THE UNIVERSITY OF TEXAS MD Anderson Cancer Center | UTHealth Houston

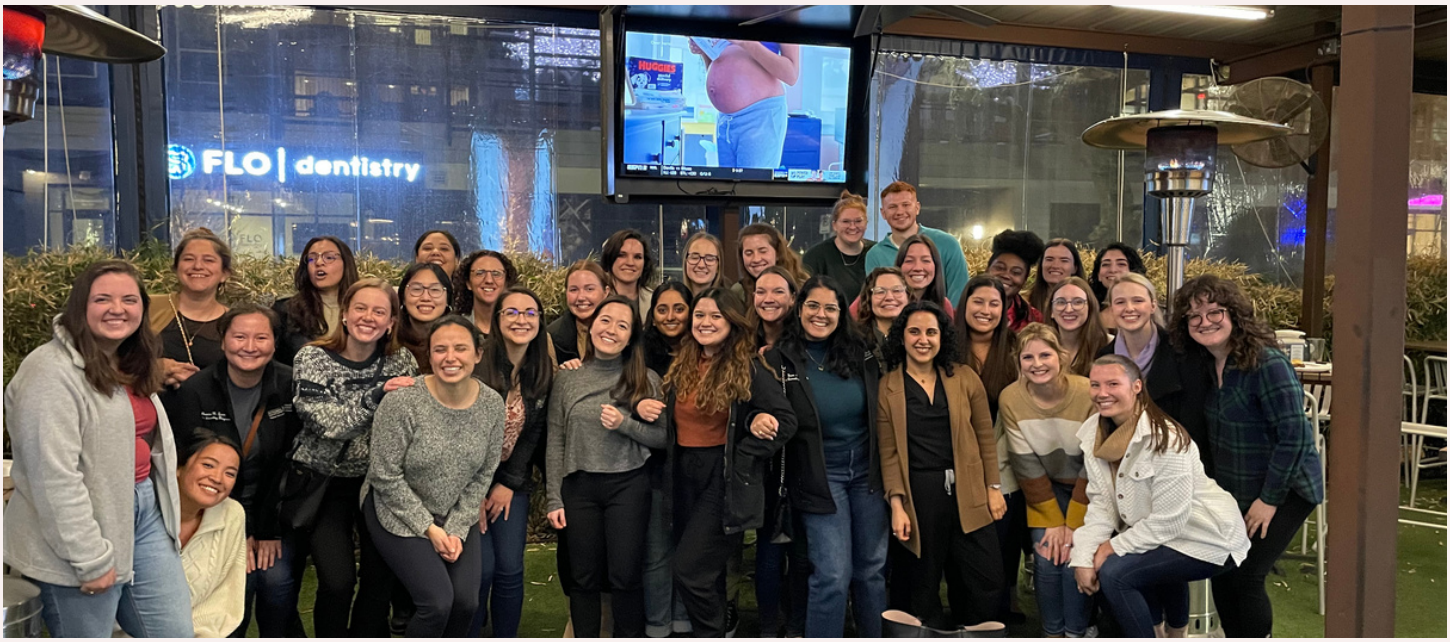
Graduate School of Biomedical Sciences

UTGCP Annual Research Newsletter

April 2023 | Issue No. 4

What's Inside

Classes of 2023 and 2024 thesis research
Alumni & faculty research & publications from calendar year 2022, hyperlinked when available



Want to learn more about UTGCP?

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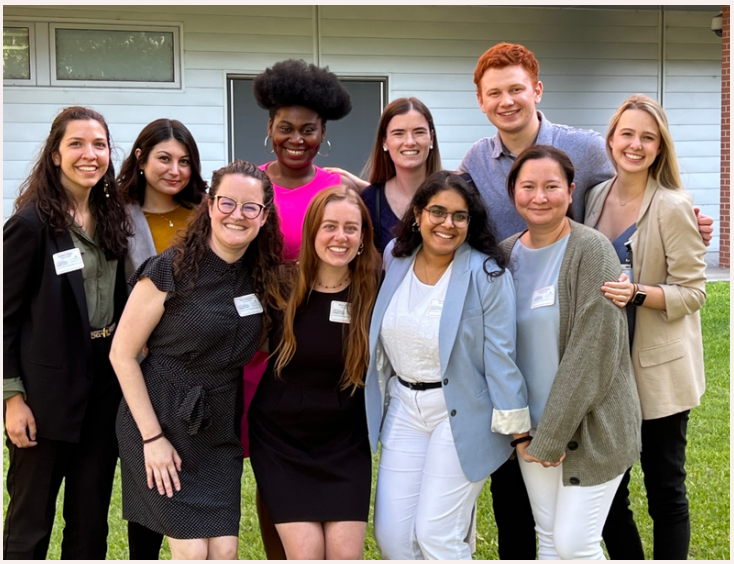


Celebrate with the class of 2023!

