Baylor College of Medicine

DEPARTMENT OF MOLECULAR & HUMAN GENETICS

10 Year Review

2014-2024

A Decade of Innovation, Service, and Achievement

Welcome

In place of our 2024 annual report, I am delighted to introduce you to our 2014-2024 ten-year review of the Department of Molecular and Human Genetics. In 2014, we restated the mission of the Department: to transform medicine with the science and practice of genetics and genomics. To achieve this, we pursued multiple strategic initiatives in our research, clinical and diagnostic laboratory divisions.

In the area of research, the Department continues to rank first among all U.S. genetics medical school departments in total awarded funding and number of grants from the National Institutes of Health, now for fourteen consecutive years. Our research program continued its long-term excellence in genomics, gene discovery, and model organism studies. During these past ten years, we established a division of clinical research and invested in team science by leading many NIH consortia including: the All of Us research program; Genomic Research to Elucidate the Genetics of Rare disease (GREGoR); the Knockout Mouse Phenotyping (KOMP) Program; the Center for Precision Medicine Models (CPMM); multiple Rare Diseases Clinical Research Network (RDCRN) consortia; and the Undiagnosed Diseases Network (UDN). We also implemented genomic medicine in underserved, rural and urban communities in Texas with NIH-supported projects TEXOME, GIVE and MAGNET. At our core, outstanding basic and translational science continues to empower genomic medicine.

No better has this been evident than in the diagnostic testing arena with Baylor Genetics, our joint venture with H.U. Group Holdings, Inc., that was established ten years ago. Baylor Genetics is thriving, having tripled its revenue and staffing over the past decade. This academic-commercial hybrid diagnostic laboratory has supported the academic mission of the Department while expanding the impact of its diagnostic genetic testing worldwide.

We continued to invest in recruiting and retaining the best faculty and trainees. We established a thriving masters program in genetic counseling and a clinical division of genetic counseling, while expanding our post-graduate clinical genetics and diagnostic laboratory training programs.

Through much uncertainty over the past decade, our faculty continued to excel and to deliver on our clinical, training, and research missions both at home and abroad through our global partnerships. Our unique integration of all mission areas within a single department imbued by a culture of intellectual generosity and collaboration sets us apart from all other genetics programs in the world.

I am privileged to be a part of this exciting and vital effort and excited to discover what the next decade will hold for us in the exciting world of genetics and genomics!

With warm regards,

Brendan Lee, M.D., Ph.D., Robert and Janice McNair Endowed Chair

Professor and Chairman, Department of Molecular and Human Genetics

Contents

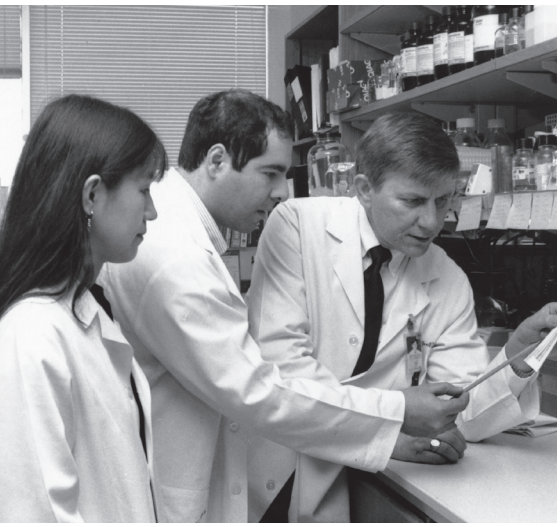
*Microsoft Co-Pilot assisted with the writing in this report.

Department By the Numbers in 2024

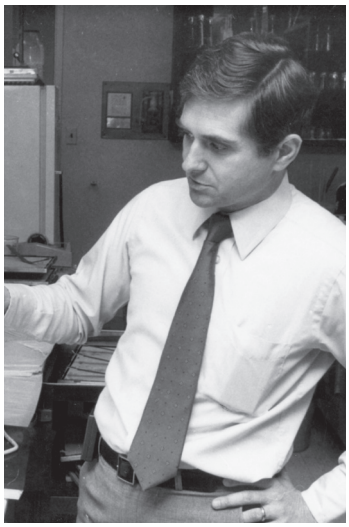
- 75 Primary Research Faculty
- 42 Clinical Faculty
- 23 Diagnostic Laboratory Faculty
- 67 Research Postdoctoral Fellows
- 211 Administrative, Clinical, and Research Staff
- 18 Medical Residents
- 5 Clinical Laboratory Fellows
- 2 Clinical Postdoctoral Fellows
- 118 Graduate Students

Mouse Cortex and Hippocampus. Image from the lab of Dr. Hsiao Tuan Chao. Sagittal section of a 6-weeks old mouse brain showing cortical and hippocampal structures. The sample is immunostained with antibodies to NeuN (blue), parvalbumin (green), and FoxP2 (magenta)

Our History







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 NIH. 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 at Baylor, 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.

Baylor Genetics

Baylor Genetics Celebrates a Decade of Innovation in Genetic Testing

Baylor Genetics, the clinical diagnostic laboratory joint venture between Baylor College of Medicine (BCM) and H.U. Group Holdings, marked its 10-year anniversary in 2025. The company offers a full spectrum of genetic tests and lab services spanning a wide variety of healthcare specialties, including neonatal and pediatric critical care, rare diseases, reproductive health, hereditary cancer and metabolic conditions. BCM and Baylor Genetics leaders look back at the company's decade of innovation.

Founding a New Venture

Baylor Genetics was formed on the foundation of the state-of-the-art genetic testing established in BCM's Department of Molecular and Human Genetics Medical Genetics Laboratory (MGL) and Human Genome Sequencing Center. In 2004, the department developed and launched a chromosomal microarray, a genetic test that analyzes DNA segments in chromosomes to identify abnormalities. Four years later, the department was the first to offer prenatal CMA testing. In the years that followed, BCM researchers led the way in other genetic testing offerings, including whole exome sequencing, cancer exome, mitochondrial genome sequencing and nuclear gene panel.

Dr. Paul Klotman, president, CEO and executive dean of the College, led the vision to leverage the BCM's genetics expertise to work with a leading clinical lab company to form a joint venture. Dr. Brendan Lee, professor and chair of the Department of Molecular and Human Genetics at BCM, led efforts on behalf of the College.

"It was important to maintain and preserve Baylor College of Medicine's academic research mission within this new commercial entity," said Lee, Robert and Janice McNair Endowed Chair in Molecular and Human Genetics at BCM and scientific advisory board member at Baylor Genetics. "We found the right partner in Miraca Holdings, now H.U. Group Holdings, because they appreciated and supported our academic mission and values." The joint venture formally launched in February 2015.

