

UTGCP ANNUAL RESEARCH NEWSLETTER

Celebrating Excellence in Genetic Counseling Research



***shout-out to our students completing their
M.S. research during a pandemic***

what's inside:

- Research from our students (classes of 2020 & 2021)
- Research submitted by alumni & faculty (Jan-Dec 2019)

Hyperlinks to the complete abstracts and conferences are included when available.

Please consider supporting our students' research efforts through the UTGCP Research & Education Fund at launch.uth.edu/utgcp

CONGRATS CLASS OF 2020!

Thesis defense looked a little different this year...



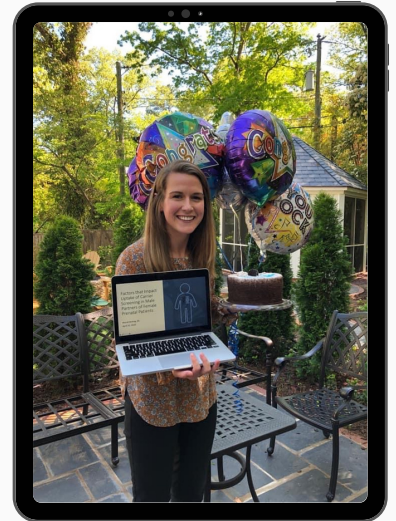
Emily Stiglich - "Genetic counselors' experiences with and approaches to discordant genotypic and phenotypic sex"

Advisor: Victoria Wagner, MS, CGC



Sarah Burke - "Understanding genetic counselors' approaches to direct-to-consumer testing for hereditary breast cancer"

Advisor: Maureen Mork, MS, CGC



Wendi Betting - "Factors that impact uptake of carrier screening by male reproductive partners of female prenatal patients"

Advisor: Meagan Choates, MS, CGC



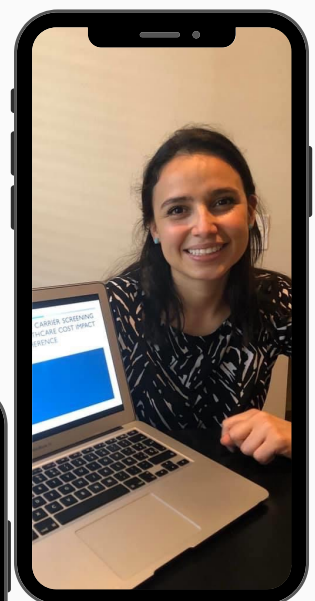
Autumn Vara - "Frequency of copy number variants involving the sex chromosomes in a clinical setting"

Advisor: David F. Rodriguez-Buritica, MD



Addison Johnson - "Assessing patient attitudes toward genetic testing for hereditary hematologic malignancies"

Advisor: Sarah Bannon, MS, CGC



Aranza Gonzalez-Cendejas - "Implementation of genetic carrier screening in the OB population: healthcare cost impact and recommendation adherence"

Advisor: Rebecca Carter, MS, CGC

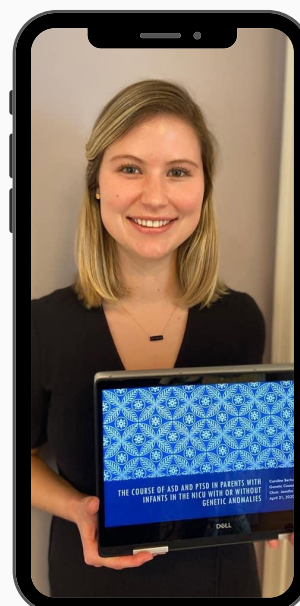


Bradley Power - "Investigating medical examiner's practices: genetic evaluation for fatal acute aortic dissection"

Advisor: Krista Qualmann, MS, CGC

Caroline Bertsch - "The course of acute stress disorder and post traumatic stress disorder in patients and infants in the neonatal intensive care unit with or without genetic anomalies"

Advisor: Jennifer Czerwinski, MS, CGC



Looking ahead to the Class of 2021

SECOND YEAR RESEARCH TOPICS



Kaitlyn Amos - "Influence of personal story on psychosocial genetic counseling practice and specialization"
Advisor: Claire N. Singletary, MS, CGC

