Baylor
College of
Medicine

DEPARTMENT OF
MOLECULAR & HUMAN
GENETICS

ANNUAL REPORT
2025

Transforming Medicine Through the Science and Practice of Genetics

Welcome

I am delighted to share the 2025 Baylor College of Medicine Department of Molecular and Human Genetics Annual Report. As we continue 2026, I'd like to briefly reflect on last year's accomplishments.

The department continues to excel in all aspects of its mission to transform medicine with the science and practice of genetics and genomics.

The department continues to demonstrate national leadership in genetics research, with the highest total number of grants from the National Institutes of Health and an average annual NIH funding level approaching $75 million over the past decade.

We continue to lead in the diagnostic testing arena with Baylor Genetics, our joint venture with H.U. Group Holdings, Inc. This jointly governed laboratory supports the academic mission and innovation of the department while promising to extend the impact of genetic diagnostic testing worldwide.

Our faculty continue to deliver on our clinical, training and research missions at home and abroad through our regional, national and global partnerships.

New and ongoing NIH and industry consortia are driving gene discovery and advancing the clinical use of genetics and genomics. These consortia include the Genomic Research to Elucidate the Genetics of Rare diseases, the Knockout Mouse Phenotyping Program, the Center for Precision Medicine Models, Clinical Genome Resource, the Somatic Mutations across Human Tissues Network, and multiple consortia in the Rare Diseases Clinical Research Network.

Our Baylor Undiagnosed Diseases Center houses the NIH Undiagnosed Diseases Network clinical site and provides clinical services, testing and analysis to assess patients who have not received a diagnosis for their condition. The NIH-supported Project TEXOME, GIVE and MAGNET studies address access to genomic care expertise for underserved families and healthcare providers in Texas. These three genomic medicine projects use the Consultagene platform to provide access to genetic evaluation, peer-to-peer consultation and genetic counseling.

Baylor's All of Us Evenings with Genetics Research Scholar Program is now in its fifth year and has completed three successful Biomedical Researcher Faculty Summits.

The future holds much promise due to the talent and dedication of our renowned faculty, trainees and staff. I consider myself privileged to be a part of this exciting and vital effort.

Warm regards,

Brendan Lee, M.D., Ph.D.
Robert and Janice McNair Endowed Chair
Professor and Chairman
Department of Molecular and Human Genetics

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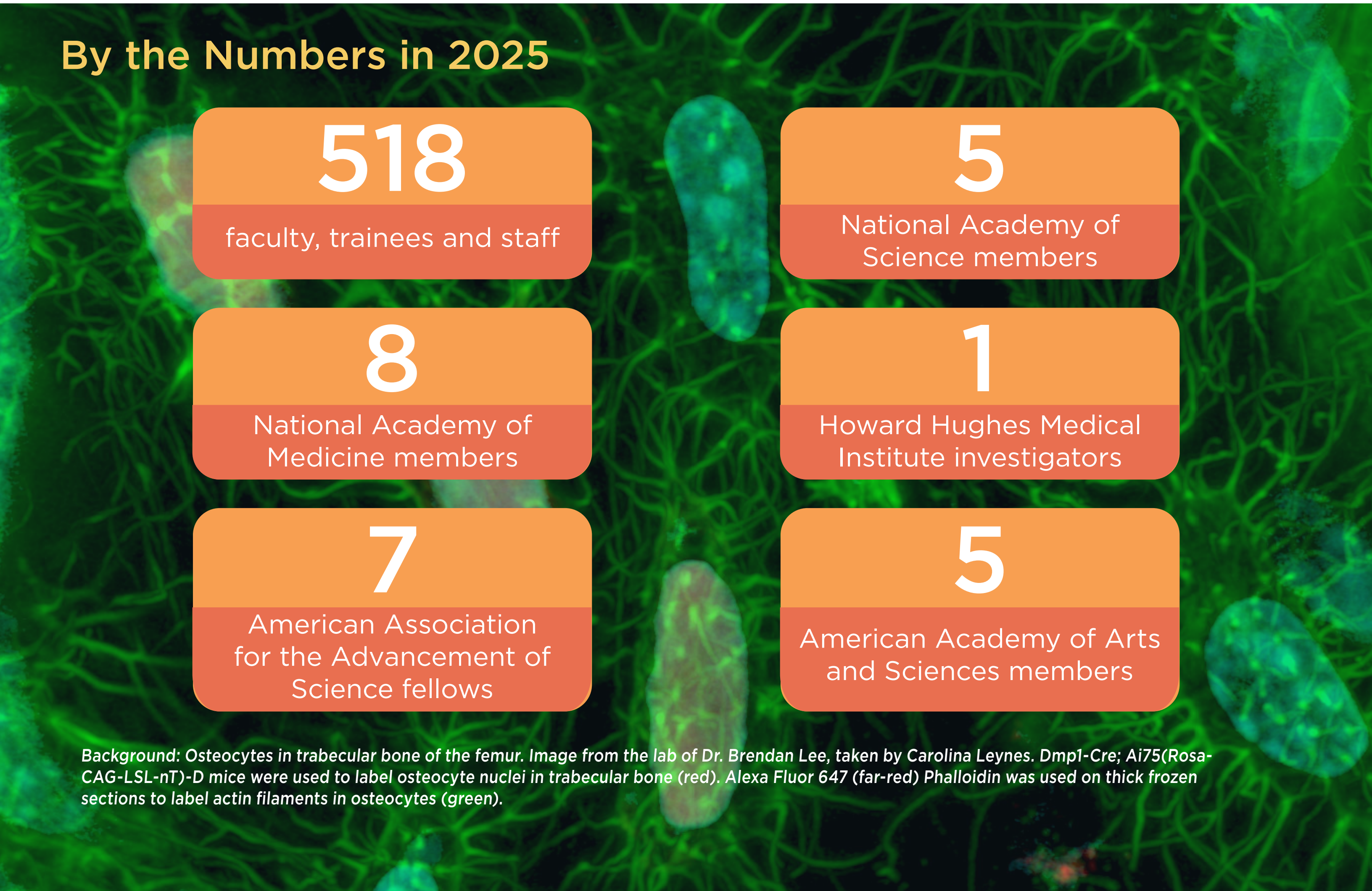
*Microsoft Co-Pilot, a service that uses artificial intelligence, assisted with this report.



By the Numbers in 2025

518
faculty, trainees and staff

5
National Academy of Science members

8
National Academy of Medicine members

1
Howard Hughes Medical Institute investigators

7
American Association for the Advancement of Science fellows

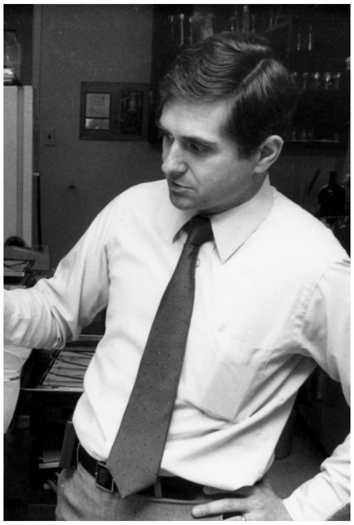
5
American Academy of Arts and Sciences members

Background: Osteocytes in trabecular bone of the femur. Image from the lab of Dr. Brendan Lee, taken by Carolina Leynes. Dmp1-Cre; Ai75(Rosa-CAG-LSL-nT)-D mice were used to label osteocyte nuclei in trabecular bone (red). Alexa Fluor 647 (far-red) Phalloidin was used on thick frozen sections to label actin filaments in osteocytes (green).

About the Department







More than half a century of genetics innovation

Research in genetics at Baylor College of Medicine began in 1971 with the recruitment of Dr. C. Thomas Caskey and Dr. Arthur Beaudet from the National Institutes of Health. Initially operating within the Departments of Internal Medicine and Pediatrics, they created a clinical training program in 1976 to educate and train top investigators in genomics and biomedical research. As the team of researchers grew in size, scope and ambition, a centralized organization was needed to unify their efforts. Consequently, in 1985, the Institute of Molecular Genetics was established, placing Baylor on the map as a genetics powerhouse. By leveraging its ability to recruit the best and brightest physicians and scientists, the institute grew substantially. In 1994, it became a full department.

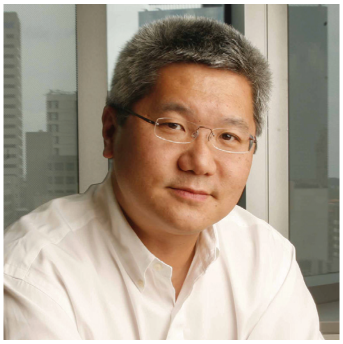
The department's success reached new heights with its selection as one of the six pilot programs for the Human Genome Project in 1996. The Human Genome Sequencing Center at Baylor College of Medicine, led by Dr. Richard Gibbs, the Wofford Cain Chair and professor of molecular and human genetics, later became one of three sites to complete the Human Genome Project. In 2000, scientists triumphantly announced they had deciphered the human genome.

Since then, the department has provided comprehensive clinical care to patients worldwide. As the largest clinical genetics program in the country, Baylor offers patients timely and expert assistance, as well as unparalleled treatment and counseling options.

The department has also expanded its reach by providing diagnostic genetic testing services to the broader medical genetics community through its laboratory, Baylor Genetics, a joint venture with H.U. Group Holdings. Baylor Genetics offers an expansive menu of genetic tests and provides leading service to practitioners worldwide.

The past 50 years have been an exciting time of growth and change. Initially focused on medical and pediatric genetics, the department has diversified into functional genomics, genome sequencing, cancer genetics, and genetic therapies, cementing its spot as the preeminent genetics department in the country, if not the world.

Department Leadership



Brendan Lee, M.D., Ph.D.
Robert and Janice McNair Endowed Chair in Molecular and Human Genetics

Laura Rosales, Ed.D., M.B.A.
Administrator

Carlos Bacino, M.D.
Vice Chair, Clinical Affairs

Christine Eng, M.D.
Vice Chair, Diagnostic Laboratory Affairs

Gad Shaulsky, Ph.D.
Vice Chair, Educational Affairs (Graduate Education)

Lorraine Potocki, M.D.
Vice Chair, Educational Affairs (Undergraduate Medical Education)

V. Reid Sutton, M.D.
Vice Chair, Educational Affairs (Graduate Medical Education)

Daniel Riconda, M.S., C.G.C.
Vice Chair, Educational Affairs (Genetic Counseling Program)

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Vice Chair, Research Affairs - Basic and Translational

Sandesh C. S. Nagamani, M.B.B.S., M.D.
Vice Chair, Research Affairs - Clinical

Debra Murray, Ph.D.
Co-director, Office of Community Engagement

Gladys Pryor, B.S.
Co-director, Office of Community Engagement

Initiatives & Partnerships

Baylor Genetics continues to advance precision diagnostics and clinical impact

In 2025, Baylor Genetics continued to strengthen its position as a leader in precision diagnostics, marked by significant advancements in testing innovation, strategic partnerships and scientific leadership. The year reflected a clear focus on expanding access to advanced genomic insights, integrating cutting-edge technologies into clinical workflows and reinforcing the organization's commitment to patients, providers and employees alike.

One of the year's most significant milestones was Baylor Genetics' expansion into federal healthcare through a multi-year national contract with the U.S. Department of Veterans Affairs. Through this agreement, Baylor Genetics began delivering pharmacogenomic (PGx) and germline genetic testing, including hereditary cancer testing, to veterans across the VA healthcare system. This initiative broadened access to precision diagnostics at a national scale and underscored the company's ability to support complex, large-scale healthcare networks with high-quality genomic services.

Technological innovation remained central to Baylor Genetics' growth. In 2025, the organization became the first laboratory to deliver Whole Genome Sequencing results through Epic Aura, a milestone that streamlined genomic results directly into electronic health records. This advancement enhanced clinical efficiency and supported more timely, informed decision-making for providers caring for patients with complex and rare conditions.


Baylor Genetics at the 2025 American Society of Human Genetics Meeting

Baylor Genetics also continued to push the boundaries of genomic science through expanded RNA sequencing (RNA-seq) and multiomics capabilities. Throughout the year, the company presented new clinical data demonstrating how RNA-seq can improve diagnostic yield, clarify variant interpretation and complement exome and genome sequencing approaches. These findings reinforced the value of integrating transcriptomic data into routine clinical practice, particularly for patients who remain undiagnosed after traditional testing.

The organization also maintained a strong presence across the scientific and clinical community. Baylor Genetics shared research and clinical insights at leading national conferences, including the American College of Medical Genetics and Genomics (ACMG), the American Society of Human Genetics (ASHG), the National Society of Genetic Counselors (NSGC) and the Child Neurology Society Annual Meeting. These engagements highlighted real-world applications of whole genome sequencing, RNA sequencing, and multiomics diagnostics across pediatric, adult and rare disease populations, reinforcing Baylor Genetics' role as a trusted partner in advancing genomic medicine.

Strategic partnerships further strengthened clinical impact during the year. Baylor Genetics joined the Child Neurology Society as its official genetic testing partner, reflecting a shared commitment to improving diagnostic pathways and outcomes for children with neurologic conditions and aligning with the company's mission to shorten the diagnostic odyssey for families facing complex genetic disorders.

