

# UTGCP Annual Research Newsletter

April 2025 | Issue No. 6



**Class of 2025 and Class of 2026 Thesis Research  
Alumni and Faculty Research and Publications from 2024**





# Congratulations Class of 2025!

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

[Click Here](#)



*Brenna Albracht*

Scientific literacy and carrier  
screening decisionmaking

Chair: Meagan Choates, MS, CGC



*Cindy Canales*

Denial of inpatient genetic testing: a study  
on yield and outcomes outpatient

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



*Jessica Clark*

"Magic Computer in the Sky" -  
participant perspectives on control  
and the genetic counseling admissions  
match

Chair: Claire Singletary, MS, CGC



*Nadia English*

Spanish-Speaking patient  
perspectives: benefits and barriers of  
telemedicine in genetic counseling

Chair: Aranza Gonzalez Cendejas, MS,  
CGC



*Hillary Esplen*

Evaluating cancer rates, cancer types, and  
variant hotspots between different races  
and ethnicities among individuals with Li-  
Fraumeni Syndrome

Chair: Jessica Corredor, MS, CGC



*Ava Henson*

"The Holy Grail of PKU Support":  
exploring the psychosocial impacts of  
camps for kids with Phenylketonuria

Chair: Megan Morand, MS, CGC



*Karli Livingston*

Impact of personal and family history of  
Huntington's Disease on the decision and  
ability to obtain life, longterm care, and  
disability insurances

Chair: Leslie Dunnington, MS, CGC



*Farren Lopez*

Achieving the Practice-Based  
Competencies: are more than 50 cases  
needed?

Chair: Aarti Ramdaney, MS, CGC



*Grace Ra*

Examination of a reproductive genetics  
module: does triage to genetic  
counseling impact prenatal care  
equitably?

Chair: Blair Stevens, MS, CGC



*Nicole Talaba*

Expanding the phenotype of SET5-related  
disorder through a Facebook support  
group

Chair: Myla Ashfaq, MS, CGC



# Introducing the Research of the Class of 2026:

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

[Click Here](#)



*Savannah Berger*

Profiling pathogenic variants and  
cancer types in patients with male  
breast cancer

Chair: Hiam Abdel-Salam, MS, CGC



*Jolie Bourek*

Do patients have preferences with  
regard to carrier screening panel  
size?

Chair: Meagan Choates, MS, CGC



*Lamia Haque*

Exploring how teratogen training  
impacts genetic counselors'  
preparedness for teratogen  
counseling

Chair: Myla Ashfaq, MS, CGC



*Katherine Ledden*

Evaluation of genetic awareness  
among ophthalmology residents

Chair: Kathryn Leal, MS, CGC



*Michelle Luong*

Genetic counselor perspectives  
and shift of practice regarding  
evolving CDH1 cancer risks

Chair: Maureen Mork, MS, CGC



*Sara Pickett*

Discussions of disordered eating  
with metabolic patients

Chairs: Danielle Vice, MS, RD, LD  
and Megan Morand, MS, CGC



*Kathryne Rojeck*

Chart review of personal and  
family histories of cancer in  
patients with moderate  
penetrance breast and ovarian  
cancer genes

Chair: Jessie Corredor, MS, CGC



*Jessica Scarlett*

Utility of amniocentesis versus  
screening tests in determining  
candidacy for prenatal surgery for  
open neural tube defects

Chairs: Rosemary Rogers, MS, CGC  
and Blair Stevens, MS, CGC



*Erin Sharkey*

Testing follow up for patients with  
pathogenic/likely pathogenic  
variants in TP53

Chair: Jessie Corredor, MS, CGC



*Marissa Williams*

Evaluation of diagnostic yield of  
genetic testing in patients involved  
in the high-risk clinic at UTHealth:

