JULY 2026

ISSUE NO. 7

UTGCP

Annual Research Newsletter

Class of 2026 and Class of 2027 Thesis Research

Alumni and Faculty Research and Publications from 2025

UT MD Anderson Cancer

UTHealth Houston

Graduate School of Biomedical Sciences

JULY 2026 UTGCP Annual Research Newsletter ISSUE NO. 7

Congratulations to the Class of 2026!

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

Savannah Berger

Profiling Genetic Testing Yield and Racial Disparities in Patients with Male Breast Cancer
Chairs: Hiam Abdel-Salam and Gabriela Chen

Sara Pickett

Conversations Regarding Disordered Eating in Metabolic Patients with Prescibed Diets
Chairs: Paige Roberts and Megan Morand

Jolie Bourek

Do Patients Have Preferences with Regard to Carrier Screening Panel Size?
Chair: Meagan Choates

Jessie Scarlett

Necessity of Diagnostic Testing for ONTD Surgical Candidacy
Chairs: Blair Stevens and Rosemary Rogers

Lamia Haque

Exploring How Teratogen Training in Graduate Programs Impacts Genetic Counselors' Self-Percieved Preparedness Towards Teratogen Training
Chairs: Myla Ashfaq and Kate Richardson

Erin Sharkey

Testing Follow up for Pathogenic/Likely Pathogenic Variants in TP53
Chair: Jessica Corredor

Michelle Luong

Genetic Counselor Perspectives and Shift of Practice Regarding Evolving CDH1 Cancer Risks
Chair: Maureen Mork

Kathryne Rojeck

Personal and Family History of Patients with RAD51C, RAD51D, and BARD1 Pathogenic/Likely Pathogenic Variants
Chair: Jessica Corredor

Katie Ledden

Evaluation of Genetics Awareness Among Ophthalmology Residents
Chairs: Katie Leal and Leslie Dunnington

Marissa Williams

Diagnostic Yield of Genetic Testing in High Risk Patients
Chair: Katie Leal and Jordan Zeiger

JULY 2026 UTGCP Annual Research Newsletter ISSUE NO. 7

Introducing the Research of the Class of 2027!

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Allison Brewer

Parental Perceptions of the Impact of Disease-Specific Camps on Children's Psychosocial Well-Being and Disease Management
Chairs: Megan Morand

Lauren Cho

Characterizing the Session Components and Psychosocial Skills Utilized in Inpatient and Outpatient Genetic Counseling Sessions
Chairs: Jordan Zeiger and Sophia Bradley

Brady Erlandson

Determining Reproductive Outcomes in Achondroplasia
Chair: Dr. David Rodriguez

Alexa Grimm

Evaluating Case Volume Adequacy for Practice-Based Competency Application: Insights from Fieldwork Supervisors
Chair: Aarti Ramdaney

Stephanie Helton

Identification the Factors that Support and Prohibit the Creation and Maintenance of Active Patient Advocacy Groups for Rare Genetic Conditions
Chairs: Leslie Dunnington and Dr. Farach

Jianna Herman

Testing the Timeline: Factors Influencing Prenatal Genetic Test Completion Times
Chairs: Meagan Choates

Emily McDonald

Value-Consistent Carrier Screening Decision-Making Following an Online Prenatal Genetics Education Module
Chairs: Blair Stevens

Faith Peterson

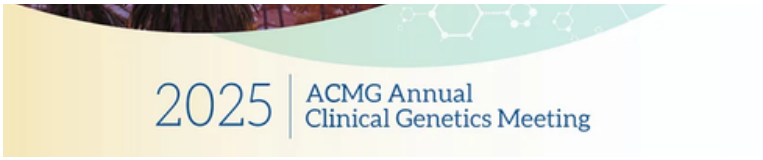
Barriers to Prenatal Care: Initial Appointment Outcome Trends in Prenatal Genetic Counseling, Maternal Fetal Medicine, and Dietitian Appointments
Chair: Samantha Montgomery

