

UTGCP Annual Research Newsletter

April 2024 | Issue No. 5



Class of 2024 and Class of 2025 Thesis Research
Alumni and Faculty Research and Publications from 2023





Congratulations Class of 2024!

Please consider supporting UTGCP
student research efforts through our
Research and Education Fund:

[Click Here](#)



Emma Billings

Inclusion of adoption as a
pregnancy management option in
prenatal genetic counseling
practice

Chair: Leslie Dunnington, MS, CGC
and Jennifer Czerwinski, MS, CGC



Carley Brueckner

Assessment of patient
empowerment in pregnancies with
fetal anomalies

Chair: Blair Stevens, MS, CGC



Laura Gorecki

Influential factors for disclosing a
Tuberous Sclerosis Complex
diagnosis to romantic partners

Chair: Kate Richardson, MS, CGC



Maria Hernandez

Dysmorphology training and utility
in genetic counseling

Chair: Katie Sheilds, MS, CGC



Mindy Kolodziejewski

Patient knowledge of fetal sex vs.
gender in the context of routine
non-invasive prenatal testing
(NIPT)

Chair: Shannon Mulligan, MS, CGC



Nicolette Murphey

Efficacy of genetic testing
methodologies for prenatal
detection of skeletal anomalies
and craniosynostosis syndromes

Chair: Theresa Wittman, MS, CGC



Rachel Notestine

Fraud in genetic testing:
swindling the system

Chair: Claire Singletary, MS, CGC
and Quinn Stein, MS, CGC



Disha Patel

Patient preferences for ultrasound
soft sign disclosure with prior
negative cfDNA screening

Chair: Meagan Choates, MS, CGC



Kirsten Risgaard

Sleep disturbances in adults with
TSC: influences of treatment and
clinical features

Chair: Kate Richardson, MS, CGC



Jordan Steffen

Motivations and attitudes for
pursuing anonymous genetic
testing for Huntington Disease in
an at-risk population

Chair: Leslie Dunnington, MS, CGC



Introducing the Research of the Class of 2025:

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Brenna Albracht

How do aspects of scientific, health, and genetic literacy impact informed decision making in a prenatal genetic counseling patient population?

Chair: Meagan Choates, MS, CGC

Cindy Canales

What is the diagnostic yield of genetic testing obtained outpatient after being denied inpatient, and is there a delay in diagnosis and subsequent referrals or services for patients whose inpatient testing was denied?

Chair: Kathleen Shields, MS, CGC and
Kathryn Leal, MS, CGC

Jessica Clark

What factors do genetic counseling applicants consider when ranking programs for the Match and how are they prioritized?"

Chair: Claire Singletary, MS, CGC

Nadia English

Spanish-Speakers Perspectives of the Benefits and Barriers of Telemedicine in Genetic Counseling

Chair: Aranza Gonzalez Cendejas, MS, CGC

Hillary Esplen

How do cancer rates vary between different ancestries among individuals with Li-Fraumeni syndrome, and are there variant hotspots unique to particular populations?

Chair: Jessica Corredor, MS, CGC

Ava Henson

How the camp PHEver experience has impacted individuals with PKU

Chair: Megan Morand, MS, CGC

Karli Livingston

How does an individual's personal or family history of Huntington's Disease impact their decision and/or ability to obtain life, long-term care, or disability insurances?

Chair: Leslie Dunnington, MS, CGC

Farren Lopez

How do levels of satisfaction with core 50 case selection impact perceived levels of preparedness

Chair: Aarti Ramdaney, MS, CGC

Grace Ra

Can an online reproductive genetics module effectively triage patients to genetic counseling?

Chair: Blair Stevens, MS, CGC

Nicole Talaba

To what extent can we expand upon the phenotype of SETD5-related disorders by a utilizing social media support group?

Chair: Myla Ashfaq, MS, CGC

UTGCP Faculty and Alumni Authorship at National and Local Meetings

Only UT-affiliated authors listed



43rd Annual Pregnancy Meeting™
February 6-11, 2023 | San Francisco, California



Blair Stevens, Chelsea Wagner, Meagan Choates, Jackie Harkenrider, Samantha Montgomery, Shannon Mulligan, Aarti Ramdaney, Jenna Lea; Informed consent for routine genetic screening: Realistic or idealistic?

