx, Novato, CA (virtual).

2021 Invited Speaker, Investigators Workshop, *New Somatic Genetics of Neocortical Epilepsy: SLC35A2 paves the way*, American Epilepsy Society Annual Meeting, Chicago, IL.

2021 Plenary Speaker, *Unraveling the mysteries of SLC35A2 associated seizures*, Frontiers in Congenital Disorders of Glycosylation 2nd Annual Scientific Symposium, La Jolla, CA.

2021 Plenary Speaker, *Epilepsy genetics – what we have learned and where we are going*, Ohio State University Neuroscience Research Institute Virtual Neurogenetics Symposium, Columbus, OH (virtual).

2021 Invited Speaker, *Gene discovery in epilepsy - paving the way towards precision therapies*, Mini Symposium in Human Genetics, Emory University, Atlanta, GA (virtual).

2021 Invited Speaker, *New Genetic Causes of the Epilepsies*, Curing the Epilepsies 2021 Conference, National Institute of Neurological Disease and Stroke, Washington, DC (virtual).

2019 Invited Speaker, *Paving the way towards precision medicine in epilepsy*, St Jude Children's Research Hospital, Memphis, TN.

2019 Invited Speaker, *Latest Development in Epilepsy Genetics*, Epigen Annual Meeting, Dulles, VA.

2019 Invited Speaker, *Paving the way towards precision medicine in epilepsy*, University of North Carolina at Chapel Hill, Eshelman School of Pharmacy, Chapel Hill, NC.

2019 Invited Speaker, *Progress towards precision medicine in epilepsy*, Cincinnati Children's Hospital, Cincinnati, OH.

2018 Invited Speaker, *Genetic bases of epilepsy disorders and relevance to Alternating Hemiplegia of Childhood*, ATP1A3 in Disease Meeting, Northwestern University, Chicago, IL.

2017 Invited Speaker, *De novo and inherited variants in MAP1B in periventricular nodular heterotopia*, American Epilepsy Society Annual Meeting, Washington, DC.

2017 Invited Speaker, *Identifying Regulatory Mutations that Influence Neuropsychiatric Disease*, Whole Genome Sequencing for Psychiatric Disorders Consortium Meeting, Boston, MA.

2016 Invited Speaker, *A Multi-electrode Array Approach to Understanding the Effects AHC-causing mutations*, Alternating Hemiplegia of Childhood Family Meeting, Indianapolis, IN.

2015 Invited Speaker, *Gene discovery in severe sporadic epilepsies*, Banbury Meeting, Scientific and Clinical Foundation for Precision Medicine in Epilepsy, Cold Spring Harbor Laboratory, Long Island, NY.

2014 Session chair, *Epilepsy Genetics in the Era of Precision Medicine Conference,* Precision Diagnostics I – Genetic Causes of Epilepsy, Half Moon Bay, CA.

2013 Invited speaker and panelist, *Genetics of Catastrophic Infantile Epilepsies: From Gene Discovery to Practical Clinical Applications*, American Epilepsy Society Annual Meeting, Washington, DC.

2013 Invited speaker and panelist, Roche Symposium, *Advancements in Target Enrichment – Improved Efficiencies in NGS Discovery and Clinical Research Workflows,* American Society for Human Genetics Annual Meeting, Boston, MA.

2013 Invited Speaker, *Somatic mutations in malformations of cortical development*, Human Genetics and Genomics Gordon Research Conference, Smithfield, RI.

2012 Invited Speaker and panelist. *Massively Parallel Sequencing in Epilepsy*, Investigator's Workshop, American Epilepsy Society Annual Meeting, San Diego, CA.

2007 Invited Speaker, *New applications for microarray data analysis, Integrating genetics with "omics"*, Cambridge Healthtech Institute, Washington, DC.

2006 Invited Speaker, *Alternative ion channel splicing in mesial temporal lobe epilepsy and Alzheimer's disease*, Splicing 2006 annual symposium, Baltimore, MD.

International

2022 Invited Speaker, *Genetics of alternating hemiplegia of childhood*, IAHCRC General Assembly 2022 (virtual).

2021 Invited Speaker, *Exome sequencing of ATP1A3-negative alternating hemiplegia of childhood reveals mutations in additional genes*, IAHCRC General Assembly 2021 (virtual).