Beyond science and technology, Baylor Genetics received meaningful recognition for its leadership and workplace culture. The company was certified as a 2025 Great Place To Work®, based on employee feedback, highlighting a culture grounded in collaboration, integrity and purpose. In addition, Dr. Christine Eng, chief medical officer of Baylor Genetics and professor of molecular and human genetics, was named Woman Scientist of the Year in the 2025 Stevie® Awards for Women in Business, and Chairman and CEO Kengo Takishima was recognized as one of the Houston Business Journal's 2025 Most Admired CEOs. Together, these honors reflect both scientific excellence and people-first leadership.

Capping the year, Baylor Genetics marked its 10th anniversary, celebrating a decade of growth as an academic–commercial hybrid and reaffirming its long-term commitment to delivering answers through precision diagnostics. With continued investment in innovation, partnerships and talent, 2025 represented a pivotal year in advancing Baylor Genetics' mission to transform healthcare through genomic insight.

Building clinical genomics capacity through multiomics

Baylor College of Medicine, in partnership with Baylor Genetics, successfully launched the Medical Genetics Multiomics Laboratory (MGML) in 2024 as an academic, Clinical Laboratory Improvement Amendments-certified clinical genetic testing laboratory dedicated to translating advanced multiomics research into clinically actionable diagnostics.

Following its launch, the MGML, directed by Dr. Pengfei Liu, associate professor of molecular and human genetics introduced its first clinical offering, Whole Transcriptomic RNA Sequencing, expanding diagnostic capabilities beyond DNA-based approaches. This assay provides transcriptome-wide analysis of gene expression and RNA structure, offering functional evidence to clarify variant interpretation and identify disease mechanisms that may be missed by whole-exome or whole-genome sequencing alone. Clinical reporting is provided in collaboration with Baylor Genetics. The RNA sequencing program is also conducted in close partnership with the NIH Undiagnosed Diseases Network (UDN), for which MGML provides transcriptome sequencing analysis for patients enrolled in the program, supporting diagnostic evaluation for some of the most challenging unsolved genetic conditions.

This past year, a major milestone was reached when the laboratory further strengthened its quality and compliance framework by achieving College of American Pathologists accreditation, reflecting the successful implementation of nationally recognized laboratory standards.

In parallel with clinical launch activities, MGML has advanced its strategic vision to integrate next-generation multiomics technologies into clinical genetic diagnostics. Ongoing development efforts include expanding long-read DNA and RNA sequencing capabilities to improve the detection of complex variants, developing approaches to identify variants present at low allele fraction, and incorporating additional multiomic testing modalities into clinical use. The laboratory is also exploring ultra-deep RNA sequencing strategies to enhance the sensitivity of transcriptome-based diagnostics and investigating the integration of patient-derived cellular models and differentiation systems to enable functional RNA analysis in clinically relevant cell types. Collectively, these efforts aim to bridge advanced diagnostic technologies with emerging therapeutic strategies, translating genomic discoveries into actionable insights for patients with rare genetic diseases.

Through the establishment of its clinical operations and the successful implementation of its initial diagnostic assay, MGML has built a strong foundation for continued growth and innovation. The laboratory will continue to expand and refine multiomic diagnostic approaches to address unmet needs in rare and complex genetic disease.

Global perspectives on clinical genetics

The sixth Baylor College of Medicine – Chinese University of Hong Kong (CUHK) – Zhejiang University (ZJU) Joint Symposium in Clinical Genetics was held Nov. 14–16, 2025, in Hangzhou, China, at the Zhejiang International Hotel. The event brought together healthcare professionals from North America and the Asia-Pacific region to discuss the role of genetics in modern medical care, with a focus on pregnancy, reproduction and children's health.

The program featured pre-conference workshops, keynote presentations and educational sessions addressing current topics in clinical genetics, including genetic testing for rare conditions, prenatal screening, reproductive genetics and genetic counseling. Sessions also explored ethical considerations


Hangzhou · China

6th BCM-CUHK-ZJU Joint Symposium in Clinical Genetics

Main Congress
15 - 16 November 2025
Pre-Congress Workshops
14 November 2025
Zhejiang Hotel, Hangzhou, China

SCAN ME

Co-Chairperson
Brendan LEE
Baylor College of Medicine

Co-Chairperson
Tak Yeung LEUNG
The Chinese University of Hong Kong

Co-Chairperson
Hefeng HUANG
Institute of Medical Genetics and Development
Zhejiang University

Jointly Organized By
Department of Molecular and Human Genetics
Baylor College of Medicine
Department of Obstetrics & Gynaecology &
Department of Paediatrics
The Chinese University of Hong Kong
Institute of Medical Genetics and Development
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and emphasized the practical integration of genetic tools into routine patient care.

Keynote and plenary presentations were delivered by leaders in the field, including Dr. Brendan Lee, professor and chair of molecular and human genetics at Baylor, Dr. Tak-Yeung Leung of CUHK, and Dr. Hefeng Huang of Zhejiang University. Speakers highlighted how genetics is increasingly influencing everyday clinical practice and underscored the importance of clear communication and patient support.

Overall, the symposium reinforced the value of international collaboration and thoughtful application of genetic advances to support high-quality, patient-centered care

Expanding access and ending diagnostic odysseys

Baylor College of Medicine and Texas Children's Hospital are leading a coordinated effort to improve access to genomic medicine for individuals and families affected by rare and undiagnosed diseases. Through innovative programs such as the Community Texome Project, the Undiagnosed Diseases Center (UDC), Project GIVE (Genetic Inclusion by Virtual Evaluation), Project MAGNET (Making Genomics Accessible to Newborns in Texas), and the PD GENEration study, these initiatives integrate cutting-edge genomic technologies, community engagement and novel service delivery models. Together, they help transform diagnostic care by shortening diagnostic odysseys and generating new insights into the genetic basis of human disease.

Community TEXOME Project

The Community Texome Project at Baylor College of Medicine and Texas Children's Hospital has been recruiting families since 2021.

The project, co-led by Dr. Michael Wangler, associate professor of molecular and human genetics, and Dr. Hugo Bellen, Distinguished Service Professor of molecular and human genetics and chair in neurogenetics at the Jan and Dan Duncan Neurological Research Institute, provides whole genome sequencing to children and adults from underserved backgrounds who have undiagnosed diseases. Study participants receive a full genetic evaluation and genomic sequencing. The Texome team follows up with participants over two years to collect additional information about their medical presentation. Participants also complete numerous surveys that provide insight into the perceptions, obstacles, and experiences associated with genomic medicine in underserved communities. The Texome Project has been funded by the National Human Genome Research Institute and receives philanthropic support through the Duncan NRI and Texas Children's.

The Texome Project has received referrals from more than 300 families, and sequencing has been completed for more than 200. The project has identified a genetic diagnosis in 30% of cases, in some ending decades-long diagnostic odysseys. The Texome team uses novel technology to reanalyze data from unsolved cases, and model organism research is conducted to assess the functional consequences of candidate variants. The project also works to identify new genes that are implicated in genetic diseases.

"The Texome Project is a win-win for all people involved. Families who are historically underserved receive access to crucial genomic care, and researchers are able to gain a better understanding of the needs, beliefs and experiences of these populations when it comes to genetics and genomics," said Dr. Chaya Murali, assistant professor of molecular and human genetics at Baylor. Murali, a clinical geneticist, leads the survey arm of the Texome Project team.

Undiagnosed Diseases Center

Established in 2022, the Undiagnosed Diseases Center incorporates the work of Baylor's Undiagnosed Diseases Network clinical site, DNA sequencing core, and model organisms screening center. The UDC is led by Dr.

Brendan Lee and Dr. Carlos Bacino, professor of molecular and human genetics, with clinical coordination provided by Jill Mokry, associate professor of molecular and human genetics. The Center provides clinical services, genetic testing and analysis for patients with rare, undiagnosed conditions and offers additional services through the Consultagene platform, which delivers educational resources and genetics services.

Project GIVE (Genetic Inclusion by Virtual Evaluation)

Funded by the National Center for Advancing Translational Sciences, Project GIVE is led by Dr. Seema Lalani, professor of molecular and human genetics. The project aims to reduce time-to-diagnosis for children with suspected genetic disorders in the Rio Grande Valley and El Paso by using the Consultagene platform to provide virtual clinical evaluation and whole-genome sequencing. The study has recruited more than 200 children and achieved a diagnostic yield of approximately 40%.

Project MAGNET (Making Genomics Accessible to Newborns in Texas)

Project MAGNET, funded in 2024, is led by Dr. Brendan Lee, Dr. Seema Lalani and Dr. Stacey Periera, associate professor in the Center for Medical Ethics & Health Policy at Baylor. The multi-year initiative seeks to expand access to genomic medicine for critically ill newborns in under-resourced neonatal intensive care units across Texas, particularly along the Texas–Mexico border. The study integrates point-of-care genetic expertise and rapid whole-genome sequencing into NICU care through the Consultagene platform and evaluates the clinical, economic and implementation impact of this approach.

PDGENE study

The PD GENEration study, funded by the Parkinson's Foundation, is led by Jamie Fong, assistant professor in molecular and human genetics; Dr. Joshua Shulman, professor of molecular and human genetics and neurology; and Dr. Brendan Lee. Baylor serves as a participating clinical site and the sole location for the Consultagene-PD GENEration Video Study, which compares disclosure of negative genetic results via pre-recorded video versus live virtual consultation.

In 2025, the study achieved its target enrollment, demonstrating that pre-recorded video is an effective and scalable model for genetic result disclosure in Parkinson's disease research.

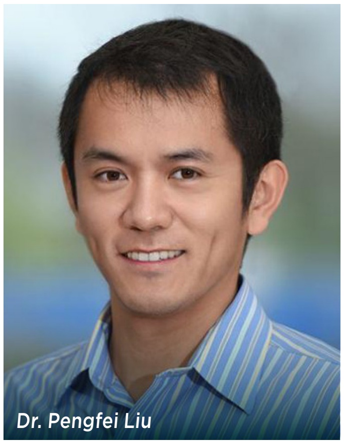
Research & Discovery

Research in the Department of Molecular and Human Genetics has led to important discoveries that increase understanding of disease and guide potential new treatments.

Baylor researchers develop and validate clinical diagnostic RNA sequencing test

Researchers have developed and validated the first whole-transcriptome RNA sequencing test for clinical genetic diagnostics, representing a major advance beyond targeted RNA testing. The work was led by the Medical Genetics and Multiomics Laboratory and published in *American Journal of Human Genetics* (DOI: 10.1016/j.ajhg.2025.02.006).

While RNA sequencing has been used in clinical settings, its application was largely limited to specific genes. This validated test analyzes the entire transcriptome, enabling detection of abnormal gene expression and RNA splicing patterns that can explain genetic disease, particularly in patients whose conditions remain unresolved after DNA sequencing, using RNA from blood or fibroblast samples for clinical diagnostic evaluation.


Dr. Pengfei Liu

The researchers validated the test using 130 samples, including individuals with known molecular diagnoses and healthy controls. Reference ranges were established using Genome in a Bottle benchmark data, and performance was evaluated with samples from the Undiagnosed Diseases Network, demonstrating detection of clinically relevant RNA abnormalities through transcriptome-driven discovery and DNA-guided analysis in clinical cases.

“This validation represents the first attempt to unleash transcriptome potential of RNA sequencing for clinical diagnostics, globally,” said Dr. Pengfei Liu, director of the MGML and associate professor of molecular and human genetics.

This RNA-seq assay is offered within the Undiagnosed Diseases Network, expanding diagnostic options for patients with rare and undiagnosed conditions.

Researchers develop new tool to improve accuracy of genetic testing

Researchers at Baylor and the Jan and Dan Duncan Neurological Research Institute at Texas Children’s Hospital have developed a new tool that improves the accuracy of genetic testing by refining how genetic variants are interpreted across diverse populations. Published in *Nature Communications* (DOI: 10.1038/s41467-025-63340-2), the study introduces the use of local ancestry inference to improve variant frequency estimates within the Genome Aggregation Database (gnomAD), a widely used global genetics resource.


Dr. Elizabeth Atkinson

Traditional genetic testing often relies on population-averaged data, which can obscure important differences in individuals with mixed ancestry backgrounds. The LAI approach breaks the genome into ancestry-specific segments, such as African, European, or Indigenous American, allowing researchers to measure how common a genetic variant truly is within each ancestral group. The team found that many variants previously thought to be rare are actually common in specific ancestry segments, with important clinical implications for determining whether a variant is benign or disease-causing.

Led by Dr. Elizabeth Atkinson, assistant professor of molecular and human genetics at Baylor, the research shows how more precise, ancestry-aware data can reduce variant misclassification and improve diagnostic accuracy across diverse patient populations.

The improved ancestry-specific data have been made publicly available through gnomAD for use by clinicians and researchers worldwide.

