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Education

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

Training

September 2005 - March 2009 Postdoctoral Fellow, Duke University, Durham, NC, Institute for Genome Sciences & Policy, Mentor: David B. Goldstein, Ph.D.

April 2004 - August 2005 Postdoctoral Fellow, University of North Carolina, Chapel Hill, NC, School of Medicine, Curriculum in Toxicology, Mentor: Richard B. Mailman, Ph.D.

Licensure

February 2002 - present Pharmacist licensure North Carolina #16251

May 2015 - present Pharmacist licensure New York #060405

Academic Appointments

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

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

Honors and Awards

Irving Institute for Clinical and Translational Research Scholar (2019)

American Society for Pharmacology and Experimental Therapeutics Young Scientist Travel Award American Association of Pharmaceutical Scientists Postdoctoral Travel Award (2005)

Division of Drug Delivery and Disposition Graduate Scholar Award (2004)

American Association of Pharmaceutical Sciences Outstanding Graduate Student Research Award in Pharmacokinetics, Pharmacodynamics, and Drug Metabolism (2003)

American Foundation of Pharmaceutical Education Pre-doctoral Fellowship (2002-2004)

Miya Endowment Fund Academic Scholarship (2000)

George T. Cornwell Academic Scholarship (1999)

Publications

Peer-Reviewed Research Publications

‡ indicates shared first or last author

1. Epi4K Consortium. Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. *Epilepsia*. 2019 Nov;60(11):2194-2203.
2. 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.
3. 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, Tagliatela M, Cilio MR. *Autism and developmental disability caused by KCNQ3 gain-of-function variants*. *Ann Neurol*. 2019 Aug;86(2):181-192.
4. Alkelai A, Greenbaum L, **Heinzen EL**, Baugh EH, Teitelbaum A, Zhu X, Strous RD, Tatarsky 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.
5. Lerche H, Berkovic SF, Lowenstein DH; EuroEPINOMICS-CoGIE Consortium; EpiPGX Consortium; Epi4K Consortium/Epilepsy Phenome/Genome Project. *Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy*. *N Engl J Med*. 2019 Apr 18;380(16):e24.
6. Epilepsy Genetics Initiative. *The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield*. *Epilepsia*. 2019 Apr 5.
7. 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, McGaughan 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 Feb 28.
8. 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*. 2019 Jan 31
9. 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, Drozd 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.

10. 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.
11. Mulhern MS, Stumpel C, Stong N, Brunner HG, Bier L, Lippa N, Riviello J, Rouhl RPW, Kempers M, Pfundt R, Stegmann APA, Kukulich 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 Sep 30.
12. 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 Sep 23.
13. 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-Yeboah 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 Sep 8.
14. Brainstorm Consortium, *Analysis of shared heritability in common disorders of the brain*. Science. 2018 Jun 22;360(6395).
15. 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.
16. **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.
17. 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.
18. 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.
19. **Epilepsy Genetic Initiative**. *De novo variants in the alternative exon 5 of SCN8A cause epileptic encephalopathy*. Genet Med. 2017 Oct 2.
20. 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.
21. Myers CT[‡], Stong N[‡], Mountier EI, Helbig KL, Freytag S, Sullivan J, Zeev BB, Nissenkorn A, Tzadok M, Heimer G, Shinde DN, Rezazadeh 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.
22. Griffin NG, Cronin KD, Walley NM, Hulette CM, Grant GA, Mikati MA, LaBrecche 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).
23. **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.
24. 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.
25. 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.
26. Depondt C, **Heinzen EL**, Goldstein DB. *Reply to Two patients with TNK2 mutations and late-onset infantile spasm*. Ann Neurol. 2016 Dec 15;
27. 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.
28. 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.
29. 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.
30. 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.
31. 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 DNMI impair synaptic vesicle endocytosis*. Neurol Genet. 2015 Apr 17;1(1):e4.
32. 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.
33. EpiPM Consortium. *A roadmap for precision medicine in the epilepsies*. Lancet Neurol. 2015 Dec;14(12):1219-28.
34. 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.
35. 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.
36. **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.
37. 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