Melana Vaughn

Investigating Sleep Disorders in the Pediatric Tuberous Sclerosis Complex Population
Chair: Kate Richardson

UTGCP Faculty and Alumni Authorship at National and Local Meetings

only UT-affilated authors listed



Megan Morand, Joseph Ray; Rare Adult Case of Stüve-Wiedemann syndrome

Carol Nowlen, Katelyn Sagaser; Genetic Etiologies of Bilateral Renal Agenesis

Nevena Krstic; Expanding the fetal phenotype of USP7-related conditions

Sarah S. Barnett; Clinical Utility of Exome and Genome Sequencing in an Adult Cohort Referred for Suspicion of Underlying Rare Disease

Sandra Darilek; A Case Series and Functional Study of Two Commonly Reported Pathogenic Variants in OTC Supports a Hypomorphic Classification

Myla Ashfaq, Paul Hillman; A case of Neurodevelopmental-Craniofacial Syndrome with Variable Renal and Cardiac Abnormalities due to a de novo heterozygous ZMYM2 pathogenic variant.

Paul Hillman, Joseph Ray, Hope Northrup; Use of Cholic Acid in Smith-Lemli-Opitz Syndrome (SLOS): Real-World Patient Outcomes

Kate Richardson, Hope Northrup, Kathleen Shields; Unpacking Genotype-Phenotype Correlation in DNM1 Encephalopathy: Two unusually mild cases with variants in the GTPase and proline-rich domains

Kathleen Shields, Shannon Mulligan, David Rodriguez-Buritica; Case Study of Yp11.31 Deletion in XXY Phenotypic Female

Kate Richardson, Hope Northrup; New Familial Variant Identified in CSDE1-Associated Neurodevelopmental Disorder

Hope Northrup; Investigating Sex Differences in the Behavioral Phenotypes of Phelan-McDermid Syndrome, Tuberous Sclerosis Complex, and PTEN Hamartoma Tumor Syndrome

David Rodriguez-Buritica; A Case Report of Meckel Gruber Syndrome: Identification of an Unreported Variant Correlating with Diagnosis

David Rodriguez-Buritica, Jacqueline Hecht; How helpful are sleep studies in determining surgical need in infants with achondroplasia?

David Rodriguez-Buritica; PTEN and WDR19 Variants in a Neonate with Brain and Cutaneous Anomalies

David Rodriguez-Buritica; Qualitative Interviews to Support the Development of a Patient-Reported Outcome (PRO) Measure for Glycogen Storage Disease Type Ia

Jenny Do, Erin Cooney; Novel UBA1 Variant Associated with Attenuated X-linked SMA Phenotype

Megan Morand, Joseph Ray; Neonate with a constellation of atypical congenital anomalies.

UTGCP Faculty and Alumni Authorship at National and Local Meetings

only UT-affilated authors listed

David Rodriguez-Buritica; Latin-American GSD family meeting: Updates on gene therapy for GSDIa and management of GSD during critical times

Chelsea Wagner, Samantha Montgomery; Evolving U.S. Abortion Laws: Implications for Genetic Counseling in Prenatal Diagnosis

Carol Nowlen; False negative cell-free DNA screening: A cautionary case series

Carol Nowlen; Expanding Horizons: Evolving Genetic Criteria for Fetal Intervention