Integrative exome sequencing and machine learning identify new genes contributing to systemic sclerosis risk

Researchers at Baylor and collaborating institutions have identified new genetic contributors to systemic sclerosis (SSc), a severe autoimmune disease with complex and poorly understood genetic causes. In a study published in *Annals of the Rheumatic Diseases* (DOI: 10.1016/j.ard.2025.05.009), the team combined exome sequencing with Evolutionary Action Machine Learning to uncover rare, protein-altering genetic variants associated with increased disease risk.

Using exome sequencing data from more than 2,500 SSc patients and nearly 900 healthy controls, the researchers conducted a genome-wide analysis to move beyond previously known risk regions. The approach revealed MICB, a gene located within the HLA region but acting independently of classical HLA genes, as a novel contributor to SSc risk. The findings were independently replicated using large European datasets, strengthening the evidence for MICB's role in the disease.

Additional analysis using machine learning further identified other genes on chromosome 6 and rare variants affecting interferon signaling, an important immune pathway in systemic sclerosis.


Dr. Shamika Ketkar

Led by Dr. Shamika Ketkar, assistant professor of molecular and human genetics at Baylor, and supported by the Evolutionary Action Machine Learning framework developed in the laboratory of Dr. Olivier Lichtarge, Cullen Chair and professor of molecular and human genetics, the study demonstrates how integrative genomic and computational approaches can reveal previously hidden disease mechanisms and identify potential new targets for future therapies.

"To solve complex diseases like SSc, we need to combine diverse approaches and apply machine learning to the analysis of large DNA, RNA and protein data sets to uncover targets for treatment that would otherwise remain hidden," said corresponding author Dr. Brendan Lee, professor and chair of molecular and human genetics.

A new treatment for patients with Alagille syndrome

Researchers at Baylor, in collaboration with the University of Massachusetts Chan Medical School and Cincinnati Children's Hospital Medical Center, have developed a promising new gene-therapy–based treatment for Alagille syndrome, a rare genetic disorder that affects multiple organs and often leads to liver failure. The study, published in Gastroenterology (DOI: 10.1053/j.gastro.2025.04.033), addresses a major unmet clinical need, as the only current definitive treatment for the disease is liver transplantation—a highly invasive option with limited donor availability and lifelong implications for patients.


Dr. Hamed Jafar-Nejad

Alagille syndrome is characterized by a severe reduction in bile ducts in the liver, which disrupts bile flow and results in progressive liver damage beginning early in life. In mouse models of the disease, the research team demonstrated that a single injection of gene therapy significantly improved bile duct development, reduced liver inflammation and scarring and restored key aspects of liver function—even when treatment was administered after liver damage had already begun.

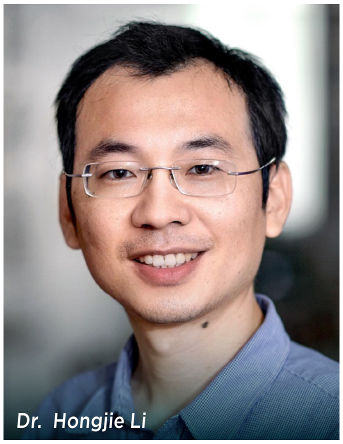
The therapy uses an adeno-associated virus (AAV) to deliver a small RNA molecule that selectively silences the Sox4 gene in liver cells. Although Sox4 was previously believed to support bile duct development, the researchers unexpectedly found that reducing Sox4 activity improved bile duct formation in Alagille syndrome models. Elevated Sox4 expression was also observed in liver samples from patients with the condition, reinforcing the translational relevance of the approach.

Led by corresponding author Dr. Hamed Jafar-Nejad, professor of molecular and human genetics at Baylor, the findings suggest that targeted gene therapy could one day offer a less invasive alternative to liver transplantation for patients with Alagille syndrome. Ongoing and future studies will focus on further clarifying the therapy's mechanism of action and rigorously evaluating safety and durability before potential advancement toward clinical trials.

Study reveals impacts of Alzheimer's disease on the whole body

While Alzheimer's disease is most often considered a disorder of the brain, growing evidence indicates that it also affects other organs throughout the body. Using the laboratory fruit fly, researchers at Baylor, the Duncan NRI, and collaborating institutions developed an Alzheimer's Disease Fly Cell Atlas—a comprehensive map of gene expression across 219 cell types in both brain and peripheral tissues. Alzheimer's-like conditions were modeled by expressing either Aβ42 or Tau proteins exclusively in adult neurons, enabling scientists to examine downstream effects on other organs without the confounding influence of developmental changes. This work was published in the journal *Neuron* (DOI: 10.1016/j.neuron.2025.04.017).

Dr. Hongjie Li, associate professor of molecular and human genetics at Baylor and member of the Dan L Duncan Comprehensive Cancer Center, is a co-corresponding author of the study. Li explained that the atlas was created to understand how Alzheimer's pathology in the brain alters gene activity throughout the body.


Dr. Hongjie Li

The study found that Aβ42 expression primarily damaged the nervous system, particularly sensory neurons involved in vision, hearing, and smell—consistent with early sensory decline seen in Alzheimer's patients. In contrast, Tau expression caused widespread changes in peripheral tissues, including disrupted fat metabolism, digestion, and reduced fertility, suggesting that Tau accelerates aging-like processes.

According to co-first authors Ye-Jin Park, a graduate student co-mentored by Li and Dr. Hugo Bellen, Distinguished Service Professor at Baylor and chair in neurogenetics at the Duncan NRI, and Dr. Tzu-Chiao Lu, postdoctoral associate in the Li laboratory, these findings highlight how Alzheimer's-related proteins in the brain can trigger systemic effects throughout the body. The research opens new avenues for identifying biomarkers and therapeutic targets beyond the brain.

Integrative approach reveals promising candidates for AD risk factors or targets for treatments

Researchers at Baylor and the Duncan NRI developed an integrative strategy to identify genes that may influence Alzheimer's disease risk or serve as potential targets for treatment. The study was published in *The American Journal of Human Genetics* (doi: 10.1016/j.ajhg.2025.03.012).


Dr. Juan Botas

The team combined large-scale computational analyses with functional testing in living models to move beyond genetic association studies. By integrating genome-wide association data with multiple computational approaches, the researchers identified genes whose altered expression predicted increased Alzheimer's disease risk in humans and behavioral impairments in fruit fly models. Importantly, they showed that reversing some of these gene alterations produced neuroprotective effects.

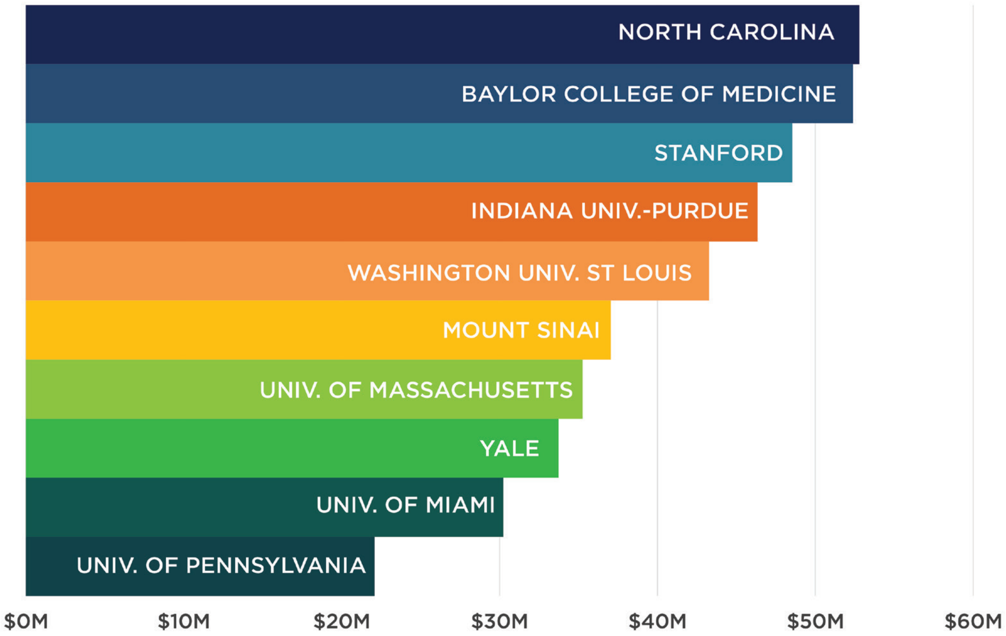
"Alzheimer's disease affects more than 50 million people worldwide and although researchers have learned a great deal about it over the years, its causes are still not fully understood and effective therapies are not yet available," said Dr. Juan Botas, professor of molecular and human genetics and molecular and cellular biology. Botas is also the director of the High-Throughput Behavioral Screening Core at the Duncan NRI.

To validate their findings, the researchers experimentally tested computational predictions by systematically altering candidate genes and examining their effects on neuronal dysfunction and hallmark Alzheimer's-related changes, including tau and beta-amyloid accumulation. Fruit fly models were used to assess motor impairments in Alzheimer's disease.

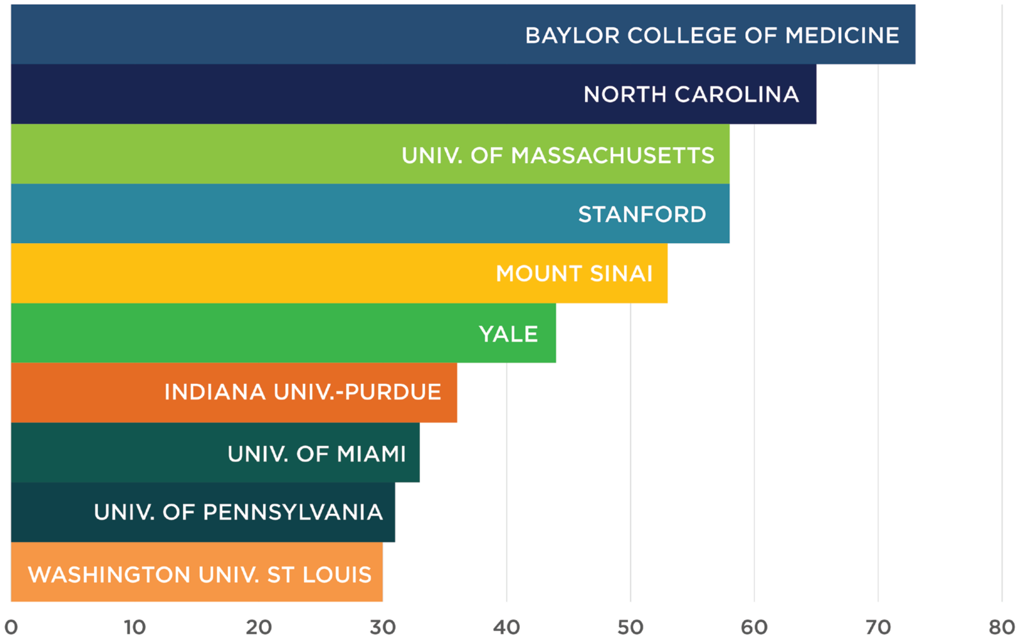
Grant awards continue to drive progress

The National Institutes of Health is the primary governmental agency responsible for biomedical and health-related research in the U.S. Success in securing NIH funding through a highly-competitive peer review process is a clear indicator of scientific excellence. By this measure, the Department of Molecular and Human Genetics stands at the forefront of the field. With the highest total number of NIH grants awarded and an average annual NIH funding level approaching $75 million, the department demonstrates unmatched breadth, sustained productivity and national leadership in biomedical discovery.

Total NIH Funding to Leading Genetics Departments for 2025



Number of NIH Grants Awarded to Leading Genetics Departments



Center for Precision Medicine Models receives $10M NIH grant

Baylor College of Medicine's Center for Precision Medicine Models (CPMM) has received a five-year, $10 million grant from the National Institutes of Health to continue and expand its research on rare genetic diseases. The center develops customized cell, fruit fly, and mouse models that replicate genetic variants found in patients, enabling researchers to better understand how specific genetic changes cause disease and to explore potential therapies for rare conditions that often lack effective treatment options.

With the new funding, CPMM will continue to strengthen collaborations across Baylor by integrating expertise in clinical genetics, animal modeling and bioinformatics. The work is led by Dr. Jason Heaney, associate professor of molecular and human genetics, with additional leadership from Dr. Lindsay Burrage, associate professor of molecular and human genetics.

Since its initial NIH funding in 2020, the center has received 156 nominations through its online portal, accepted 63 for modeling, and generated more than 200 precision models, contributing to the discovery of new rare disease genes and improved understanding of known genetic conditions.

NIH awards more than $15M to fund two rare diseases consortia

Baylor College of Medicine has received more than $15 million in funding from the NIH to support two consortia within the Rare Diseases Clinical Research Network. The awards include $7.4 million over five years to renew the Brittle Bone Disorders Consortium (BBDC) and $8.2 million over five years to establish the Rare Organic Acidemias Research (ROAR) Consortium, supporting research aimed at improving care for individuals with rare genetic and metabolic disorders.

The BBDC focuses on osteogenesis imperfecta, a genetic disorder characterized by fragile bones that affect both children and adults. Baylor and Texas Children's Hospital lead 12 clinical sites nationwide.