38. EuroEPINOMICS-RES Consortium; Epilepsy Phenome/Genome Project; Epi4K Consortium. *De Novo Mutations in Synaptic Transmission Genes Including DNMI Cause Epileptic Encephalopathies*. *Am J Hum Genet*. 2014 Oct 2;95(4):360-70.
39. 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.
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. 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.
43. 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, Rivielo 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.
44. Epi4K Consortium and Epilepsy Phenome/Genome Project. *De novo mutations in epileptic encephalopathies*. *Nature*. 2013 Sep 12;501(7466):217-21.
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. 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
47. 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.
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. 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.
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. 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;91(2):303-12.
52. **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.
53. The Epi4K Consortium. *Epi4K: Gene discovery in 4,000 genome*. *Epilepsia*. 2012 Aug;53(8):1457-1467.

54. Poduri A, Evrony GD, Cai X, Elhosary PC, Hills L, Beroukhir 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.
55. 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.
56. 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.
57. 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.
58. 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. *SVA: Software for Annotating and Visualizing Sequenced Human Genomes*.
59. 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.
60. 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.
61. 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.
62. 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.
63. Kasperavičiūtė 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.
64. 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.
65. **Heinzen EL**[‡], Radtke RA[‡], Urban TJ[‡], Cavalleri GL, Depondt C, Need AC, Walley NM, Nicoletti P, Ge D, Catarino CB, Duncan JS, Kasperavičiūtė 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.
66. 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

67. **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.
68. 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.
69. 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.
70. Need AC, Ge D, Weale ME, Maia J, Feng S, **Heinzen EL**, Shianna KV, Yoon W, Kasperaviciute 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.
71. **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.
72. 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.
73. **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 Biology* 2007; 8: R32.
74. **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*. *American Journal of Human Genetics* 2007; 80: 876-883.
75. **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.
76. **Heinzen EL** and Pollack GM. *Pharmacodynamics of morphine-induced neuronal nitric oxide production and antinociceptive tolerance development*. *Brain Res* 2004; 1023:175-184.
77. **Heinzen EL** and Pollack GM. *The development of morphine antinociceptive tolerance in nitric oxide synthase-deficient mice*. *Biochemical Pharmacology* 2003; 67, 735-741.
78. **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.
79. **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.
80. Naderer OJ, Dupuis RE, **Heinzen EL**, Wiwattanawongsa K, Johnson MW, Smith PC. *Influence of antibiotics on the disposition of mycophenolic acid in healthy volunteers*. *J Clin Pharmacol* 2005; 45: 219-226.
81. 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.

Book Chapters

Ruzzo EK, Radtke RA, Goldstein DB, and **Heinzen EL**. *Practical Epilepsy: Genetics*; Aatif Husain, Demos Medical Publishing, New York City; 2016; 11-27.

Abstracts

1. Lai D, Sosicka P, Ressler AK, Boland MJ, Freeze HH, **Heinzen EL**. *Characterizing the Mechanisms of SLC35A2 Epilepsy in Human iPSC-Derived Neurons*. *ASHG*, 2019.
2. Alkelai, Greenbaum L, **Heinzen EL**. *A New Pipeline Designed for Reliable and Accurate Short Tandem Repeat Realignment*. *ASHG*, 2019.
3. CSNK2B Working Group. *CSNK2B: A Novel Cause of Neurodevelopmental Disease and Epilepsy*. *ACMG*, 2019.