2019 Invited Speaker, *Epilepsy genetics and the role somatic mosaicism*, Third Workshop on Ring14 Syndrome, Franciacorta, Italy.

2017 Invited Speaker, *Challenges and opportunities in somatic genetics*. AstraZeneca Genomic Partners Workshop. Cambridge, England.

2016 Invited Speaker, *Somatic mutations in hypothalamic hamartoma,* International Hypothalamic Hamartoma Symposium, London, England.

2014 Invited Speaker, *How it all started: ATP1A3 as the major gene for AHC,* Symposium of ATP1A3 in Disease. Lieden, Netherlands.

2013 Invited Speaker, *Genetic variation in ATP1A3 in neurological, developmental, and psychiatric diseases,* Symposium of ATP1A3 in Disease, Rome, Italy.

2012 Invited Speaker, *De novo mutations in ATP1A3 cause alternating hemiplegia of childhood*, Symposium of ATP1A3 in Disease, Brussels, Belgium.

2011 Invited Speaker, *Small-scale exome-sequencing followed by large-scale follow-up to detect genetic variants that increase the risk of idiopathic generalized epilepsy.* Neurogenetics IV: Schizophrenia and Epilepsy, 12th International Congress of Human Genetics, Montreal, Canada.

2010 Invited Speaker, XVIII World Congress on Psychiatric Genetics, *Symposium on genetic control of expression in human brain,* Athens, Greece.

Conference session Moderator

2022 Moderator, Genetics Sig, *Prophecy or Empiricy? Clinical value of predicting versus determining genetic variant dysfunction*, Annual Epilepsy Society Annual Meeting, Nashville, TN.

2020 Moderator, Investigators Workshop, *New vistas in the genetic landscape of epilepsy: explaining the genetically unexplained*, American Epilepsy Society Annual Meeting (virtual).

MEDIA FEATURES

2019 Neurology Today. Polygenic risk scores may help identify patients at higher risk of common types of epilepsy.

2019 Epilepsy Advocate. Advances in epilepsy's genetic makeup.

2017 Science Daily. Common epilepsies share genetic overlap with rare types.

2013 Science Daily. Newly identified genetic factors drive severe childhood epilepsies.

2012 Science Daily. Gene discovery set to help with mysterious paralysis of childhood.

2008 ABCnews.com. Genes Seem to Affect Tissues Differently.

2008 PhysOrg.com. Gene Expression and Splicing Vary Widely from One Tissue to the Next.

6. TEACHING ACTIVITIES

COURSES AND LECTURES

University of North Carolina at Chapel Hill

Eshelman School of Pharmacy

Spring 2023	PHCY 624: Research and Scholarship in Pharmacy I (3.0 credit hrs), Role: Division Co-Director (6.0 facilitator contact hrs), 4 students
Spring 2023	PHCY 726: Research and Scholarship in Pharmacy III (3.0 credit hrs), Role: Division Co-Director (4.5 facilitator contact hrs), 5 students
Fall 2022	DPET 873: Precision Therapeutics Through Genomics (3.0 credit hrs), Role: Course Co-Director and Lecturer (3.0 contact hrs), Enrollment: 10 students, Title of lecture: Rare disease genetics and genetically-guided pharmacotherapy
Fall 2022	PHCY 725: Research and Scholarship in Pharmacy II (3.0 credit hrs), Role: Division Co-Director (3.0 facilitator contact hrs), 5 students
Summer 2022	Innovations and Transformations in Pharmaceutical Sciences (ITPS), Role: Lecturer (1.0 contact hr), Approximately 75 attendees, Title of lecture: Advancements in Genetically Guided Pharmacotherapy
Spring 2022	PHRS 899: Division of Pharmacotherapy and Experimental Therapeutics Seminar (1.0 credit hr), Role: Faculty Director, Enrollment: 16 students
Spring 2022	PHCY 624: Research and Scholarship in Pharmacy I (3.0 credit hrs), Role: Division Co-Director (6.0 facilitator contact hrs), 3 students
Spring 2022	PHCY 726: Research and Scholarship in Pharmacy III (3.0 credit hrs), Role: Division Co-Director (4.5 facilitator contact hrs), 3 students
Fall 2021	PHCY 725: Research and Scholarship in Pharmacy II (3.0 credit hrs), Role: Division Co-Director (3.0 facilitator contact hrs), 3 students
Fall 2021	PHRS 899: Division of Pharmacotherapy and Experimental Therapeutics Seminar (1.0 credit hr), Role: Faculty Director, Enrollment: 21 students
Summer 2021	Innovations and Transformations in Pharmaceutical Sciences (ITPS), Role: Lecturer (1.0 contact hr), Approximately 75 attendees, Title of lecture: Genetics Guiding Pharmacotherapy
Spring 2021	PHCY 624: Research and Scholarship in Pharmacy I (3.0 credit hrs), Role: Division Co-Director (6.0 facilitator contact hrs), 3 students
Summer 2020	Innovations and Transformations in Pharmaceutical Sciences (ITPS), Role: Lecturer (1.0 contact hr), Approximately 75 attendees, Title of lecture: Genetics Guiding Pharmacotherapy