TSGC 2023 Annual Education Conference

Saturday, March 4, 2023
8:00 AM - 6:00 PM

Claire Singletary, Salma Nassef; And then there were three: Status of genetic counseling programs in Texas

Haley Streff; The road becoming more travelled: Genetic counselors in laboratory stewardship



SOCIETY FOR INHERITED METABOLIC DISORDERS

44th Annual Meeting, March 18 - 21, 2023
Hyatt Regency, Salt Lake City, Utah

Kate Richardson, Paul Hillman; Expanding the phenotype of MT-ATP6: Diagnosis of multiple family members after an abnormal newborn screen



2023

ACMG Annual Clinical Genetics Meeting

MARCH 14-18 • EXHIBIT DATES: MARCH 15-17
SALT PALACE • SALT LAKE CITY, UTAH
MARCH 18 | ACMG/SIMD JOINT SESSION

Leslie Dunnington, Laura Farach; Cognitive declined, Parkinson-like features, and mutism lead to diagnosis of MECP2 in a 46 year old male

Paul Hillman; Novel de novo intronic KAT6B variant in 9-year-old female adds to the wide phenotypic spectrum of KAT6B disorders

Paul Hillman, Kathryn Leal; Report on a patient with a 28 Mb chromosome 2p duplication at 2p22.2p14: The largest 2p duplication described to date

Katie Leal, Paul Hillman; A case of multiple hemangiomas and arteriovenous malformations in a patient with known Greig cephalopolysyndactyly syndrome

Kathryn Leal, David Rodriguez-Buritica; Donor Organ Failure: The importance of donor heterozygosity in a case of primary hyperoxaluria type I

Kate Richardson, Jenny Do, Hope Northrup; Triple diagnoses with overlapping phenotypes: Recommendations and reflections

Kathleen Shields, Hope Northrup; Fifth case of MYHII-associated megacystis-microcolon-intestinal hypoperistalsis syndrome identified in patient with missense variant and whole gene deletion

Kathleen Shields, David Rodriguez-Buritica, Laura Farach; Dual diagnoses of mosaic SRY deletion and HSD17B3 deficiency in a patient with discordant fetal sex

Kathleen Shields, David Rodriguez-Buritica, Laura Farach, Paul Hillman; Heterozygous ACTB pathogenic variant causing Baraister-Winter syndrome presenting with tracheal ring, intestinal atresia, and neonatal diabetes

Haley Streff; Genetic test stewardship for clinical exome sequencing: Review of critical care orders at Texas Children's Hospital

Colorado Genetic Counseling Symposium, Spring 2023

May 12, 2023

Samantha Montgomery, Aranza Gonzalez Cendejas; Impact of abortion bans on genetic counseling in Texas and beyond



Emily Moura da Silva; Homozygous protein S deficiency in siblings

Emily Moura da Silva; Novel thrombophilia - Prothrombin belgrade variant

OTIS Annual Education Meeting

Join us for the 35th Annual OTIS Education Meeting, June 24-27, 2023

Emily Daykin, Syed Hashmi, Kate Richardson, Katie Shields, Molly Daniels, Myla Ashfaq; Investigation of teratogenic concerns in the COVID-19 era

UTGCP Faculty and Alumni Authorship at National and Local Meetings

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LITTLE PEOPLE of AMERICA
Empowerment • Awareness • Community

Amanda Gerard; Parental discussion of medical and genetic information with pediatric patients and siblings



2023 International TSC Research Conference

Fueling the Future

Washington, DC
September 7-9, 2023

Jenny Do; Parental stress in tuberous sclerosis complex

2023 ASCO ANNUAL MEETING

June 2-6, 2023 • McCormick Place • Chicago, IL & Online



Autumn Vara; Overcoming barriers and improving genetic testing completion rates for hereditary cancer in an underserved population at a large safety net hospital



OCT. 26-29, 2023



Kate Richardson; Impact of dosage frequency of propranolol on sleep patterns in patients with infantile hemangiomas



PLUGS[®] SUMMIT

Haley Streff; A review of mitochondrial DNA orders at a tertiary children's hospital: is this the next test to steward?