Renee Bardini - "Isolated absent cavum septum pellucidum: a single center's outcomes and review of the literature"
Advisor: Blair Stevens, MS, CGC

Grant Bonestelle - "Assessing genetic counselor attitudes and practices concerning variant reclassification downgrades"
Advisor: Jessica Corredor, MS, CGC

Lauren Douglas - "Disability awareness education: what's missing? Perspectives from genetic counselors and the disability community"
Advisors: Chelsea Wagner, MS, CGC

Katlyn Frane - "Evaluating insurance approval rates of whole exome sequencing and its effects on minority patients' access to genetic care"
Advisor: Victoria Wagner, MS, CGC

Katie Huang - "Barriers experienced by underrepresented minority applicants in becoming a competitive genetic counseling applicant"
Advisor: Claire Singletary, MS, CGC

Lukas Kruidenier - "Assessing patient coping strategies and decision-making factors related to genetic testing after an abnormal ultrasound finding"
Advisor: Blair K. Stevens, MS, CGC

Emily Martin - "Breast cancer risk for female relatives of male breast cancer patients with negative *BRCA1/2* testing"
Advisor: Meagan Kaulfus, MS, CGC

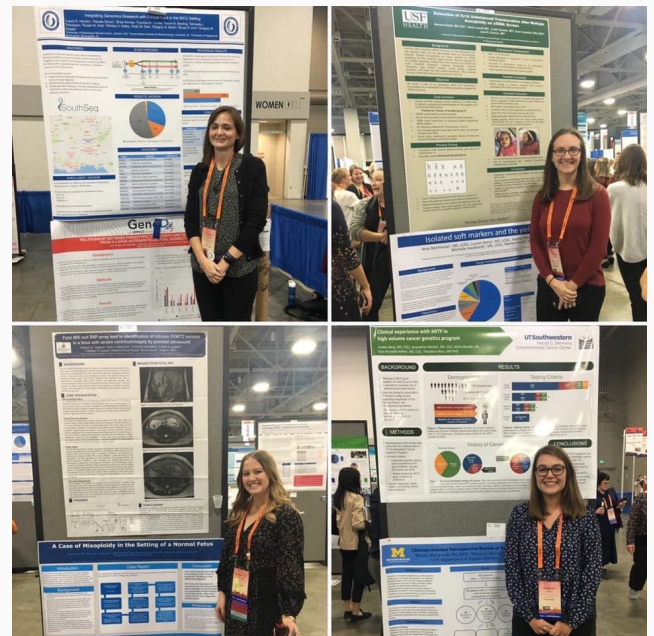
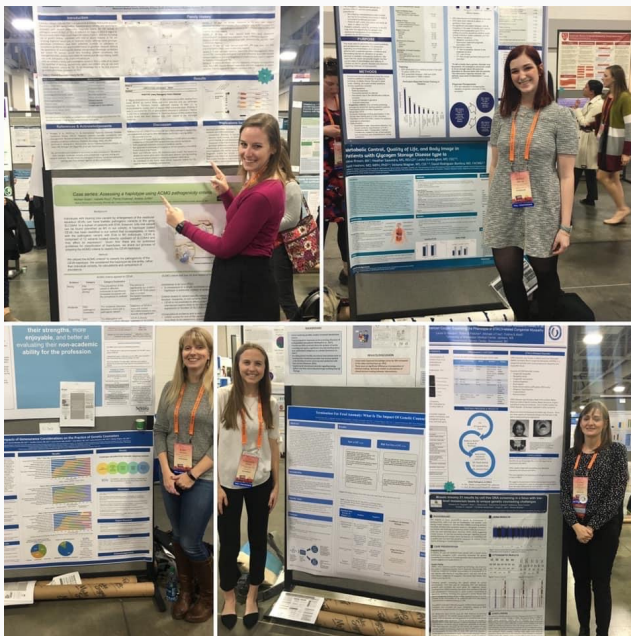
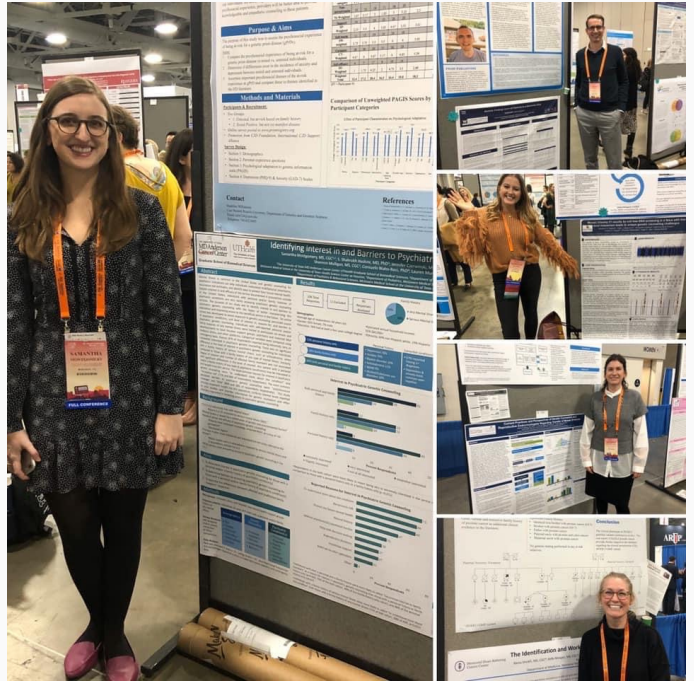
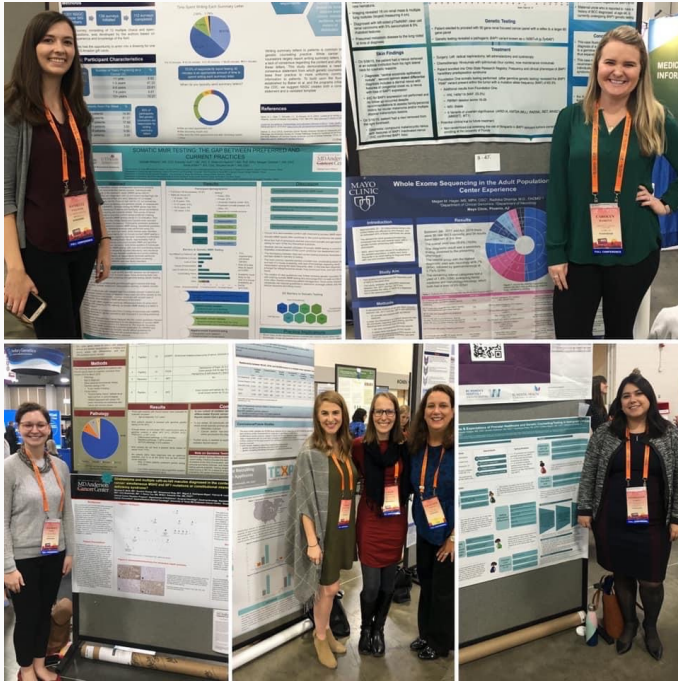
Meaghan Mones - "Dermatological concerns for women and girls with Turner syndrome"
Advisor: David F. Rodriguez-Buritica, MD