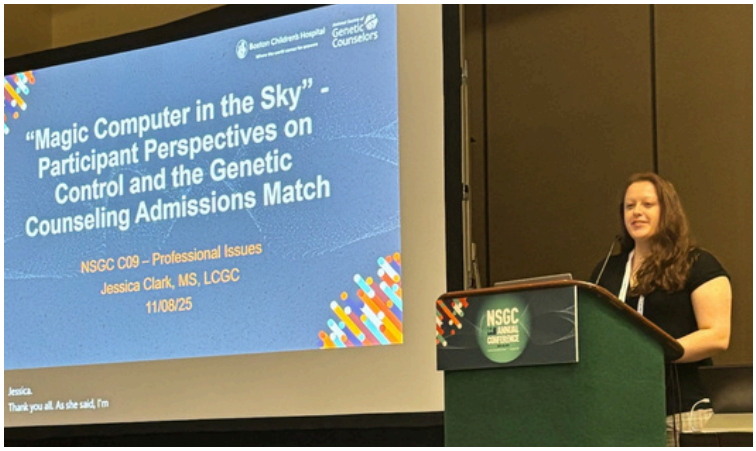
ICIEM 2025 KYOTO

David F Rodriguez-Buritica; Results From a Pivotal Phase 3 Double-blind Placebo-controlled Trial of DTX401 for the Treatment of Individuals With Glycogen Storage Disease Type Ia

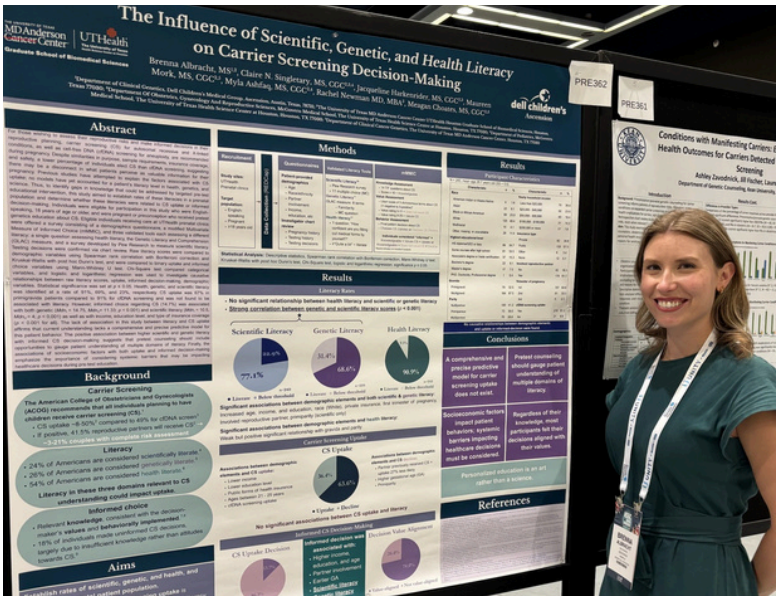
ISPOR Europe 2025
9 - 12 November

David F Rodriguez-Buritica; The Clinical Burden and Complications Associated with Glycogen Storage Disease Type Ia (GSDIa): Results From a Systematic Literature Review

UTGCP Faculty and Alumni Authorship at National and Local Meetings
only UT-affiliated authors listed

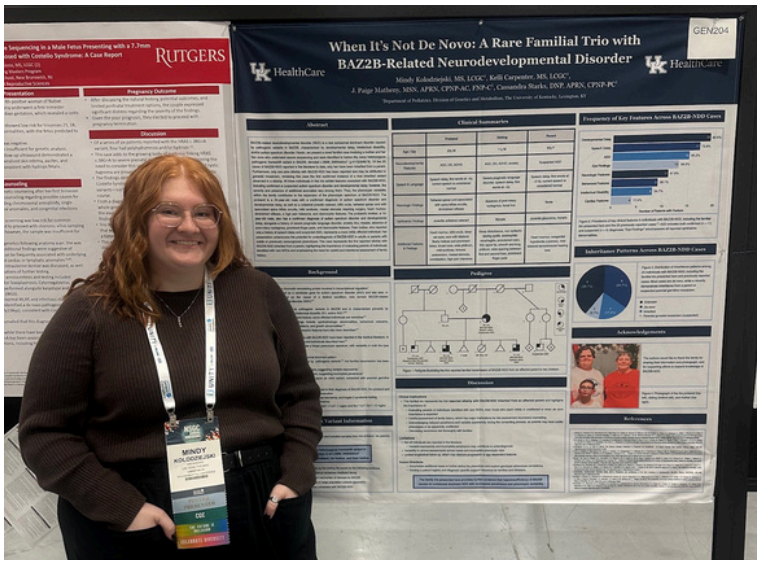


Jessica Clark, Meagan Choates, Megan Morand, Laura Farach, Luana Goulet, Claire Singletary; Magic Computer in The Sky – Participant Perspectives on Control and the Genetic Counseling Admissions Match