Eshelman School of Pharmacy (prior to faculty position)

2010	Lecturer, Pharmacogenetics
2005 – 2007	Lecturer, Short course on Integrated and Organ Systems Pharmacology
2001 – 2002	Lecturer, Advanced Pharmacokinetics I
2002	Lecturer, Clinical Pharmacokinetics I
2003	Lecturer, Clinical Pharmacokinetics II
2002	Lecturer, Advanced Pharmacokinetics II

School of Medicine

Fall 2022	GNET 703: Curriculum Genetics and Molecular Biology Seminar (1.0 credit hr), Role: Faculty Director, Enrollment: 33 students
Summer 2022	Educational Pathways to Increase Diversity in GEnomics, Role: Lecturer, Approximate enrollment: 20 students, Title of lecture: Induced pluripotent stem cell derived cellular model systems
Spring 2022	GNET 703: Curriculum Genetics and Molecular Biology Seminar (1.0 credit hr), Role: Faculty Director, Enrollment: 27 students
Fall 2021	GNET 703: Curriculum Genetics and Molecular Biology Seminar (1.0 credit hr), Role: Faculty Director, Enrollment: 27 students
Spring 2021	BBSP 902: Biological and Biomedical Science Program (BBSP) First Year Group (2 credit hrs), Role: First-year group mentor, Enrollment: 36 students

Fall 2020 BBSP 902: BBSP First Year Group (2 credit hrs), Role: First-year group mentor, Enrollment: 36 students

Columbia University

2019 GU4305: Lecturer, Seminar in Biotechnology
2019 Lecturer, Systems Pharmacology
2016 – 2018 Lecturer, Genomic Approaches II
2017 – 2018 Lecturer, Introduction to Precision Medicine
2016 – 2019 Lecturer, Introduction to Neuromuscular Diseases
2016 – 2018 Lecturer, Molecular Pharmacology

Duke University

2012 Lecturer, Human Genetics
2011 – 2014 Lecturer, Pharmacogenetics

Guilford College

2021 Lecturer, Drugs of Addiction

ADVISING AND MENTORSHIP

Faculty

Supervisor

2023 – present Dulcie Lai, PharmD., Ph.D, UNC Research Assistant Professor of Pharmacy

Career Development Mentor

2021 – present Senyene Hunter, MD, PhD, UNC Assistant Professor of Neurology
American Epilepsy Society Sergievsky Scholar Award (awarded)
Child Neurologist Career Development Program-K12 (awarded)

Postdoctoral Fellows

2021 – 2022 Nelson Manzanza, M.D., Ph.D., Postdoctoral fellow (UNC)
Current position: Instructor Western Governors University
2020 – 2021 Sarah Dugger, Ph.D., Postdoctoral fellow (UNC)
Current position: Clinical Genomic Scientist at Invitae
2018 – 2022 Dulcie Lai, PharmD., Ph.D, Postdoctoral fellow (UNC/Columbia University)
2017 – 2021 Jinfeng Lu, Ph.D., Postdoctoral fellow (UNC/Columbia University)
Current position: Postdoctoral fellow (Columbia University)
2013 – 2018 Nicole G. Griffin, Ph.D, Postdoctoral fellow (Duke University/Columbia University)
Current position: Bioinformatic Scientist (Syntensor)