A chart review

Chair: Kathryn Leal, MS, CGC



# UTGCP Faculty and Alumni Authorship at National and Local Meetings

\*\*\*Only UT-affiliated authors listed\*\*\*

## TSGC Annual Educational Conference 2024 "The Value of Your Network"

**Sandra Darilek**; One case through the specialties: Zellweger spectrum disorder

7<sup>th</sup> ANNUAL  
HEREDITARY CANCER SYMPOSIUM  
for Health Care Providers  
VIRTUAL ENVIRONMENT APRIL 5 – APRIL 6, 2024

THE UNIVERSITY OF TEXAS  
MDAnderson  
Cancer Center  
Making Cancer History®

**Sandra Darilek**; Preimplantation genetic testing (PGT) for hereditary cancer syndromes

2024 | ACMG Annual  
Clinical Genetics Meeting  
MARCH 12-16 • EXHIBIT DATES: MARCH 13-15  
METRO TORONTO CENTER • TORONTO, CANADA  
1994-2024 | 30TH ACMG ANNUAL CLINICAL GENETICS MEETING

**Erin Cooney, Megan Morand, Joseph Ray**; Outcomes in 14 live births resulting from Pegvaliase-treated pregnancies in PKU-affected females

**Laura Farach**; Use of dextrose-containing fluids and intralipids in a patient with KCNAI-related neuromyotonia

**Nevena Krstic**; After Dobbs: Impact of abortion laws on genetics and genomics training & practice

**Kathleen Shields, Laura Farach, Paul Hillman**; Triple diagnosis of X-linked chondrodysplasia punctata, MECP2-related disorder, and mosaic Jacobs syndrome in a male infant via rapid WGS

**Kathleen Shields, Samantha Montgomery, David Rodriguez-Buritica**; Clinical overlap in two patients with 46,XY DSD and limb abnormalities with identical variant in candidate gene LHX9

**Ginger Tsai**; Prenatal diagnosis of a novel biallelic ARFGF1-related disorder due to uniparental isodisomy: A case report

**Ginger Tsai**; Optimizing diagnostic potential: Impact of familial sample configurations on exome sequencing

AMERICAN ACADEMY OF  
NEUROLOGY®



AAN  
2024  
Annual Meeting

**Brad Rolf**; Neurogenetics: Diagnostic dilemmas and therapeutic implications

2024 ASCO®  
ANNUAL MEETING

May 31 – June 4, 2024

McCormick Place | Chicago, IL & Online  
am.asco.org

#ASCO24

**Catherine Skefos**; On the right TRACK: Operationalized a national, patient-centric fully remote precision trial offering comprehensive genomic profiling and a molecular tumor board for rare cancers

73<sup>rd</sup>  
Pediatrics  
by the Gulf | Annual  
Conference  
on Advances  
in Pediatrics

**Erin Cooney, Megan Morand, Jenny Do**; Enhancing trainee education in medical genetics rotations: Exploring the efficacy of video-based learning modalities for core genetics concepts



THE POWER OF SHARED PURPOSE:  
Transforming Gynecologic Cancer Care



ANNUAL MEETING  
ON WOMEN'S CANCER  
San Diego, CA • 2024

**Erica Bednar**; Hereditary cancer screening



28th International Conference on  
Prenatal Diagnosis and Therapy



7-10 July 2024  
Boston, Massachusetts, USA

**Meagan Choates, Claire Singletary, Theresa Wittman, Samantha Montgomery, Luana Goulet, Blair Stevens**; Prenatal karyotype: Relevant or relic?

# UTGCP Faculty and Alumni Authorship at National and Local Meetings

\*\*\*Only UT-affiliated authors listed\*\*\*

**Sandra Darilek;** Influencing factors and patient decisions regarding mosaic and aneuploid embryo transfer

**Sandra Darilek;** Perinatal genetic testing uptake and transfer outcomes following mosaic and aneuploid embryo transfer



**43rd ANNUAL CONFERENCE • Sept. 17-21, 2024**  
Ernest N. Morial Convention Center • New Orleans, LA

**Hiam Abdel-Salam;** Development of a culturally targeted educational intervention to increase cancer genetic counseling utilization among Iranian and Arab populations in the United States

**Yusra Aziz;** Heterozygous loss of function germline variants in ELPI: A case report of pediatric SHH-activated medulloblastoma

**Emma Billings, Leslie Dunnington, Meagan Choates, Syed S. Hashmi, Megan Morand, Jennifer Czerwinski;** Inclusion of adoption as a pregnancy management option in prenatal genetic counseling practice

**Rachel Bluebond;** Goodbye 96040, Hello 9X100: A genetic counseling CPT update for all genetic counselors

**Sophia Bradley;** Supporting medical examiners' roles in identifying inherited cardiac disease in cases of sudden cardiac death in the young.