District XI Annual District Meeting
Obstetrics and Gynecology: The Next Frontier

Aarti Ramdaney; Genetic screening & testing: Updates for the OB/GYN practice



27th International Conference on Prenatal Diagnosis and Therapy
18-21 JUNE 2023 EDINBURGH, UK

Carol Nowlen; Dehydrated hereditary stomatocytosis: A disorder that may present with profound, life-threatening fetal anemia



Latonya Alexander, Jennifer Czerwinski, Erica Bednar, Syed Hashmi, Carla McGruder, Claire Singletary; Pregnant Black/African Americans' experiences in prenatal genetic counseling



Taylor Beecroft, Alana Cecchi; Cardiovascular outcomes and survival in patients with early onset Marfan syndrome

Taylor Beecroft; Standardizing cardiovascular genetics evaluation through a dedicated consult team, a quality improvement initiative

Erin Atkinson, Blair Stevens, Meagan Choates, Syed Hashmi, Luana Goulet, Aranza Gonzalez Cendejas; Prenatal screening decisions facilitated through an online education module

Yusra Aziz, Syed Hashmi, Aarti Ramdaney, Myla Ashfaq, Victoria Wagner, Claire Singletary; The need for racial and ethnic health disparity in genetic counseling programs;
Winner of Best Student Abstract Award

UTGCP Faculty and Alumni Authorship at National and Local Meetings

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<p>Erica Bednar; Exploring the experiences of family communication and cascade genetic testing for hereditary predisposition to cancer in medically-underserved populations: A qualitative study</p> <p>Erica Bednar; Implement this! Learn how to use implementation science to improve your clinic and advance your research through facilitated project-based learning</p> <p>Sarah Burke; Case report of BICRA-related neurodevelopmental disorder with novel clinical features</p> <p>Aarti Ramdaney; Specialization in prenatal genetic counseling: Defining the role of the fetal therapy genetic counselor</p> <p>Jack Colleran, Blair Stevens, Meagan Choates, Syed Hashmi, Brittanie Shelton, Aranza Gonzalez Cendejas; Perceived utility of genetic carrier screening in a diverse patient population</p> <p>Jessica Corredor; Clinical application of a Li-Fraumeni Syndrome Risk Assessment tool (LFSPRO)</p> <p>Deanna Darnes, Brad Rolf, Carla McGruder; “Say my name, Say my name”: It’s time to discuss the problem with the name “genetic counselor”</p> <p>Emily Daykin, Syed Hashmi, Kate Richardson, Katie Shields, Molly Daniels, Myla Ashfaq; Investigation of teratogenic concerns in the COVID-19 era</p> <p>Carolyn Haskins; An unusual presentation of apparently de novo MLHI-associated Lynch syndrome</p> <p>Tessa Heller, Megan Morand, Syed Hashmi, Hope Northrup, Christina Falugi, Kate Richardson; The impact of treatment with Palynziq on quality of life for individuals with PKU</p> <p>Cindy Hernandez, Meagan Choates, Aranza Gonzalez Cendejas, David Rodriguez-Buritica, Victoria Wagner, Myla Ashfaq, Kathryn Leal; Monolingual Spanish-speaking patient satisfaction and comfort when comparing Spanish-speaking provider to an interpreter</p> <p>Nevena Krstic; Prenatal detection of Muenke syndrome via ultrasound and single gene cell-free DNA screening</p>	<p>Karli Livingston; Evaluation of patients with ATM and/or CHEK2 germline likely pathogenic/pathogenic variants and hematological malignancies</p> <p>Carla McGruder; Rolling the dice on pharmacogenetics: A gamified workshop</p> <p>Jaqueline Mersch; How referral characteristics impact patient scheduling outcomes</p> <p>Salma Nassef, Haley Streff; Clinical supervisor feedback growth: A GCP’s experience with increasing supervisor engagement</p> <p>Brad Rolf; Translating polygenic scores into clinical practice: Insights from genetic counselors</p> <p>Blair Stevens; Reproductive carrier screening today: Evolving guidelines, novel treatments, and disparities in care</p> <p>Matt Tschirgi; You Can Do It Too! Genetics Content Creation and Science Communication via TikTok, Instagram, Podcasting, YouTube, LinkedIn, and More</p> <p>Elise Watson, Jacqueline Mersch; How referral characteristics impact patient scheduling outcomes</p> <p>Elise Watson, Jacqueline Mersch; I. The view from 30,000 patients: eight years of referral data in a cancer genetics program</p> <p>Cathy Wicklund; Assessing genetic counselors’ perspectives on acceptability, feasibility, and readiness of chatbots in genetic counseling</p> <p>Cathy Wicklund; Does microeducation improve knowledge of cardiogenomic risk assessment? An exploratory analysis of online learner data</p>
	<div> <div>  </div> <div> <p>October 27–28, 2023 Boston, MA & Online</p> </div> <div> <p>#ASCOQLTY23</p> </div> </div> <p>Jessica Corredor; Development of provider ordered genetic testing pathway and electronic medical record best practice advisory (BPA) to aid in germline genetic testing for patients with metastatic breast cancer</p>