4. 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. *AES*, 2019.
5. 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. *AES*, 2019.
6. 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.
7. **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 Meeting*, 2018.
8. Goldstein D, **Heinzen EL**, Wapner R, Anyane-Yeboah 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. *ACMG*, 2018.
9. Alkelai A, Greenbaum L, **Heinzen EL**, Goldstein D, Lerer B. Potential Role of Rare Variants in the Genetics of Tardive Dyskinesia. *ASHG*, 2018.
10. 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. *AES*, 2019.
11. **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). *AES*, 2018
12. 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, McBrien 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**. *AES*, 2018.
13. Song S, Dhindsa R, Williams D, **Heinzen EL**, Goldstein D, Boland M. Modeling an Epilepsy-Associated Cortical Malformation Using hiPSC-Derived Organoids. *J. Neurosci*, 2018.
14. Poduri A, Winawer MR, Crino PB, **Heinzen EL**. Somatic Mutation in SLC35A2 Leads to Focal Epilepsy. *J. Neurosci*, 2018.
15. 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.
16. **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
17. Mulhern M, Lippa L, Ernst M, Bazil C, Rivello J, Stong N, **Heinzen EL**, Goldstein D. Review of Previous Genetic Testing in a Cohort of Neurology Patients with Genetic Diagnoses Made through Research Whole Exome Sequencing. *ACMG*, 2017.
18. 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. *ACMG*, 2017.
19. 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. *ASHG*, 2017.
20. Griffin N, Yang E, Garcia-Tarodo S, Von Allmen G, Bhattacharjee M, Grant G, Porter BE, Mikati MA, Walley N, Muh CR, LaCoursiere C, Keever K, Shain C, Crino PB, Poduri A, **Heinzen EL**. Exome Sequencing of a Pediatric Refractory Status Epilepticus Cohort (the pSERG Cohort). *AES*, 2017.

21. 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. *AES*, 2017.
22. **Heinzen, EL**. De novo and inherited variants in MAP1B in periventricular nodular heterotopia. *AES*, 2017.
23. 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. *AES*, 2017.
24. McSweeney K, **Heinzen EL**, Boland M, Goldstein D. Mutations Causing Alternating Hemiplegia of Childhood Disrupt Normal Neural Network Activity Patterns. *J Neurosci*, 2017.
25. 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. *AES*, 2016.
26. 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. *ASHG*, 2016.
27. 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 Epilepsy. *AES*, 2015.
28. 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). *AES*, 2014.
29. 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. *AES*, 2014.
30. **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.
31. **Heinzen EL**, Poduri A, Epi4K and EPGP Investigators. Trio Sequencing in Malformations of Cortical Development. *ASHG*, 2013.
32. 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. *AES*, 2013.
33. Poduri A, Evrony GD, Cai X, Elhosary PC, Beroukhir 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. *AES*, 2012.
34. 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. *ASHG*, 2012.
35. Poduri A, Evrony GD, Cai X, Elhosary PC, Beroukhir 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. *ASHG*, 2012.
36. **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. *ASHG*, 2012.
37. 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. *ASHG*, 2012.
38. 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. *AES*, 2010..
39. **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 Myoclonic Epilepsy Patients. *AES*, 2010.
40. 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. *AES*, 2009.
41. 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.
42. **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. *ASPET*, 2005.

43. **Heinzen EL**, Booth RG, Pollack GM. Morphine-induced Neuronal Nitric Oxide (NO) Production Modulates Constitutive Activit. *AAPS*, 2004.
44. **Heinzen EL**, Pollack GM. The Role of Nitric Oxide as a Mediator of Morphine Antinociceptive Tolerance. *AAPS*, 2003.
45. **Heinzen EL**, Pollack GM. Pharmacokinetics and Pharmacodynamics of L-Arginine in Rats: A Model of Stimulated Nitric Oxide Synthesis. *AAPS*, 2002.
46. 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.
47. Wiwattanasongsa K, **Heinzen EL**, Dupuis RE, Smith PC. Influence of Antibiotics on the Disposition of Mycophenolic Acid in the Guinea Pig. *AAPS*, 1997.