**Carley Brueckner, Meagan Choates, Theresa Wittman, Blair Stevens;** Impact of genetic counseling on patient empowerment after diagnosis of fetal anomaly

**Danielle Carr;** Genetic services uptake for prostate cancer patients stratified by referring physician and disease stage

**Danielle Carr;** Landscape of molecular screening program at a community hospital system: Increasing appropriate referrals through alternative service delivery

**Danielle Carr;** Landscape of molecular screening program at a community hospital system: Yield of germline genetic results

**Sandra Darilek;** At face value - When carrier screening results may not be what they appear

**Sandra Darilek;** Genetic counselor value unity: A pilot study

**Sandra Darilek;** Perceptions of the need and utility of a genetic counselor value unit: An exploratory study

**Callie Diamondstein;** From conception to cradle: Enhancing perinatal care workflows

**Amanda Gerard;** Comparing social determinants of health and initial genetics evaluation between inpatient and outpatient settings at Texas Children's Hospital

**Laura Hendon;** Cell-free DNA results indicating mosaic monosomy X of likely maternal origin: Impact on genetic counseling practices and patient experiences

**Laura Hendon;** Prenatal genetic counseling challenges with indeterminate SMA results

**Ava Henson;** Errors in genome sequencing result disclosures by genetic counselors and trained neonatologists: A qualitative content analysis

**Maria Hernandez, Meagan Choates, Myla Ashfaq, Megan Morand, Syed S. Hashmi, David Rodriguez-Buritica, Kathleen Shields;** Dysmorphology training and utility in genetic counseling

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

**Nevena Krstic;** Understanding the landscape of psychiatric medications in pregnant patients: Implications for prenatal genetic counselors

**Michelle Luong;** Assessing criteria for germline evaluation based on somatic genetic test results

**Jacqueline Mersch;** Improving our worth: Impact of billing interventions on reimbursement for preventative cancer genetic counselor services

# UTGCP Faculty and Alumni Authorship at National and Local Meetings

\*\*\*Only UT-affiliated authors listed\*\*\*

**Jacqueline Mersch**; Working with the "in" crowd: Novel oncology genetic testing inpatient services

**Nichollette Murphey, Blair Stevens, Myla Ashfaq, Aranza Gonzalez Cendejas, Theresa Wittman**;

Comparison of methodologies for prenatal diagnosis of skeletal anomalies

**Rachel Notestine, Claire Singletary, Meagan Choates, Molly Daniels**; Fraud in genetic testing: Swindling the system

**Carol Nowlen**; Experiences with genetic services for cystic fibrosis: Perspectives among Latine caregivers of children with cystic fibrosis

**Gina Nuccio**; A landscape assessment of state medicaid coverage for genetic counseling

**Disha Patel, Shannon Mulligan, Natalie Stoner, Gina Sanchez, Meagan Choates**; Patient preferences for ultrasound soft sign disclosure with prior negative cfDNA screening

**Aarti Ramdaney, Samantha Montgomery, Aranza Gonzalez Cendejas, Brenna Albrecht, Meagan Choates**; Client decision-making following carrier screening results with variable reproductive risk

**Jordan Steffen, Jessie Corredor, Leslie Dunnington**; Motivations and attitudes for pursuing anonymous genetic testing for Huntington's disease

**Matt Tschirgi**; Increasing supply to meet demand: One laboratory's experience expanding student rotation opportunities