Megan Pope - "Understanding adolescent and young adult cancer patient decision-making to pursue genetic counseling and/or testing"
Advisor: Jessica Corredor, MS, CGC

Rose Rogers - "Assessing the spectrum of therapy-related and de novo hematologic malignancies in germline *BRCA1* and *BRCA2* mutation carriers"
Advisor: Sarah Bannon, MS, CGC

Katie Shields - "NICU nurses perceived and desired genetics knowledge"
Advisor: Kathryn Gunther, MS, CGC

UTGCP Alumni & Faculty at National Meetings



UTGCP Alumni & Faculty Authorship at National Meetings

submitted presentations from 2019

(only UT authors listed below)

National Society of Genetic Counselors Annual Conference

Laura Amendola ('09). Improving your communication with all your patients: techniques to communicate across literacy and language. (educational breakout session)

Erica Bednar (faculty). Assessing readiness for hereditary cancer cascade testing. (poster)

Jordan Berg ('17) & Jackie Mersch ('14). Clinical experience with MITF in high volume cancer genetics program. (poster)

Rebecca Carter ('09), Callie Jenevein Diamonstein ('15) &

Katie Sagaser ('15). Perinatal palliative care and the genetic counselor: optimizing multi-disciplinary collaboration for holistic care of critically ill infants and their families. (pre-conference symposium)