UTGCP Faculty and Alumni Authorship at National and Local Meetings

Only UT-affiliated authors listed



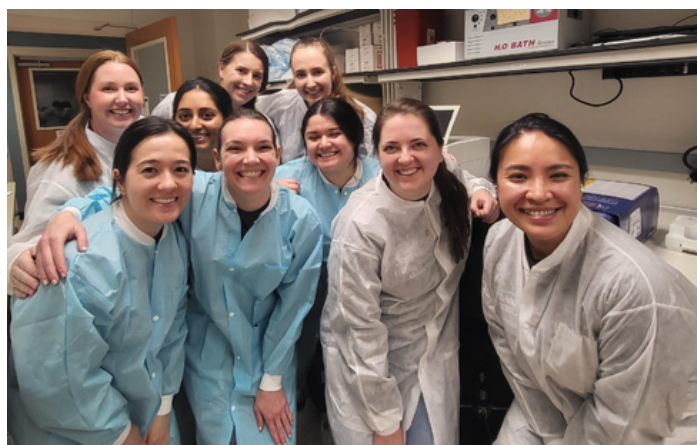
Myla Ashfaq; APMHG undergraduate medical education core competencies in genetics and genomics: A framework and implementation example

Sandra Darilek, Salma Nassef, Sarah Huguenard;

Consultagene: Design and experience of a tele-genetic academic platform supporting genetic consultation, patient & provider education, & research engagement

Christina Falugi, Syed Hashmi, Claire Singletary; Implicit bias in genetics education and healthcare: Traditional textbook photographs elicit negative student responses while natural photographs are educational and stimulate positive quality of life perceptions

Michelle Zelnick; Applying ACMG/AMP variant classification guidelines for SCID – ClinGen SCID VCEP recommendations



Class of 2024 at the Molecular Workshop



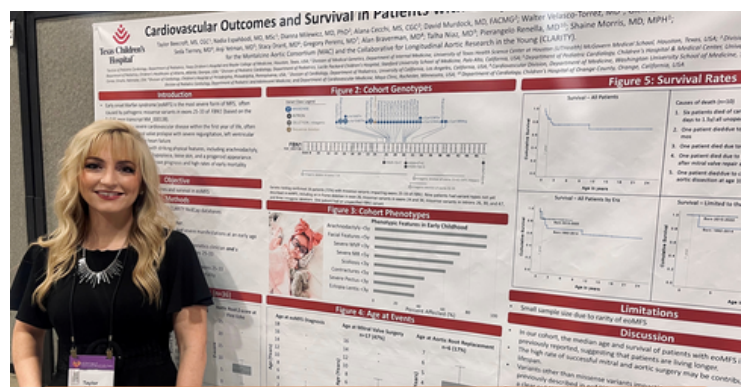
Medical Genetics Faculty at ACMG



APR 14

#232 Entrepreneurship in Genetics with Matt Tschirgi

Matt Tschirgi on the DNA Today Podcast



Taylor Beecroft at World Congress of Pediatric Cardiology and Cardiac Surgery

UTGCP Faculty and Alumni Scientific Publications

Only UT-affiliated authors listed

AJHG

The American Journal of Human Genetics

Amanda Gerard. Monoallelic variation in DHX9, the gene encoding the DEXH-box helicase DHX9, underlies neurodevelopment disorders and Charcot-Marie-Tooth disease. American Journal of Human Genetics. 2023 Aug 13