Over its first decade, the BBDC, led by Dr. Brendan Lee, professor and chair of molecular and human genetics, built the world's largest longitudinal cohort of patients with the condition, launched interventional clinical trials addressing bone and dental health, and published nearly 50 research studies.

The ROAR Consortium will study organic acidemias, a group of rare metabolic disorders. ROAR is led by Baylor and Texas Children's, with principal investigators Dr. V. Reid Sutton, professor of molecular and human genetics, and Dr. Lindsay Burrage, overseeing five clinical sites.

Other Grants/Awards

The Department is proud to receive generous funding from many agencies and foundations, some of which are listed below:

The Howard Hughes Medical Institute
The Robert and Janice McNair Foundation
The Cancer Prevention and Research Institute of Texas
The Welch Foundation
The Simons Foundation
The Huffington Foundation
The Doris Duke Foundation
The American Heart Association
Autism Speaks

Research Centers

Baylor College of Medicine is home to one of the largest biomedical research programs in the nation. The Department of Molecular and Human Genetics works closely with the aforementioned Center for Precision Medicine Models and the following centers, each focused on specialized areas of biomedical research. Led by primary faculty within the department, these centers collectively advance the frontiers of scientific discovery and medical innovation.

Human Genome Sequencing Center

The Baylor Human Genome Sequencing Center (HGSC), led by Dr. Richard Gibbs, Wofford Cain Chair and professor of molecular and human genetics, has been a cornerstone of genomic discovery for more than two decades. Established in 1996 to participate in—and ultimately help complete—the Human Genome Project, the center has grown into an internationally recognized hub for large-scale DNA sequencing and analysis. Today, the HGSC serves as a Center for Complex Disease Genomics supported by the National Institutes of Health and the National Human Genome Research Institute, with a mission focused on translating genomic discovery from bench to bedside.

Dr. Richard Gibbs in Baylor's Human Genome Sequencing Center.

Located in the Texas Medical Center, the HGSC employs more than 180 staff members and occupies over 36,000 square feet in the Margaret M. and Albert B. Alkek Building at Baylor. Its core activities include high-throughput DNA sequencing and advanced genomic analysis using multiple technology platforms, supported by sophisticated bioinformatics pipelines and broad data sharing with the global research community. A central focus of the HGSC is defining the genetic basis of common complex diseases—including cardiovascular disease, neurodegeneration and cancer predisposition—work that underpins national precision medicine efforts. To support clinical translation, the HGSC also operates the HGSC Clinical Lab (HGSC-CL), a CAP-accredited and CLIA-certified facility that delivers clinical-grade genomic data for diagnostic use and return of results to physicians

In 2025, the HGSC continued to demonstrate national leadership across rare disease genomics, somatic mutation biology, infectious disease sequencing and precision medicine. The GREGoR program advanced efforts to solve previously unsolved Mendelian conditions through innovative analytical and sequencing approaches. The NIH Common Fund supported SMaHT initiative entered its third year, with HGSC investigators playing a central role in generating deep, multi-tissue genomic datasets designed to establish a foundational reference for somatic variation in human health and disease, alongside new collaborations extending these insights to thyroid, liver and vascular tissues. Through the NIAID Genome Center for Infectious Disease program, the HGSC continued to provide critical sequencing and genomic characterization technologies supporting studies of mucosal infections and microbial diversity, while TOPMed activities sustained large-scale proteomics and long-read WGS data generation driving discovery in complex disease biology.

The HGSC partners with Texas Children's Hospital to solve undiagnosed pediatric conditions with support from the Chan Zuckerberg Initiative (CZI). The CZI program leverages whole genome sequencing and functional genetic studies to elucidate the affected genes and biological pathways. Additionally, the TRISH Space Omics and Biobank program advanced comparative analyses of astronaut pre-flight and post-flight samples, supported by the HGSC-developed GENESTAR framework, providing novel insights into gene expression responses to extreme environmental stressors with implications for both space biology and terrestrial medicine. Complementing these national programs, the HGSC-led local precision medicine program, IMAGINE, continued to scale recruitment in Baylor Medicine departments, and launched return of results in August 2025. Within these programs, the HGSC advanced research into AI applications to benefit the return of genetic results to patients and research participants. This work was presented at a National Academy of Sciences event in October 2025 entitled: Exploring Applications of AI in Genomics and Precision Health: A Workshop.

Together, these efforts reflect the HGSC's sustained impact and evolution—from its foundational role in the Human Genome Project to its current leadership in large-scale genomics, precision medicine and clinical translation.

Jan and Dan Duncan Neurological Research Institute (NRI) at Texas Children's Hospital

Jan and Dan Duncan Neurological Research Institute

The Jan and Dan Duncan Neurological Research Institute (Duncan NRI) at Texas Children's Hospital opened in December 2010 as the first facility in the U.S. dedicated to pediatric brain disorder research. The Duncan NRI is now a leader in neurogenetic discovery, with research advances that extend well beyond pediatrics to improve our understanding of neurodevelopmental, neurodegenerative and neuro-psychiatric disease across the lifespan.

Under the direction of founding director Dr. Huda Zoghbi, Distinguished Service Professor at Baylor and Howard Hughes Medical Institute Investigator, the Duncan NRI fosters a uniquely collaborative research environment designed to shape the future of neurological medicine. Investigators work across disciplines—including genetics, neurobiology, engineering, biochemistry, bioinformatics and artificial intelligence—to promote the exchange of ideas, data and resources. This integrative approach remains central to the Duncan NRI's culture, resulting in profound scientific impact.

The Duncan NRI is now home to 43 principal investigators and 400-plus trainees and scientific staff. Duncan NRI investigators have published more than 1,900 scientific studies in top-tier journals, discovered over 100 disease-causing genetic mutations, obtained FDA approval for the treatment of a previously-intractable epilepsy, and advanced multiple discoveries into ongoing clinical trials. These discoveries span a wide spectrum of often rare neurological and neuropsychiatric conditions, including autism, epilepsy, Rett syndrome, MECP2 duplication syndrome, EBF3-HADDS, Alzheimer's disease, Parkinson's disease, bipolar disorder, OCD, eating disorders and addiction.

Faculty excellence remains a hallmark of the Duncan NRI, with investigators receiving numerous competitive research grants from federal, state and private philanthropic organizations, as well as prestigious honors nationally and internationally. And the Duncan NRI serves as a hub for scientific exchange, hosting lectures, symposia and workshops with the very best scientists, clinicians, industry leaders, patients and their families to devise the best path forward for previously intractable conditions.

Community engagement is integral to the Duncan NRI's mission. School outreach, including in-school hands-on experiences, field trips to the Duncan NRI, and hosting the student-led Epilepsy Youth Forum, inspired a new generation of potential biomedical scientists and physicians. Duncan NRI faculty, staff and trainees also participated in initiatives such as the Walk to End Alzheimer's, demonstrating their commitment to public awareness.

Together, these efforts reflect a year of meaningful progress built on 15 years of scientific leadership, reinforcing the Duncan NRI's role as a premier center for neurological discovery and translational impact.

Baylor Rice Genome Editing Testing Center

Established in 2023 with more than $3.9 million in funding over five years from the NIH's Office of Research Infrastructure Programs, the Baylor/Rice Genome Editing Testing Center (GETC) was created to help researchers nationwide advance promising somatic genome editing technologies toward clinical application. The center addresses a critical gap in translation by providing rigorous preclinical testing and mouse model resources needed to evaluate whether new genome editing tools are effective, precise and safe before they are considered for use in humans. The GETC is co-led by Dr. Jason Heaney, associate professor of molecular and human genetics, Dr. William Lagor, professor of integrative physiology, and Dr. Christopher Walkey, instructor of integrative physiology.

In its first year, the GETC quickly established itself as a national resource for *in vivo* genome editing research, offering a broad range of services that support genome editing experiments in mouse models. These services include evaluating how genome editing tools are delivered and distributed within tissues, validation of novel viral vectors, testing gene editing–based therapeutic strategies and assessing potential off-target effects using complementary computational and laboratory approaches.

The center has already provided testing services for seven academic research teams, delivering critical support through specialized assays, mouse models, and technical expertise. While early efforts focused on understanding delivery and tissue distribution, much of the first year's work evolved toward early-stage evaluation of genome editing strategies themselves. This included developing and validating viral vectors for somatic genome editing and responding to growing demand for careful measurement of editing accuracy and safety.

During the past year, the GETC supported a wide range of genome editing approaches, including CRISPR-based activation systems, engineered viral vectors, lipid nanoparticle delivery of CRISPR/Cas9 components and donor templates, and comprehensive analysis of both intended and unintended genome edits. Research enabled by the GETC contributed to conference presentations and peer-reviewed publications in 2025, highlighting the center's role in accelerating impactful scientific discovery.

Looking ahead, the GETC aims to expand its national reach, strengthen collaborative partnerships and continue refining methods that make genome editing more effective, precise, and safe—helping ensure that today's scientific advances can become tomorrow's therapies.


Dr. Brendan Lee, director of the Center for Skeletal Medicine and Biology with Nele Haelterman, Ph.D.

Center for Skeletal Medicine and Biology

The Center for Skeletal Medicine and Biology (CSMB), co-directed by Dr. Brendan Lee, professor and chair of molecular and human genetics, and Dr. Florent Elefteriou, professor of molecular and human genetics and orthopedic surgery, aims to advance the understanding, prevention and treatment of congenital and degenerative skeletal disorders. Its research spans skeletal dysplasias, osteoporosis, osteoarthritis, low back pain and bone cancers.

The CSMB leverages the Lawrence Family Bone Disease Program of Texas (BDPT), a contractual collaboration among Baylor, The University of Texas MD Anderson Cancer Center and The University of Texas Health Science Center at Houston, to foster team science across the Texas Medical Center and, since 2024, beyond Houston. The Center provides Baylor investigators with specialized resources for musculoskeletal research and creates opportunities for faculty and trainees to connect, collaborate and share expertise.

This year, Dr. Angela Bachim and Dr. Simon Kelley, assistant professors of orthopedic surgery, joined the Center's faculty, bringing additional clinical expertise and expanding translational research opportunities. The 2025 annual BDPT retreat convened more than 120 scientists and trainees from Texas Medical Center institutions and universities across Texas, including UT Southwestern Dallas, UT Health Science Center at San Antonio, UT Austin, and Texas A&M. The keynote speaker, Dr. Aris Economides, vice president of research at Regeneron Pharmaceuticals, presented his work on molecular disease mechanisms and the development of therapies for fibrodysplasia ossificans progressiva. The monthly seminar series hosted six speakers from external U.S. institutions and drew in-person and virtual participation from investigators in Houston and research centers statewide.

Bioinformatics Research Laboratory

The Bioinformatics Research Laboratory, directed by Dr. Aleksander Milosavljevic, Henry and Emma Meyer professor of molecular and human genetics, develops data-intensive and computational methods that accelerate genomic discovery and advance genomic medicine. The laboratory collaborates with more than a dozen research groups across genomics, epigenomics, extracellular RNA (exRNA) communication, and brain, heart, and tumor biology, and is supported by a team of software engineers pioneering modern web- and cloud-based technologies.

The laboratory is recognized for innovative information-theoretic approaches to biological network analysis, deconvolution of complex tissue omics profiles, and identification of pathogenic variants. As part of the NIH Roadmap Epigenomics Program, the group constructed the Human Epigenome Atlas, now applied with computational deconvolution and single-cell profiling to study Alzheimer's disease, heart failure, and cancer progression.

The laboratory also pioneered the use of epigenomic allelic imbalances to define cis-regulatory effects of genetic variation, leading to a unifying model linking regulatory variation, stochastic gene regulation, and disease risk. Building on this work, the laboratory leads method and infrastructure development within the NIH Common Fund Ecosystem project to enable discovery of disease-causing regulatory variants from whole-genome sequencing.

In addition, the laboratory developed core informatics infrastructure for the FDA-recognized Clinical Genome Resource (ClinGen), including globally used tools such as the ClinGen Allele Registry, Criteria Specification Registry, and Evidence Repository. Through the NIH Extracellular RNA Communication Consortium, the group constructed the exRNA Atlas and identified extracellular RNA-binding proteins and their RNA cargo, uncovering a novel exRNA-mediated mechanism of vascularization in glioblastoma that explains resistance to anti-VEGF therapies.

Computational and Integrative Biomedical Research Center

The Computational and Integrative Biomedical Research (CIBR) Center is directed by Dr. Olivier Lichtarge, Cullen Chair and professor of molecular and human genetics.

The CIBR Center is comprised of more than 100 affiliate faculty members from different Houston institutions. The CIBR Center helps the College bridge the translational gap from data to models, and from models to drug discovery and personalized therapy by fostering collaborations among scientists and developing original quantitative approaches to biological and clinical problems.

To assist students and faculty, the CIBR Center provides the resources to help address the broad range of analytical problems posed by the complexity of high throughput biological datasets. The center organizes the Current Topics in Computational Biomedicine Course where students keep abreast of active quantitative research among the CIBR faculty. To date, the Current Topics course has hosted more than 160 seminars and approximately 40 journal clubs.