Georgeann Garza ('19), Sarah Jane Noblin ('99), Priscilla Hodges (faculty), Jen Hoskovec ('03) & Chelsea Wagner ('16). Exploring experiences and expectations of prenatal healthcare and genetic counseling/testing in immigrant Latinas. (poster)

Laura Godfrey Hendon ('11). Recurrent non-immune fetal hydrops (NIFH) due to Native American Myopathy (NAM) in an African-American couple: expanding the phenotype of STAC3-related congenital myopathy. (poster)
Integrating genomics research with clinical care in the NICU setting. (poster)

Sarah (Mayes) Huguenard ('15) & Sarah Jane Noblin ('99). Webinar outreach: an effective tool for recruiting potential genetic counseling program applicants. (poster)

Emily Krossschell ('19), Lauren Murphy ('09), Jennifer Lemons (faculty), Laura Farach (faculty), Ashley Woodson ('11) & Chelsea Wagner ('17). Impacts of Genesurance considerations on genetic counselors' practices and attitudes. (poster)

Peyton Nunley ('19), Syed Hashmi (faculty), Myla Ashfaq (faculty), Laura Farach (faculty), Claire Singletary (faculty) & Blair Stevens ('08). Exploring the potential yield of prenatal testing by evaluating a postnatal population with structural abnormalities. (poster)

Katie Sagaser ('15) & Katie Rock Forster ('12). Fetal MRI and SNP-array lead to identification of intronic *POMT2* variants in a fetus with severe ventriculomegaly by prenatal ultrasound. (poster)
Mosaic trisomy 21 results by cell free DNA screening in a fetus with low-level mosaicism leads to unique genetic counseling challenges. (poster)

Katie Sagaser ('15). "A calming reassurance": the role of patients' religion/spirituality in prenatal genetic counseling appointments at religiously-affiliated and non-religiously affiliated institutions. (poster)

Haley Streff ('15) & Hope Northrup (faculty). Genotype vs phenotype: pathogenic variant in TSC1 in a three-generation family without clinical evidence of tuberous sclerosis complex. (poster)

Matthew Tschirgi ('08). Utilizing genetic counseling assistants to ease the burden of multi-state genetic counselor licensure. (poster)

Victoria Wagner ('16) & Myla Ashfaq (faculty). Hot topics in teratology: Zika, marijuana, and more. (educational break-out session)

Danielle Williams ('19), Syed Hashmi (faculty), Meagan Choates ('16), Sarah Jane Noblin ('99), Maureen Mork (faculty). Somatic MMR testing: the gap between preferred and current practices. (poster)

Julia Wynn ('06). Non-traditional genetic counseling: weighing the alternatives. (educational break-out session)

A qualitative study of Latinx parents' experiences of clinical exome sequencing. (poster)

American Society of Human Genetics

Laura Amendola ('09). Results from clinical exome sequencing for cancer risk assessment in primary care patients who screen positive for genetic risk. (poster)

Sarah (Mayes) Huguenard ('15), Salma Nassef ('12), Sandra Darilek ('03), Andi Lewis ('14) & Haley Streff ('15). Integrating an alternative genetic services delivery model using an online platform. (poster)

American College of Medical Genetics & Genomics Annual Clinical Genetics Meeting

Laura Amendola ('09). Approaches to secondary findings across the clinical sequencing evidence-generating research (CSER) consortium. (poster)
Returning results using a literacy-focused genetic counseling approach: early experiences from the CHARM study. (workshop)

Haley Streff ('15) & Amanda Gerard ('17). Pre-authorization for whole exome sequencing: one center's experience. (poster)

Matt Tschirgi ('08). Cell-free DNA screening detects maternal blood chimerism - a case report. (poster)

Society for Maternal Fetal Medicine

Katie Sagaser ('15). Impact of maternal and paternal factors on fetal nasal bone visualization during first trimester screening. (poster)

Katie Sagaser ('15) & Katie Rock Forster ('12). Clinical vs novel single nucleotide variants identified by whole exome sequencing in fetuses with pleural effusions. (poster)

International Society of Prenatal Diagnosis

Salma Nassef ('12), Sarah (Mayes) Huguenard ('15) & Sandra Darilek ('03). Patient testing decisions following transfer of an aneuploid embryo. (poster)

Katie Sagaser ('15) & Katie Rock Forster ('12). Autosomal dominant hypophosphatasia (HPP): a condition overlooked on expanded carrier screening? (poster)

International Fetal Medicine and Surgery Society Meeting

Katie Sagaser ('15) & Katie Rock Forster ('12). Fool me once: the dilemma of fetal long bone bowing.