Kate Richardson, Hope Northrup. SRSF1 haploinsufficiency is responsible for a syndromic developmental disorder associated with intellectual disability. The American Journal of Human Genetics. 2023 Apr 17

AMERICAN JOURNAL OF medical genetics

Jack Colleran, Emily Daykin, Cindy Hernandez, Megan Morand. Novel mosaic TRAF7 likely pathogenic variant in an African American family. American Journal of Medical Genetics. 2023 April 17

Haley Streff. A familial deletion of 10p12.1 associated with thrombocytopenia. American Journal of Medical Genetics. 2023 Sep 25

Haley Streff. A novel de novo pathogenic variant in TBLIXR1 as a new proposed cause of Pierpont syndrome. American Journal of Medical Genetics. 2023 Feb 26

Haley Streff. Novel phenotype of aortic root dilatation and late-onset metabolic decompensation in a patient with TMEM70 deficiency. American Journal of Medical Genetics. 2023 Feb 7

AJOG

American Journal of Obstetrics & Gynecology

Taylor Beecroft, Leslie Dunnington. Prenatal vs postnatal diagnosis of 22q11.2 deletion syndrome: cardiac and noncardiac outcomes through 1 year of age. American Journal of Obstetrics and Gynecology. 2023 Sep 16



children

Jessica O'Shea. Breaking Barriers to Rapid Whole Genome Sequencing in Pediatrics: Michigan's Project Baby Deer. Children. 2023 Jan 4

Clinical Obstetrics and Gynecology

Rosemary Rogers. Chorionic Villous Testing Versus Amniocentesis After Abnormal Noninvasive Prenatal Testing. Clinical Obstetrics and Gynecology. 2023 Sep 1

Blair Stevens. Incidental detection of malignancies with cell-free DNA screening. Clinical Obstetrics and Gynecology. 2023 Jul 14

Cytogenetic and Genome Research

Autumn Vara, S. Shahrukh Hashmi, Victoria F. Wagner, Kathryn Gunther, David F. Rodriguez-Buritica. Frequency of Sex Chromosome Involvement in a Large Cohort of Subjects with Two Copy Number Variants. Cytogenetic Genome Research. 2023 May 18

Frontiers in Medicine

David Rodriguez-Buritica, Meaghan Mones, Kate Richardson. Dermatological concerns for women and girls with turner syndrome. Frontiers in Medicine. 2023 Sep 20

UTGCP Faculty and Alumni Scientific Publications

Only UT-affiliated authors listed

Genetics in Medicine

An Official Journal of the ACMG

Laura Godfrey Hendon. Poison exon annotations improve the yield of clinically relevant variants in genomic diagnostic testing. Genetics in Medicine. 2023 Aug 25

Haley Streff. Clinical genome sequencing: Three years' experience at a tertiary children's hospital. Genetics in Medicine. 2023 Oct 25

Haley Streff. Cascade testing after exome sequencing: Retrospective analysis of linked family data at 2 US laboratories. Genetics in Medicine. 2023 Feb 24

GYNECOLOGIC ONCOLOGY

OFFICIAL PUBLICATION OF THE SOCIETY OF GYNECOLOGIC ONCOLOGY

Erica Bednar. Outcomes of the BRCA Quality Improvement Dissemination Program: An initiative to improve patient receipt of cancer genetics services at five health systems. Gynecologic Oncology. 2023 Mar 31

ELSEVIER

Human Genetics and Genomics Advances



Blair Stevens. Project inclusive genetics: Protecting reproductive autonomy from bias via patient-centered counseling. Human Genetics and Genomics Advances. 2023 Aug 1

JCO™ Clinical Cancer Informatics

Jessica Corredor. LFSPROShiny: An interactive R/Shiny app for prediction and visualization of cancer risks in families with deleterious germline TP53 mutations. JCO Clinical Cancer Informatics. 2023 Aug 15

JCO® Oncology Practice

An American Society of Clinical Oncology Journal

Amie Blanco. Implementation of a telehealth genetic testing station to deliver germline testing for men with prostate cancer. JCO Oncology Practice. 2023 May 19

The Journal of Continuing Education in Nursing®

CONTINUING COMPETENCE
FOR THE FUTURE

Kathleen Shields, Jennifer Czerwinski, Kate Richardson, Syed Hashmi, Paul Hillman, Kathryn Leal. Genetics in the NICU: Nurses' Perceived Knowledge and Desired Education. The Journal of Continuing Education in Nursing. 2023 January 1.