Nahki Kitamura, managing executive officer and CFO of H.U. Group Holdings, led the effort on behalf of his company. "Through numerous negotiations regarding the joint venture agreement with Dr. Klotman, Dr. Lee, and Mr. Robert Corrigan (senior vice president and general counsel at BCM), we were able to start the joint venture, Baylor Genetics, with a deep understanding of each other, which I believe strengthened the trust between both parties," Kitamura said. "Additionally, during the initial phase of the joint venture, I have good memories of going to see a rodeo with BCM members during my six-month stay in Houston. Understanding not only each other's perspectives but also the cultural differences, as well as the personal trust built with Dr. Lee over 10 years, has contributed to the growth and success of Baylor Genetics, a unique joint venture between U.S. academia and a Japanese corporation."

Persevering through challenges

The ensuing years presented various challenges as Baylor Genetics navigated a highly competitive landscape for diagnostic laboratories. A commitment to its core values as an academic/commercial hybrid helped pave the way to success.

"Long-term investments in developing new technologies like whole-genome sequencing and growing partnerships in the reproductive healthcare space have helped Baylor Genetics grow and succeed," Lee said.

"In a market that has seen dramatic change, Baylor Genetics has thrived for a decade. I'm incredibly proud of the dedicated team behind our growth and grateful for the trust of our expanding roster of customers and partners," said Kengo Takishima, president and chief executive officer of Baylor Genetics. "As pioneers in precision diagnostics, we continue to push the boundaries of science and innovation with an unwavering commitment to our mission of delivering genetic answers that matter."

Leading genetic testing and research

Today, Baylor Genetics' whole-genome sequencing (WGS) and whole-exome sequencing (WES) tests are among the fastest and most accurate precision diagnostic tools available. In 2017, the company introduced a non-invasive prenatal sequencing panel for *de novo* dominant disorders, and in 2024, it introduced targeted RNA sequencing, which advances the detection of certain qualified variants. Last year, Baylor Genetics became the first to offer whole-transcriptomic RNA sequencing through the Medical Genetics Multi-Omics Laboratory, an academic clinical genetic testing laboratory at BCM in partnership with Baylor Genetics.

The company continues to embody the original academic research mission. All lab directors hold faculty appointments at BCM, and the lab supports BCM's American Board of Medical Genetics and Genomics-accredited clinical laboratory fellowship training programs. Baylor Genetics labs also support BCM's research studies. For example, Baylor Genetics together with BCM is the lead genetic sequencing partner for the National Institutes of Health's Undiagnosed Diseases Network (UDN), with approximately 30% of the 2,470 UDN participants affirmatively diagnosed using WGS and WES testing. The company also is increasing access to genetic testing and reducing healthcare disparities through BCM's NIH-funded genomic medicine initiatives like the Texome Project, which provides genomic testing for medically underserved populations in Texas, and Project GIVE, which is addressing disparities in access to genetic services for

Current and former staff of Baylor Genetics gathered at a formal event to mark the tenth anniversary.

children with rare diseases along the Texas-Mexico border.

New Medical Genetics Multi-Omics Lab translates research into clinical diagnostics

We have partnered with Baylor Genetics to launch an academic clinical genetic testing laboratory focused on integrating multi-omics research to enhance clinical diagnostics in medical genetics. The lab supports the academic mission of Baylor College of Medicine and provides CLIA/CAP approved clinical services. The laboratory will leverage the power of cutting-edge multiomic technologies to deliver accurate and personalized insights into genetic diagnostics, driving advancements in patient care and therapeutic innovation. Here is the current status and what is on the horizon for the laboratory:

- Whole transcriptomic analyses (RNA sequencing)
- Long-read sequencing (DNA and RNA) to detect structural variation (SV) and phasing
- Implementation of T2T reference genome
- Detection of low-level mosaicism in DNA
- Epigenetics – Global DNA methylation, etc.
- Higher order DNA interaction – HiC WGS, e.g., mutations that affect promoter/enhancer/repressor interactions
- Finding the missing heritability in genetic diagnostics

Baylor Genetics Milestones

2014
- Entered a unique academic/commercial joint venture
- First to offer metabolomics

2015
- First to offer prenatal trio exome
- Introduced universal carrier screening

2017
- Launched PreSeek, the first clinical noninvasive prenatal sequencing screen
- Launched ClariFind, a next-gen somatic tumor sequencing test

2019
- Launched clinical whole genome sequencing

2020
- Launched its official COVID-19 test

2022
- Introduced rapid whole genome sequencing

2024
- Introduced targeted RNA sequencing
- Launched Whole Transcriptomics for the Undiagnosed Diseases Network

Consultagene

Consultagene is a personalized web and app-based platform that integrates research and clinical care through genetic counseling, peer-to-peer consultation, patient and provider education and diagnostic interpretation of clinical genomic data. It operates through a three-party interaction system involving the referrer, client and provider, and includes modules for client engagement, video education, health history intake and telegenetic counseling.

Consultagene: A Journey of Innovation and Expansion in Patient Access

2016

Consultagene introduced at the American Society of Human Genetics meeting.

2018

Educational journeys launched in Texas Childrens Hospital genetics clinics.

2019

Consultagene Telegenetic Counseling Clinic for reproductive health launched.

2020

Expansion of clinical services to neurology and cancer.

2021

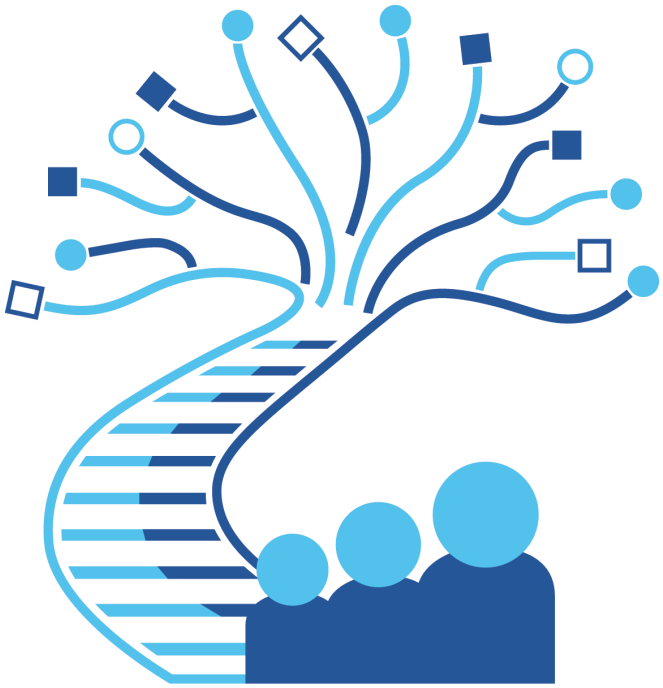
Over 1,800 clients served.

2022

Expansion to other states, upgrades to allow patient self referral, and launch of Consultagene for the BCM Undiagnosed Diseases Center.

2023

Launch of Consultagene as part of NIH-funded Project GIVE and Project MAGNET to accelerate genetic diagnoses in underserved areas in Texas.