PhD Students

Primary Advisor

2022 – present Ana Berglind, B.S., Graduate student (UNC, BBSP, Curriculum in Genetics and Molecular Biology)
2020 – present Meethila Gade, M.S., Graduate student (UNC, Pharmaceutical Science Program)
Thesis title: *A genomics driven approach to precision medicine in hemimegalencephaly*

Rotation Students

2022 Ryan Videgar-Laird (UNC, BBSP, Curriculum in Bioinformatics and Computational Biology)
2022 Kalynn Van Voorhies (UNC, BBSP, Neuroscience Curriculum)
2021 Jessica McAfee (UNC, BBSP, Curriculum in Genetics and Molecular Biology)
2020 Brandon Pratt (UNC, BBSP, Curriculum in Genetics and Molecular Biology)

Dissertation Committees

2023 – present Rachel Sharp, B.S. (UNC, BBSP, Neuroscience Curriculum Ph.D. Program, Dissertation Committee)
2022 – present Tom Collins, B.S. (UNC, Pharmacology M.D./Ph.D. Program, Dissertation Committee)

2021 – present Ricardo Gonzalez, B.S. (UNC, Pharmaceutical Science (DPET) Ph.D. Program, Eshelman School of Pharmacy, Dissertation Committee Chair)
2021 – present Soad Elziny, B.S. (University of Maryland, Neuroscience Ph.D. Program, Dissertation

Masters Students

Internship Advisor

2018 – 2019 Shih Hua Yu, B.S. (Columbia University, Masters in Data Science Program)

Thesis Advisor

2017 Ceylan Hasan, B.S. (Columbia University, Masters in Biotechnology Program)
2016 Rami Kathuda, B.S. (Columbia University, Masters in Biotechnology Program)

Visiting Scholars

Host Laboratory

2017 Thomasso Pippucci, Ph.D., EMBO Short-Term Fellowship

PharmD Students

Research Advisor

2022 – present MaryAnn Boyer (UNC)
Project title: *Evaluating an in vitro model system to predict SLC35A2 variant localization*
2022 – present Grace Paleracio (UNC, RASP Program)
Project title: *Evaluating an in vitro model system to utilize SLC35A2 variant localization to predict response to exogenous galactose therapy*
2021 – 2022 Jonathan Thulson (UNC, RASP Program)
Project title: *Effects of Smith-Kingsmore Syndrome associated MTOR variants on the mTOR signaling cascade*
2021 Amelia Ryan (University College London, PharmD candidate mentored research)
Project title: *Establishing a model system to study the effects of pathogenic MTOR variants on mTOR1 and mTOR2 signaling*

Undergraduate Students

Research Advisor

2021 – 2022 Ethan Lee, Undergraduate research assistant (UNC)
2020 – 2021 Eric Norloff, Undergraduate research assistant (UNC Work Study Program)
2020 – 2021 Lasya Kambhampati, Undergraduate research assistant (UNC)

High School Students

Research Advisor

2019 Shrey Shah (Jericho High School)
2018 – 2019 Rachel Tiersky (Bergin County Technical Schools High School, Senior Experience Internship Program)
2017 – 2018 Deborah Haimowitz, (The Abraham Joshua Heschel School, Summer Research Program)

COMMUNITY EDUCATION

2008 - 2012 Judge, National DNA Day Essay Contest, American Society for Human Genetics, GenEdNet