Journal of Genetic Counseling

The official Journal of the
National Society of Genetic Counselors



Myla Ashfaq, Syed Hashmi. Medical Student Knowledge and Clinical Comfort with Medical Genetics in Pakistan. Journal of Genetic Counseling. 2023 Nov 13

UTGCP Faculty and Alumni Scientific Publications

Only UT-affiliated authors listed

Erica Bednar. Implementation and Outcome Evaluations of a Multi-Site Improvement Program in Cancer Genetics. Journal of Genetic Counseling. 2023 Feb

Lauren Douglas, Jennifer Czerwinski, Myla Ashfaq, Rebecca Lunstroth, Chelsea Wagner. Disability education and implications for genetic counselor training. Journal of Genetic Counseling. 2023 Oct 25.

Emily Daykin, Jenny Do. An exploration of knowledge, risk perceptions, and communication in a family with multiple genetic risks for Parkinson's disease. Journal of Genetic Counseling. 2023 Jan 8

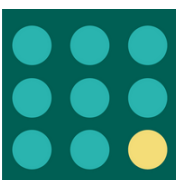
Sarah Huguenard, Haley Streff, Salma Nassef. Perceived impact of ethnocultural competency training on genetic counselors' clinical interactions. Journal of Genetic Counseling. 2023 Aug 10

Matthew L Tschirgi. Abandoning the word Caucasian. Journal of Genetic Counseling. 2023 May 26

Matthew L Tschirgi. Perceived barriers, recommendations, and resources for multistate licensure in the United States: A mixed-methods study of laboratory genetic counselors. Journal of Genetic Counseling. 2023 Oct 5



Myla Ashfaq, Kathryn Leal, Hope Northrup, David Rodriguez-Buritica, Laura Farach. Arginase deficiency masked by cerebral palsy and coagulopathy—Three varied presentations of Latin American origin. Journal of Inherited Metabolic Disease. 2023 Oct 21



Journal of
*Personalized
Medicine*

Laura Godfrey Hendon. Parents' perspectives on the utility of genomic sequencing in the neonatal intensive care unit. Journal of Personalized Medicine. 2023 Jun 21

JAHA

Journal of the American Heart Association

Taylor Beecroft, Alana Cecchi. Vertebral Tortuosity Is Associated With Increased Rate of Cardiovascular Events in Vascular Ehlers-Danlos Syndrome. J Am Heart Assoc. 2023 Oct 3



Jenny Do. Variants in the WDR44 WD40-Repeat Domain Cause a Spectrum of Ciliopathy by Impairing Ciliogenesis Initiation. Nature Communications. 2023 Dec

Haley Streff. FOXL3 pathogenic variants cause one form of craniofacial microsomia. Nature Communications. 2023 Apr 11



Taylor Beecroft. Evaluating Variation in the Cardiac Management of Children with Hereditary Thoracic Aortic Disease in the United States. Pediatr Cardiol. 2023 Sep 27

TP TRANSLATIONAL PEDIATRICS

Aarti Ramdaney. Challenges in the clinical understanding of genetic testing in birth defects and pediatric diseases. Translational Pediatrics. 2023 May 30

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 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.

- **Latonya Alexander (Class of 2023)** Pregnant Black/African Americans' experiences with prenatal genetic counseling
- **Cindy Hernandez (Class of 2023)** Monolingual Spanish speaking patient satisfaction and comfort when comparing Spanish speaking provider to an interpreter
- **Yusra Aziz (Class of 2023)** The need for racial and ethnic health disparity curriculum in genetic counseling programs
- **Tessa Heller (Class of 2023)** The impact of treatment with Palynziq on quality of life for individuals with PKU
- **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

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 our students' research efforts!

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