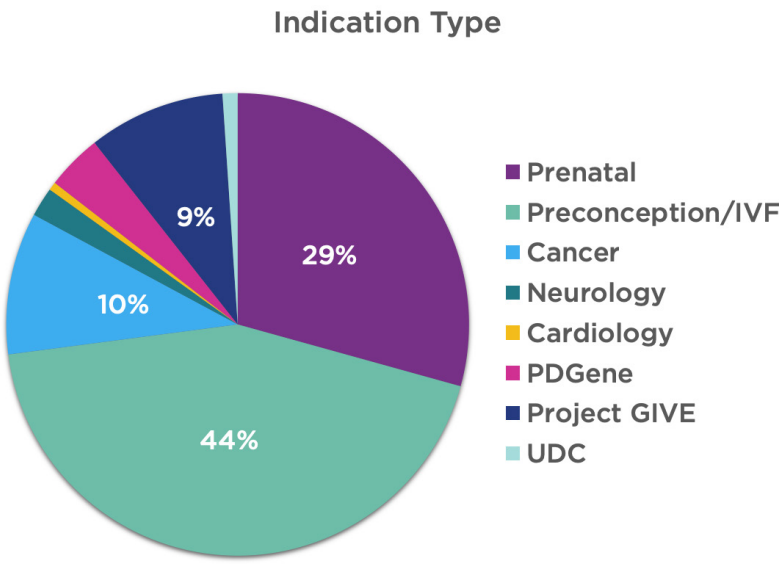
Consultagene in Research

Undiagnosed Diseases Center

Established in 2022, the UDC incorporates the work of Baylor's Undiagnosed Diseases Network clinical site, DNA sequencing core, and model organisms screening center. It provides clinical services, genetic testing, and analysis for patients with rare, undiagnosed conditions. The center also offers services through the Consultagene platform, which provides educational resources and genetics services.

Project GIVE (Genetic Inclusion by Virtual Evaluation)

Funded by the National Center for Advancing Translational Sciences, this project aims to reduce time-to-diagnosis for children with suspected genetic disorders in the Rio Grande Valley. It uses the Consultagene platform for virtual clinical evaluation and genome sequencing. The project has recruited over 200 children in the study, with a diagnostic yield of around 40%.



PDGENE study

The PD GENEration study, funded by the Parkinson's Foundation, offers genetic testing and counseling to people with Parkinson's disease. Baylor is a participating clinical site and the sole location for the PD GENEration Video study, which compares pre-recorded video via Consultagene to in-person counseling for returning negative results. Findings suggest video disclosure is a viable option, supporting scalable genetic testing for future Parkinson's trials.

Genetic Counseling

The Division of Genetic Counseling has made significant strides in multiple key areas over the past 10 years, expanding our ability to provide high-quality genetic counseling services to our patients and families while furthering the Department's clinical, education and research missions.

Billing for Services

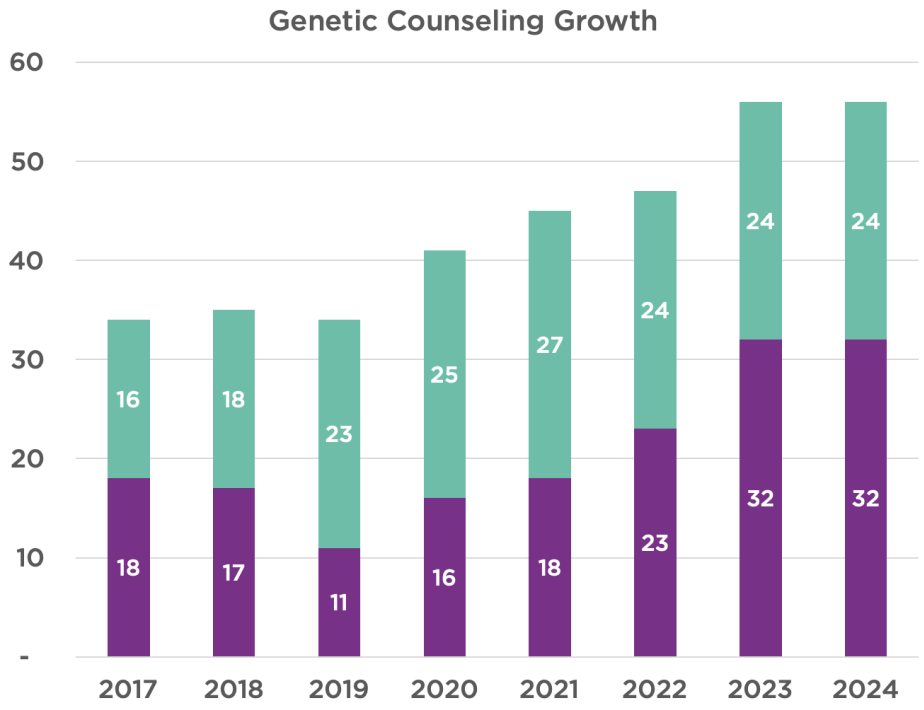
The division has successfully implemented structured billing practices for genetic counseling services at Texas Children's Hospital, ensuring proper reimbursement and financial sustainability. This initiative has streamlined the process, improved revenue generation and enhanced overall operational efficiency, while recognizing genetic counselors for the services they provide.

Development of an Academic RVU

We developed and integrated an Academic Relative Value Unit (RVU) system, enabling a more effective way to measure and recognize the academic and clinical contributions of our genetic counselors. The GCVU (Genetic Counselor Value Unit) is the first of its kind in the field of genetic counseling and takes into account clinical, administrative, education, scholarship, research and laboratory skills components. The GCVU has supported better tracking of workload, facilitated performance assessments and fostered a clearer pathway for career advancement. In addition, this tool has been piloted at other academic institutions throughout the country where our research has shown that there is significant need and utility for this type of measure in the field.

Workforce Growth

The division has experienced a significant expansion of its workforce, attracting and retaining talented genetic counselors. We have over 60 genetics counselors, which is more than double from the number of genetic counselors from 10 years ago. This growth has not only helped meet the increasing demand for genetic counseling services in multiple departments and divisions such as cardiology, neurology, hematology, oncology, allergy and immunology, pulmonology, endocrinology, but also fostered a collaborative and dynamic



work environment that contributes to the ongoing development of the field.

None of the accomplishments of the Division would be possible without the genetic counselors themselves who serve in many different capacities and continue to demonstrate their expansive leadership abilities, their dedication to excellence in teaching and their commitment to improving the quality of genetic counseling services.

Addressing a Growing Demand for Genetic Counselors

The genetic counseling profession has experienced significant growth over the years. Since 2006, the number of genetic counselors has more than doubled, and the U.S. Bureau of Labor Statistics projects a growth rate of over 25 percent for genetic counseling positions through 2029. This surge in growth underscores an urgent need to train more professionals to meet the rising demand for genetic counseling services.

To meet this need, a 21-month Master of Science program was launched within the School of Health Professions, supported both financially and logistically by the Department of Molecular and Human Genetics and its Genetic Counseling Division. In 2018, the program earned Recognized New Program accreditation status from the Accreditation Council for Genetic Counseling (ACGC) and welcomed its inaugural cohort of eight students.

Since its inception, the program has graduated 43 students, 16 graduates of the program have secured jobs at Baylor or related affiliates and more than a third of the graduates have had their thesis projects published in peer reviewed journals.