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

Click Here

M.S. Thesis Topics

Latonya Alexander

Pregnant Black/African Americans' Experiences with Prenatal Genetic Counseling
Chair: Claire Singletary

Madeline Alpar

Clinical Expectations & Psychosocial Concerns Impact on the Diagnostic Odyssey of Hypermobile Ehlers-Danlos Syndrome (hEDS) Patients

Chair: Kathryn Leal

Erin Atkinson

Prenatal decision-making facilitated through an online patient education module

Chair: Aranza Gonzalez

Yusra Aziz

The Need for Racial and Ethnic Health Disparity Curriculum in Genetic Counseling Programs

Chair: Claire Singletary

Jack Colleran

Perceived Utility of Genetic Carrier Screening in Diverse Patient Populations

Chair: Blair Stevens

Emily Daykin

Investigation of Teratogenic Concerns in the COVID-19 Era

Chair: Myla Ashfaq

Tessa Heller

The Impact of Treatment with Palynziq on Quality of Life for Individuals with PKU

Chair: Kate Richardson

Cindy Hernandez

Monolingual Spanish Speaking Patient Satisfaction and Comfort when Comparing Spanish Speaking Provider to an Interpreter

Chair: Kathryn Leal

Jasmine Tyson

Impact of Recent Abortion Legislation on Patient Trust in Healthcare Providers

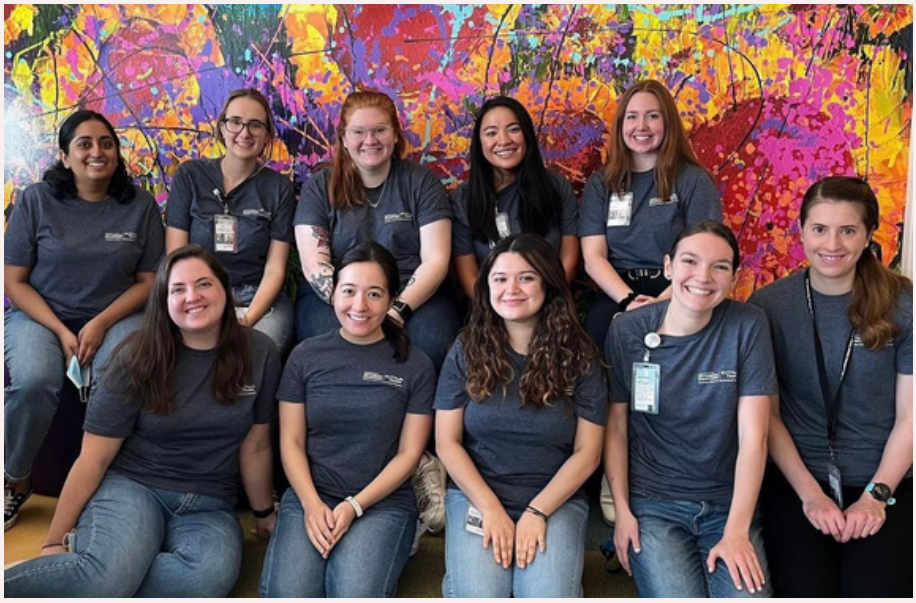
Chairs: Aarti Ramdaney and Samantha Montgomery

Jordan Zeiger

Saliva Kit Returns after Consenting for Genetic Cancer Testing in a Genetic Counseling Setting

Chair: Maureen Mork

Introducing the Research of the Class of 2024!



