UTGCP Annual Research Newsletter

May 2022 | Issue No. 3

what's inside

- *classes of 2022 and 2023 thesis research*
- *alumni & faculty research & publications from 1/2020-12/2021, hyperlinked when available*

Support UTGCP student research

Please consider financially supporting our students' research efforts through the MD Anderson UT Health Graduate School by selecting "Genetic Counseling Research & Education Endowed Fund" as the designation

Congrats to the class of 2022!

If you would like to support student research efforts of the UTGCP through our Research & Education Fund, please

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M.S. Thesis Topics

Émile Moura Coelho Da Silva

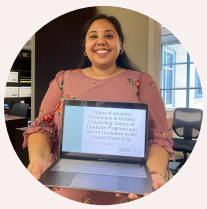
Experiences of racial and ethnic minority patients with genetic counseling

Advisors: Myla Ashfaq & Brittanie Morris

Emilyn Banfield

Analysis of Huntington disease caregiver quality of life using the Enroll-HD population

Advisor: Leslie Dunnington



Gina Sanchez

Status of abortion curriculum in genetic counseling: Survey of graduate programs and recent graduates in the United States

Advisor: Aarti Ramdaney

Jenna Lea

Non-invasive prenatal screening (NIPS) testing motivations and informed decision making in the low-risk population

Advisor: Meagan Choates

Jenny Do

Parental stress in Tuberous Sclerosis Complex

Advisor: Kate Mowrey

Katie Baudoin

Investigating the attitudes of the d/Deaf community towards genetic counseling and the utilization of genetic services

Advisor: Jennifer Czerwinski & Chelsea Wagner

Mandy Chamberlain

Perceptions and roles of genetic counselors in perinatal palliative care: Where are we now?

Advisors: Callie Diamonstein & Theresa Wittman

Michelle Appel

Prenatal testing decisions and motivations in pregnancies conceived via IVF

Advisors: Aarti Ramdany & Lauren Murphy

Michelle Zelnick

Assessing the impact of Camp PHEver on serum phenylalanine levels

Advisor: Kate Mowrey

Natalie Stoner

Invisible diversities, academic capital, and competitiveness of genetic counseling applicants

Advisor: Claire Singletary

Introducing the Research of the Class of 2023!

M.S. Thesis Topics

Latonya Alexander: Pregnant Black/African Americans and Their Experiences in Prenatal Genetic Counseling.
Advisor: Claire Singletary

Maddi Alpar: The Impact of Clinical Expectations and Psychosocial Concerns on the Diagnostic Odyssey of Hypermobile Ehlers-Danlos Syndrome (hEDS) Patients.
Advisor: Katie Leal

Erin Atkinson: Perceived Barriers to Cascade Testing of At-Risk Relatives Located in Latin America.
Advisors: Amanda de Leon and Aranza Gonzalez Cendejas

Yusra Aziz: What topics related to racial and ethnic health disparities (REHD) should be taught in genetic counseling programs?
Advisor: Claire Singletary

Jack Colleran: Perceived Utility of Genetic Carrier Screening in Diverse Patient Populations.
Advisor: Blair Stevens

Emily Daykin: Investigation of Teratogen Concerns in the Covid-19 Era.
Advisor: Myla Ashfaq

Tessa Heller: The Impact of Palynziq on Quality of Life for Individuals with PKU.
Advisor: Kate Mowrey

Cindy Hernandez: Monolingual Spanish Speaking Patient Satisfaction and Comfort when Comparing Spanish Speaking Provider to an Interpreter.
Advisor: Katie Leal

Jasmine Tyson: The Impact of Restrictive Abortion Legislation Tactics on the Trust Reproductive-Aged Individuals Assigned Female at Birth Have in their Healthcare Providers.
Advisors: Aarti Ramdaney and Samantha Montgomery

Jordan Zeiger: Saliva Kit Returns for Hereditary Cancer Genetic Testing After Genetic Counseling.
Advisor: Maureen Mork

UTGCP Alumni & Faculty: Authorship at National Meetings

Submitted presentations from 2020-2021; only UT affiliated authors listed

American Academy of Pediatrics National Conference & Exhibition 2020

Taylor Beecroft - Outcomes of Branch Pulmonary Artery Stenosis in Williams Syndrome and Non-Williams Supravalvar Aortic Stenosis. Accepted for oral presentation at American Academy of Pediatrics National Conference and Exhibition, October 2020.