In addition to the Current Topics course, the CIBR Center coordinates workshops and access to cluster computing for its faculty members. The center provides site licenses to scientific software (Mathworks MATLAB and Wolfram Mathematica) and regular consultation on data organization and analysis through its Data Clinics (16 sessions per year).

Intellectual and Developmental Disabilities Research Center

The Intellectual and Developmental Disabilities Research Center (IDDRC) at Baylor College of Medicine, led by Dr. Sandesh Nagamani and Dr. David L. Nelson, professors of molecular and human genetics, is one of 15 centers nationwide funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD). Continuously supported since 1988, the IDDRC has played a vital role in advancing translational research in intellectual and developmental disabilities (IDDs) and in improving the lives of individuals and families affected by these conditions.

The Baylor IDDRC has discovered many genetic causes of IDDs, developed robust preclinical models to enable clinical translation, established best practices for rigorous and reproducible model-organism research and trained multiple generations of scientists and physicians dedicated to IDD research and care. Its six core facilities—the Clinical Translational Core (CTC), Molecular and Expression Analysis (MEA) Core, Cell and Tissue Pathogenesis (CTP) Core, Circuit Analysis and Modulation (CAM) Core, Preclinical and Clinical Outcomes (PCO) Core and the Administrative Core—provide critical infrastructure that transforms basic scientific discoveries into biomarkers and novel therapeutic approaches.

Each year, the Center supports more than 80 investigators across 11 departments engaged in basic, translational and clinical studies. From 2020–2025, IDDRC investigators produced 331 publications supported directly by the Center's grant. Over the past 5 years, collectively, the IDDRC investigators have published 2,285 papers and secured 205 NIH grants across 14 NIH institutes, including 115 R01 awards—demonstrating the Center's broad and sustained impact across the biomedical research community.

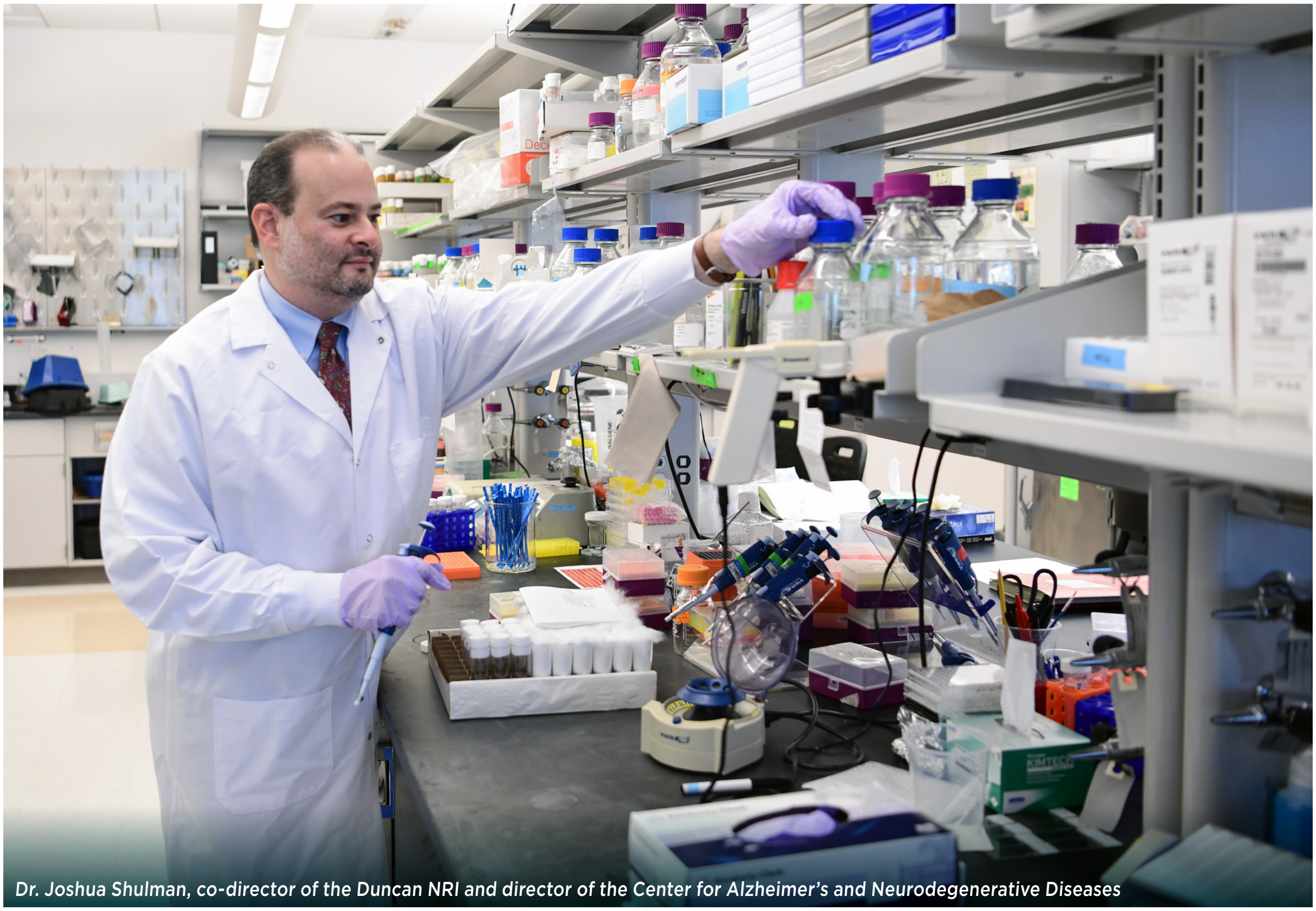
Since the start of the current funding cycle in 2020, the Center has also recruited 38 new faculty members and expanded its capabilities with advanced spatial gene expression platforms, enhanced mass spectrometry for single-cell proteomics and machine-learning–based behavioral analysis systems. The Center's Signature project identified neurophysiological, and molecular biomarkers that discriminate MECP2 dosage, leading to commencement of the gene therapy clinical trial for MECP2 Duplication Syndrome.

Center for Alzheimer's and Neurodegenerative Diseases

Neurodegenerative diseases, including Alzheimer's, Parkinson's and Lou Gehrig's disease, are currently incurable conditions characterized by the progressive deterioration of neurons (brain cells) that are vital for cognitive, motor and other functions. More than 400,000 Texans and nearly 6 million Americans suffer from Alzheimer's. At least 50,000 in Texas and 1 million in the U.S. have Parkinson's disease. As our population grows older, the prevalence of these and related conditions are anticipated to swell unless effective treatments or preventive approaches are developed. The Center for Alzheimer's and Neurodegenerative Diseases (CAND), directed by Dr. Joshua Shulman, co-director of the Duncan NRI and professor of neurology, molecular and human genetics, and neuroscience at Baylor, integrates cross-disciplinary clinical research and educational programs to advance precision diagnosis and personalized therapies for Alzheimer's, Parkinson's and other neurodegenerative diseases.

In the last year, CAND precision medicine core faculty integrated neurologic and neuropsychiatric evaluations, blood and neuroimaging biomarkers, along with cutting edge genetic testing to provide research volunteers with personalized dementia risk assessments. Our comprehensive risk disclosure includes polygenic risk scores incorporating genetic information from more than 100,000 different sites in the genome—this assay was developed together with the HGSC. In further collaboration with the Center for Medical Ethics and Health Policy, CAND is studying how to effectively and responsibly communicate findings and to document their impact.

CAND resources, including clinical and genomic data along with biospecimens, are available to enhance research across Baylor. A growing collection of induced pluripotent stem cells derived from well-characterized research participants adds to CAND precision disease models available from fruit flies and mice. In partnership with UT Health San Antonio, CAND has also developed a brain banking resource, making postmortem brain tissues available for Baylor faculty studying neurodegenerative disease. Lastly, CAND Pilot grants and Scholarships, along with an annual symposium, are further attracting new investigators and trainees to help elucidate mechanisms of Alzheimer's, Parkinson's and related neurodegenerative diseases, pointing the way to more effective therapies.



Dr. Joshua Shulman, co-director of the Duncan NRI and director of the Center for Alzheimer's and Neurodegenerative Diseases

Clinical Research

The Clinical Research Division within the Department of Molecular and Human Genetics at Baylor College of Medicine, led by Dr. Sandesh Nagamani, professor of molecular and human genetics, oversees the design and execution of clinical studies and trials focused on rare disorders. The Division's portfolio spans gene discovery studies, natural history studies, proof-of-concept investigations that translate discoveries from bench to bedside, early-phase clinical trials and pivotal registration trials that support FDA approval of novel therapies for genetic diseases.

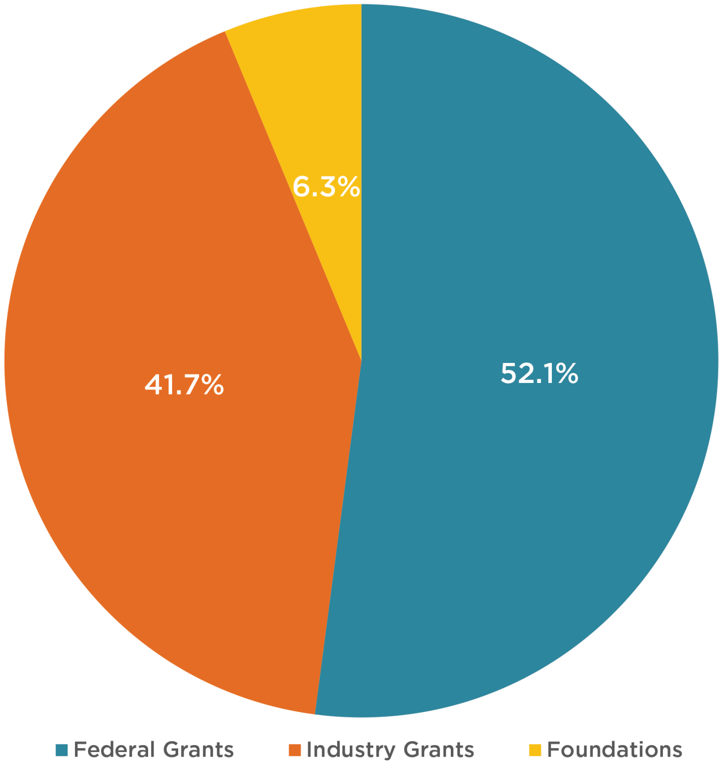
The Department and its Clinical Research Division have served as a primary coordinating or lead site for numerous NIH Rare Diseases Clinical Research Network (RDCRN) consortia, including the Urea Cycle Disorders Consortium, Brittle Bone Disorders Consortium, North American Mitochondrial Disease Consortium, Global Leukodystrophy Initiative Clinical Trials Network, and the Frontiers in Congenital Disorders of Glycosylation.

In 2025, the Division also became the primary coordinating site for the Rare Organic Acidemias Research Consortium. In addition, Baylor's Undiagnosed Diseases Center, Center for Precision Medicine Models, GREGoR Consortium and Intellectual and Developmental Disabilities Research Center conduct research in partnership with the Division.

Over the past five years, the Division has conducted an average of 54 studies annually, supported by $50.3M in total funding across 125 federal grants ($33.3M), 116 industry grants ($16.5M) and 28 private nonprofit grants ($0.6M).

In 2025 alone, 48 active studies were supported by $11.1M in funding from 25 federal grants ($4.6M), 20 industry grants ($6.4M) and 3 private nonprofit grants.

2025 Clinical Studies



Clinical Services

Improving patients’ lives with unmatched clinical services

Baylor College of Medicine’s clinical genetics program is the largest program of its kind in the country, with clinics spanning across multiple genetics-based disciplines. The clinical program takes a collaborative approach that provides patients with the highest quality, individualized care available. Clinical activities take place across several sites.

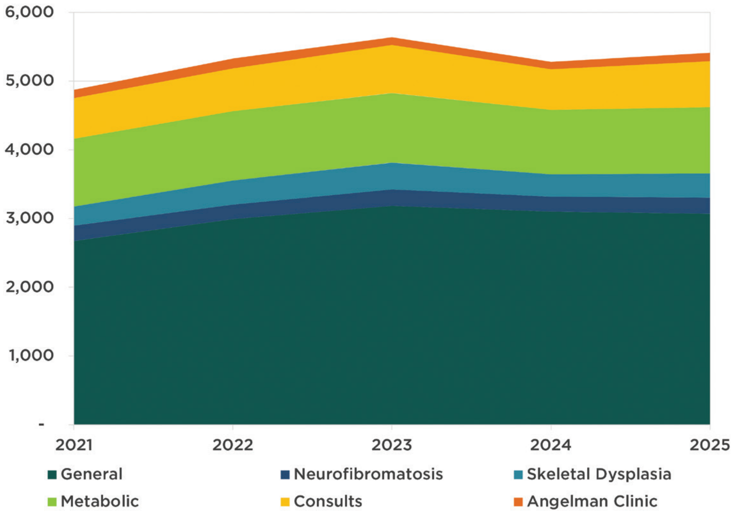
Pediatric Genetics

The Department of Molecular and Human Genetics’s pediatric genetics clinic service provides inpatient care to complex and/or critically ill patients at Texas Children’s Hospital and several other hospitals within the Texas Medical Center and just outside Houston (Texas Children’s Hospital West Campus and Texas Children’s Hospital The Woodlands). The outpatient pediatric genetics clinics are among the largest genetics clinics in the U.S. and see more than 5,000 patients annually.