M.S. Thesis Topics

Emma Billings: Adoption education in GC training programs; Advisor: Jennifer Czerwinski

Carley Brueckner: Assessing the impact of diagnostic testing on patient empowerment in pregnancies with fetal anomalies; Advisor: Blair Stevens

Laura Gorecki: Factors associated with diagnosis disclosure with romantic partners among individuals with TSC; Advisor: Kate Richardson

Maria Hernandez: Comfort and perceived utility of the dysmorphology exam in medical genetics among students and genetic counselors; Advisor: Katie Shields

Mindy Kolodziejski: Patients' understanding of fetal sex vs. gender in the context of NIPT; Advisor: Shannon Mulligan

Nicolette Murphey: Pregnancy yields and screening methodologies for skeletal dysplasias and craniosynostosis syndromes; Advisor: Theresa Wittman

Rachel Notestine: Fraud, waste, and abuse in the genetic testing industry; Advisor: Claire Singletary

Disha Patel: Patient preferences for soft sign disclosure with prior negative NIPS; Advisor: Meagan Choates

Kiki Risgaard: The relationship between TSC and sleep disorders; Advisor: Kate Richardson

Jordan Steffen: Attitudes and motivations for anonymous predictive testing in the Huntington's Disease population; Advisor: Leslie Dunnington

UTGCP Alumni & Faculty: Authorship at National Meetings

Submitted presentations in 2022.
only UT affiliated authors listed

American Academy of Pediatrics, October 2022

Taylor Beecroft, Yield of Systematic Inpatient Genetic Testing of Neonates with Conotruncal, Right-sided and Laterality Heart Defects. Accepted for Oral Presentation in the Young Investigator Award Competition.

Taylor Beecroft. Phenotypic Features Associated with Yield of Clinical Genetic Testing Among Patients with Bicuspid Aortic Valve Yield. Accepted for Oral Presentation in the Young Investigator Award Competition.

Taylor Beecroft. A Preliminary Guide for Clinicians. Accepted for Oral Presentation in the Young Investigator Award Competition.

American College of Medical Genetics, March 2022

Amanda Gerard and Haley Streff. Genetic Testing Uptake in a Large Metropolitan Pediatric Outpatient Genetics Clinic. Accepted Poster.

American Society of Human Genetics, October 2022

Sandra Darilek. A multiomics approach to resolving small supernumerary marker chromosomes. Accepted Poster.

Baylor College of Medicine Annual Cardiovascular Research Institute Symposium, April 2022

Taylor Beecroft. Yield of Systematic Inpatient Genetic Testing of Neonates with Conotruncal and Laterality Heart Defects. Accepted for presentation.

Taylor Beecroft. Evaluating Preterm Birth as A Biomarker Of Major Events In Children With Vascular Ehlers-Danlos Syndrome. Accepted for presentation.

Taylor Beecroft. Genetic Testing for Patients with Non-Williams Supravalvar Aortic Stenosis: A Preliminary Guide for Clinicians. Accepted for presentation.

Collaborative Group of the Americas- Inherited Gastrointestinal Cancer, November 2022

Amie Blanco. The case for germline testing in all patients with endometrial cancer: lessons learned from universal somatic tumor testing. Accepted for presentation.

Amie Blanco. A step towards patient centered care: Combined endometrial biopsy and colonoscopy for Lynch syndrome. Accepted for presentation.

UTGCP Alumni & Faculty: Authorship at National Meetings

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Children's Hospital of Philadelphia Cardiology Conference, September 2022

Taylor Beecroft. Genetic Testing for Patients with Non-Williams Supravalvar Aortic Stenosis: A Preliminary Guide for Clinicians. Accepted for Oral Presentation.

Digestive Disease Week®, May 2022

Amie Blanco. Increasing Referrals to Genetic Counseling in Patients at Elevated Risk for Lynch Syndrome. Accepted for presentation.

Ethical, Legal Social Implications Congress, June 2022

Julia Wynn. Family Experiences of Genetic Testing for Autism: A Study of SPARK Participants.

HADDS Conference, July 2022

Haley Streff. Genetics 101, EFB3 HADDS Conference, Houston, Texas. July 2022.

International Society of Prenatal Diagnosis, June 2022

Laura Godfrey Hendon. Expanded NIPT: Genetic Counseling Concerns.

Sandra Darilek. Side Effects: When preimplantation genetic testing for aneuploidy (PGT-A) results lead to discovery of an unexpected chromosome abnormality in a patient. Accepted poster

International Symposium on Marfan, Loeys-Dietz, and Related Conditions, August 2022

Taylor Beecroft. Cardiovascular Outcomes and Survival in Patients with Early Onset Marfan Syndrome. Accepted for Poster Presentation.

Taylor Beecroft. Case Report: Longest Reported Survival for Autosomal Recessive Cutis Laxa Type 1B in 31-year-old Patient with Large Ascending Aortic Aneurysm and Extensive Arterial Tortuosity. Accepted for Poster Presentation.

Taylor Beecroft. Gestational Age as a Proxy for Arterial Fragility in Vascular Ehlers-Danlos Syndrome. Accepted for Poster Presentation.