American Academy of Pediatrics National Conference & Exhibition 2021

Taylor Beecroft -Outcomes of Branch Pulmonary Artery Stenosis in Williams Syndrome and Non-Williams Supravalvar Aortic Stenosis. Accepted for poster presentation at American Academy of Pediatrics National Conference and Exhibition, March 2021.

American College of Cardiology

Taylor Beecroft - Vertebral Artery Tortuosity Is A Biomarker For Arterial Events In Children And Young Adults With Vascular Ehlers-Danlos Syndrome. Accepted for poster presentation at the American College of Cardiology Scientific Sessions, March 2020.

American College of Gastroenterology Meeting

Taylor Beecroft - Improving Identification of Patients Meeting Lynch Syndrome Testing Criteria in Endoscopy. Blaisdell et al. 2021 American College of Gastroenterology Meeting, Presidential Poster Award. 2021 Oct.

American College of Medical Genetics & Genomics

Katie Sagaser & Katie (Rock) Forster - Not just a carrier: hypophosphatasia (HPP) presentations of 12 ALPL heterozygotes identified through routine expanded carrier screening (ECS). Oral presentation; recipient of 2020 Carolyn Mills Lovell Genetic Counselor Award.

Katie Sagaser & Katie (Rock) Forster - A case of apparently non-mosaic trisomy 7: prenatal natural history in the midtrimester. Poster presentation, 2020

Haley Streff, Amanda Gerard, & Andi Lewis - Outcomes Of Insurance Pre-authorization Requests For Genomic Testing In An Outpatient Pediatric Genetics Clinic [Abstract]. Poster presentation, 2020.

American Heart Association

Taylor Beecroft - Cardiovascular outcomes in Williams syndrome and non Williams supravalvar aortic stenosis. Accepted for poster presentation, 2020.

American Society of Clinical Oncology Meeting

Kate Principe, Trisha Nichols, & Ann Bunnel - Germline testing in community oncology patients with somatic BRCA1/2 mutations. American Society of Clinical Oncologists Annual Meeting, 2021.

Amie Blanco - Streamlining the genetics pipeline to increase testing for patients at risk for hereditary prostate cancer. American Society of Clinical Oncology Meeting, 2020.

UTGCP Alumni & Faculty: Authorship at National Meetings

Submitted presentations from 2020-2021; only UT affiliated authors listed

American Society of Human Genetics

Haley Streff, Amanda Gerard, & Andi Lewis - Outcomes of insurance pre-authorizations and influence of ICD-10 codes in pediatric genetics clinics. Poster at ASHG 2020.

American Society of Reproductive Medicine Scientific Congress & Expo

Sandra Darilek - When preimplantation genetic testing for aneuploidy (PGT-A) results lead to discovery of an unexpected chromosome abnormality in a patient. Poster presentation, 2021.

Annual Meeting of the Collaborative Group of the Americas on Hereditary Gastrointestinal Cancer 2020

Amie Blanco - Founder CDKN2A variant in individuals of Hispanic ancestry – Implications for Pancreatic Cancer Surveillance. 2020 Nov 14-15.

Amie Blanco - Clinical characteristics of siblings with bi-allelic MSH3 pathogenic variants. 2020 Nov 14-15.

Amie Blanco - Are you SURE? Decisional Conflict in Patients with Pancreas Cancer Presenting to the Genetic Testing Station. 2020 Nov 14-15.

Annual Meeting of the Collaborative Group of the Americas on Hereditary Gastrointestinal Cancer 2021

Amie Blanco - NTHL1-asociated polyposis: A case solved by multi-gene panel testing. 2021 Nov 13-15.

Amie Blanco - Creation of a cloud-based database to facilitate multi-institutional collaborative research on universal screening for Lynch syndrome: a partnership between Kintalk, UCSF, LSSN and CGA. 2021 Nov 13-15.