In 2021, a postdoctoral research fellowship was established by the department that allowed for a second-year student to receive tuition coverage for their second year and a 12-month salaried research position at Baylor.

Genetic Counseling Program Milestones

2018

Received Recognized New Program accreditation status from the ACGC

2020

Graduated its first class

2021

Blake Vuocolo (pictured here with Ryan German) and Ray Belanger Deloge were the first recipients of the postgraduate research fellowship

2024

Increased admissions to 10 students each year

The BCM-CUHK Joint Center for Medical Genetics

Established in 2016, the BCM-CUHK Joint Center for Medical Genetics is a collaborative partnership between Baylor College of Medicine (BCM) and the Chinese University of Hong Kong (CUHK). This center represents a significant milestone in advancing professional training in clinical genetics and genomics and fostering collaborative research globally.

Professional Training

The center offers professional training in clinical genetics, including the development of the first Master of Science in Medical Genetics Programme in Hong Kong.

Accreditation

The center has been working with the ACGME International Office to provide accreditation to CUHK's Faculty of Medicine and the Genetics Program. This includes assessments and the potential for board certification through ABMS-I.

Collaborative Research

The center is involved in various collaborative research projects, such as evaluating metabolomics signatures in pregnant women with preeclampsia and conducting metabolomics on inborn errors of metabolism (IEMs).

Joint Symposium

The center organizes an annual conference to facilitate collaboration and knowledge exchange. Initially designated as the BCM-CUHK Joint Symposium in Clinical Genetics, the event has seen the following locations:

- **2017:** Hong Kong with CUHK
- **2018:** Hong Kong with CUHK
- **2019:** Beijing with CUHK and Peking Union Medical College Hospital
- **2023:** Singapore with CUHK and National University of Singapore
- **2024:** Kuala Lampur, Malaysia with CUHK and Universiti Malaya

Invited speakers and attendees of the 4th BCM-CUHK-NUS Joint Symposium in Clinical Genetics.

Education & Training

Graduate Program Transition

The Genetics and Genomics Graduate Program at Baylor College of Medicine transitioned to be under the Graduate School of Biomedical Sciences in 2015.

The transition was part of a broader effort to integrate and centralize the various research and educational initiatives within the College. This move aimed to enhance collaboration, streamline operations and leverage the strengths of the different departments to create a more cohesive and effective educational environment. The transition had several significant impacts:

- **Enhanced Collaboration:** The integration facilitated better collaboration among various departments, leading to a more cohesive and effective educational environment.
- **Streamlined Operations:** The centralized organization helped streamline operations, making it easier to manage and coordinate research and educational initiatives.
- **Increased Visibility:** The transition placed Baylor on the map as a genetics powerhouse, attracting top researchers and students to the program.
- **Improved Resources:** The program benefited from the resources and support of the Graduate School of Biomedical Sciences, enhancing its ability to conduct cutting-edge research and provide high-quality education.

The transition has led to a notable increase in student enrollment. The program offers one of the most competitive annual stipends in the country, with full coverage of tuition and health benefits.

Scholarships and Publications

In the past decade, Genetics and Genomics graduates received

34 scholarships

Awarded by:
National Institutes of Health, American Heart Association, National Institute of Neurological Disorders and Stroke, American Cancer Society, the Howard Hughes Medical Institute and other institutions

Genetics and Genomics graduate students also contributed to

585+ Publications

including

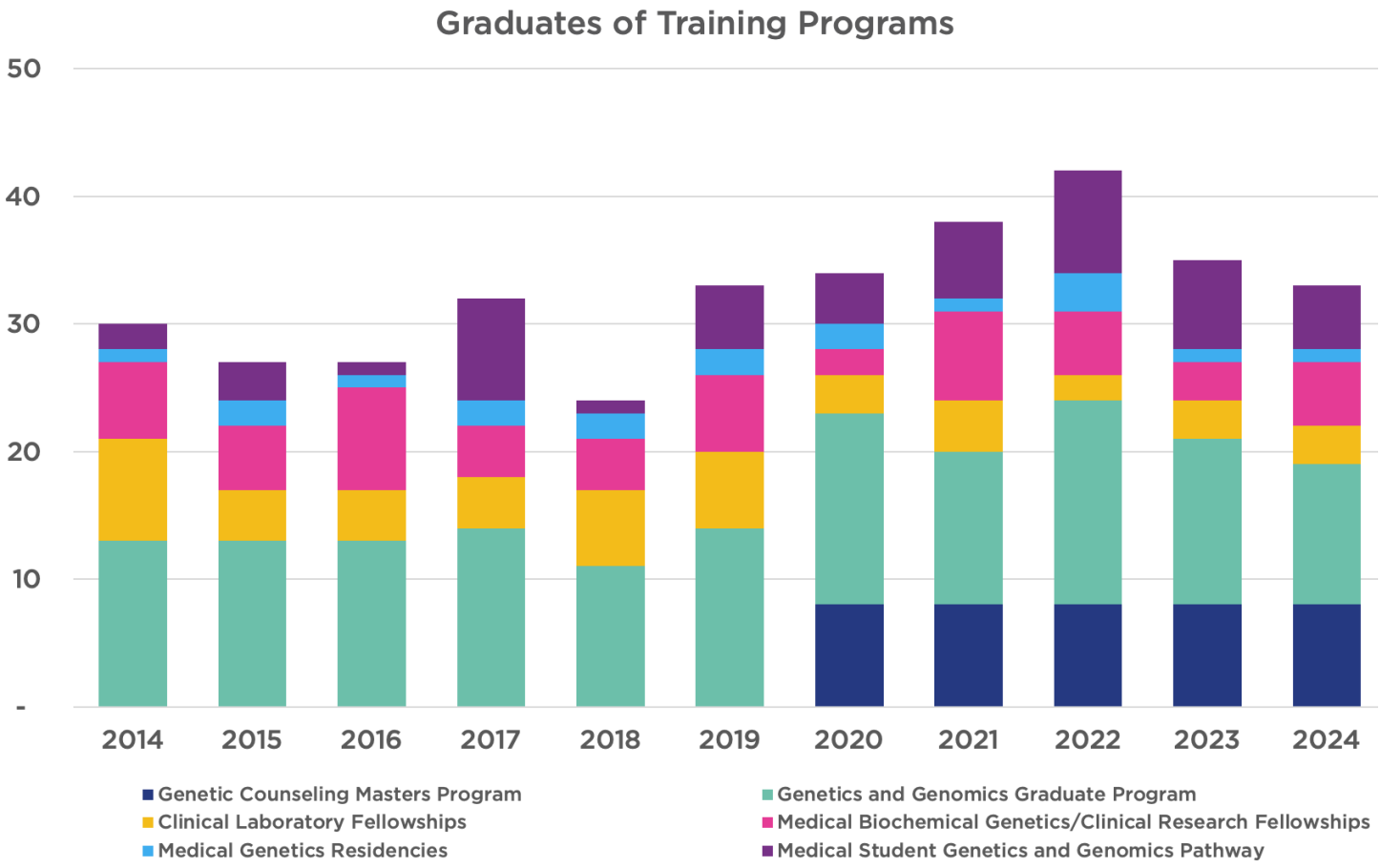
134 First-Author Papers

New Pathways in Residency and Clinical Fellowships

Over the past decade, Baylor College of Medicine has strategically expanded its genetics training programs, establishing the Internal Medicine-Genetics Residency Program in 2016 and the Maternal-Fetal Medicine and Medical Genetics and Genomics Clinical Fellowship in 2017—both designed to advance clinical expertise and research in adult and maternal-fetal medical genetics.