Taylor Beecroft. Arterial Tortuosity Syndrome: A Longitudinal Assessment of Cardiovascular Features. Accepted for Poster Presentation.

Kaiser Permanente, Regional Pediatric Aortopathy Grand Round Lecture Series, November 2022

Taylor Beecroft. Creating a Pediatric Aortopathy Program. Virtual Webinar

Mountain States Regional Genetics Network webinar, May 2022

Haley Streff. Genetic Testing: It's Not Rocket Science.

UTGCP Alumni & Faculty: Authorship at National Meetings

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National Society of Genetic Counselors, November 2022

Megan Choates, Carla McGruder, Claire Singletary, Natalie Stoner, Theresa Wittman, Sara Wofford. Invisible Diversities, Academic Capital and Competitiveness of Genetic Counseling Applicants.

Callie Diamonstein. Genetic Counseling for Critically Ill Patients: Roadmap for Professional Development and Patient Centered Care. Pre-conference symposium.

Meagan Choates, Shannon Mulligan and Blair Stevens. Non-Invasive Prenatal Screening (NIPS) Testing Motivations and Informed Decision Making in the Low-Risk Population.

North American Neuroendocrine Tumor Society Multidisciplinary NET Medical Symposium, October 2022

Amie Blanco. Germline Pathogenic Variants in Patients with High-Grade (G3) Metastatic Gastroenteropancreatic (GEP). Accepted for presentation.

Amie Blanco. Variants of Uncertain Significance (VUS) are More Common in Non-Caucasian Patients with Neuroendocrine Neoplasms (NENs). Accepted for presentation.

San Antonio Breast Cancer Symposium, October 2022

Amie Blanco. Hereditary vs Sporadic Invasive Lobular Carcinoma and Impact of Locoregional Therapy on Disease-Free Survival. Accepted for presentation.

Amie Blanco. The impact of streamlined processes and patient-directed messaging to improve enrollment in a remote, pragmatic clinical trial. Accepted for presentation.

Amie Blanco. The WISDOM study: Reducing sequential steps and implementing parallel workflows in pragmatic trials. Tomiyuri Lewis, Amie Blanco et al. Accepted for presentation.

Society for Gynecologic Oncology 2022 Winter Meeting, January 2022

Amie Blanco. Germline testing for endometrial cancer in the age of next generation sequencing.

UTGCP Alumni & Faculty: Authorship at National Meetings

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Texas Children's Hospital, Events in 2022

Taylor Beecroft. Heart Center Performance Rounds. March 2022. TCH Cardiovascular Genetics Team: 2022 Update in Volumes, Outcomes, and QI Initiatives.

Taylor Beecroft. Evening with Genetics Program, November 2022.Marfan Syndrome: Current Cardiac Management and New Research Updates.

Taylor Beecroft. Adult Congenital Heart Disease and Women's Heart Health Conference, April 2022. Genetic Considerations for Mother and Baby.

Workshop on Malformations and Morphogenesis, August 2022

Jessica O'Shea. Something Old (Silver-Russell Syndrome), Something New (Unusual 11p15.5 Duplication), and Something Borrowed. Accepted Poster.

UTGCP Alumni & Faculty: Scientific Publications

Submitted publications from 2022;
Only UT affiliated authors listed

American Journal of Human Genetics

Leslie Granger. ADGRL1 haploinsufficiency causes a variable spectrum of neurodevelopmental disorders in humans and alters synaptic activity and behavior in a mouse model. Am J Hum Genet. 2022 Aug 4;109(8):1436-1457.

American Journal of Medical Genetics Part A

Megan Morand. GABRG1 variant as a potential novel cause of epileptic encephalopathy, hypotonia, and global developmental delay. American Journal of Medical Genetics Part A. 2022 Dec;188(12):3546-3549.

Kate Mowrey. NEXMIF pathogenic variants in individuals of Korean, Vietnamese, and Mexican descent. Am J Med Genet Part A. 2022 Jun;188(6):1688-1692.

Birth Defects Research

Nevena Krstic. A novel clinic structure for exposure counseling during pregnancy. Birth Defects Research. 2022 Sep 1;114(15):855-862.

Clinical Genetics

Sarah Burke and Carolyn Haskins. Six case reports of NTHL1-associated tumor syndrome further support it as a multi-tumor predisposition syndrome. Clin Genet. 2023 Feb;103(2):231-235. Epub 2022 Oct 17.