Amie Blanco - Does family history of pancreatic cancer in pathogenic variant carriers identify patients who are diagnosed with pancreatic cancer: Results of a multi-site collaboration. 2021 Nov 13-15.

Amie Blanco - Therapy associated polyposis: An acquired and under-recognized polyposis syndrome. 2021 Nov 13-15.

Amie Blanco - Carriers Of Cystic Fibrosis From A Diverse Background Are At An Increased Risk Of Pancreatic Cancer. 2021 Nov 13-15.

Amie Blanco - Universal tumor screening for Lynch syndrome through the lens of diversity, equity, and inclusion. 2021 Nov 13-15.

UTGCP Alumni & Faculty: Authorship at National Meetings

Submitted presentations from 2020-2021; only UT affiliated authors listed

Association of Women's Health, Obstetric, and Neonatal Nurses Annual Convention

Jen Czerwinski - Maximum Safety Zone: A Qualitative Exploration of Maternal Patient and Family Perspectives of Patient Safety, 2020.

Jen Czerwinski - Safer Culture: A Developing Roadmap to Safer Maternal Care, 2020.

CHOP Pediatric Cardiology Sessions

Taylor Beecroft - Cardiovascular Characteristics in Children with Vascular Ehlers-Danlos Syndrome. Accepted for poster presentation, January 2021

Collaborative Group of the Americas on Inherited Colorectal Cancer

Sarah Burke - Adolescent diffuse gastric cancer and Li Fraumeni syndrome: A case report. November 2021.

European Society of Human Genetics

Amanda Gerard - Delineation of the clinical and molecular spectrum or QRICH1 associated neurodevelopmental disorder [Abstract]. Poster, 2021.

Genitourinary Cancers Symposium

Amie Blanco - Streamlining the genetics pipeline to increase testing for patients at risk for hereditary prostate cancer, 2021 Feb 20.

International Society for Prenatal Diagnosis 2020

Katie Sagaser & Katie (Rock) Forster - Follow your nose: the impact of repeat nasal bone evaluation on Down syndrome risk assessment by first trimester screening. Poster, 2020 (meeting cancelled).

Katie Sagaser & Katie (Rock Forster) - More than just a carrier: ALPL heterozygotes identified through routine expanded carrier screening (ECS) display signs and symptoms of hypophosphatasia (HPP). Poster, 2020 (meeting cancelled).

Katie Sagaser - Genetic etiologies for early pregnancy renal anhydramnios: Updates from the Renal Anhydramnios Fetal Therapy (RAFT) trial. Poster, 2020 (meeting cancelled).

International Society for Prenatal Diagnosis 2021

Patti Robbins-Furman, Sandra Darilek, Salma Nassef - Prenatal Genetic Counseling and Telemedicine during the COVID-19 Pandemic. Poster, June 2021.

Patti Robbins-Furman, Sandra Darilek, Salma Nassef - The Effect of COVID-19 Pandemic and Telemedicine Counseling on Prenatal Screening and Diagnosing Procedures. Poster, June 2021.

Katie Sagaser & Katie (Rock) Forster - More than just a carrier: ALPL heterozygotes identified through routine expanded carrier screening (ECS) display signs and symptoms of hypophosphatasia (HPP). Virtual poster, 2021.

Katie Sagaser & Katie (Rock) Forster - Chromosome analysis at the time of fetoscopic laser surgery yields significant abnormalities in monochorionic (MC) twin pregnancies with twin-to-twin transfusion syndrome (TTTS). Virtual poster, 2021.

UTGCP Alumni & Faculty: Authorship at National Meetings

Submitted presentations from 2020-2021; only UT affiliated authors listed

Michigan Association of Genetic Counselors

Jessie (Omark) O'Shea - An Evolving Practice Area: Inpatient Genetic Counselor Panel. Michigan Association of Genetic Counselors Annual Education Conference, 2021.

Multidisciplinary Management of Cancers Conference

Amie Blanco - A Case Based Approach: Supportive Care Panel, 21st Multidisciplinary Management of Cancers Conference (Virtual). 2021 Mar 12-13.