Recently, the Department of Molecular and Human Genetics has partnered with Texas Children’s Hospital to establish inpatient and outpatient clinical genetics services at the new Texas Children’s Hospital North Austin Campus.

Specialty clinics within the Texas Children’s Genetics Clinic include the metabolic clinic, neurofibromatosis clinic and the skeletal dysplasia clinic. There are many multidisciplinary team clinics like the Angelman Syndrome Clinic, and the Mitochondrial Medicine Clinic. The department’s clinical and genetic counseling faculty also staff joint clinics with other departments such as oncology (cancer genetics), otolaryngology (otogenetics) and neurology (neurogenetics/tuberous sclerosis).

Clinical Genetics Patient Volume (Pediatric)



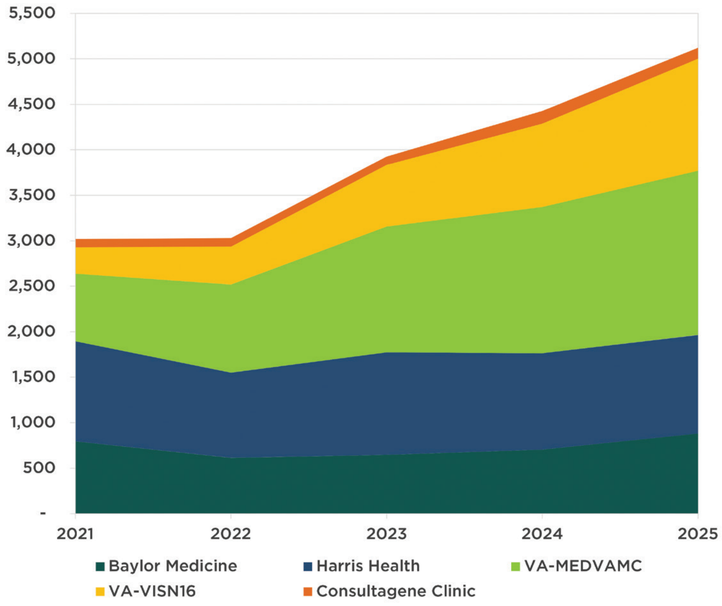
Adult Genetics

The Department’s adult genetics clinical service is one of the largest genetics clinics of its kind, providing inpatient and outpatient care and genetic counseling exclusively for adult patients in four different healthcare settings (Baylor Medicine, Harris Health, the Michael E. DeBakey VA Medical Center and through the virtual Consultagene Clinic). In addition, the service sees patients via telemedicine throughout Veteran Integrated Service Network 16 (VISN16). The U.S. is divided into 18 VISNs that provide care for veterans. VISN16 includes eight veterans affairs medical centers (VAMCS) and community clinics in the Southeast U.S.

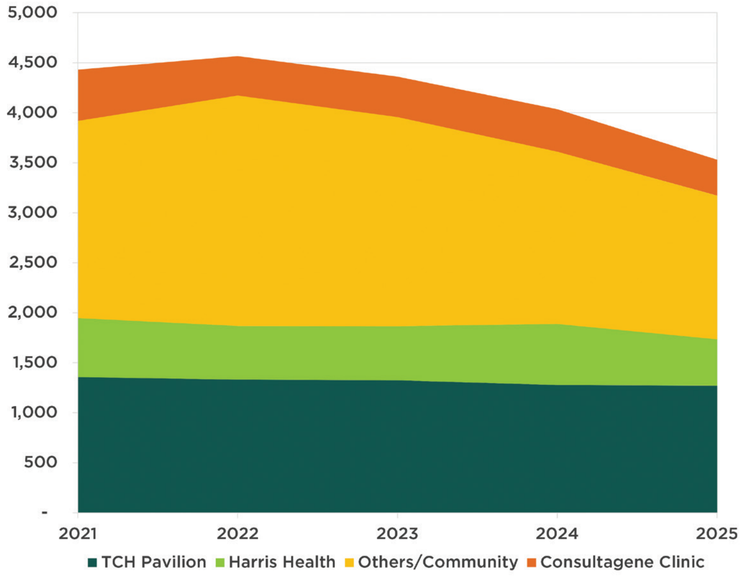
This service sees patients for a wide variety of indications including, but not limited to, intellectual disability, neurological conditions, cardiovascular conditions, connective tissue disorders and for a personal or family history of cancer.

In addition to our general genetics clinic, we have specialty clinics such as the Metabolic and Genetic Disorders of the Bone Clinic, Cancer Genetics Clinic, Neurogenetics Clinic, Cardiovascular Genetics Clinic and Mitochondrial Medicine Clinic.

Clinical Genetics Patient Volume (Adult)



Clinical Genetics Patient Volume (Prenatal and Reproductive)



Prenatal and Reproductive Genetics

The physicians and genetic counselors of the Baylor Prenatal and Reproductive Genetic Counseling Service specialize in prenatal and reproductive genetic risk assessment and the latest genetic testing technologies. Through partnership with the Department and the Texas Children's Fetal Center, they offer world-renowned clinical and research expertise in prenatal and preconception genetic screening, diagnostic testing and counseling.

Services are offered at TCH through in-person and virtual visits at the Pavilion for Women, seven Houston community maternal-fetal medicine clinics, and the Austin campus maternal fetal medicine clinic. We also offer genetic counseling at the Ben Taub Tower Specialty Clinics, and virtually through Baylor's Consultagene Clinic.

The Consultagene Clinic

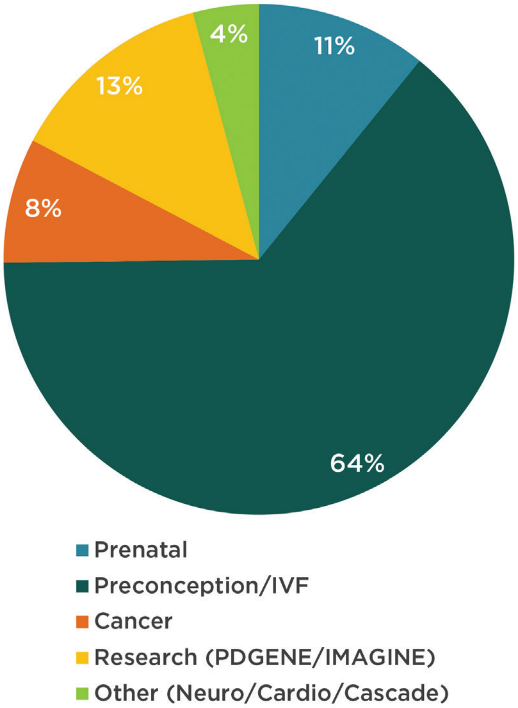
The Consultagene Clinic is now starting its 7th year of operation as a fully virtual genetic counseling clinic. In 2025, a total of 480 patients were seen. Since its launch in 2019, a total of 3,883 patients have received genetic counseling through the Consultagene Clinic.

Preconception/IVF referrals continue to make up the majority of referrals seen, making up 64% of referrals in 2025. Cancer and cardiology referrals continue to remain stable and research use of Consultagene continues to expand with new collaboration with the BCM IMAGINE study for ACMG secondary findings disclosures. The clinic also continues to see requests for genetic counseling from patients outside of Texas.

To address the continued interest, expand its reach and make genetic counseling more accessible, the clinic now has genetic counselors licensed in Arkansas, California, Illinois, Louisiana, New Mexico, Oklahoma and Washington. The clinic has continued outreach to IVF practices in these states and states without genetic counseling licensure and also performed outreach to local practices.

Patients seen in the clinic are provided access to the Consultagene platform, allowing patients to watch educational videos, explore online resources, communicate with their provider and access documentation from their consultations. The Consultagene website makes it easy for providers to refer patients for genetic counseling as well as patients to self-refer.

Patients are asked to participate in a survey to gauge their experience with the clinic and the platform. Of the 362 patients who have participated in the survey, 50% used the resources provided in the patient portal; 99% said the genetic counseling met or exceeded their expectations; 96% agreed or strongly agreed that virtual genetic counseling was equivalent to an in-person appointment; and 96% would recommend virtual genetic counseling.



2025 Consultagene Consultations By Indication

Education & Community Engagement

The Genetics and Genomics Graduate Program provides outstanding educational opportunities for students who wish to pursue careers in the broad and exciting field of genetics. In addition to their work in genetics, graduate students receive rigorous training in modern biology, bioinformatics, DNA replication and repair and other diverse fields. They also participate in cutting-edge research and publish their work in highly respected peer-reviewed scientific journals.

Awards and Special Recognitions for Genetics and Genomics Graduate Program Students

- Ruu Hsu received the Emerging Scientist Grant from Kids Beating Cancer.
- Matthew McAlister earned the 2nd Place Speaker Award at the BCM Molecular and Human Genetics Annual Retreat.
- English Laserna received multiple honors, including: 1st Place Talk at the 35th Annual Research Symposium at BCM; International Scholar Award ($25,000) from the Philanthropic Educational Organization (PEO); 66th McKusick Short Course Educational Travel Award from The Jackson Laboratory; People's Choice Award in the American Society of Microbiology–Texas Medical Center Image Contest; 1st Place Speaker Award at the BCM Molecular and Human Genetics Annual Retreat
- Brooke Horist received the 3rd Place Speaker Award at the BCM Molecular and Human Genetics Annual Retreat.
- Nathalie Aceves was selected as a Predoctoral Finalist for the 2025 ASHG Trainee Research Excellence Award
- Anna Burkhalter was named a 2025 BP America Biomedical Scholar through the BP America Biomedical Scholar Endowment.
- Professor John J. Trentin Scholarship Award from Baylor College of Medicine Graduate School of Biomedical Sciences 2025 recipients included Daniel Brock, Jasmine Brown, Zachary Castles, Yifan Chen, Adamantia Koulouri, Harita Mani, Soraya Nevin, Chloe Sands, Aanya Subramaniam, Katrine Taran and Kancheng Yin.

Dr. Brendan Lee with Genetics and Genomics Graduate Student, English Laserna and recent Genetics and Genomics Graduate, Morgan Stephens

2025 Genetics and Genomics Graduate Program Dissertations

Student Name	Title	Preceptor/Advisor
Matthew Avalos	Targeting SERPINs as a therapeutic strategy to ameliorate neurodegenerative disease	Juan Botas, Ph.D.
Matthew Cooke	Transcriptional Determinants of Microbial Genome Stability	Christophe Herman, Ph.D.
Colleen Dupuis	Characterizing cells in the mouse utricle and their regenerative potential through single-cell RNA sequencing	Andrew K. Groves, Ph.D.
Duncan Fox	The Role of SOX4 in Biliary Development and as an Effective Therapeutic Target in Alagille Syndrome	Hamed Jafar-Nejad, M.D.
Jordan Fox	Loss of Mitochondrial Fusion Results in Iron Imbalance and Nuclear Genome Instability	Grzegorz Ira, Ph.D.
Brandon Garcia	Retrieval-Augmented Phenotype Extraction with Large Language Models	Jennifer Posey, M.D., Ph.D.
Katarzyna Kent	Unraveling protease-mediated mechanisms of spermatogenesis: from gene editing models to translational advances in male fertility	Martin M. Matzuk, M.D., Ph.D.
Bree Lege	Single-Cell Analysis of Circulating Tumor Cells to Clarify Mechanisms of Metastasis in Breast Cancer	Chonghui Cheng, M.D., Ph.D.
Howard Lin	Defining mechanisms of pathogenesis and exploring novel therapeutic avenues in Langerhans Cell Histiocytosis	William Parsons, M.D., Ph.D.
Khushali Patel	Hyaluronic acid mediates circulating tumor cell clustering in triple-negative breast cancer metastasis	Chonghui Cheng, M.D., Ph.D.
Morgan Stephens	Retrieval-Augmented Phenotype Extraction with Large Language Models	Juan Botas, Ph.D.
Venkatasubramaniam Sundaramurthy	Role of clonal hematopoiesis in the development of cardiovascular diseases	Margaret Goodell, Ph.D.
Nicole Wang	Unveiling the mechanisms of circulating tumor cell clustering	Chonghui Cheng, M.D., Ph.D.
Alice Wen	Antibiotics Stimulate Proteome Transfer to Persister Cells	Christophe Herman, Ph.D.
Kevin Wilhelm	A Meta-Evolutionary Framework for the Discovery and Functional Prioritization of Complex Disease Risk Genes	Olivier Lichtarge, M.D., Ph.D.
Eder Xhako	Study of the Contributions of Wildtype Ataxin-1 to the Pathogenesis of Spinocerebellar Ataxia Type 1	Huda Zoghbi, M.D.

Residency and Clinical Laboratory Fellowship Programs

Throughout the evolution of the Department of Molecular and Human Genetics, there has been a strong emphasis on training and education. The residency and clinical lab fellowship programs began in 1987 and are among the Department's highest priorities. The growth and development of the Department and its clinical enterprises are linked to the excellence of the training programs and its trainees.