Brenna Albracht, Meagan Choates, Myla Ashfaq, Jacqueline Harkenrider, Maureen Mork, Claire Singletary; The Influence of Scientific, Genetic, and Health Literacy on Carrier Screening Decision-Making

Ava Henson, Kate Richardson, Colin Bixler, Hope Northrup, Jessica Corredor, Megan Morand; "The Holy Grail of PKY Support" Exploring the Psychosocial Impacts of Camps for Kids with PKU

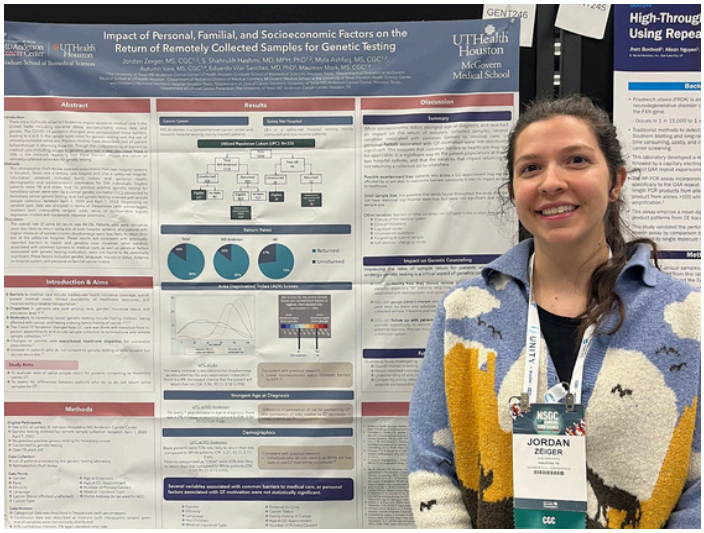


Mindy Kolodziejski; When It's Not De Novo: A Rare Familial Trio with BAZ2B-Related Neurodevelopmental Disorder

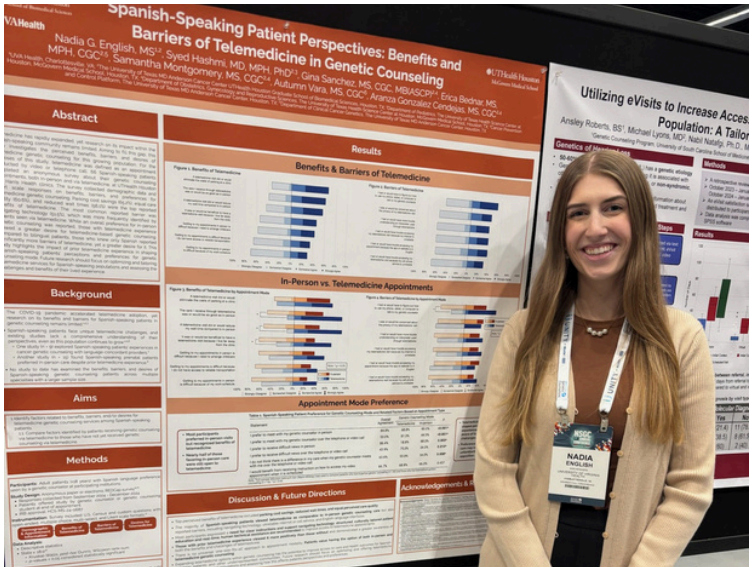
UTGCP Faculty and Alumni Authorship at National and Local Meetings