National Association of Neonatal Nurses

Katie Shields, Jen Czerwinski, Syed Hashmi, Kate Mowrey, & Katie Gunther - Genetics in the NICU: Nurses' Perceived Knowledge and Desired Education. 2021 Sept 13.

National Society of Genetic Counselors, 2020

Amie Blanco - Adapting Genetic Testing Results Disclosures to Patient Preferences at the Prostate Cancer Genetic Testing Station. (Virtual), Nov 18-22 2020.

Amie Blanco - Streamlining the genetics pipeline to increase testing for patients at risk for hereditary prostate cancer: the UCSF Prostate Cancer Genetic Testing Station. (Virtual), 2020 Nov 18-22.

Amie Blanco - Measuring Decisional Conflict and Genetic Counseling Satisfaction for a Video-Based Alternative Cancer Genetic Counseling and Testing Model. (Virtual), 2020 Nov 18-22.

National Society of Genetic Counselors, 2020, cont'd

Amie Blanco - Creative Cancer Counseling: Use of Alternate Service Delivery Models to Facilitate Genetic Testing for Therapeutic Decisions: The Genetic Testing Station for Patients with Pancreatic Cancer. (Virtual) 2020 Nov 18-22.

Sandra Darilek - An Academic RVU for Genetic Counselors: Development of a Tool to Measure Productivity. Poster, Virtual Conference November 2020.

Sarah Huguenard, Sandra Darilek, & Salma Nassef - Patient Knowledge of, Attitudes Toward, and Perceived Utility of Carrier Screening in an Obstetric Setting. Poster, Virtual Conference November 2020.

Laura Hendon - Genome Sequencing and the Challenges of the Rare Diagnosis in the NICU Setting. Poster, Virtual Conference November 2020.

Jordan Berg - Mutation-Positive Rate and Genetic Testing Panels: Is Bigger Really Better? Platform Presentation, Virtual, November 2020.

Aranza Gonzelez Cendejas, Chelsea Wagner, Theresa Wittman - Implementation of genetic carrier screening in the OB population: Healthcare cost impact and recommendation adherence. Poster, Virtual, November 2020.

Chelsea Wagner, Blair Stevens, & Katie Sagaser - ECS: When does Expanded Carrier Screening become Excessive? Educational Breakout Session, Virtual, November 2020.

UTGCP Alumni & Faculty: Authorship at National Meetings

Submitted presentations from 2020-2021; only UT affiliated authors listed

National Society of Genetic Counselors, 2020, cont'd

Haley Streff - Assessment of Understanding and Satisfaction of Clinical Exome Sequencing: Video Consent vs Telephone Consent." Poster at NSGC 2020.

Katie Sagaser & Katie (Rock) Forster - Omphalocele and monosomy X: expanding the phenotype of 45,X through first trimester ultrasound. Poster, Virtual, November 2020.

National Society of Genetic Counselors, 2021

Amie Blanco - Better together: the importance of both germline genetic testing and somatic tumor profiling. (Virtual), 2021 Sept 22-26.

Sandra Darilek & Salma Nassef - Prenatal genetic counselors' practices and confidence when counseling on inconclusive NIPT results. Poster, September 2021.

Laura Hendon - Whole genome sequencing and the identification of dual genetic diagnoses in complex phenotypes in the NICU. Poster, September 2021.

Erica Bednar, Aarti Ramdaney, & Jessie (Ross) Corredor - Factors Impacting Adolescent and Young Adult Cancer Patients' Decision to Purse Genetic Counseling and Testing. Poster, September 2021.

Jordan Berg - Impact of Service Delivery Model on Test Uptake and Completion Rates in a Cancer Genetics Clinic. Poster, September 2021.

National Society of Genetic Counselors, 2021, cont'd

Katie Huang, Meagan Choates, Grace Tran, Chelsea Wagner, & Claire Singletary - Barriers Experienced by Underrepresented Minorities in Becoming a Competitive Genetic Counseling Applicant. Platform presentation, Virtual, September 2021.

Lauren Douglas, Jen Czerwinski, Myla Ashfaq, & Chelsea Wagner - Disability Education: What's missing? Poster, virtual, September 2021.