Invited Presentations

Regional

1. 2020 Invited Speaker, Discovery and functional characterization of *SLC35A2* epilepsy, UNC Neuroscience Seminar Series, Chapel Hill, NC.
2. 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.
3. 2019 Invited Speaker, Identification and functional characterization of somatic variants in epilepsy, Department of Neurology, Columbia University Medical Center, New York, NY.
4. 2018 Invited Speaker, Pediatric status epilepticus research meeting, Exome sequencing of a pediatric status epilepticus cohort, Boston Children's Hospital, Boston, MA.
5. 2017 Invited Speaker, Med into Grad Symposium, Elucidating the genetic basis of malformations of cortical development, Columbia University, New York, NY.
6. 2017 Invited Speaker, The expanding de novo paradigm in severe sporadic epilepsies, Langone Comprehensive Epilepsy Center, New York University, New York, NY.
7. 2013 Invited Speaker, The role of post-zygotic mutations in malformations of cortical development, Human Genetics Evening. Duke University School of Medicine, Durham, NC.
8. 2012 Guest Lecturer, 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.
9. 2008 Invited Speaker, Genetic Regulation of Splicing & Expression in Human Tissue, Affymetrix Integrated Genomics Seminar Series, Durham, NC.

National

1. 2019 Invited Speaker, Paving the way towards precision medicine in epilepsy, St Jude Children's Research Hospital, Memphis, TN.
2. 2019 Invited Speaker, Latest Development in Epilepsy Genetics, Epigen Annual Meeting, Dulles, Virginia.
3. 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.
4. 2019 Invited Speaker, Progress towards precision medicine in epilepsy, Cincinnati Children's Hospital, Cincinnati, OH.
5. 2018 Invited Speaker, ATP1A3 in Disease, Genetic bases of epilepsy disorders and relevance to Alternating Hemiplegia of Childhood, Northwestern University, Chicago, IL.
6. 2017 Invited Speaker, De novo and inherited variants in MAP1B in periventricular nodular heterotopia, American Epilepsy Society Annual Meeting, Washington, DC.
7. 2017 Invited Speaker, Identifying Regulatory Mutations that Influence Neuropsychiatric Disease, Whole Genome Sequencing for Psychiatric Disorders Consortium Meeting, Boston, MA.
8. 2016 Invited Speaker, A Multi-electrode Array Approach to Understanding the Effects AHC-causing mutations, Alternating Hemiplegia of Childhood Family Meeting, Indianapolis, IN.
9. 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.
10. 2014 Session chair, Epilepsy Genetics in the Era of Precision Medicine Conference, Precision Diagnostics I – Genetic Causes of Epilepsy, Half Moon Bay, CA.

11. 2013 Invited speaker and panelist, Genetics of Catastrophic Infantile Epilepsies: From Gene Discovery to Practical Clinical Applications, American Epilepsy Society Annual Meeting, Washington, DC.
12. 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
13. 2013 Invited Speaker, Somatic mutations in malformations of cortical development, Human Genetics and Genomics Gordon Research Conference, Smithfield, RI.
14. 2012 Invited Speaker and panelist. Investigator’s Workshop, Massively Parallel Sequencing in Epilepsy, American Epilepsy Society Annual Meeting, San Diego, CA.
15. 2007 Invited Speaker, Cambridge Healthtech Institute, New applications for microarray data analysis, Integrating genetics with “omics”
16. 2006 Invited Speaker, Alternative ion channel splicing in mesial temporal lobe epilepsy and Alzheimer's disease, Splicing 2006 annual symposium

International

1. 2019 Speaker, Epilepsy genetics and the role Somatic mosaicism, Third Workshop on Ring14 Syndrome, Franciacorta, Italy.
2. 2017 Speaker, Challenges and opportunities in somatic genetics. AstraZeneca Genomic Partners Workshop. Cambridge, England.
3. 2016 Invited Speaker, Somatic mutations in Hypothalamic Hamartoma, International Hypothalamic Hamartoma Symposium, London, England.
4. 2014 Speaker, How it all started: ATP1A3 as the major gene for AHC, Symposium of ATP1A3 in Disease. Lieden, Netherlands.
5. 2013 Speaker, Genetic variation in *ATP1A3* in neurological, developmental and psychiatric diseases, Symposium of ATP1A3 in Disease, Rome, Italy.
6. 2012 Speaker, Symposium of ATP1A3 in Disease, Brussels, Belgium.
7. 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.
8. 2010 Speaker, XVIII World Congress on Psychiatric Genetics, Symposium on Genetic control of expression in human brain, Athens, Greece.