only UT-affiliated authors listed

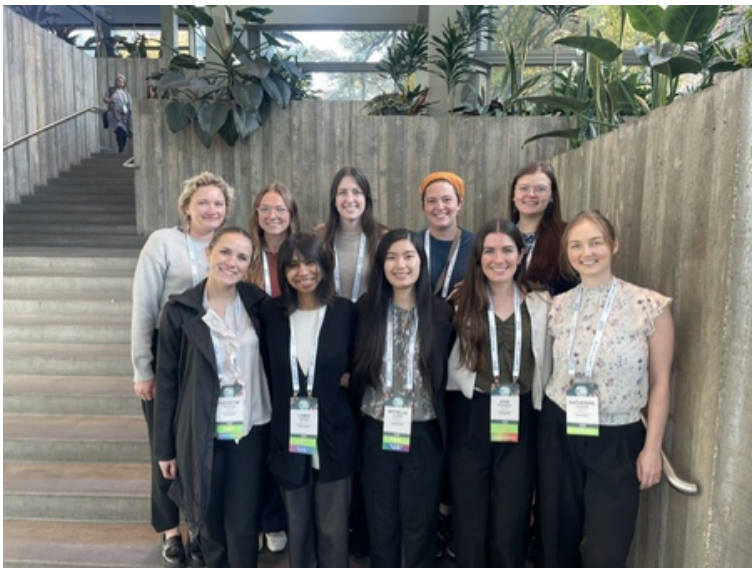
Rosemary Rogers, Meagan Choates, Claire Singletary, Kristen Kelly, Gina Sanchez, Aarti Ramdaney, Blair Stevens; Diagnostic testing following atypical NIPT result: A retrospective cohort study



Jordan Zeiger, Syed Hashmi, Myla Ashfaq, Autumn Vara, Maureen Mork; Impact of Personal, Familial, and Socioeconomic Factors on the Return of Remotely Collected Samples for Genetic Testing



Nadia English, Syed Hashmi, Gina Sanchez, Erica Bednar, Aranza Gonzalez Cendejas; Spanish Speaking Patient Perspectives: Benefits and Barriers of Telemedicine in Genetic Counseling



Class of 2026 at NSGC in Seattle

UTGCP Faculty and Alumni Authorship at NSGC*
only UT-affiliated authors listed*

Jacqueline Mersch: Hereditary Melanoma: Who Needs Genetic Testing?

Nevena Krstic: A Familial Case of PPP1R12A-Related Urogenital and/or Brain Malformation Syndrome

Brad Rolf: Assessing Genetic Testing Confidence and Practices Among Puerto Rican Physicians

Karli Livingston, Leslie Dunnington, Syed Hashmi, Christina Falugi, Emile Moura Coelho da Silva, Jordan Zeiger: Impact of Personal and Family History of Huntginton's Disease on the Decision and Ability to Obtain Life, Long-Term Care, and Disability Insurance

Erica Bednar, Autumn Vara: Effects of Hereditary Cancer Educational Videos on Safety-Net Hospital Patients' Knowledge and Self-Efficacy

Cathy Sullivan, Salma Nassef: Examining the Accuracy of ChatGPT-Generated Information on Genetic Conditions

Lukas Kruidenier: SeqFirst-neo- Utilizing a Disruptive Approach to Increase Access to a Precise Genetic Diagnosis in the NICU

Nevena Krstic: Fetal Diagnosis of DOK7-Related Fetal Akinesia Deformation Sequence: a Case Series Expanding the Data on Phenotype and Mutational Spectrum

Lukas Kruidenier: SeqFirst-neo- Utilizing a Disruptive Approach to Increase Access to a Precise Genetic Diagnosis in the NICU

Roya Mostafavi: The Importance of Family and Provider Communication for a Patient with Multiple Congenital Anomalies: A Case Report

Karli Livingston, Leslie Dunnington, Syed Hashmi, Christina Falugi, Emile Moura Coelho da Silva, Jordan Zeiger; Achieving the Practice-Based Competencies: Are More Than 50 Cases Needed?