Katlyn Frane, Victoria Wagner, Chelsea Wagner, Leslie Dunnington, Carol Nowlen, & Claire Singletary - Evaluating Payer Approval Rates of Exome Sequencing and its Effect on Minority Patients' Access to Genetic Care. Poster, virtual, September 2021.

Katie Sagaser - Advocates for Autonomy: Genetic counselors as champions for comprehensive reproductive health. Educational breakout session, September 2021.

Philadelphia Prenatal Diagnosis: Prenatal Genetics & Ultrasound, Obstetrics/MFM Conference

Katie Sagaser - Expanded carrier screening for hypophosphatasia and skeletal dysplasias. Oral presentation; Virtual meeting (2020).

Project Baby Deer Case Review Conference

Jessie Omark O'Shea - Case Review: ASXL3 and PURA-related disorders. February 2021

UTGCP Alumni & Faculty: Authorship at National Meetings

Submitted presentations from 2020-2021; only UT affiliated authors listed

San Antonio Breast Cancer Virtual Symposium

Amie Blanco - Personalized breast cancer screening in a population-based study: Women informed to screen depending on measures of risk (WISDOM). 2020 Dec 8-11.

Society for Maternal-Fetal Medicine, 2021

Katie Sagaser - Molecular genetic diagnosis for early pregnancy renal anhydramnios. Poster presentation, virtual meeting (2021).

Katie Sagaser - Chromosome analysis at the time of fetoscopic laser surgery yields significant abnormalities in monochorionic (MC) twin pregnancies with twin-to-twin transfusion syndrome (TTTS). Poster presentation, virtual meeting (2021).

Katie Sagaser - Incidental diagnoses from expanded carrier screening: Lessons from hypophosphatasia. Oral presentation as part of Genetics Scientific Forum; virtual meeting (2021).

Texas Society of Genetic Counselors

Haley Streff & Hope Northrup - Genotype vs Phenotype: Pathogenic variant in TSC1 in a three-generation family without clinical evidence of tuberous sclerosis complex. Best abstract and poster at TSGC 2020.

University of Michigan & Wayne State University Supervisors Workshop 2021

Jessie Omark O'Shea - Lessons Learned in Telehealth and Remote Supervision. 2021 May.

United Mitochondrial Disease Foundation Meeting

Amanda Gerard - Intracranial calcifications simulating Aicardi-Goutieres syndrome in PARS2-related mitochondrial cytopathy [Abstract]. Poster, 2021.

UTGCP Alumni & Faculty: Scientific Publications

Submitted publications from 2020-2021; only UT affiliated authors listed

American Journal of Human Genetics

Marisa Vineyard Andrews - De novo and bi-allelic pathogenic variants in NARS1 cause neurodevelopmental delay due to toxic gain-of-function and partial loss-of-function effects. Am J Hum Genet. 2020;107(2):311-324.

Haley Streff - De novo variants in the ATPase module of MORC2 cause a neurodevelopmental disorder with growth retardation and variable craniofacial dysmorphism. Am J Hum Genet. 2020;107(2), 352-363.

American Journal of Medical Genetics Part A

Katie Sagaser - Deletion rescue resulting in segmental homozygosity: A mechanism underlying discordant NIPT results. Am J Med Genet A. 2020; 182(11), 2666–2670.

Lauren Westerfield & Amanda Gerard - Clinical characterization of individuals with the distal 1q21. 1 microdeletion. Am J Med Genet A. 2021;185(5), 1388-1398.

Sandra Darilek - The sixth international RASopathies symposium: Precision medicine-From promise to practice. Am J Med Genet A. 2020 Mar;182(3):597-606.

Sandra Darilek - A rare description of pure partial trisomy of 16q12.2q24.3 and review of the literature. Am J Med Genet A. 2021 Oct;185(10):2903-2912.

Nevena Krstic -Report of a novel variant in the FAM111A gene in a fetus with multiple anomalies including gracile bones, hypoplastic spleen, and hypomineralized skull. Am J Med Genet A. 2021 Jun;185(6):1903-1907.

American Journal of Obstetrics & Gynecology

Katie Sagaser - Society for Maternal-Fetal Medicine Special Statement: Maternal-fetal medicine subspecialist survey on abortion training and service provision. American Journal of Obstetrics and Gynecology. 2021 Jul;225(1):B2-B11.