**Chelsea Wagner**; D-[A]I-Y: Practical Implementation of AI into GC practice

**Chelsea Wagner, Samantha Montgomery**; Navigating the new landscape: A review of evolving abortion legislation impacting genetic counselors' scope of practice

**Cathy Wicklund**; Assessing the impact of the decreasing workforce of geneticists on the roles and responsibilities of general pediatric genetic counselors



**Erin Cooney, Megan Morand, Jenny Do**; Enhancing trainee education in medical genetics rotations: Exploring the efficacy of video-based learning modalities for core genetics concepts



**Emile Moura Coelho da Silva**; Clinical characteristics of 1,000+ epilepsy patients with genetically-assessed etiology at a tertiary care center

## Other Notable Talks

**Emilyn Banfield**; Recruitment and engagement on an international scale: Culture, community, and common ground; Lecture for NIH Genomics and Health Disparities Interest Group

**Sandra Darilek, Blair Stevens**; Expanded carrier screening: What, when and why is it this complicated? Lecture for UCSF Basic Training in Reproductive Medical Genetics Course

**Sandra Darilek, Salma Nassef**; Celebrating women's health: Taking control through carrier and cancer screening; Lecutre for Evenings with Genetics Seminar Series at Baylor College of Medicine

**Sandra Darilek**; Classification investigation - Examining pathogenicity of DMD variants detected on carrier screening; Webinar for American College of Medical Genetics and Genomics Genetics Academy



# UTGCP Faculty and Alumni Scientific Publications

\*\*\*Only UT-affiliated authors listed\*\*\*

# AJHG

The American Journal of Human Genetics

**Jessica Omark O'Shea.** Comprehensive EHMTI variants analysis broadens genotype-phenotype associations and molecular mechanisms in Kleeftstra syndrome

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

## AMERICAN JOURNAL OF medical genetics

**Sandra Darilek.** Family Lore, a variant of uncertain significance and CADASIL

**Amanda Gerard.** Intracranial calcifications simulating Aicardi-Goutières syndrome in PARS2-related mitochondrial disease

**Megan Morand, Joseph Ray, Colin Bixler.** Expanded prenatal phenotype of ALG12-associated congenital disorder of glycosylation including bilateral multicystic kidneys

**Hope Northrup, Laura Farach.** Common epilepsy variants from the general population are not associated with epilepsy among individuals with tuberous sclerosis complex

## ANNALS OF SURGICAL ONCOLOGY

THE LEADING RESOURCE IN SURGICAL ONCOLOGY

**Catherine Skefos.** SDHB-associated pheochromocytomas: What is their clinical behavior?

## Birth Defects Research



**Nevena Krstic.** Impact of perinatal exposure counseling on patient reported emotional outcomes and decisional empowerment

## CANCER PREVENTION RESEARCH

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

# ENT

## EAR, NOSE & THROAT JOURNAL

**Laura Hendon.** Middle ear fibrosis contributes to hearing loss in patients with Myhre syndrome

## ENDOCRINE ONCOLOGY

**Catherine Skefos.** SDHA secondary findings in germline testing: counseling and surveillance considerations

# Familial Cancer


Quarterly Journal on Cancer Genetics

**Erica Bednar.** Cascade genetic testing: An underutilized pathway to equitable cancer care?

**Gina Nuccio.** A content analysis of parents' reflections on pathogenic and uncertain pediatric oncology germline sequencing results

# UTGCP Faculty and Alumni Scientific Publications

\*\*\*Only UT-affiliated authors listed\*\*\*



Frontiers in  
**Pediatrics**

**Nichollette Murphey, Kate Richardson, Paul Hillman.**  
Case report: Early acute kidney failure in an 11-year-old boy with Dent disease type I

**Genetics  
in  
Medicine**  
An Official Journal of the ACMG

**Myla Ashfaq.** Developing medical genetics in a low-income country: Unveiling the journey of the Pakistani Society of Medical Genetics and Genomics (PSMG)