Cathy Wicklund: Perspectives of Prenatal Genetic Counseling Clinical Supervisors in Educating Genetic Counseling Graduate Students About Reproductive Rights

Jianna Herman, Yusra Aziz; Hereditary Heme Mailgnancy Woes: Will Your Variants be Reclassified?

Cathy Wicklund: Exploring the Unmet Needs of Siblings of Individuals Diagnosed with Smith Magenis Syndrome

Matthew Tschirgi: A Tau-tally New Era: Genetic Counseling Meets Alzheimer's Biomarker Testing

Farren Lopez, Syed Hashmi, Myla Asfaq, Aarti Ramdaney; Achieving the Practice-Based Competencies: Are More Than 50 Cases Needed?

Jacqueline Mersch: Hereditary Melanoma: Who Needs Genetic Testing?

Matthew Tschirgi: Think Big, Start Small: A Practical Guide to Entrepreneurship

Laura Hendon: The Evolving Landscape of Screening for Inherited Conditions: From Carrier Status to Clinical Manifestations NIPT

Kaylene Ready Beyond Expanded Carrier Screening: Detection of Inherited Disorders Using Preimplantation Genetic Testing with Whole Genome Sequencing

Angie Jacobson: GCA-IGA Presents: Soecialized Technologies for Challenging Gastrointestinal Cancer Genetics Cases

Tricia Page: Breaking the Mold: Transforming Industry Careers

UTGCP Faculty and Alumni Scientific Publications
only UT-affiliated authors listed

Journal of Genetic Counseling

Deanna Darnes. Shifting ownership, shifting protections: Patient privacy and genetic data ownership in the era of mergers and acquisitions

Deanna Darnes. Shifting ownership, shifting protections: Patient privacy and genetic data ownership in the era of mergers and acquisitions

Chelsea Wagner, Samantha Montgomery. Navigating the new landscape: A review of evolving abortion legislation impacting genetic counselors' scope of practice in the United States

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

Erin Salo-Mullen. Call to action for genetic counseling research in hereditary cancer: Considerations from the evidence-based guidelines development process

Erin Salo-Mullen. Call to action for genetic counseling research in hereditary cancer: Considerations from the evidence-based guidelines development process

Jennifer Lemoine . Incidental finding of maternal sex chromosome aneuploidy from DMD carrier screening and single-nucleotide polymorphism (SNP)-based prenatal cell-free DNA screening

Natalie Stoner, Meagan Choates, Carla McGruder, Theresa Wittman, Sara Wofford, Claire N Singletary. Invisible diversities, academic capital, and competitiveness of genetic counseling applicants

Laura Hendon. Prenatal genetic counseling challenges with indeterminate SMA results

Mindy Kolodziejski, Natalie Stoner, Samantha Montgomery, Shannon Mulligan. Patient understanding of fetal sex versus gender in the context of routine cell-free DNA screening

Regina Nuccio. A landscape assessment of Medicaid recognition for genetic counselors

Julia Wynn. Inclusive genetic counseling for LGBTQ+ clients: A qualitative study of reproductive genetic counselors' experiences and perspectives

AJMG American Journal of Medical Genetics Part A

Claire Singletary. Self-Reported Access to Specialized Genetics Providers Among Families of Young Children With Birth Defects in Texas

Amanda Gerard. Unraveling the Molecular and Clinical Consequences of an Intragenic TRIP12 Duplication Using Genomic and RNA Analyses

Justine Pickarski. Genetic Services in Appalachia Conference Series

TH OPEN
Companion journal to Thrombosis and Haemostasis

Emile Moura Coelho da Silva. Identification of Prothrombin Belgrade Variant in a Mexican-American Family with Recurrent Deep Vein Thrombosis

Additional UTGCP Faculty and Alumni Scientific Publications
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Kristen Kelly. Genome Sequencing for All Pregnant Persons: Navigating the Next Frontier in Prenatal Diagnosis Through Patient Reflections

Laura Hendon. Cell-Free DNA Results Indicating Mosaic Monosomy X of Likely Maternal Origin: Impact on Genetic Counseling Practices and Patient Experiences

