Elena Marie Kessler (Infante)

Education

University of Pittsburgh, Pittsburgh, PA **2008-2010**
Master of Science, Genetic Counseling, Department of Human Genetics
Thesis: Phenotypic and Genetic Analysis of Indices of Liability to Addiction

Westminster College, New Wilmington, PA **2004-2008**
Bachelor of Science, Biology – *Magna Cum Laude*

Certifications

Certified Genetic Counselor, American Board of Genetic Counseling, August 19, 2011 - present
Licensed Genetic Counselor, Commonwealth of Pennsylvania, December 23, 2013 - present

Work Experience

Pediatric Genetic Counselor, departments of Genetics and Genomics and Hematology/Oncology
Children's Hospital of Pittsburgh of University of Pittsburgh Medical Center
February 4, 2013 – present
Director: Gerard Vockley, MD, PhD

Genetic Counselor and Research Coordinator, department of Gastroenterology
University of Pittsburgh
July 12, 2010 - January 24, 2013
Director: Randall Brand, MD

Research Coordinator, Center for Education and Drug Abuse Research, University of Pittsburgh
September 1, 2008 - July 11, 2010
Director: Michael Vanyukov, PhD
Study Coordinator of the Pittsburgh Registry of Infant Multiplets

Academic Experience

University of Pittsburgh, Adjunct Faculty Appointment (July 2010-present)
Genetic Counseling Program, Graduate School of Public Health

- HUGEN 2035 and HUGEN 2038 - Principles of Genetic Counseling and Intervention Skills for Genetic Counselors
  Provide yearly lectures on pediatric genetics, pediatric oncology
  Previously provided lectures on metabolic genetics, clinical internship, supervising students, post graduate school, certification, job placement, national meetings, societies
- HUGEN 2036 - Genetic Counseling Internship
  Provide clinical supervision to 11 genetic counseling students for a 10-month period annually.
  Provide mentoring, admission interviews, and administration of oral examinations.
- HUGEN 2047 - Clinical Genetics Case Conference
  Provide supervision and mentoring of genetic case presentations in the Fall and Spring terms for second year genetic counseling students, participate with own presentations
- Genetic Counseling Program Master's Thesis advisor for one genetic counselling student (currently Oncology project, previously Sickle cell project)
- Interviewer for University of Pittsburgh Genetic Counseling program applicants
- Member of University of Pittsburgh Genetic Counseling Program Advisory Board, 2014-2020
- Member of University of Pittsburgh Genetic Counseling Program Reaccreditation Unit, Clinical Training Evaluation Committee, 2018-present

Doctor of Physical Therapy program

- "Genetics and Physical Therapy" – yearly lecture, 2016 - present

Westminster College, New Wilmington, PA

- Biology department Seminar series “Genetic Counseling; a dynamic and gratifying field,” April 12, 2019; “Genetic Counseling: An Expanding Career Field,” February 10, 2017; “Is Genetic Counseling a Profession For You?” October 23, 2009
- Panelist for Professional Networking Symposium, “Science Careers,” April 1, 2017
- Invited speaker for young women’s organization Zeta Tau Alpha. “Hereditary Breast Cancer: Awareness, Proactivity, and Support,” November 16, 2010