**Rachel Notestine, Claire Singletary, Meagan Choates, Molly Daniels.** Fraud in genetic testing: Swindling the system



International Journal of  
**Neonatal Screening**

**Brad Rolf.** One size does not fit all: A multifaceted approach to educate families about newborn screening

**JCI** The Journal of  
Clinical Investigation

**Leslie Granger.** Spliceosome malfunction causes neurodevelopmental disorders with overlapping features

**Nevena Krstic.** Application of the RIME framework in genetic counseling fieldwork training to assess practice-based competencies

**JOURNAL OF  
CLINICAL  
LIPIDOLOGY**  
OFFICIAL JOURNAL OF THE NATIONAL LIPID ASSOCIATION

**Jenny Do.** Genetically and clinically confirmed atypical cerebrotendinous xanthomatosis with normal cholesterol and marked elevations of bile acid precursors and bile alcohols

Journal of  
**Community  
Genetics**

**Erica Bednar.** Feasibility of an environmental scan-based approach to collecting information about factors impacting cancer genetics services in Latin American countries

Journal of  
**Genetic  
Counseling** The official Journal of the  
National Society of Genetic Counselors



**Jenna Lea, Blair Stevens, Shannon Mulligan, Syed S. Hashmi, Meagan Choates.** Non-invasive prenatal screening: testing motivations and decision making in the low-risk population

**Gina Sanchez, Syed S. Hashmi, Erica Bednar, Katie Sagaser, Claire Singletary, Aarti Ramdaney.** Status of abortion curriculum in genetic counseling: Survey of graduate programs and recent graduates in the United States



# UTGCP Faculty and Alumni Scientific Publications

\*\*\*Only UT-affiliated authors listed\*\*\*

## IJGC INTERNATIONAL JOURNAL OF GYNECOLOGICAL CANCER

**Erica Bednar.** Delivery of hereditary cancer genetics services to patients newly diagnosed with ovarian and endometrial cancers at three gynecologic oncology clinics in the United States, Brazil, and Mexico

## Journal of Immunotherapy and Precision Oncology

**Carol Nowlen, Molly Daniels, Jacqueline Harkenrider.** Limited independent follow-up with germline testing of variants detected in BRCA1 and BRCA2 tumor-only sequencing

## THE JOURNAL OF PEDIATRICS

**Amanda Gerard.** Transient generalized osteosclerosis in a newborn mimicking congenital osteoporosis with negative comprehensive genetic workup: A case report

## JAMIA A SCHOLARLY JOURNAL OF INFORMATICS IN HEALTH AND BIOMEDICINE

**Carla McGruder.** Sounding out solutions: Using SONAR to connect participants with relevant healthcare resources

## MCE Molecular and Cellular Endocrinology

**Catherine Skefos.** Metastatic pheochromocytoma and paraganglioma: Integrating tumor biology in clinical practice

## Molecular Genetics and Metabolism

**Megan Morand, Erin Cooney, Joseph Ray.** Outcomes in 14 live births resulting from Pegvaliase-treated pregnancies in PKU-affected females

## nature COMMUNICATIONS

**Amanda Gerard.** Variants in the WDR44 WD40-repeat domain cause a spectrum of ciliopathy by impairing ciliogenesis initiation

## Journal of Pediatric Neurology

**Jenny Do, Syed S. Hashmi, Hope Northrup, Laura Farach, Kate Richardson.** Parenting stress in tuberous sclerosis complex

**Laura Farach.** Drug-resistant epilepsy in tuberous sclerosis complex is associated with TSC2 genotype: More findings from the preventing epilepsy using vigatrin (PREVeNT) trial

## PRENATAL DIAGNOSIS

**Sandra Darilek.** Telegenetics to provide comprehensive prenatal diagnosis

**Laura Hendon.** Fetal presentation of MYRF-related cardiac urogenital syndrome: An emerging and challenging prenatal diagnosis



