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

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Congratulations Class of 2024!

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

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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 Shields, MS, CGC

Mindy Kolodziejski
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
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<th>Student Name</th>
<th>Research Question</th>
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<td>Brenna Albracht</td>
<td>How do aspects of scientific, health, and genetic literacy impact informed decision making in a prenatal genetic counseling patient population?</td>
<td>Meagan Choates, MS, CGC</td>
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<td>Cindy Canales</td>
<td>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?</td>
<td>Kathleen Shields, MS, CGC and Kathryn Leal, MS, CGC</td>
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<td>Jessica Clark</td>
<td>What factors do genetic counseling applicants consider when ranking programs for the Match and how are they prioritized?</td>
<td>Claire Singletary, MS, CGC</td>
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<td>Nadia English</td>
<td>Spanish-Speakers Perspectives of the Benefits and Barriers of Telemedicine in Genetic Counseling</td>
<td>Aranza Gonzalez Cendejas, MS, CGC</td>
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<td>Hillary Esplen</td>
<td>How do cancer rates vary between different ancestries among individuals with Li-Fraumeni syndrome, and are there variant hotspots unique to particular populations?</td>
<td>Jessica Corredor, MS, CGC</td>
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<td>Ava Henson</td>
<td>How the camp PHEver experience has impacted individuals with PKU</td>
<td>Megan Morand, MS, CGC</td>
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<td>Karti Livingston</td>
<td>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?</td>
<td>Leslie Dunnington, MS, CGC</td>
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<td>Farren Lopez</td>
<td>How do levels of satisfaction with core 50 case selection impact perceived levels of preparedness</td>
<td>Aarti Ramdaney, MS, CGC</td>
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<td>Grace Ra</td>
<td>Can an online reproductive genetics module effectively triage patients to genetic counseling?</td>
<td>Blair Stevens, MS, CGC</td>
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<td>Nicole Talaba</td>
<td>To what extent can we expand upon the phenotype of SETD5-related disorders by a utilizing social media support group?</td>
<td>Myla Ashfaq, MS, CGC</td>
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Kate Richardson, Jenny Do, Hope Northrup; Triple diagnoses with overlapping phenotypes: Recommendations and reflections

Kathleen Shields, Hope Northrup; Fifth case of MYH11-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

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

Emily Moura da Silva; Homozygous protein S deficiency in siblings

Emily Moura da Silva; Novel thrombophilia - Prothrombin belgrade variant

Emily Daykin, Syed Hashmi, Kate Richardson, Katie Shields, Molly Daniels, Myla Ashfaq; Investigation of teratogenic concerns in the COVID-19 era
Amanda Gerard; Parental discussion of medical and genetic information with pediatric patients and siblings

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

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

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

Carol Nowlen; Failure of Bladder Emptying (FOBE) and what single gene disorders are teaching us: Not all lower urinary tract obstructions (LUTOs) on prenatal ultrasound have LUTO

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

Jenny Do; Parental stress in tuberous sclerosis complex

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

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

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

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
Erica Bednar; Exploring the experiences of family communication and cascade genetic testing for hereditary predisposition to cancer in medically-underserved populations: A qualitative study

Erica Bednar; Implement this! Learn how to use implementation science to improve your clinic and advance your research through facilitated project-based learning

Sarah Burke; Case report of BICRA-related neurodevelopmental disorder with novel clinical features

Aarti Ramdaney; Specialization in prenatal genetic counseling: Defining the role of the fetal therapy genetic counselor

Jack Colleran, Blair Stevens, Meagan Choates, Syed Hashmi, Brittanie Shelton, Aranza Gonzalez Cendejas; Perceived utility of genetic carrier screening in a diverse patient population

Jessica Corredor; Clinical application of a Li-Fraumeni Syndrome Risk Assessment tool (LFSPRO)

Deanna Darnes, Brad Rolf, Carla McGruder; “Say my name, Say my name”: It’s time to discuss the problem with the name “genetic counselor”

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

Carolyn Haskins; An unusual presentation of apparently de novo MLH1-associated Lynch syndrome

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

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

Nevena Krstic; Prenatal detection of Muenke syndrome via ultrasound and single gene cell-free DNA screening

Karli Livingston; Evaluation of patients with ATM and/or CHEK2 germline likely pathogenic/pathogenic variants and hematological malignancies

Carla McGruder; Rolling the dice on pharmacogenetics: A gamified workshop

Jaqueline Mersch; How referral characteristics impact patient scheduling outcomes

Salma Nassef, Haley Streff; Clinical supervisor feedback growth: A GCP’s experience with increasing supervisor engagement

Brad Rolf; Translating polygenic scores into clinical practice: Insights from genetic counselors

Blair Stevens; Reproductive carrier screening today: Evolving guidelines, novel treatments, and disparities in care

Matt Tschirgi; You Can Do It Too! Genetics Content Creation and Science Communication via TikTok, Instagram, Podcasting, YouTube, LinkedIn, and More

Elise Watson. Jacqueline Mersch; How referral characteristics impact patient scheduling outcomes

Elise Watson, Jacqueline Mersch; I. The view from 30,000 patients: eight years of referral data in a cancer genetics program

Cathy Wicklund; Assessing genetic counselors’ perspectives on acceptability, feasibility, and readiness of chatbots in genetic counseling

Cathy Wicklund; Does microeducation improve knowledge of cardiogenomic risk assessment? An exploratory analysis of online learner data

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
Myla Ashfaq; APHMG 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

Taylor Beecroft at World Congress of Pediatric Cardiology and Cardiac Surgery

#232 Entrepreneurship in Genetics with Matt Tschirgi

Matt Tschirgi on the DNA Today Podcast

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


Haley Streff. A novel de novo pathogenic variant in TBL1XR1 as a new proposed cause of Pierpont syndrome. American Journal of Medical Genetics. 2023 Feb 26


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

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

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


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

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

Jessica Corredor. LFS PRO Shiny: 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

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

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.

Myla Ashfaq, Syed Hashmi. Medical Student Knowledge and Clinical Comfort with Medical Genetics in Pakistan. Journal of Genetic Counseling. 2023 Nov 13
Erica Bednar. Implementation and Outcome Evaluations of a Multi-Site Improvement Program in Cancer Genetics. Journal of Genetic Counseling. 2023 Feb


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


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


Haley Streff. FOXI3 pathogenic variants cause one form of craniofacial microsomia. Nature Communications. 2023 Apr II


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.