Nevna Krstic. Identification and Prenatal Evaluation of Suspected Congenital Cataracts: Three Very Different Cases

Nevna Krstic.The evolution of teratology: Historical perspectives and lessons learned

Julia Wynn. Clinical Performance of Cell-Free DNA for Fetal RhD Detection in RhD-Negative Pregnant Individuals in the United States

Natalie Stoner, Jack Colleran. Seeking answers, getting questions: Genome-wide prenatal cell-free DNA screening in a case of multiple congenital anomalies

Laura Amendola. ACMG SF v3.3 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG)

Laura Amendola. Data-driven consideration of genetic disorders for global genomic newborn screening programs

Laura Amendola. Development of a comprehensive cardiovascular disease genetic risk assessment test

Laura Amendola. When families bridge the research-clinical divide: An exploration of values regarding cascade screening in genomic research

Lukas Kruidenier. Expanding implementation of pediatric whole-genome sequencing: Insights from SeqFirst providers to inform equitable access to a precise genetic diagnosis

Additional UTGCP Faculty and Alumni Scientific Publications
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Case Reports in Genetics

Laura Amendola. Long-Read Sequencing as a Diagnostic Tool for Primary Ciliary Dyskinesia

JAMA Journal of the American Medical Association

Laura Amendola, Julia Wynn. Expanded Newborn Screening Using Genome Sequencing for Early Actionable Conditions

Molecular Genetics and Metabolism

Lauren Youngborg. The PKU Patient Registry: Development of a patient-driven registry and initial outcomes

JACI The Journal of Allergy and Clinical Immunology IN PRACTICE

Shelly Zelnick. Genetic Counseling in Pediatric Inborn Errors of Immunity: Perspective Piece on Current Practice and Considerations

THE JOURNAL OF PEDIATRICS

Lukas Kruidenier. Implementation of First-Line Rapid Genome Sequencing in Non-Critical Care Pediatric Wards

PEDIATRIC NEUROLOGY

Laura Gorecki, Syed Hashmi, Jenny Do, Laura Farach, Hope Northrup, Deobrah Pearson, Kate Richardson. Influential Factors for Disclosing a Tuberous Sclerosis Complex Diagnosis to Romantic Partners

ELSEVIER American Journal of Human Genetics AJHG

Jessica O'Shea. Discovery of a DNA methylation profile in individuals with Sifrim-Hitz-Weiss syndrome

SLEEP and Breathing

International Journal of the Science and Practice of Sleep Medicine

David F Rodriguez-Buritica, Jacqueline Hecht, Syed Hashmi. Evolution of sleep disordered breathing in infants with achondroplasia

IJCMCR Open Access International Journal of Clinical Studies & Medical Case Reports

Megan Morand, Joseph Ray. Birth Trauma in a Patient with Prader-Willi Syndrome: A Diagnostic Dilemma.

UTGCP Faculty and Alumni Scientific Publications
only UT-affiliated authors listed

BRAIN COMMUNICATIONS — OXFORD UNIVERSITY PRESS

Alice Schindler. CSF and blood neuronal injury biomarkers in spinal bulbar muscular atrophy and amyotrophic lateral sclerosis 4

CTS Clinical and Translational Science — Open Access

Carley Brueckner. Multi-Gene Pharmacogenomic Testing in a Community-Based Setting Is Feasible and Reduces Total Healthcare Costs

JIMD JOURNAL OF INHERITED METABOLIC DISEASE

David F Rodriguez-Buritica. Safety and Efficacy of DTX401, an AAV8-Mediated Liver-Directed Gene Therapy, in Adults With Glycogen Storage Disease Type I a

David F Rodriguez-Buritica. State of the Art and Consensus Statements by Healthcare Providers, Patients, and Caregivers on Continuous Glucose Monitoring in Liver Glycogen Storage Diseases

nature COMMUNICATIONS

Sara Pickett. Urinary Complement proteome strongly linked to diabetic kidney disease progression