Peer Reviewed Literature

- Ernst ME, Baugh EH, Madan-Khetarpal S, **Infante E,** et al. (2021) *CSNK2B*: A broad spectrum of neurodevelopmental disability and epilepsy severity. *Epilepsia.*
- Marbach F, Schaaf C, Sklirou E, **Kessler E**, et al. (2021) Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia and insensitivity to pain. *Genetics in Medicine*.
- Hansen A, Madan-Khetarpal S, **Infante E,** et al. (2020) Phenotypic Expansion in *KIF1A*-related Dominant Disorders: A Description of Novel Variants and Review of Published Cases. *Human Mutation.* 41(12): 2094-2104.
- Johnson BV, Kumar R, Madan-Khetarpal S, **Infante E,** et al. (2020) Partial loss of *USP9X* function leads to a male neurodevelopmental and behavioural disorder converging on TGFβ signalling. *Biol Psychiatry*. 15;87(2): 100-112.
- Nielsen M, **Infante E**, Brand R. *MUTYH* Polyposis. 2012 Oct 4 [Updated 2019 Oct 10]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. Available from: https://www.ncbi.nlm.nih.gov/books/NBK107219/
- Venier RE, Maurer LM, **Kessler E**, et al. (2019) A germline *BARD1* mutation in Ewing Sarcoma: implications for familial testing and counseling. *Peds Blood & Cancer*. 66(9):e27824.
- Aarabi M, **Kessler E**, Madan-Khetarpal S, Yatsenko S et al. (2018) Autism spectrum disorder in females with *ARHGEF9* alterations and a random pattern of X chromosome inactivation. *EJMG*. S1769-7212(18)30285-4.
- Marcogliese P, Shashi V, Ortiz D, **Infante E** et al. (2018) *IRF2BPL* Is Associated with Neurological Phenotypes. *AJHG*. 103(2): 245-260.
- Van Dijck A, et al. (2018) Clinical presentation of a complex neurodevelopmental disorder caused by mutations in *ADNP*. *Biological Psychiatry*. ADNP consortium member and contributor.
- Bostwick B, McLean S, Madan-Khetarpal S, **Infante E**, et al. (2017) Phenotypic and Molecular Characterization of *CDK13*-Related Congenital Heart Defects, Dysmorphic Facial Features and Intellectual Developmental Disorders. *Genome Medicine.* 14;9(1): 73-81.
- Hu, J Ou Z, **Infante E**, Kochmar S, Madan-Khetarpal S, Hoffner L, Parsazad S, Surti U. (2017) Chromosome 12q13.13q13.13 Microduplication and Microdeletion: A Case Report and Literature Review. *Molecular Cytogenetics.* 10: 24.
- **Infante E**, Alkorta-Aranburu G, and El-Gharbawy A. (2017) Rare Form of Autosomal Dominant Familial Cornelia de Lange Syndrome due to a Novel Duplication in *SMC3*. *Clinical Case Reports*. 5(8): 1277-1283.
- Makadia P, Srinath A, Madan-Khetarpal S, McGuire M, **Infante E**, Zhang J, Felgar RE, Davis AW, Chong HJ, Windreich RM. (2017) Aplastic anemia and cytotoxic T lymphocyte antigen-4 haploinsufficiency treated with bone marrow transplantation. *The Journal of Allergy and Clinical Immunology: In Practice*. 5(5): 1445-1447.
- Tokita, Mari J., **Infante E**, et al. (2016) De Novo Truncating Variants in *SON* Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. *The American Journal of Human Genetics.* 99(3): 720-727.
- **Infante E et al.** NSGC Cancer SIG Pediatric Cancer Syndrome Fact Sheets, published to NSGC website January 16, 2014
- **Infante E**. (2012) Confusion and Strength. *Journal of Genetic Counseling*. 21(2):199-200.

Poster Presentations

- Wimmer, K, Meade J, **Kessler EM.** Constitutional POLE variants known to be somatic driver mutations in cancer cause a phenotype reminiscent of Constitutional Mismatch Repair Deficiency, SIOP Europe 2021, abstract presentation, (2021).
- Venier RE, Grubs R, **Kessler EM**, Meade J, Bailey KM. Evaluation of barriers to referral for cancer predisposition syndromes in pediatric oncology patients, ACMG Annual Meeting, virtual poster, (2021).