The **Internal Medicine-Genetics Residency Program** at Baylor is a new five-year initiative aimed at training medical students in adult medical genetics, focusing on cancer, connective tissue disease and cardiovascular disease. As one of only seven such programs in the U.S., it seeks to enhance patient care and develop new clinical techniques. Ultimately, it aims to produce physician-scientists who will advance adult medical genetics globally.

The **Maternal-Fetal Medicine and Medical Genetics and Genomics Clinical Fellowship** at Baylor is a fully accredited program that combines 18 months of clinical training in both maternal-fetal medicine and medical genetics, along with 12 months of research training. This comprehensive program aims to equip fellows with the necessary skills to manage complex genetic and maternal-fetal conditions.



Department's Lectureships

The Department's lectureships foster academic enrichment by featuring leading experts and emerging voices who share research, spark dialogue and inspire innovation across medicine and science.


2018 Frank Greenberg Lecturer Brenda Finucane (seated, 2nd from left) pictured with the Greenberg Family and Department Faculty

Frank Greenberg Memorial Lectureship

Established in memory of Dr. Frank Greenberg, a faculty member of molecular and human genetics and pediatrics at Baylor, who helped advance the clinical understanding of several chromosomal abnormalities, including Williams syndrome.

- **2015:** Dr. Kym Boycott
- **2016:** Dr. Stefan Mundlos
- **2017:** Brenda Finucane, M.S.
- **2018:** Dr. Cynthia Morton
- **2019:** Dr. James Lupski


2023 Oshman Efron Lecturer Dr. Leslie Vosshall (4th from left) pictured with the Oshman Efron Family and Department Faculty

Jeanette Oshman Efron Lecture in Molecular Genetics

A distinguished lecture series established at Baylor in 1989 to honor Jeanette Oshman Efron's passion and commitment to the advancement of medical education and biomedical research.

- **2015:** Dr. Susan Lindquist
- **2017:** Dr. Chris Walsh
- **2018:** Dr. George Church
- **2022:** Dr. Ardem Patapoutian
- **2023:** Dr. Leslie B. Vosshall


2022 Fernbach Lecturer Taylor Harris (Right)


2019 Beaudet Lecturer Dr. Hugo Bellen (2nd from left)

Arthur L. Beaudet Lecture for Outstanding Mentorship

This annual lecture was established in 2019 to recognize exemplary mentorship at Baylor College of Medicine.

- **2021:** Dr. Arthur L. Beaudet
- **2022:** Dr. Hugo Bellen
- **2023:** Dr. Malcolm K. Brenner
- **2023:** Dr. Mary Estes
- **2024:** Dr. Jeffrey M. Rosen

Fernbach Lecture for Humanism in Genetics

The lecture honors Susan Fernbach, who retired in July 2022 after nearly 40 years of service.

- **2022:** Taylor Harris, author of "This Boy We Made"

Community Engagement

The Department of Molecular and Human Genetics at Baylor College of Medicine has made significant contributions to community engagement and education during the past decade through various initiatives.

- **Evenings with Genetics**, an ongoing seminar series for the general public hosted by the Department of Molecular and Human Genetics at Baylor College of Medicine and Texas Children's Hospital featured genetics topics and expert speakers and attracted hundreds of attendees each year.
- **The Race and Genetics - Perspectives on Precision Medicine seminar series**, which began in 2021 honored Black History Month by addressing disparities in genetics research and educating the public on the history of race and genetics. It emphasized that the construct of race is independent of genetics and ancestry.
- The Department has held annual **Rare Disease Day events** in collaboration with organizations like the National Organization for Rare Disorders and Texas Rare Action Network, featuring booths and expert sessions.
- Collaborative efforts with local organizations have led to **genetic seminars and resource fairs in underserved communities across the state**. The Department along with the UT Texas Center for Disability Studies and Early Childhood Intervention Services also organizes **genetic webinars for healthcare providers** in Texas. These initiatives are supported with funding from the Department of State Health Services.
- Efforts to **recruit students to genetics** have been a focus, with events at various conferences and the establishment of the Medical Genetics Visiting Students Program in 2019.
- The department celebrates **National DNA Day** facilitating tours for students at research laboratories.
- **From Stress to Strength Program** is based on the Positive Adult Development curriculum and aims to help parents of individuals with intellectual and developmental disabilities (IDD) manage stress.
- The department has presented at local health fairs and continues efforts to **introduce students to various career pathways in medical genetics** with the "A Whitecoat and Genes" town hall and the "Careers in Genetics and Genomics" series.

Discover the Impact

The Office through its Evenings with Genetics seminar series and statewide outreach acquired

over $150,000

in annual funding from the UT-Austin/ Texas Department of State Health Services, and has built two robust YouTube Playlists containing...