TEACHING CERTIFICATIONS

2021 Faculty Mentor Training for Biomedical Researchers

7. GRANTS

EXTERNAL

Grant Number	Agency	Title	Role	Total Directs	Percent Effort Funded	Dates
R01-NS115017S1	NIH - NINDS	Defining disease mechanisms in SLC35A2 epilepsy - Diversity Supplement	MPI - contact PI	$54,075	0%	May 2022 – April 2023
NA	Malan Syndrome Foundation	Moving from gene to treatment for Malan syndrome	MPI	$30,000	0%	July 2022 – June 2024
NA	Uplifting Athletes	Elucidating the molecular and cellular pathologies in an iPSC model of Malan syndrome to identify transformative therapies for modifying disease progression	MPI	$20,000	0%	May 2020 – Sept 2022
R01-NS115017	NIH - NINDS	Defining disease mechanisms in *SLC35A2* epilepsy	MPI - contact PI	$3,300,255	25%	July 2020 – May 2025
R01-NS114122	NIH - NINDS	Somatic Mutation in Intractable Focal Epilepsy	MPI	$2,805,821	10%	Aug 2020 – July 2025
R01-EY028203	NIH - NEI	Precision medicine for ABCA4 disease: modifier alleles	Co-Investigator	$739,096	5%	July 2018 – Dec 2019
R01-NS094596	NIH - NINDS	Identification and molecular characterization of somatic mutations in MCD	MPI - contact PI	$2,636,042	20%	Sept 2016 – June 2022*
R01-NS097876	NIH - NIA	Statin neuroprotection & cognitive dysfunction after carotid endarterectomy, feasibility, & outcomes	Co-Investigator	$1,534,557	5%	Sept 2016 – Dec 2019
NA	AstraZeneca UK Limited	Genomics Alliance Agreement	Co-Investigator /MPI**	$2,884,680	10%	Aug 2016 – Dec 2019
NA	CURE AHC	Columbia Alternating Hemiplegia of Childhood Multielectrode Array Project Plan	PI	$100,000	5%	Dec 2015 – Dec 2017
R01-NS089552	NIH - NINDS	Discovery of novel molecular abnormalities underlying non-lesional focal epilepsy	Co-Investigator	$1,334,148	10%	Sept 2014 – July 2019
U01-NS077303S1	NIH - NINDS	3 of 7 Epi4K: Sequencing, Biostatistics & Bioinformatics Core - Epilepsy Genetics Initiative	MPI	$200,000	5%	Sept 2014 – July 2018
NA	Citizens United for Research in Epilepsy (CURE)	Epilepsy Genetic Initiative	Co-Investigator	$352,063	5%	May 2014 – Dec 2019
R21-NS078657	NIH - NINDS	Exploratory genomic investigations of mesial temporal lobe epilepsy	PI	$270,625	25%	March 2012 – Feb 2015
U01-NS077303	NIH - NINDS	3 of 7 Epi4K: Sequencing, Biostatistics & Bioinformatics Core	MPI	$7,927,321	20%	Sept 2011 – July 2016
RC2-NS070344	NIH - NINDS	Whole-genome sequencing in multiplex epilepsy families	Co-Investigator	$1,099,524	20%	Sept 2009 – Aug 2011

** 5-year renewal to be awarded by NINDS in early 2023; ** Temporary PI role due to a conflict of interest of PI*

INTERNAL

Grant Number	Agency	Title	Role	Total Directs	Percent Effort Funded	Dates
NA	UNC Core Facilities Advocacy Committee (CFAC) voucher program	Genotype-informed single-cell transcriptomic profiling in mosaic brain tissue	PI	$9,886	0%	2023
NA	Irving Institute for Clinical and Translational Research		PI	$30,000	0%	July 2019 – Dec 2019
NA	Kathleen Price Bryan Alzheimer's Disease Research Center	The role of ion channel splice variation in Alzheimer's disease	PI	$30,000	10%	Sept 2009 – Aug 2010

8. PROFESSIONAL SERVICE

WITHIN DISCIPLINE

Committees/Advisory Boards

2023 – 2025 American Epilepsy Society, Investigators Workshop Committee
2020 – 2022 American Epilepsy Society, Basic Science Committee
August 2018 – 2020 ILAE Consortium on Complex Epilepsies Task Force of the Commission on Genetics
December 2016 – present Epi25 Consortium, Steering and Analysis Committees
January 2015 – 2018 Epilepsy Precision Medicine (EpiPM) Consortium, Steering Committee
April 2014 – 2020 Epilepsy Genetics Initiative (EGI), Steering Committee
January 2013 – present Pediatric Status Epilepticus Research Group, Member
September 2011 – present ILAE Consortium on Genetics of Complex Epilepsies, Steering Committee
July 2011 – present Steering Committee, Epi4K Consortium
April 2010 – present EpiGen Consortium, Scientific Advisory Board