The Medical Genetics and Genomics Residency Programs at Baylor College of Medicine are designed to prepare individuals for an academic career by providing an integrated experience in both clinical and experimental genetics. Training activities in clinical genetics and research are coordinated through the Department of Molecular and Human Genetics. The programs prepare trainees to care for both pediatric and adult patients with cytogenetic, biochemical and developmental diseases. Residents also gain laboratory experience in a chosen area of medical genetics and genomics.

The Department's residency programs enjoy preeminence in the genetics community. They are approved by the Accreditation Council for Graduate Medical Education and supported by a training grant from the National Institute of General Medical Sciences.

Trainees in the Department's clinical laboratory fellowship programs train at Baylor's genetics diagnostic laboratory, Baylor Genetics, for 24 months.

The Department also offers a one-year Medical Biochemical Genetics Fellowship program that provides additional training in the diagnosis and management of inborn errors of metabolism.

After completion of these programs, trainees are eligible for American Board of Medical Genetics and Genomics certification.

Residencies

- Medical Genetics and Genomics
- Pediatrics/Medical Genetics and Genomics
- Internal Medicine/Medical Genetics and Genomics
- Maternal-Fetal Medicine/Medical Genetics and Genomics Fellowship

Clinical Laboratory Fellowships

- Laboratory Genetics and Genomics
- Clinical Biochemical Genetics

2025 MHG Trainee Awards

Amber Nakar Weinstein, M.D.
Clinical Resident Award

Aaron Williams, M.D.
Clinical Resident Award

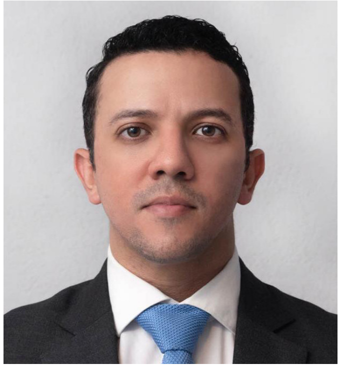
Haowei Du, Ph.D.
Laboratory Fellow Award

2025 Graduating Class of Residents and Fellows

Ryan Difalco, M.D.
Pediatric Genetics Residency

Dan Brooks M.D.
Pediatric Genetics Residency

Taylor Kerrins, M.D.
Medical Genetics Residency



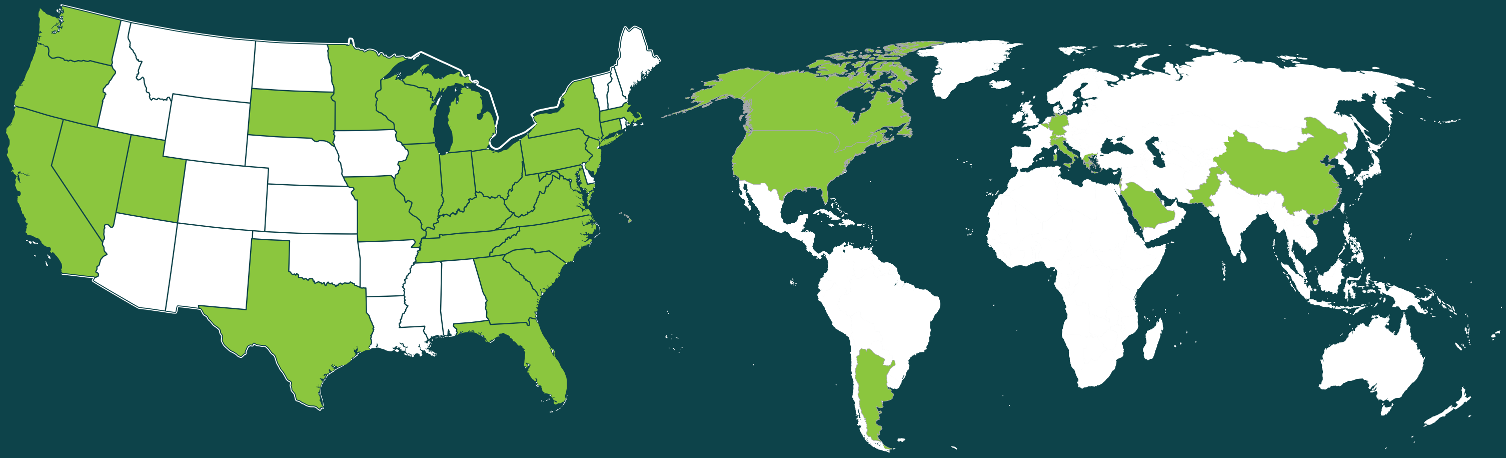
Andres Caceres Salgado, M.D.
Medical Genetics Residency

Amir Hossein Saeidian, Ph.D.
Laboratory Genetics and Genomics Fellowship

Chelsi Rose, D.O.
Medical Biochemical Genetics Fellowship

Veronica Ortiz, Ph.D.
Laboratory Genetics and Genomics Fellowship

Locations of Medical Genetics Alumni



Genetic Counseling Program

The Baylor College of Medicine Genetic Counseling Program was established in 2018 with financial and logistical support from the Department of Molecular and Human Genetics. The 22-month Master of Science program integrates rigorous didactic instruction, extensive clinical rotations and student-driven research to prepare graduates for a wide range of genetic counseling careers. This comprehensive training model has contributed to strong educational outcomes and program stability within the School of Health Professions.

Over the past three years, graduates have achieved a 75% first-time pass rate on the American Board of Genetic Counseling (ABGC) Certification Examination. Employment outcomes have remained particularly strong, with 94% of graduates securing positions within three months of graduation across clinical, laboratory, research and industry settings. Notably, the program has maintained a 0% attrition rate since its inception, reflecting strong student support and alignment between program expectations and trainee preparedness.

Students benefit from training within the Texas Medical Center and access to the nation's largest clinical genetics program, including rotations in prenatal, pediatric, adult and cancer genetics. Overall, the Baylor College of Medicine Genetic Counseling Program continues to demonstrate excellence and measurable success, positioning it for ongoing impact and leadership in genetic counseling education.

2025 Graduates

Claudia Cerezo, Zoe Epstein, Ryan German, Katherine McCormick, Aeran Melancon, Ashley Ray, McKay Schumway, Sonja Singelstad and Julia Volpi

Introducing the Class of 2027

Zoe Wayne, Grace Stinson, Danielle Schwalk, Amanda Pastrano, Hannah Namgoog, Grace Hanson, McKenna Groessl, Delaney Gaston, Jiaqi (Kiki) Ding and Satya Das.

The Genetic Counseling Program Class of 2027 pictured with program leadership Dan Riconda, M.S., C.G.C., Salma Nassef, M.S., C.G.C. and Rachel Franciskovich, M.S., C.G.C.

Distinguished Lectures

Jeanette Oshman Efron Lecture in Molecular Genetics

Dr. Brendan Lee and Dr. Huda Zoghbi with 2025 Jeanette Oshman Efron Lecture in Molecular Genetics Lecturer, Dr. Erich D. Jarvis

Jeanette Oshman Efron, who died in 2009 at 98, was an ardent supporter of science and the arts and a generous friend to Baylor College of Medicine. The Oshman Lectureship in Molecular Genetics was established at Baylor in 1989 by her daughters, Marilyn Oshman and Judy Margolis, and her grandchildren, Karen Desenberg, Gary Gerson, Jay Gerson and Andrew Lubetkin, to honor Jeanette's passion and commitment to the advancement of medical education and biomedical research.

This lecture series, which is held once every two years, brings internationally renowned scientists to Baylor to present seminars on important developments in genetics. This year, Erich D. Jarvis was the featured speaker. The title was "Evolution of Brain Pathways for Vocal Learning and Spoken Language."

Jarvis is a professor at The Rockefeller University and a Howard Hughes Medical Institute (HHMI) Investigator, where he heads the Laboratory of Neurogenetics of Language. His research focuses on the neural, genetic and genomic mechanisms underlying vocal learning and spoken language, using song-learning birds and other species as models to understand how complex communication behaviors are generated, learned and represented in the brain.

Jarvis is also a leader in large-scale genomics and evolutionary biology. He serves as chair of the international Vertebrate Genomes Project, which aims to generate high-quality reference genomes for all vertebrate species, and has played a central role in global collaborations that have reshaped understanding of avian evolution and brain organization. He earned his B.A. from Hunter College and his Ph.D. from The Rockefeller University, and his work has been recognized with numerous honors, including the National Science Foundation (NSF) Alan T. Waterman Award and the NIH Director's Pioneer Award.

Arthur L. Beaudet Lecture for Outstanding Mentorship

In 2019, Dr. Huda Zoghbi, Distinguished Service Professor at Baylor College of Medicine and director of the Jan and Dan Duncan Neurological Research Institute, established an endowment in the Department of Molecular and Human Genetics to recognize outstanding mentorship at Baylor.

The award is open to any outstanding faculty mentor from any discipline and all academic ranks. An eligible candidate must have demonstrated a sustained career of exemplary mentorship at the graduate, postdoctoral, residency, fellowship or junior faculty level across the educational, clinical and research

Dr. Huda Zoghbi, Dr. Arthur L. Beaudet, Dr. Sharon Plon and Dr. Brendan Lee at the 2025 Arthur L. Beaudet Lecture.

missions of the College. The award is named after its first recipient, Dr. Arthur L. Beaudet.

The awardee receives a plaque and a monetary award of $10,000 and is invited to speak or host an annual lecture. In 2025, the recognition went to Dr. Sharon Plon.

Plon is a professor of molecular and human genetics and pediatrics (Hematology–Oncology) and holds the Dan L. Duncan Comprehensive Cancer Center Professorship. She is co-leader of the pediatric cancer program at the Dan L. Duncan Comprehensive Cancer Center and directs the cancer genetics clinical and research programs at Texas Children's Hospital, where she founded the Cancer Genetics Clinic in 1995.

Her research focuses on inherited cancer predisposition syndromes and the clinical implementation of genomic sequencing in pediatric cancer care. She has served as a principal investigator on multiple NIH-funded studies, including the BASIC3 and Texas KidsCanSeq trials, which evaluated the integration of clinical exome sequencing into the care of children with newly diagnosed cancers.

She is a principal investigator of the NIH-supported Clinical Genome Resource (ClinGen) and co-chairs the Hereditary Cancer Clinical Domain, helping to establish national standards for the interpretation of cancer susceptibility genes.

Fernbach Lecture for Humanism in Genetics

In 2022, after many years of service, Susan Fernbach, former co-director of the Office of Community Outreach retired. Susan founded the *Evenings with Genetics* Program and served as a role model to many within the department. In recognition of her service, as well as her extensive community engagement and outreach efforts within the department and the genetics community at large, an annual lecture was established in her honor.

In 2025, the lecture featured, James G. Robinson, author of *More Than We Expected: Five Years With a Remarkable Child*. Robinson spoke about his family's experience parenting a child with a congenital heart defect that required extensive surgery.

Dr. Chaya Murali and Susan Fernbach with 2025 Fernbach Lecturer, James G. Robinson

Community Engagement

The Department of Molecular and Human Genetics Office of Community Engagement, co-directed by Dr. Debra Murray, associate professor of molecular and human genetics, and Gladys Pryor, works to provide education and resources regarding genetics services to the community at large.

Evenings with Genetics

Evenings with Genetics is a free virtual seminar series hosted by the Department and Texas Children's Hospital which is open to the public. Each seminar features a Baylor faculty member paired along with a parent speaker, to provide both clinical and lived perspectives. The series explores a wide range of topics, including genetics and autism as well as genetic-associated cardiac conditions in both adults and children.

For its 19th anniversary in February 2025, Evenings with Genetics honored Black History Month with its fifth annual Perspectives on Precision Medicine program. This webinar series examines the historical relationship between race and genetics, emphasizing that race is a social construct independent of genetics and ancestry. The event featured two distinguished speakers, Dr. Vanessa Northington Gamble—physician, historian, bioethicist and the first African American woman to hold a University Professorship at George Washington University—and Dr. April Adams, assistant professor of molecular and human genetics and obstetrics and gynecology.

Statewide Genetic Outreach

Statewide Genetics Outreach efforts in 2025 reached thousands of professionals and families through a combination of virtual training and in-person engagement.

Continuing-education webinars, offered in partnership with the Texas Department of State Health Services, the Texas Health and Human Services Department of State Health Services Newborn Screening Program, the University of Texas at Austin Center for Disability Studies, Baylor, and Texas Children's Hospital, consistently attracted 150 or more live professional attendees per session across Texas and beyond. Expanded credentialing opportunities allowed participants from multiple disciplines—including nursing, social work, behavioral health and early childhood education—to earn professional development credit. Topics included autism, cardiovascular genetic conditions, updates on Down syndrome and more.

In-person community conferences and resource fairs further strengthened regional access to genetics education. Events held in areas such as Laredo, El Paso and South Texas drew more than 100 family attendees per conference, with over 20 partner agencies frequently participating in concurrent resource fairs. Survey feedback from families and professionals consistently reflected high satisfaction, strong relevance to daily practice and increased awareness of referral pathways and local genetic resources. Families also visited on-site vendors who shared information about community-based resources, telehealth options and referral programs.