# The Faces of UTGCP Research



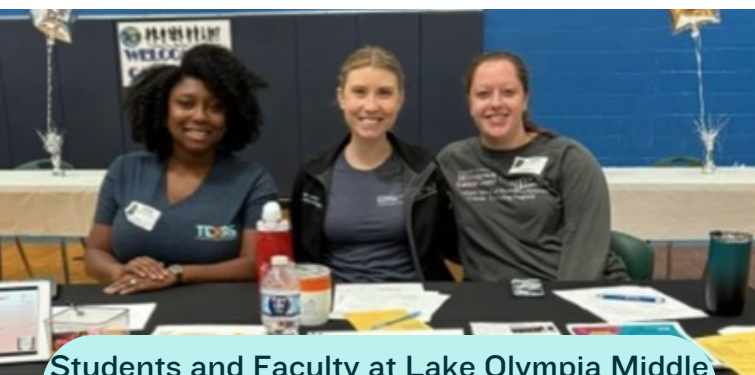
UTGCP Students and Alumni at the UTGCP Reunion Lunch  
NSGC 2024, New Orleans, Louisiana



Class of 2025 with Claire at TSGC



Students and Faculty at Camp PHEver



Students and Faculty at Lake Olympia Middle  
School Career Fair



Class of 2025 at Lab Workshop

# 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 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

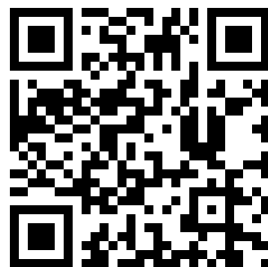
## **Class of 2022**

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**



## Alumni, Students, and Faculty Featured In this Issue

### **Class of 1993**

Cathy Wicklund

### **Class of 2003**

Sandra Darilek

### **Class of 2006**

Jennifer Czerwinski

### **Class of 2008**

Blair Stevens (Faculty)

Matt Tschirgi

### **Class of 2010**

Leslie Dunnington (Faculty)

Gina Nuccio

### **Class of 2011**

Laura Hendon

### **Class of 2012**

Nevena Krstic

Salma Nassef

### **Class of 2013**

Brad Rolf

### **Class of 2014**

Jacqueline Mersch

Shannon Mulligan (Faculty)

Aarti Ramdaney (Faculty)

### **Class of 2015**

Callie Diamondstein

Carla McGruder

Katie Sagaser

### **Class of 2016**

Meagan Choates (Faculty)

Jessie Corredor (Faculty)

Ginger Tsai

Theresa Wittman (Faculty)

### **Class of 2017**

Amanda Gerard

Leslie Granger

Chelsea Wagner

### **Class of 2018**

Jessica Omark O'Shea

Kate Richardson (Faculty)

# Alumni, Students, and Faculty Featured In this Issue

## **Class of 2019**

Danielle Carr  
Carol Nowlen  
Samantha Montgomery (Faculty)

## **Class of 2020**

Aranza Gonzalez Cendejas (Faculty)

## **Class of 2021**

Megan Morand (Faculty)  
Kathleen Shields

## **Class of 2022**

Emilyn Banfield  
Jenny Do (Faculty)  
Jenna Lea  
Emile Moura Coelho da Silva (Faculty)  
Gina Sanchez (Faculty)  
Natalie Stoner

## **Class of 2023**

Yusra Aziz

## **Class of 2024**

Emma Billings  
Carley Breuckner  
Maria Hernandez

Mindy Kolodzieski  
Nicholette Murphey  
Rachel Notestine  
Disha Patel  
Jordan Steffen

## **Class of 2025**

Brenna Albrecht  
Ava Henson

## **Class of 2026**

Michelle Luong

## **Faculty**

Haim Abdel-Salam  
Myla Ashfaq  
Erica Bednar  
Colin Bixler  
Rachel Bluebond  
Sophia Bradley Kulkarni  
Erin Cooney  
Molly Daniels  
Laura Farach  
Luana Goulet  
Jacqueline Harkenrider  
Syed S. Hashmi  
Paul Hillman



## Alumni, Students, and Faculty Featured In this Issue

Katie Leal  
Joseph Ray  
David Rodriguez-Buritica  
Claire Singletary  
Catherine Skefos

### **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!**