European Journal of Haematogy

Addison Johnson and Chelsea Wagner. Assessing patient attitudes toward genetic testing for hereditary hematologic malignancy. Eur J Haematol. 2023 Jan;110(1):109-116. Epub 2022 Oct 21.

Familial Cancer

Julia Wynn. Questioning the validity of clinically available breast cancer polygenic risk scores: comparison of two labs reveals discrepancies. Fam Cancer. 2022 Apr;21(2):125-127.

Frontiers in Genetics

Kate Mowrey. Case Report: Novel pathogenic variant in NFIX in two sisters with Malan syndrome due to germline mosaicism. Front Genet. 2022 Nov 9;13:1044660.

Frontiers in Reproductive Health

Kate Mowrey. Expanding Our Knowledge of Menstrual Irregularities Reported by Females With Tuberous Sclerosis Complex. Front Reprod Health. 2022 Feb 18;4:798983.

UTGCP Alumni & Faculty: Scientific Publications

Accepted publications from 2022;
Only UT affiliated authors listed

Genetics in Medicine

Taylor Beecroft. Evaluating perinatal and neonatal outcomes among children with vascular Ehlers-Danlos syndrome. Genet Med. 2022 Oct;24(10):2134-2143.

Sandra Darilek. Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: An evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2023 Feb;25(2):100336. Epub 2022 Dec 16.

Haley Streff. De Novo ZMYND8 variants result in an autosomal dominant neurodevelopmental disorder with cardiac malformations. Genet Med. 2022 Sep;24(9):1952-1966.

Jen Hoskovec and Julia Wynn. Maternal carrier screening with single-gene NIPS provides accurate fetal risk assessments for recessive conditions. Genet Med. 2023 Feb;25(2):100334. Epub 2022 Dec 1.

Haley Streff. Retrospective analysis of a clinical exome sequencing cohort revealed the mutational spectrum and identified novel disease-associated loci of BAFopathies. Genet Med. 2022 Feb;24(2):364-373.

Julia Wynn. Challenges of variant reinterpretation: Opinions of stakeholders and need for guidelines. Genet Med. 2022 Sep;24(9):1878-1887.

Julia Wynn. The reckoning: The return of genomic results to 1444 participants across the eMERGE3 Network. Genet Med. 2022 May;24(5):1130-1138.

Genome Medicine

Laura Godfrey Hendon. Return of non-ACMG recommended incidental genetic findings to pediatric patients: considerations and opportunities from experiences in genomic sequencing. Genome Medicine. 2022 Nov 21;14(1):131.

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. Hum Mol Genet. 2022 May 4;31(9):1430-1442.

Leslie Granger. SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance. Hum Mol Genet. 2022 Sep 29;31(19):3325-3340.

International Journal of Neonatal Screening

Julia Wynn. Newborn Screening for Duchenne Muscular Dystrophy: First Year Results of a Population-Based Pilot. Int J Neonatal Screen. 2022 Sep 22;8(4):50.

Julia Wynn. Improving Recruitment for a Newborn Screening Pilot Study with Adaptations in Response to the COVID-19 Pandemic. Int J Neonatal Screen. 2022 Mar 22;8(2):23.

UTGCP Alumni & Faculty: Scientific Publications

Accepted publications from 2022;
Only UT affiliated authors listed

Journal of Genetic Counseling

Kaitlyn Amos, Jennifer Czerwinski, Lauren Murphy, Claire Singletary, Chelsea Wagner. Influence of Genetic Counselor Medical History on Specialty Choice and Psychosocial Practice in North America. Journal of Genetic Counseling. 2022 June 31(2):663-676.

Sarah Huguenard, Salma Nassef, Tamara Solomon, Cathy Sullivan. Prenatal genetic counseling practices regarding recommendations for cancer genetic counseling: A retrospective chart review from two academic institutions. Journal of Genetic Counseling, 2022 Oct;31(5):1062-1070.

Salma Nassef. Lack of consensus among healthcare professionals at a large academic medical center on the use of exome sequencing for prenatal diagnosis. Journal of Genetic Counseling, 2022 Dec;31(6):1330-1340

Kate Principe. An evidence-based practice guideline of the National Society of Genetic Counselors for telehealth genetic counseling. Journal of Genetic Counseling. 2023 Feb;32(1):4-17. Epub 2022 Aug 30

Julia Wynn. Measuring quality and value in genetic counseling: The current landscape and future directions. J Genet Couns. 2022 Nov 17 Epub ahead of print.