Review Panels

June 2022 NINDS Study Section, ad hoc reviewer for DBD: Developmental Brain Disorders
2020 – 2022 NINDS Study Section, ad hoc reviewer for ANIE: Acute Neural Injury and Epilepsy
March 2019 American Association for the Advancement of Science, Review Panel, International Collaboration Grant - Saudi Arabian Ministry of Education's Research Development Office
January 2017 Department of Defense, Review Panel, Peer Reviewed Medical Research Program - Epilepsy Research Program - Idea Development
July 2016 NINDS Study Section, Review Panel, Centers Without Walls for Collaborative Research in the Epilepsies: Developing Transformative Therapies for Modifying or Preventing Epilepsy
December 2014 Department of Defense, Pre-application Review Panel, Peer Reviewed Medical Research Program – Epilepsy
June 2014 NINDS Study Section, Review Panel, Centers Without Walls for Collaborative Research in the Epilepsies: Sudden Unexpected Death in Epilepsy (SUDEP)
August 2013 Department of Defense, Review Panel, Peer Reviewed Medical Research Program - Epilepsy
August 2013 The Channel 7 Children's Research Foundation, Grant Reviewer
January 2004 – December 2006 Awards Committee for the Pharmacokinetics, Pharmacodynamics, and Drug Metabolism (PPDM) Section of the American Association of Pharmaceutical Sciences
2004 – 2005 Grant application reviewer, United States Environmental Protection Agency/University of North Carolina Toxicology Research Program

Meeting/Event Organizer

December 2021, 2022 Women in Epilepsy Symposium, American Epilepsy Society Annual Meeting
November 2015 Banbury Meeting, Scientific and Clinical Foundation for Precision Medicine in Epilepsy

August 2015 Scientific Organizer, Symposium of *ATP1A3* in Disease

Professional Organizations and Societies

2022 – present Member, American Association for the Advancement of Science
2010 – present Member, American Epilepsy Society
2007 – present Member, American Society of Human Genetics
2003 – 2010 Member, American Society of Pharmacology and Experimental Therapeutics
2002 – 2005 Member, Rho Chi Honor Society
2001 – 2008 Member, Research Triangle Park Drug Metabolism Discussion Group
1999 – 2008 Member, American Association of Pharmaceutical Scientists
1999 – 2005 Member, International Society for the Study of Xenobiotics
1998 – 2001 Member, American Society of Health System Pharmacists

Journal Reviewer

Alzheimer's and Dementia
American Journal of Human Genetics
Annals of Neurology
Annals of Clinical and Translational Neurology
BMC Genomics
Brain Research
Cell Systems Reviews
Developmental Medicine and Child Neurology
eBioMedicine
Epilepsia
Epilepsy Research
Genetics in Medicine
Genome Biology
Genome Medicine
Genome Research
Journal of Pharmacology and Experimental Therapeutics
Molecular Case Studies
Molecular Pharmacology
Nature Communications
Nature Genetics
Nature Medicine
Neuron
Neurotherapeutics
Pediatrics
PLoS Genetics
PLoS Biology
Scientific Reports

Editorial Boards

2022 – present Guest Editor, Neurobiology of Disease, Special Issue Title: Brain Somatic Mosaicism in Epilepsies
2019 – present Deputy Editor, Cambridge Elements, Genetics of Epilepsy
2016 Guest Editor, PLoS Genetics
2010 – 2011 Associate Editor Journal of Alzheimer's Disease

UNC

2022 – present Division of Pharmacotherapy and Experimental Therapeutics PhD Curriculum Review Committee
2022 – present Neuroscience Center Executive Committee
2021 – present Advisor Board, UNC CRISPR Genetic & Epigenetic Screening Center, Drug Discovery Initiative, Eshelman School of Pharmacy
2021 – present Research and Scholarship in Pharmacy (RASP) Oversight Committee
2021 Neuroscience Faculty Search Committee, Division of Pharmacoengineering and Molecular Pharmaceutics, Eshelman School of Pharmacy
2020 Eshelman School of Pharmacy, Scholarship Committee
2020 – 2022 Curriculum in Neuroscience Graduate Program Search Committee
2020 – 2022 Division of Pharmacotherapy and Experimental Therapeutics Graduate Program Search Committee

EXTERNAL

Jan 2016 Chair, Basic Science Faculty Search Committee, Columbia University Medical Center, Institute for Genomic Medicine
Jan – Dec 2016 Committee member, A Culture of Collaboration & Common Mission, Clinical/ Research Subcommittee, Columbia University Medical Center
July 2013 Organizer, Human Genetics Evening, Duke University Medical Center
2006 – 2007 Scientific consultant, NIH Short Course on Integrated and Organ Systems Pharmacology, University of North Carolina at Chapel Hill, School of Pharmacy