- **Kessler E** and Meade J. Development and Outcomes of a Multidisciplinary Pediatric Cancer Predisposition Program in its first two years, NSGC Annual Education Conference, poster presentation, (2019).
- Venier RE, **Kessler EM**, Meade J, Bailey KM. Somatic Tumor Testing Identifies Germline *BARD1* Mutation in a Patient with Ewing Sarcoma: Implications for Familial Testing and Genetic Counseling, NSGC Annual Education Conference, poster presentation, (2019).
- Amodei K, **Kessler E,** Meade J, McAllister-Lucas L, Chang W. MD Screening Tool for Pediatric Cancer Predisposition Reveals Low Rate of Referral in At-Risk Population, Children's Oncology Group Annual Meeting Young Investigator poster session, (2019).
- **Kessler E** and Meade J. Development and Outcomes of a Multidisciplinary Pediatric Cancer Predisposition Program in its first two years, 2nd Annual Pitt Genetics retreat, poster presentation, (2019).
- Russell C, Hillery C, Durst A, **Kessler E.** Assessment of Hemoglobin Trait Notification in Western Pennsylvania Newborn Screening, American Public Health Association Annual meeting, poster presentation, (2019).
- Russell C, Hillery C, Durst A, **Kessler E**. Assessment of Hemoglobinopathy trait notification in Pennsylvania Newborn screening, University of Pittsburgh Graduate School of Public Health Dean's Day, poster presentation, (2019).
- ***CSNK2B* Working Group**, Ernst M. *CSNK2B:* A Novel Cause of Neurodevelopmental Disease and Epilepsy, ACMG Annual Meeting, poster presentation, (2019).
- Aarabi M, Baumann J, **Kessler E** et al. Genotype-phenotype correlation of patients with copy number variants in 2q12.3-2q13 region reveals new hotspots for pathogenic variants, ACMG Annual Meeting, poster presentation, (2019).
- Russell C, Hillery C, Durst A, **Kessler E**. Assessment of Hemoglobinopathy trait notification in Pennsylvania Newborn screening, University of Pittsburgh Health Disparities competition, poster presentation, (2019).
- Jolly L, **USP9X Neurodevelopmental Disorder Consortium**, et al. *USP9X* missense variants associated with neurodevelopmental disorders disrupt signaling pathways critical to brain development, Society for Neuroscience Annual meeting, poster presentation, (2018).
- Besnard T, Ebstein F, Fuqua L, **Infante E** et al. Ubiquitin-proteasome system impairment and intellectual disability: the *CUL4B* example, ESHG Annual Conference, poster presentation, (2018).
- Spillmann R, Stong N, Mokry J, Ortiz D, **Infante E**, et al. Nonsense Variants in the Gene *IRF2BPL* are Associated with a Neurodegenerative Course, ACMG Annual Meeting, poster presentation, (2018).
- Besnard T, Ebstein F, Lefebvre M, **Infante E**, McGuire M, Küry S et al. Déficience intellectuelle et système ubiquitine-protéasome: l'exemple de *CUL4B.* les Assises de Génétique Humaine et Médicale, poster presentation, (2018).
- Bostwick B, McLean S, Madan-Khetarpal S, **Infante E**, et al. A Growing Need for 'Reverse Clinical Genomics:' Demonstrated by Phenotypic Characterization of *CDK13*-Related Disorders, ASHG Annual Meeting, poster presentation, (2017).
- **Infante E**, Alkorta-Aranburu G, and El-Gharbawy A. Familial Cornelia de Lange Syndrome due to a Novel Duplication in the *SMC3* Gene, NSGC Annual Education Conference, poster presentation, (2017).
- Aarabi M, Madan-Khetarpal S, **Kessler E**, et al. Intragenic ARHGEF9 microdeletion in Xq11.1 is associated with global developmental delay and neurobehavioral problems in Females. ACMG Annual Clinical Genetics Meeting, poster presentation, (2017).
- Madan-Khetarpal S, **Infante E**, et al., Mitigating the cost of the diagnostic odyssey through exome sequencing. ACMG Annual Clinical Genetics Meeting, poster presentation, (2015).
- **Infante, E**, Binion D, Baidoo L, Brand R, Regueiro M. Introduction of Genetic Counseling Services to an IBD Clinic, Crohn's and Colitis Foundation of America's Annual Professional meeting, poster presentation, (2011).