National DNA Day

National DNA Day commemorates the successful completion of the Human Genome Project in 2003. This year was the 22nd anniversary of the project's completion.

The Department celebrated National DNA Day by arranging in-person tours of 11 research laboratories, providing students with first-hand exposure to genetics research and scientific career pathways. Twenty-five visitors were made up of high school students from Paetow High School and college students from the University of Houston–Downtown, Houston Community College, Prairie View A&M University and San Jacinto South Community College.

Rare Disease Day

Rare Disease Day 2025 included two community-focused events designed to raise awareness, connect families with resources and strengthen partnerships between clinical programs and patient advocacy organizations.

The first event, held on February 27 at the Texas Children's Hospital Auxiliary Bridge, welcomed more than 100 attendees and featured participation from 15 clinical laboratories and patient support organizations. These partners provided educational materials, research information and direct family support resources.

A second Rare Disease Community Event was held on April 12 at The Health Museum, drawing more than 150 families for an afternoon of educational presentations and interactive engagement. Featured speakers included Dr. Brendan Lee, professor and chair of molecular and human genetics, and Darby Gavin, M.S., director of education at the National Organization for Rare Disorders, who shared insights on rare disease research, advocacy and care navigation. The event also hosted 27 community vendors and organizations, offering families access to local and national support services while encouraging exploration of museum exhibits within a family-friendly learning environment.

Collectively, these events generated more than 250 direct community interactions and reinforced the department's commitment to visibility, education and meaningful support for individuals and families affected by rare diseases.

Genetic Counselors and Genetic Counseling Students at a Rare Disease Day event at the The Health Museum.

The fourth annual Biomedical Researcher Faculty Summit Research scholars.

Undiagnosed Diseases Network (UDN) Outreach

In 2025, the Office of Community Engagement conducted 24 UDN-aligned outreach activities across West and South Texas, integrating Undiagnosed Diseases Network (UDN) awareness into professional trainings, community conferences and partner-led events.

These efforts generated more than 2,000 direct family and professional interactions, connecting individuals experiencing unresolved or complex medical conditions with telehealth genetic evaluation options, research participation opportunities and clinical referral pathways.

UDN outreach efforts were supported through collaboration with regional partner organizations, including Paso del Norte Children's Development Center and the Help Me Grow coalition, as well as education service centers, nonprofit family-support agencies, healthcare providers, and early childhood intervention networks. Programming frequently occurred in communities with limited access to in-person genetic specialists, helping to reduce geographic and informational barriers while strengthening long-term regional partnerships.

Feedback from families consistently emphasized the importance of access to remote evaluation options and research participation resources, reinforcing the value of sustained UDN visibility in underserved areas.

All of Us Evenings with Genetics Research Program

NIH's *All of Us* Research Program is an ambitious effort to gather biomedical data from 1 million or more individuals living in the U.S., especially those who are traditionally excluded from biomedical research studies, to support scientific discoveries and advance precision medicine.

The *All of Us Evenings with Genetics* Research Program held its fourth annual Biomedical Researcher Faculty Summit in Houston. Dr. Erez Lieberman Aiden, professor of molecular and Human Genetics at Baylor and adjunct professor of Computer Science at Rice University, delivered the keynote address.

The purpose of the summit is to train early career faculty and senior postdoctoral researchers on the NIH's *All of Us* Researcher Workbench. During the summit, participants form research teams around a common research question, receive data science training on how to set up cohorts on the workbench and professional development. Two types of mentors are present: invited distinguished faculty for one-on-one sessions, and Baylor research faculty for guidance with projects.

Faculty

Faculty Awards and Recognitions

Dr. Graham Erwin receives NIH New Innovator Award

Dr. Graham Erwin, assistant professor of molecular and human genetics, received the prestigious NIH Director's New Innovator Award in 2025. This highly competitive award from the NIH Common Fund supports exceptional early-career investigators pursuing bold, high-risk research.

Erwin was awarded $2.4 million to develop a new class of small, cell-permeable molecules designed to permanently correct disease-causing DNA repeat expansions. Building on his prior work in Friedreich ataxia, his research aims to create an alternative to viral-based genome editing technologies by using precisely engineered small molecules that can safely and durably restore normal gene function, with broad potential applications across numerous genetic disorders


Dr. Graham Erwin

Li and Zhang Receive 2025 Michael E. DeBakey Excellence in Research Awards

Dr. Hongjie Li and Dr. Lilei Zhang were named recipients of the 2025 Michael E. DeBakey Excellence in Research Award. The award recognizes Baylor College of Medicine faculty for outstanding, high-impact scientific contributions published over the past three years and is supported by the DeBakey Medical Foundation.

Li, an associate professor of molecular and human genetics, was recognized for his pioneering work in single-cell sequencing and multi-omics approaches to study aging, including the development of comprehensive cell atlases that have transformed understanding of cellular aging and age-related disease. Li was also awarded the Nancy Chang, Ph.D. Award for Research Excellence and a Hevolution/AFAR New Investigator Award in Aging Biology and Geroscience Research.

Zhang, an associate professor of molecular and human genetics, integrative physiology, and internal medicine, was honored for her research on circadian gene regulation in the heart and rare inherited cardiac diseases, advancing translational approaches that bridge fundamental discovery with clinical applications in cardiovascular medicine. She was also elected to the American Society of Clinical Investigation this year.

Cheung receives ACGA Lifetime Achievement Award

Dr. Sau Wai Cheung, professor of molecular and human genetics, received the 2025 Association of Chinese Geneticists in America (ACGA) Lifetime Achievement Award in recognition of her four decades of transformative contributions to clinical genetics.


Dr. Sau Wai Cheung

As director of the Cytogenetics Laboratory at Baylor College of Medicine, Cheung led the development and clinical adoption of chromosomal microarray analysis, establishing it as a first-tier diagnostic test and fundamentally reshaping genetic testing worldwide. Her work has advanced

2025 Michael E. Debakey Research Award Winners: Dr. Anthony Maresso (not pictured), Dr. Alastair Thompson, Dr. Na Li, Dr. Anna Mandalakas, Dr. Lilei Zhang, and Dr. Hongjie Li with President Dr. Paul Klotman and Senior Vice President and Dean of Research, Dr. Carolyn Smith

the understanding of microdeletion and microduplication syndromes, influenced global clinical practice, and trained generations of leaders in medical genetics.

Jafar-Nejad receives Harrington Scholar-Innovator Award

Dr. Hamed Jafar-Nejad, professor of molecular and human genetics, received the 2025 Harrington Scholar-Innovator Award from the Harrington Discovery Institute.

The award recognizes physician-scientists whose discoveries show strong potential for translation into new therapies. Jafar-Nejad was selected for his innovative work advancing gene-based approaches to treat rare genetic disorders, including Alagille syndrome, with the goal of accelerating novel treatments toward clinical impact.

Dr. Hamed Jafar-Nejad

Yamamoto receives Genetics Society of America Early Career Medal

Dr. Shinya Yamamoto, associate professor of molecular and human genetics, received the 2025 Genetics Society of America Early Career Medal in recognition of his outstanding contributions to the field of genetics. The award honors exceptional early-career scientists whose research has had a significant impact on advancing genetic discovery. Yamamoto is recognized for pioneering the use of *Drosophila* models to identify and functionally characterize human

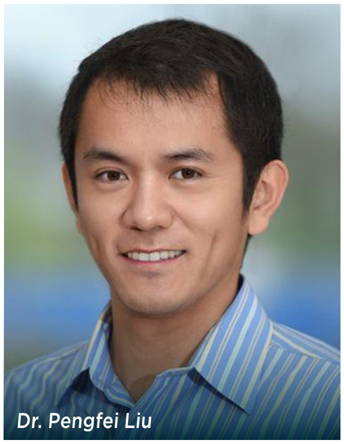
Dr. Shinya Yamamoto

disease genes, accelerating diagnosis and improving understanding of rare genetic disorders.

In addition to this honor, Yamamoto was named to the 2025 Selection of Japan's National Institute of Science and Technology Policy (NISTEP) and received the Texas Children's Mark A. Wallace Endowment Award. Together, these recognitions highlight the international impact and translational significance of his research program.

Dr. Anthony Zoghbi also received the Texas Children's Mark A. Wallace Endowment Award this year

Liu receives Baylor Young Alumnus Award


Dr. Pengfei Liu

Dr. Pengfei Liu, associate professor of molecular and human genetics, received the Baylor College of Medicine Young Alumnus Award in recognition of his exceptional contributions to medical genetics early in his career.

An internationally recognized leader in genomic medicine, Liu's work has advanced the understanding of complex genetic variation and improved the diagnosis of rare genetic disorders, helping to solve some of the most challenging cases in human health. Liu also received the Oxford-Harrington Rare Disease Centre Rare Disease Scholar Award.

Graduate School of Biomedical Sciences Awards

Best Course in Development, Disease Models & Therapeutics
Evolutional Conservation of Developmental Mechanisms" Directors: Dr. Benjamin Arenkiel & Dr. Blair Benham-Pyle

Genetics & Genomics Outstanding Lecturer
Dr. Jason Heaney.

Best Course in Genetics and Genomics
Method & Logic in Genetics & Genomics Directors: Dr. Ross Poché & Dr. Shinya Yamamoto

Best Course in Quantitative & Computational Biosciences
"Computational Project Design & Grant Writing" Director: Dr. Chad Shaw

Quantitative & Computational Biosciences Program Outstanding Lecturer
Dr. Chad Shaw

MS in Biomedical Sciences Program Outstanding Lecturer
Dr. Herman Dierick

Genetics & Genomics Outstanding Teaching Assistant
Melanie Mew

Department Awards

Excellent Attending
Dr. Sanmati Rao Cuddapah (Metabolic)
Dr. Chaya Murali (Pediatric)
Dr. Mir Reza Bekheirnia (Adult)
Dr. April Adams (Reproductive Genetics)

Excellent Metabolic Dietician
Brandy Rawls-Castillo

Excellent Research Mentor
Dr. Fernando Scaglia

Excellent Genetics Educator
Dr. Lindsay Burrage

Excellent Genetics Counselor
Emily Magness Bland, M.S., C.G.C. (Pediatric)
Darwin Argueta, M.S., C.G.C. (Adult)
Sandra Darilek, M.S., C.G.C. (Prenatal)

Baylor Genetics Service Award
Dr. Xiaonan Zhao

Rolanette and Berdon Lawrence Awards
Dr. Lindsay Burrage (Faculty)
English Laserna (Trainee)
Sherri Weaver (Administration)

Kenneth Scott Graduate Mentor Award
Dr. Shinya Yamamoto

Shan and Lee-Jun Wong Fellowship
Dr. Shenglan Li

Faculty Appointments

In 2025, the Department of Molecular and Human Genetics at Baylor College of Medicine welcomed Dr. Xiao Li as a new tenure-track primary research faculty member. An assistant investigator at the Texas Heart Institute at Baylor, Dr. Li leads a multidisciplinary research program focused on genomics, cardiac biology and innate immunity.


Dr. Xiao Li

Dr. Li earned his B.S. from East China Normal University, his Ph.D. from the University of Iowa, and completed postdoctoral training at the University of California, San Diego. His lab studies cardiac remodeling, immune-driven regenerative repair, and 3D genomics in heart regeneration, using single-cell and multi-omics approaches, machine learning and genome architecture modeling. He is internationally recognized for his work on RNA–chromatin interactions, including development of GRID-seq, and has published in journals such as Nature Biotechnology, Immunity, Circulation, and Nature Communications.

Clinical Division

Julie Mathew, M.S.N., R.N., FNP-C
Assistant Professor

Diagnostic Division

Veronica Ortiz, Ph.D.
Assistant Professor, non-tenure-track

Amir Hossein Saeidian, Ph.D.
Assistant Professor, non-tenure-track

Promotions

Distinguished Emeritus Professor, tenured
Richard Lewis, Ph.D.

Associate Professor, tenured
Mir Reza Bekheirnia, M.D.
Hongjie Li, Ph.D.
Fritz Sedlazeck, Ph.D.

Professor, non-tenured
Qin Sun, Ph.D.

Associate Professor, non-tenured
Sanmati Cuddapah, M.D.
Tiemo Klisch, Ph.D.
Yue Wang Ph.D.

Assistant Professor, non-tenure-track
Amanda Gerard, M.S., C.G.C.
Rachel Franciskovich, M.S., C.G.C.
Liz Mizerik, M.S., C.G.C.

Instructor, non-tenure-track
Ashley Spector, M.S., C.G.C

In Memoriam

While this report was in production, we were deeply saddened by the unexpected passing of Dr. Mir Reza Bekheirnia, a cherished member of our genetics community. An associate professor in our clinical division and a leader in renal genetics, Reza was known for his compassion and excellence as a physician and colleague.

A former trainee who joined our faculty, he practiced adult and pediatric general genetics across Harris Health System, the Michael E. DeBakey VA Medical Center and Texas Children's Hospital. He also served as a vital link to the Department of Pediatrics at Baylor, leading the renal genetics clinic within the Division of Nephrology.

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