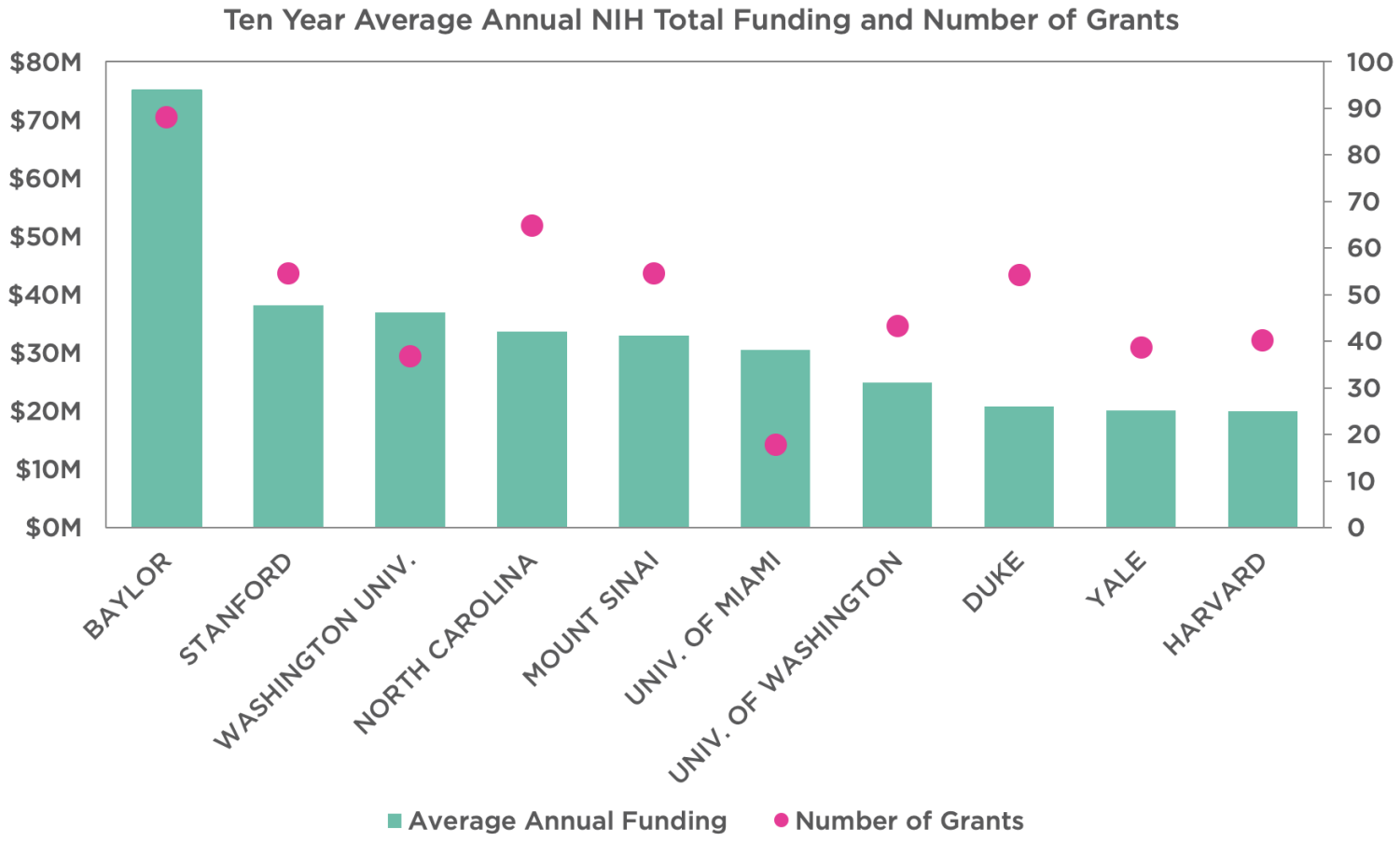
60 educational videos

receiving

80,000 views

NIH Rankings

1 ranked U.S. Genetics Department in NIH awarded grants and total funding for past 14 years



Other Awards/Grants

The Department is proud to receive generous funding from many agencies and private 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 Assocation
Autism Speaks

Clinical Research Division

The Clinical Research Division within the Department of Molecular and Human Genetics was established in 2014 to bridge the gap between groundbreaking genetic and genomic advancements and their clinical applications.

Under the leadership of Dr. Sandesh Nagamani, the division has been instrumental in designing and implementing clinical studies focused on genetic disorders. The primary mission of the Clinical Research Division is to translate basic science discoveries from the "bench-to-the-bedside" and to drive laboratory research based on clinical phenotypic observations.

The division's robust administrative and research infrastructure has enabled basic science researchers, clinical researchers, physicians, nurse practitioners, genetic counselors and research coordinators to form meaningful collaborations with patients and advocacy groups. These collaborations have facilitated a wide array of studies, ranging from individual investigator-initiated proof-of-concept studies aimed at identifying potential biomarkers and therapeutic targets, to collaborative multi-institutional gene discovery studies and multicenter natural history studies. Additionally, pivotal clinical trials conducted by the division have led to FDA approval of medications for treating rare genetic conditions.

The Clinical Research Division serves as a primary or lead site for several consortia within the NIH Rare Diseases Clinical Research Network, including:

- Urea Cycle Disorders Consortium
- Brittle Bone Disorders Consortium
- North American Mitochondrial Disease Consortium
- Global Leukodystrophy Initiative Clinical Trials Network
- Frontiers in Congenital Disorders of Glycosylation

Furthermore, Baylor's Undiagnosed Diseases Network, Center for Precision Medicine Models, GREGoR Consortium and the Intellectual and Developmental Disabilities Research Center are all integrated into and benefit from the division's resources.