Educational Contributions

Teaching

- Seminar in Biotechnology*, GU4305, Columbia University, 2019
- Analysis of genetic variation in humans*, Genomic Approaches II, Columbia University, 2017, 2018
- Pharmacogenetics*, Introduction to Precision Medicine, Columbia University, 2017
- Next generation sequencing/bioinformatic tools for genomics*, Introduction to Precision Medicine, Columbia University, 2017, 2018
- Molecular diagnosis of neuromuscular*, Introduction to Neuromuscular Diseases, Columbia University, 2016-2019
- Pharmacogenetics*, Molecular Pharmacology, Columbia University, 2016-2018
- Application of next-generation sequencing in complex diseases*, Human Genetics, Duke University, 2012
- Pharmacogenetics in neurological diseases*, Pharmacogenetics, Duke University, 2011- 2014
- Epilepsy Pharmacogenetics*, Pharmacogenetics, Doctor of Pharmacy Curriculum, University of North Carolina at Chapel Hill, School of Pharmacy, 2010
- Complex Trait Genetics*, IGSP/NSF Explorations in the Genomes Sciences, Undergraduate Summer Fellowship Program, Duke University, 2008-2009
- Stepwise Nonlinear Regression*, Advanced Pharmacokinetics I, Graduate School Curriculum, University of North Carolina at Chapel Hill, School of Pharmacy, 2002
- Kinetics of Pharmacological Response*, Clinical Pharmacokinetics I, Doctor of Pharmacy Curriculum University of North Carolina at Chapel Hill, School of Pharmacy, 2002
- Pharmacokinetic Alterations in Pregnancy*, Clinical Pharmacokinetics II, Doctor of Pharmacy Curriculum, University of North Carolina at Chapel Hill, School of Pharmacy, 2003

Effect Compartment Modeling, Advanced Pharmacokinetics II, Graduate School Curriculum, University of North Carolina at Chapel Hill, School of Pharmacy, 2002

Convolution/Deconvolution, Advanced Pharmacokinetics II, Graduate School Curriculum, University of North Carolina at Chapel Hill, School of Pharmacy, 2002

Phenytoin Pharmacokinetics, Clinical Pharmacokinetics I, Doctor of Pharmacy Curriculum, University of North Carolina at Chapel Hill, School of Pharmacy, 2001

Drugs of Addiction, Undergraduate Sports Medicine Class, Guilford College, April 2001

Short course on Integrated and Organ Systems Pharmacology, University of North Carolina at Chapel Hill, School of Pharmacy, July 2005-2007, Lectures and demonstrations of rat venous cannulations and *in vivo* microdialysis

Advising and Mentorship

2018 – present	Dulcie Lai, PharmD., Ph.D, Postdoctoral fellow
2018 - 2019	Shih Hua Yu, Masters in Data Science student, Internship Advisor
2017 – present	Jinfeng Lu, Ph.D., Postdoctoral fellow
2017	Ceylan Hasan, Biotechnology Masters student in the Biological Sciences Department at Columbia University, Thesis advisor
2016	Rami Kathuda, Biotechnology Masters student in the Biological Sciences Department at Columbia University, Thesis advisor
2013 – 2018	Nicole G. Griffin, Ph.D, Postdoctoral fellow

Community Education

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

Present

September 2016 – June 2021	Principal investigator [MPI], NIH, NINDS, 1R01NS094596, Identification and molecular characterization of somatic mutations in malformations of cortical development
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Pending

July 2020 – June 2025	Principal investigator, [MPI], NIH, NINDS, R01, Defining disease mechanisms in SLC35A2 epilepsy
July 2020 – June 2025	Principal investigator, [MPI], NIH, NINDS, R01, Somatic Mutation in Intractable Focal Epilepsy
July 2020 – June 2022	Principal investigator, NIH, NINDS, R03, Single cell transcriptomic profiling of mosaic human brain tissue