American Journal of Perinatology

Nevena Krstic - N95 Filtering Facepiece Respirator Use during Pregnancy: A Systematic Review. American Journal of Perinatology. 2020 Aug;37(10):995-1001.

Annals of Surgical Oncology

Jordan Berg, Sara Pirzadeh-Miller - Clinicopathological features and outcomes in individuals with breast cancer and ATM, CHEK2, or PALB2 mutations. Ann Surg Oncol. 2021;28(6):3383-3393.

Birth Defects Research

Nevena Krstic - Current landscape of prenatal genetic screening and testing. Birth defects research. 2020 Mar 1;112(4):321-331.

Breast Cancer Research

Amie Blanco - The WISDOM study: a new approach to screening can and should be tested. Breast Cancer Research and Treatment. 2021 Oct;189(3):593-598.

UTGCP Alumni & Faculty: Scientific Publications

Submitted publications from 2020-2021; only UT affiliated authors listed

Cancer Journal

Deanna Darnes & Danielle Williams - Challenges and Errors in Genetic Testing: The Fifth Case Series. Cancer J. 2021 Nov-Dec 01;27(6):417-422.

Clinical Case Reports

Katie Gunther, Kate Mowrey, & Lara Farach - Two new reported cases of 16q22.3q23.3 duplication syndrome highlight intrafamilial variability and potential sex expression differences within a rare duplication syndrome. Clin Case Rep. 2021 Jan 27;9(3):1629-1633.

Clinical Genetics

Haley Streff - PPP3CA truncating variants clustered in the regulatory domain cause early-onset refractory epilepsy. Clinical Genetics. 2021 Aug 100(2), 227-233.

eLife

Amanda Gerard - KDM5A mutations identified in autism spectrum disorder using forward genetics. eLife. 2020 Dec 22;9:e56883.

Frontiers in Cell and Developmental Biology

Haley Streff - OTUD5 variants associated with X-linked intellectual disability and congenital malformation. Frontiers in Cell and Developmental Biology. 2021 Mar 3;9:631428.

Frontiers in Neurology

Kate Mowrey, Hope Northrup, Syed Hashmi, David Rodriguez-Buritica - Frequency, Progression, and Current Management: Report of 16 New Cases of Nonfunctional Pancreatic Neuroendocrine Tumors in Tuberous Sclerosis Complex and Comparison With Previous Reports. Front Neurol. 2021 Apr 9;12:627672.

Genetic Counseling Practice Advanced Concepts and Skills

Claire Singletary - Risk Assessment and Communication: A Complex Process. In N. Callanan, B LeRoy, P McCarthy-Veach (Eds), Genetic Counseling Practice Advanced Concepts and Skills, Wiley-Blackwell: Hoboken, New Jersey. ISBN 9781119529859

Genetics in Medicine

Marisa Vineyard Andrews - Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia and insensitivity to pain. Genet Med. 2021;23(8):1465-1473.

Marisa Vineyard Andrews - Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genet Med. 2021;23(8):1474-1483.

Amie Blanco - Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genet Med. 2020 Oct;22(10):1653-1666.

Laura Hendon - Genome sequencing as a first-line diagnostic test for hospitalized infants. Genet Med. Epub 2021 Nov 27.

Haley Streff, Amanda Gerard & Andi Lewis - Outcomes of prior authorization requests for genetic testing in outpatient pediatric genetics clinics. Genet Med. 2021 23(5), 950-955.

Haley Streff - Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Gene Med. 2021 23(7), 1234-1245.

UTGCP Alumni & Faculty: Scientific Publications

Submitted publications from 2020-2021; only UT affiliated authors listed

Handbook of Clinical Adult Genetics & Genomics: A Practice-Based Approach

Sandra Darilek & Sarah (Mayes) Huguenard - Preconception and prenatal care; pp 71-81; In Handbook of Clinical Adult Genetics and Genomics: A Practice-Based Approach; Academic Press; San Diego, CA; 2020; Dhar SU, Nagamani SCS, Eble TN, ed.; 1st edition.