10 Year Totals

206 studies

563 total grant accounts

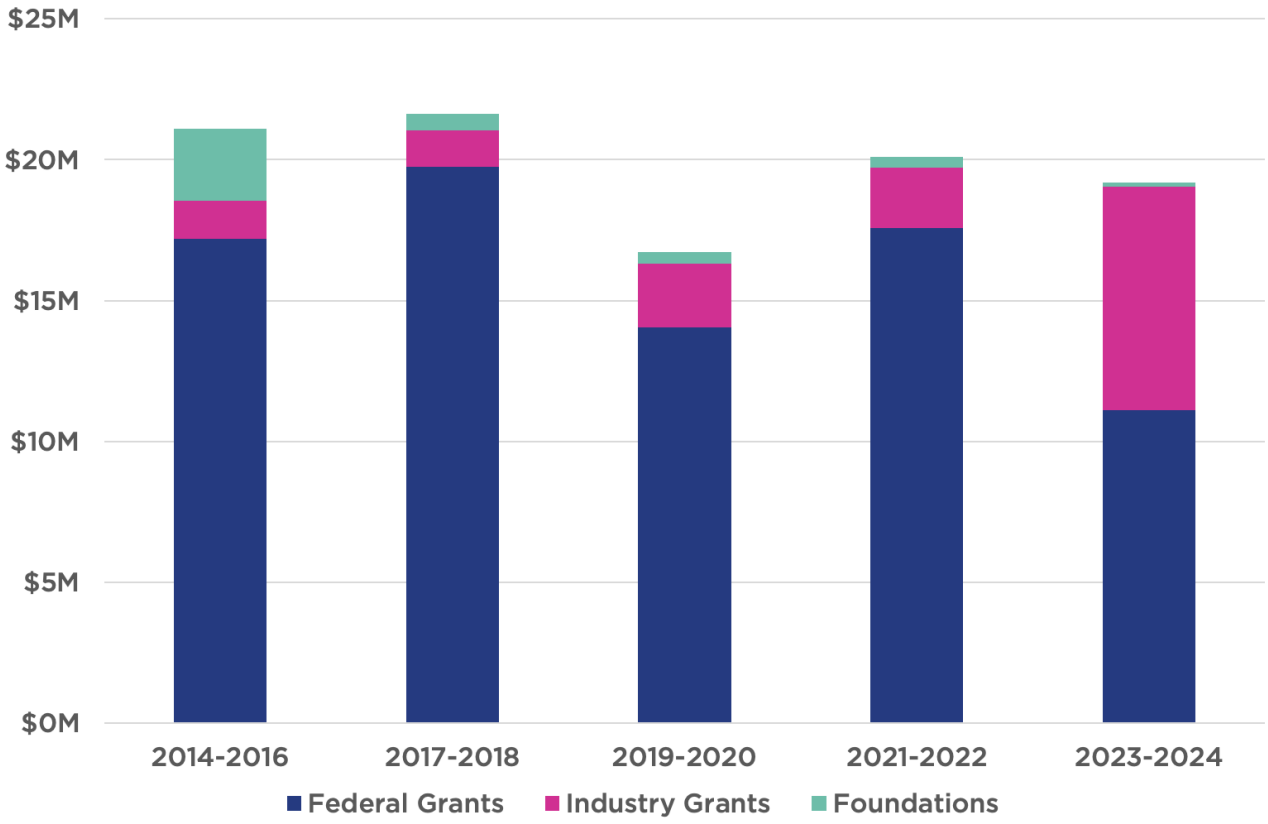
273 federal grants

182 industry grants

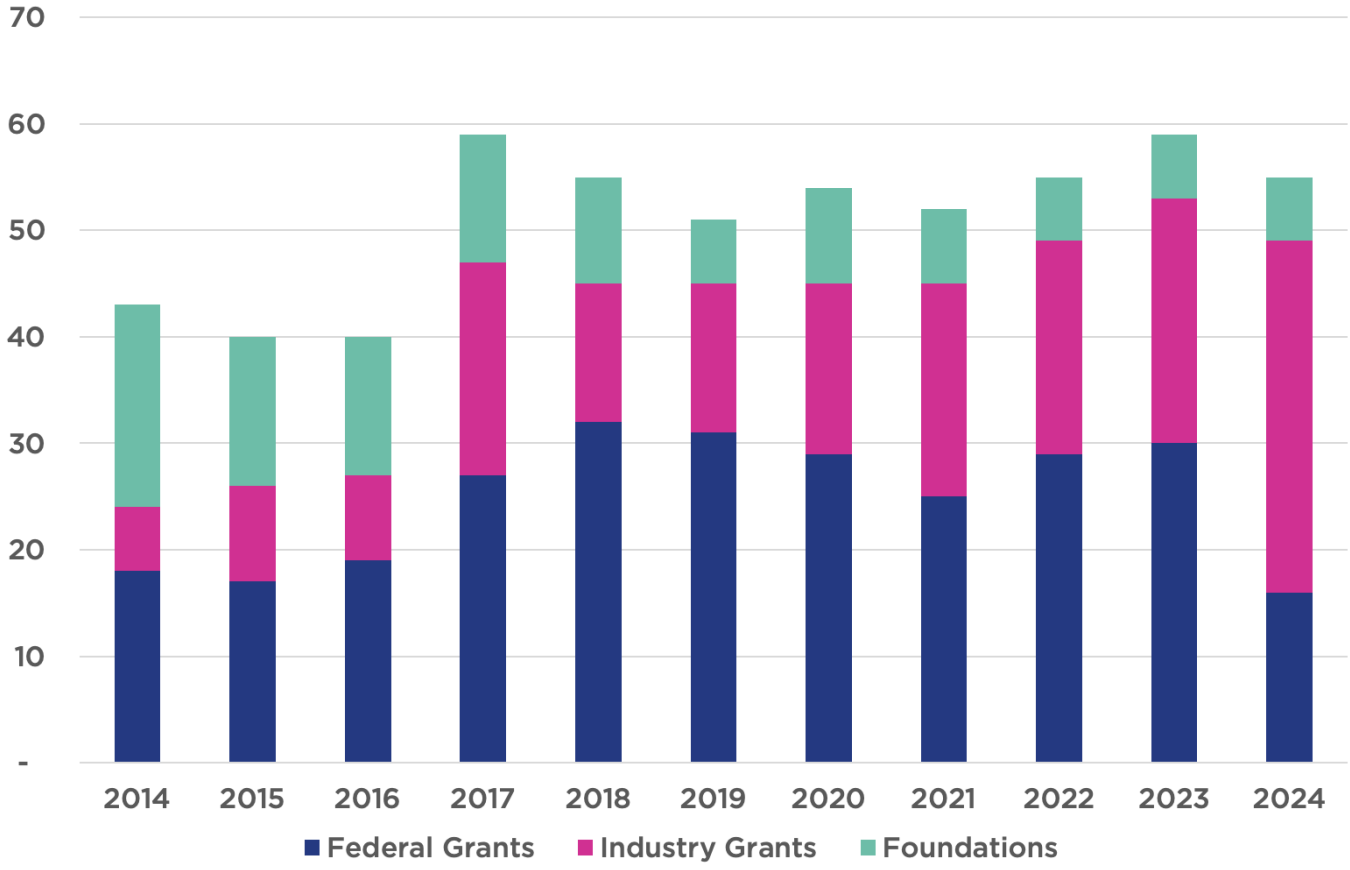
108 foundations

totaling **$99 Million**

Clinical Research Funding



Clinical Research Grants

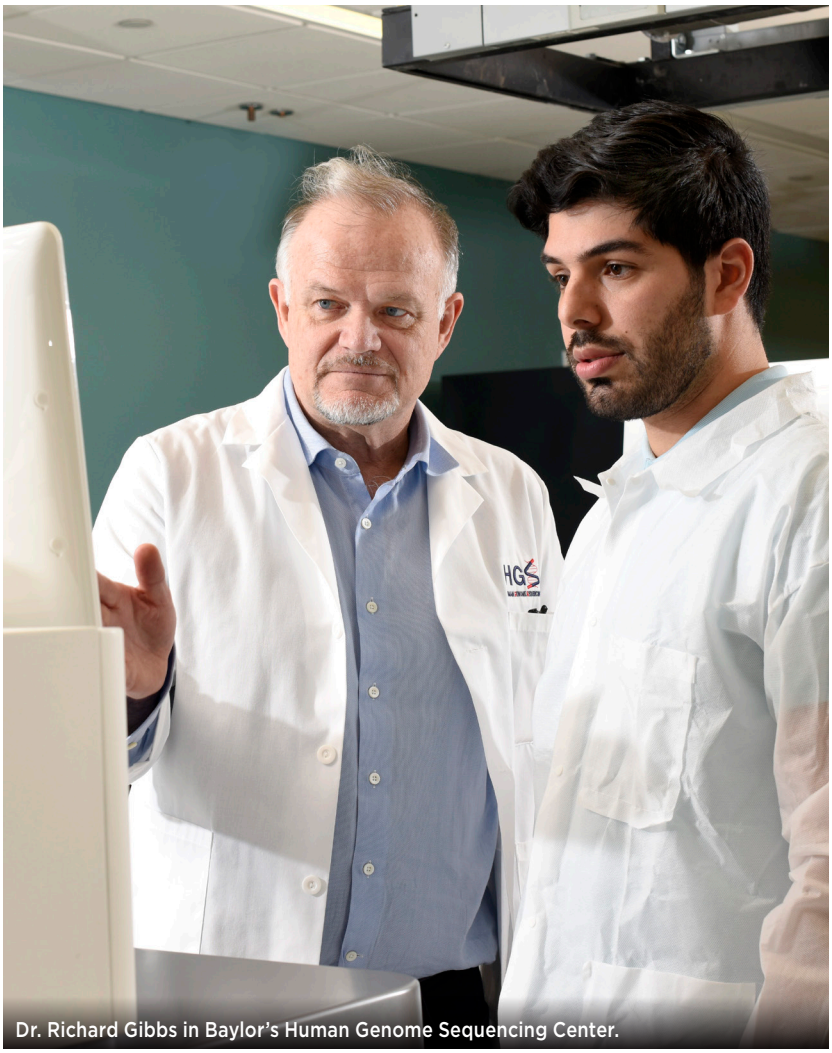


New Projects

2014

Baylor continued and established new consortia as part of the **National Institutes of Health (NIH) Rare Diseases Clinical Research Network**. Department faculty renewed funding in the Urea Cycle Disorders Consortium (UCDC) and the North American Mitochondria Consortium (NAMDC), and led establishment of the Brittle Bone Disorders Consortium (BBDC).