The Children's Tumor Foundation 2019 NF Conference

Leslie (Durham) Granger ('17). Phenotype-genotype correlations with Noonan syndrome like features in Neurofibromatosis type 1. (poster)

Simpósio Internacional em Oncogenética

Erica Bednar (faculty). Universal genetic testing quality improvement initiative in gynecologic cancer.

CSER/eMERGE Joint Winter Meeting

Julia Wynn ('06). Videos for scalable genomic education.

ASCO Genitourinary Cancers Symposium

Annelise Pace ('19), Ashley Woodson ('11), Molly Daniels (faculty), Jen Hoskovec ('03). Real-world outcomes of genetic testing in a genitourinary genetics clinic and evaluation of current guidelines.

UTGCP Alumni & Faculty Scientific Publications

submitted peer-reviewed publications from 2019 - only UT authors listed below

Journal of Genetic Counseling

Danielle Williams ('19), S. S. Hashmi (faculty), Meagan Choates ('16), Sarah Jane Noblin ('99), Maureen Mork (faculty). Somatic Mismatch Repair Testing in Evaluations of Lynch Syndrome: The Gap Between Preferred and Current Practices.

Blair Stevens ('08), Sarah Jane Noblin ('99), Jennifer Czerwinski ('03), Chelsea Wagner ('17). Introduction of cell-free DNA screening is associated with changed in prenatal genetic counseling indications.

Lauren Fledderman ('19), S. S. Hashmi (faculty), Blair Stevens ('08), Lauren Murphy ('08), David Rodriguez-Buritica (faculty). Current Genetic Counseling Practice in the United States Following Non-Invasive Prenatal Testing For Sex Chromosome Abnormalities.

Meagan Kaulfus ('17), S.S. Hashmi (faculty), Blair Stevens ('08) & Rebecca Carter ('09). Attitudes of Clinicians Toward Cardiac Surgery and Trisomy 18.

Ellie Gould ('19), S.S. Hashmi (faculty), Victoria Wagner ('16), Jennifer Czerwinski ('03). Examining genetic counselors' implicit attitudes toward disability.

Laura Amendola ('09). Insurance coverage does not predict outcomes of genetic testing: the search for meaning in payer decisions for germline cancer tests.

American Journal of Medical Genetics Part A

Victoria Wagner ('16) & Laura Farach (faculty). A de novo *HDAC2* variant in a patient with features consistent with Cornelia de Lange syndrome phenotype.

Marisa Vineyard Andrews ('09). De novo variants disrupting the HX repeat motif of *ATN1* cause a recognizable non-progressive neurocognitive syndrome.

Haley Streff ('15). Biallelic variants in *COX4I1* associated with a novel phenotype resembling Leigh syndrome with developmental regression, intellectual disability, and seizures.

Contemporary Clinical Trials

Laura Amendola ('09). Clinical exome sequencing vs usual care for hereditary colorectal cancer diagnosis: a pilot comparative effectiveness study.

Neurology: Genetics

Haley Streff ('15). *GNA11* brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis.

Prenatal Diagnosis

Meagan Choates ('17), Blair Stevens ('08), Chelsea Wagner ('17), Lauren Murphy ('08), Claire Singletary (faculty), Theresa Wittman ('16). It takes two: uptake of carrier screening among male reproductive partners.

Leslie Durham ('18), Blair Stevens ('08), Sarah Jane Noblin ('99), David Rodriguez-Buritica (faculty), S. S. Hashmi (faculty), Nevena Krstic ('12). The Utilization of Prenatal Microarray: A Survey of Current Genetic Counseling Practices and Barriers.

European Journal of Human Genetics

Marisa Vineyard Andrews ('09). Pathogenic variants in *DOCK3* cause developmental delay and hypotonia.