Hereditary Cancer Clinical Practice

Jess (Omark) O'Shea, Leslie Dunnington, Sarah Noblin, Blair Stevens, & Maureen Mork - Patients with unexplained mismatch repair deficiency are interested in updated genetic testing. Hered Cancer Clin Practice. 2020 Sep 21; 18:19.

Human Genetics and Genomics Advances

Jess (Omark) O'Shea - AHDC1 missense mutations in Xia-Gibbs syndrome. HGG Adv. 2021 Oct 14;2(4):100049.

Human Molecular Genetics

Haley Streff - Mutations of the Histone Linker H1-3 in Neurodevelopmental Disorders and Functional Characterization of Neurons Expressing C-terminus Frameshift Mutant H1.4. Human Molecular Genetics. 2022 May 4;31(9):1430-1442

JAMA Open Network

Amie Blanco - Yield and Utility of Germline Testing Following Tumor Sequencing in Patients With Cancer. JAMA Network Open. 2020 Oct 1;3(10):e2019452.

Journal of Clinical Oncology

Amie Blanco - The marginal diagnostic benefit of pancreatic cancer molecular profiling after germline testing. Journal of Clinical Oncology, 2021 39(15) supplemental.

Amie Blanco - Implementation of Germline Testing for Prostate Cancer: Philadelphia Prostate Cancer Consensus Conference 2019. Journal of Clinical Oncology. 2020 Aug 20;38(24):2798-2811.

Journal of Extracellular Vesicles

Taylor Beecroft - Extracellular microRNAs in blood differentiate between ischaemic and haemorrhagic stroke subtypes. J Extracell Vesicles. 2020 Jan 24;9(1):1713540.

Journal of Genetic Counseling

Kaitlyn Amos, Chelsea Wagner, Jen Czerwinski Lauren Murphy, Maureen Mork, & Claire Singletary - Influence of genetic counselor medical history on specialty and psychosocial practice in North America. Journal of Genetic Counseling. 2021 Nov 26. Online ahead of print

Jordan Berg & Sara Pirzadeh-Miller - Adapting Genetic Counseling Operations Amidst the COVID-19 Pandemic. J Genet Couns. 2021;30(4):949-955.

Sarah Burke, Maureen Mork, Krista Qualmann, & Ashley Woodson - Genetic counselor approaches to BRCA1/2 direct to consumer genetic testing results. Journal of Genetic Counseling. 2021 Jun;30(3):803-812.

UTGCP Alumni & Faculty: Scientific Publications

Submitted publications from 2020-2021; only UT affiliated authors listed

Journal of Genetic Counseling (cont'd)

Caylene Smith, Syed Hashmi, Jen Czerwin - The impact of genetic counseling on women's grief and coping following termination of pregnancy for fetal anomaly. J Genet Couns. 2021 Apr;30(2):522-532.

Matthew Tschirgi - Commentary from a commercial laboratory: The unexpected benefits of a more virtual world during the COVID-19 pandemic. J Genet Couns. 2021 Aug;30(4):956-957.

Matthew Tschirgi - Easing the burden of multi-state genetic counseling licensure in the United States: Process, pitfalls, and possible solutions. J Genet Couns. Epub 2021 Jul 12.

Journal of Human Genetics

Jess Omark O'Shea - Kagami-Ogata syndrome in a patient with 46, XX,t(2;14)(q11.2;q32.32)mat disrupting MEG3. J Hum Genet. 2021 Apr;66(4):439-443.

Journal of Maternal-Fetal and Neonatal Medicine

Katie Sagaser & Katie (Rock) Forster - Molecular testing strategies in the evaluation of fetal skeletal dysplasia. J Matern Fetal Neonatal Med . 2020 Aug 4;1-7.

Journal of Personal Medicine

Laura Hendon - Genomic Sequencing Results Disclosure in Diverse and Medically Underserved Populations: Themes, Challenges, and Strategies from the CSER Consortium. J. Pers. Med. 2021 Mar; 11(3): 202.

Journal of Perinatology

Laura Hendon - Asynchronous Telemedicine for Clinical Genetics Consultations in the NICU: A Single Center's Solution. J Perinatol. Epub 2021 Jul 23.