Past

May 2014 – Dec 2019	Principal investigator [MPI], Citizens United for Research in Epilepsy (CURE). Epilepsy Genetics Initiative
September 2015 – July 2019	Co-investigator, NIH, NINDS, 1R01NS089552, Discovery of novel molecular abnormalities underlying non-lesional focal epilepsy. PIs: Melodie Winawer/Peter Crino
August 2016 – July 2018	Principal investigator, AstraZeneca UK Limited - Genomics Alliance Agreement
September 2011 – July 2018	Principal investigator [MPI], NIH, NINDS, 1U01NS077303, 3 OF 7 Epi4K: Sequencing, Biostatistics & Bioinformatics Core
January 2015 - December 2017	Principal investigator, Cure AHC, Gene discovery in Alternating Hemiplegia of Childhood/Multi-electrode array screening for Candidate Drugs in ATP1A3 Mutations D801N & E815K
March 2012 – February 2014	Principal investigator, NIH, NINDS, 1R21NS078657, Exploratory genomic investigations of mesial temporal lobe epilepsy.
September 2009 – August 2012	Co-investigator, NIH, NINDS, 5RC2NS070344, Whole-genome sequencing in multiplex epilepsy families, PIs: David Goldstein/Ruth Ottman
July 2007 – July 2008	Principal investigator, Kathleen Price Bryan Alzheimer's Disease Research Center Pilot Grant Funding

Academic Service*Local*

2020	Committee Member, Division of Pharmacotherapy and Experimental Therapeutics Graduate Program Search Committee
January 2016	Chair, Basic Science Faculty Search Committee, Columbia University Medical Center, Institute for Genomic Medicine
January 2016 - December 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
August 2006 – August 2007	Scientific consultant, NIH Short Course on Integrated and Organ Systems Pharmacology, University of North Carolina at Chapel Hill, School of Pharmacy
April 2004 - September 2005	Grant application reviewer, United States Environmental Protection Agency/University of North Carolina Toxicology Research Program

National/International

March 2019	American Association for the Advancement of Science, Review Panel, International Collaboration Grant - Saudi Arabian Ministry of Education's Research Development Office
August 2018 – present	ILAE Consortium on Complex Epilepsies Task Force of the Commission on Genetics
January 2017	Department of Defense, Review Panel, Peer Reviewed Medical Research Program - Epilepsy Research Program - Idea Development
December 2016 – present	Epi25 Consortium, Steering and Analysis Committees
July 2016	NINDS Study Section, Review Panel, Centers Without Walls for Collaborative Research in the Epilepsies: Developing Transformative Therapies for Modifying or Preventing Epilepsy
November 2015	Organizer, Banbury Meeting, Scientific and Clinical Foundation for Precision Medicine in Epilepsy
August 2015	Scientific Organizer, Symposium of ATP1A3 in Disease
January 2015 - present	Epilepsy Precision Medicine (EpiPM) Consortium, Steering Committee
April 2014 - present	Epilepsy Genetics Initiative (EGI), Steering Committee
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 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
January 2004 - December 2006	Awards Committee for the Pharmacokinetics, Pharmacodynamics, and Drug Metabolism (PPDM) Section of the American Association of Pharmaceutical Sciences

Professional Organizations and Societies*Memberships*

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	Epilepsy Research	Nature Communications
Annals of Neurology	Genome Biology	Nature Genetics
American Journal of Human Genetics	Journal of Pharmacology and Experimental Therapeutics	Neuron
Brain Research	Molecular Case Studies	Pediatrics
Cell Systems Reviews	Molecular Pharmacology	PLoS Genetics
Epilepsia		PLoS Biology

Editorial Boards

2010-2011	Associate Editor Journal of Alzheimer's Disease
2016	Guest Editor, PLoS Genetics