Haley Streff ('15). A de novo variant in the human *HIST1H4J* gene causes a syndrome analogous to the *HIST1H4C*-associated neurodevelopmental disorder

Pediatric Clinics of North America

Katie Sagaser ('15). Prenatal genetic testing options.

Women's Health Across the Lifespan, 2nd Ed.

Shannon Mulligan ('14) & Sarah Jane Noblin ('99). Prenatal Diagnosis.

JCO Precision Oncology

Sarah Azram ('18) & Krista Qualmann (faculty). Lynch syndrome with germline MSH2 mutation in a patient with primary anaplastic glioneuronal tumor.

Leukemia & Lymphoma

Maggie Clifford ('17), Sarah Bannon (faculty), Erica Bednar (faculty), Jen Czerwinski ('03), Leslie Dunnington ('10), S. S. Hashmi (faculty). Clinical applicability of proposed algorithm for identifying individuals at risk for hereditary hematologic malignancies.

GeneReviews

Victoria Wagner ('16) & Hope Northrup (faculty). Mucopolysaccharidosis type III.

Journal of Child Neurology

Sara Wofford ('19), Sarah Jane Noblin ('99), Jessica Davis ('13), Laura Farach (faculty), S.S. Hashmi (faculty), & Victoria Wagner ('16). Genetic testing practices of genetic counselors, geneticists, and pediatric neurologists with regard to childhood-onset neurogenetic conditions.

Gynecologic Oncology

Erica Bednar (faculty). Disparities in gynecologic cancer genetics evaluation.

Erica Bednar (faculty). Dissemination of the universal genetic testing initiative to a diverse, indigent patient population at a county hospital gynecologic oncology clinic.

Genetics in Medicine

Marisa Vineyard Andrews ('09). Variants in *TCF20* in neurodevelopmental disability: description of 27 new patients and review of the literature.

Julia Wynn ('06). Psychological outcomes related to exome and genome sequencing result disclosure: a meta-analysis of seven Clinical Sequencing Exploratory Research (CSER) Consortium studies.

Julia Wynn ('06). Evaluation of the cost and effectiveness of diverse recruitment methods for a genetic screening study.

Laura Amendola ('09). Secondary findings from clinical genomic sequencing: prevalence, patient perspectives, family history assessment, and health-care costs from a multisite study.

American Journal of Human Genetics

Julia Wynn ('06). Harmonizing clinical sequencing and interpretation for the eMERGE III Network.

Haley Streff ('15). Aberrant function of the C-terminal tail of *HIST1H1E* accelerates cellular senescence and causes premature aging.

Leslie (Durham) Granger ('18). Homozygous missense variants in *NTNF2*, encoding a presynaptic netrin-G2 adhesion protein, lead to a distinct neurodevelopmental disorder.

Laura Amendola ('09). Rates of actionable findings in individuals with colorectal cancer or polyps ascertained from a community medical setting.

Journal of Clinical Oncology

Jessie (Ross) Corredor ('16). Improving access to genetic counseling services in an adolescent and young adult (YA) clinic.

Biological Psychiatry

Laura Godfrey Hendon ('11). Clinical presentation of a complex neurodevelopmental disorder caused by mutations in *ADNP*.

ASCO Educational Book: Cancer

Prevention, Hereditary Genetics, and Epidemiology

Erica Bednar (faculty). Genetic counseling and testing in a community setting: quality, access, and efficiency.

Obstetrics & Gynecology

Katie Sagaser ('15) & Katie Rock Forster ('12). Re: Chromosomal microarray results from pregnancies with various ultrasonographic anomalies.

WANT TO JOIN THE UTGCP?

For more information regarding the
University of Texas Genetic Counseling Program
at Houston, please contact
Meagan Choates, MS, CGC
(Coordinator of Recruitment & Outreach)
(713) 500-5599
gsbs.gcpinfo@uth.tmc.edu

ALUMNI - KEEP US IN THE LOOP!

If you didn't see your research featured in this
issue, we hope to include you next year.
A call for publications and presentations will be
distributed in summer 2021. We look forward to
seeing your accomplishments (and photos)!

**Find out more about supporting the UTGCP Research & Education fund at
launch.uth.edu/utgcp**