Mitochondrion

Myla Ashfaq, Hope Northrup, Claire Singletary, Syed Hashmi, David Rodriguez-Buritica - Hypoglycemia in mitochondrial disorders. Mitochondrion. 2021 May;58:179-183.

Molecular Genetics & Genomic Medicine

Kate Mowrey - A De Novo case of autosomal dominant mitochondrial membrane protein-associated neurodegeneration. 2021 Jul;9(7):e1706.

Kate Mowrey, Shannon Mulligan, Hope Northrup - Two different genetic etiologies for tuberous sclerosis complex (TSC) in a single family. 2020 Jul;8(7):e1296.

Nature Communications

Marisa Vineyard Andrews - Loss-of-function mutations in UDP-glucose 6-dehydrogenase cause recessive developmental epileptic encephalopathy. Nat Commun. 2020;11(1):595.

Nature Medicine

Deanna Darnes - Responsible use of polygenic risk scores in the clinic: potential benefits, risks and gaps. Polygenic Risk Score Task Force of the International Common Disease Alliance. Nat Med. 2021;27(11):1876-1884.

UTGCP Alumni & Faculty: Scientific Publications

Submitted publications from 2020-2021; only UT affiliated authors listed

Nature Genetics

Amie Blanco - Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nat Genet. 2020 Jan;52(1):56-73.

npj Genomic Medicine

Amanda Gerard - De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. NPJ Genom Med. 2021 Dec 7;6(1):104.

The Oncologist

Amie Blanco - Implementation of an Embedded In-Clinic Genetic Testing Station to Optimize Germline Testing for Patients with Pancreatic Adenocarcinoma. Oncologist. 2021 Nov;26(11):e1982-e1991

Prenatal Diagnosis

Blair Stevens, Peyton Nunley, Chelsea Wagner, Lauren Murphy, Theresa Wittman, Aarti Ramdaney, Malorie Jones, Meagan Choates - Utility of expanded carrier screening in pregnancies with ultrasound abnormalities. Prenat Diagn. 2022 Jan;42(1):60-78.

Peyton Nunley, Syed Hashmi, Lara Farach, Claire Singletary, & Blair Stevens - Exploring the predicted yield of prenatal testing by evaluating a postnatal population with structural abnormalities using a novel mathematical model. Prenat Diagn. Epub 2021 Nov 18

Prenatal Diagnosis (cont'd)

Katie Sagaser & Katie (Rock) Forster - The utility of exome sequencing for fetal pleural effusions. Prenat Diagn. 2020 Apr. 40(5), 590–595.

Katie Sagaser & Katie (Rock) Forster - Etiology and management of early pregnancy renal anhydramnios: Is there a place for serial amnioinfusions? Prenat Diagn. 2020 Apr. 40(5), 528–537.

Scientific Data

Taylor Beecroft - Extracellular circular RNA profiles in plasma and urine of healthy, male college athletes. Sci Data. 2021 Oct 28;8(1):276.

Scientific Reports

Haley Streff - Molecular characterisation of rare loss-of-function NPAS3 and NPAS4 variants identified in individuals with neurodevelopmental disorders. 2021 Mar 23;11(1):6602

Supportive Care in Cancer

Erica Bednar, Aarti Ramdaney, & Jessie (Ross) Corredor - Factors Impacting Adolescent and Young Adult Cancer Patients' Decision to Purse Genetic Counseling and Testing. Epub 2022 Mar 19.

Texas Children's Hospital Handbook of Congenial Heart Disease

Taylor Beecroft - Connective Tissue Disorders. In: Texas Children's Hospital Handbook of Congenital Heart Disease, edited by Mery CM, Bastero P, Hall AGCSR. Texas Children's Hospital; 2020; p368-375. ISBN 1734272112, 9781734272116.

want to join the UTGCP?

contact us

Phone: (713) 500-5599
Email: *gsbs.gcpinfo@uth.tmc.edu*

to our alumni community:

If you didn't see your research in this issue, we hope to include you next year.

We look forward to seeing your accomplishments and photos!

Support the Research and Education Fund online here!