OXFORD JOURNALS — Human Molecular Genetics

Kate Richardson, Hope Northrup. KDM2B variants in the CxxC domain impair its DNA-binding ability and cause a distinct neurodevelopmental syndrome

ELSEVIER Kidney Medicine — Kidney Medicine

Catherine Wicklund. Development and Evaluation of a Culturally Targeted Apolipoprotein L1 Counseling Training Program for Transplant Clinicians: A Pilot Study

ELSEVIER Molecular Genetics and Metabolism Reports — MGM Reports

Sara Pickett. Application of high-resolution mass spectrometry profiling towards the diagnosis and acute management of maple syrup urine disease

nature portfolio

Laura Hendon. Pathogenic UNC13A variants cause a neurodevelopmental syndrome by impairing synaptic function

Journal of Community Genetics — Springer — springer.com

Erin Atkinson. Genetics services in Latin America: a descriptive study of availability and utilization of genetics in healthcare

Additional UTGCP Faculty and Alumni Scientific Publications
only UT-affiliated authors listed

JBMR Journal of Bone and Mineral Research

David F Rodriguez-Buritica. Switching from active vitamin D and phosphate supplementation to burosumab significantly corrects lower limb malalignment in pediatric X-linked hypophosphatemia

The Oncologist — OXFORD UNIVERSITY PRESS

Rachel Bluebond, Autumn Vara, Banu Arun. Improving genetics equity: identifying women eligible for genetic care services using mammography clinics in underserved areas as screening hubs

GENEReviews

Alice Schindler. Autosomal Dominant TRPV4- Related Disorders

Molecular Genetics & Genomic Medicine MGGM

Jacqueline Mersch. Electronic Patient Portals as a Modality for Returning Reclassified Genetic Test Results

Hiam Abdel-Salam. Characterization of the Germline Pathogenic Mutational Landscape and Oncologic Outcomes Among 877 Patients with Invasive Lobular Carcinoma

Volume 44, Number 3, March 2026
UROLOGIC ONCOLOGY
Seminars and Original Investigations

Jacqueline Mersch. Prostate cancer-related genetic counseling in a safety-net healthcare setting

JCO® Precision Oncology
An American Society of Clinical Oncology Journal

Hiam Abdel-Salam. Unveiling the Origins of Pathogenic TP53 Variants in Clinical Context: A Molecular Tumor Board Case Discussion

Justine Cooper Pickarski. Germline POT1 Variants in a Pan-Cancer Cohort

Regina Nuccio. Contribution of Germline Predisposition to Pediatric Thyroid Cancer

UTGCP Research and Education Fund Awardees

Distributions from the Genetic Counseling Research and Education Endowed Fund are used to support students enrolled in the genetic counseling program at the MD Anderson UTHealth Houston Graduate School. This past year, distributions directly supported student research projects. These students completed short-term projects focused on clinical care or professional development in genetic counseling. Endowment resources enabled the students to provide incentives to assist with recruiting participants. Those who completed the surveys were entered into a drawing for an Amazon gift card. Gifts to this fund are fundamental to the success of student projects and the completion of their degree program.

Class of 2026

Katie Ledden
Sara Pickett
Jolie Bourek
Michelle Luong
Lamia Haque

Class of 2025

Brenna Albracht
Karli Livingston
Farren Lopez
Jessica Clark
Ava Henson

Class of 2024

Emma Billings
Carley Brueckner
Laura Gorecki
Maria Hernandez
Mindy Kolodzieski
Disha Patel
Kirstin Risgaard
Jordan Steffen

Class of 2023

Latonya Alexander
Cindy Hernandez
Yusra Aziz
Tessa Heller

Class of 2022

Katie Baudoin
Emile Moura Coelho da Silva
Gina Sanchez
Natalie Stoner

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.

To Our Alumni and Faculty Communities:

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!

And if you want to join in and assist with the creation of the newsletter next year, please let us know!

Samantha.Montgomery@uth.tmc.edu