Julia Wynn. Do research participants share genomic screening results with family members? J Genet Couns. 2022 Apr;31(2):447-458.

Journal of Orthopaedic Case Report

Kate Mowrey. Metaphyseal Dysplasia, Spahr Type; A Case Report of Variable Expressivity in Non-Consanguineous Filipino Siblings. Journal of Orthopaedic Case Reports. 2022 Sep;12(9):20-25.

Journal of Pediatrics

Julia Wynn. Mother and Daughter Perspectives on Genetic Counseling and Testing of Adolescents for Hereditary Breast Cancer Risk. J Pediatr. 2022 Dec;251:113-119.e7.

Nature Communications

Julia Wynn. SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. Nat Commun. 2022 Jul 15;13(1):4112.

Personalized Medicine

Julia Wynn. Development of Competency-based Online Genomic Medicine Training (COGENT). Per Med. 2022 Nov 23 [Epub ahead of print] Review.

UTGCP Alumni & Faculty: Scientific Publications

Accepted publications from 2022;
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Molecular Genetics & Genomic Medicine

Kate Mowrey. Identification of a novel microdeletion causative of Nance- Horan syndrome. Mol Genet Genomic Med. 2022 Mar;10(3):e1879.

Obstetrics & Gynecology

Blair Stevens. Incidental Detection of Maternal Malignancy by Fetal Cell-Free DNA Screening. Obstetrics & Gynecology. 2022 Jul 1;140(1):121-131.

Pancreas

Amie Blanco. The Additional Diagnostic Benefit of Pancreatic Cancer Molecular Profiling After Germline Testing. Pancreas. 2022 April 51(4): 302-304.

Prenatal Diagnosis

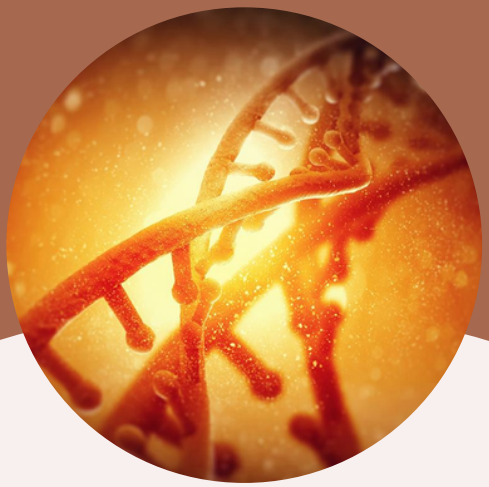
Meghan Choates, Malorie Jones, Lauren Murphy, Peyton Nunley, Aarti Ramdaney, Blair Stevens, Chelsea Wagner, Theresa Wittman. Utility of expanded carrier screening in pregnancies with ultrasound abnormalities. Prenat Diagn. 2022 Jan;42(1):60-78.

Shannon Mulligan, Aarti Ramdaney, Chelsea Wagner, Theresa Wittman. First trimester ultrasound in the age of cell-free DNA screening: What are we missing? Prenat Diagn. 2022 May;42(5):542-548.

Julia Wynn. Information is power: The experiences, attitudes and needs of individuals who chose to have prenatal genomic sequencing for fetal anomalies. Prenat Diagn. 2022 Jun;42(7):947-954.

Public Health Genomics

Julia Wynn. Diverse Parental Perspectives of the Social and Educational Needs for Expanding Newborn Screening through Genomic Sequencing. Public Health Genomics. 2022 Sep 15:1-8 [Epub ahead of print].



Congratulations to the UTGCP Research and Education Fund Awardees

The Research and Educational Fund was fully funded in 2021, which allows for the distribution of funds to begin! Below are the first three recipients from the fund. The leadership of UTGCP are key in identifying the needs of current students and helping them through the application process. Awardees can use the fund in support of their research and/or educational needs.

- **Katie Baudoin (Class of 2022)** Investigating the attitudes of the Deaf community towards genetic counseling and utilization of genetic counseling services for indications other than deafness
- **Emile Moura Coelho da Silva (Class of 2022)** Racial concordance between patient and genetic counselor and reported levels of trust and satisfaction
- **Gina Sanchez (Class of 2022)** Status of Termination Curriculum in Genetic Counseling Programs: Survey of Program Directors and Recent Graduates
- **Natalie Stoner (Class of 2022)** Invisible Diversity, Intersectionality, Academic Capital and Barriers to Being a Competitive Genetic Counseling Applicant

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

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!