Baylor became one of the seven initial **Undiagnosed Diseases Network (UDN)** nationwide clinical sites. The UDN aimed to diagnose and ultimately cure rare genetic disorders by bringing together experts from across the country to share clinical and laboratory data. The Baylor Human Genome Sequencing Center (HGSC) and Baylor Genetics served as one of the two DNA sequencing cores for the UDN. Baylor also housed the Model Organism Screening Center (MOSC).



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

2016

The Knockout Mouse Project at Baylor College of Medicine led by Dr. Arthur Beaudet and Dr. Mary Dickinson received nearly $28 million in renewed funding from the NIH to generate and characterize lines for 1,000 new mouse genes using Cas9/CRISPR technology. The primary goal of the project is to understand how genes work and what functions they are responsible for by knocking out or deleting genes in animal models.

2018

The Baylor HGSC was named one of three centers responsible for generating clinical grade genomic data for the **NIH *All of Us* Research Program**. The program is a historic effort to collect data from 1 million or more people living in the United States to support a wide range of scientific discoveries and support precision medicine research.

2020

Dr. Jason Heaney and colleagues received a $10 million NIH grant to establish the **Center for Precision Medicine Models**. This center uses precision animal models to study rare genetic diseases and develop potential treatments.

2021

Baylor and Stanford University received an award for more than $25 million over five years from the NIH to continue building the **Clinical Genome (ClinGen) Resource**. This effort aims to create expert curated knowledge about clinically relevant genes and genomic variants for use in precision medicine. The award is one of three NIH grants totaling $73.2 million over five years for the project.

The NIH also awarded nearly $80 million to establish the **Genomics Research to Elucidate the Genetics of Rare Diseases (GREGoR) Consortium**. This consortium aims to develop novel methods and approaches that help researchers identify the genetic causes of single-gene diseases. Baylor College of Medicine is one of five clinical sites included in the consortium.

Baylor College of Medicine received a five year grant from the NIH to develop an educational program, designated the ***All of Us* Evenings with Genetics Research Program**, to train biomedical researchers in utilizing the NIH's *All of Us* Research Program's data resources to advance precision medicine.

Funded by the National Human Genome Research Institute (NHGRI), **the TEXOME Project** focuses on providing genetic testing to underserved populations in Texas. It leverages whole exome sequencing (WES) technology and innovative bioinformatics analysis to find answers for patients with undiagnosed diseases.

2023

The **Somatic Cell Genome Editing Consortium**, also funded by the NIH, aims to accelerate genome editing research and the development of new gene-editing related technologies and therapeutic approaches. This collaborative initiative includes 45 projects from 38 institutions and 72 principal investigators, including researchers from Baylor and Rice University.

2024

Baylor received a $3.5 million grant from the NHGRI for the **MAGNET (Making Genomics Accessible to Newborns in Texas)** program. This initiative aims to improve genetic diagnoses for sick newborns in neonatal intensive care units (NICUs) across West and South regions of Texas. The program will utilize low-cost whole genome and RNA sequencing technologies and adapt Baylor's Consultagene platform for in-patient genetic services, including virtual genetic counseling and provider consultation. The program will also establish a consortium of clinical partner sites in West and South Texas, training on-site providers to integrate genomic data into clinical decision-making for personalized treatments.

Discoveries

Genomics and Medicine

Genome Surgery (2015)

A study led by Baylor's Center for Genome Architecture achieved the first successful genome surgery, altering how the genome is folded inside the nucleus. This groundbreaking advance could lead to new methods for understanding and treating genetic diseases. The research was published in the *Proceedings of the National Academy of Sciences*.

3-D Mapping of the Genome (2016)

The Center for Genome Architecture at Baylor, led by Dr. Erez Lieberman Aiden, introduced Juicer, an open-source tool used in three-dimensional genome sequencing (Hi-C) processes. This tool facilitates the visualization and analysis of DNA dara into 3-D maos showing how DNA folds insides cells. The findings were published in *Cell Systems.*

Diagnosing Diseases with Multiple Genetic Causes (2016)

A study published in the *New England Journal of Medicine* by scientists at Baylor College of Medicine, Baylor Genetics, the University of Texas Health Science Center at Houston and Texas Children's Hospital highlighted advancements in diagnosing complex diseases involving multiple genes. By combining clinical features with genetic information, scientists achieved more precise diagnoses, leading to more effective treatments and better counseling for patients. The study involved whole exome sequencing of nearly 7,400 unrelated patients, identifying genetic causes in 28% of them, with approximately 5% having two or more disease genes involved.

4-D Genome Folding (2017)

A multi-institutional team, including researchers from the Center of Genome Architecture, created the first high-resolution 4-D map of genome folding, tracking an entire human genome as it folds over time. The study, published in *Cell*, revealed that distant DNA elements rapidly interact by forming loops enabling cells to regulate gene activity in response to stimuli. Through observing genome folding like a movie, The team identified two key mechanisms that organize the genome: loop extrusion and compartmentalization.

Exome Sequencing in ICUs (2017)

A study conducted by physicians and researchers from Baylor College of Medicine, Texas Children's Hospital, and Baylor Genetics demonstrated the efficacy of exome sequencing in diagnosing genetic disorders in infants in intensive care units. The research showed that exome sequencing could provide a molecular diagnosis in a significant number of cases, leading to changes in medical management for many infants. The study was published in *JAMA Pediatrics*.

Reanalysis of Molecular Data Yields New Genetic Diagnoses (2019)

A team led by Dr. Pengfei Liu implemented a genomic strategy to increase the molecular diagnostic rate of undiagnosed diseases. By reanalyzing exome sequencing data of two patient cohorts, the team nearly doubled the diagnostic yield in one cohort and significantly increased it in another. The reanalysis included newly identified disease-causing genes and genetic knowledge since the original study. The team also developed a computational pipeline to semi-automate the reanalysis process, which proved to be efficient and beneficial for patients and their clinical management. Findings were published in the *New England Journal of Medicine.*

Sequencing African Genomes (2020)

The department participated in the Human Heredity and Health in Africa (H3Africa) Consortium, which sequenced genomes from various African regions and countries. This research, published in *Nature*, revealed over 3 million novel genetic variants and provided insights into population history, environmental adaptation and disease susceptibility.

RNA Sequencing for Genetic Disease Diagnosis (2021)

In a study published in the *Journal of Clinical Investigation*, researchers at Baylor College of Medicine demonstrated the effectiveness of RNA sequencing as a diagnostic tool for genetic diseases. This approach revealed how genetic variants impact gene expression and splicing—insights often missed by traditional exome and genome sequencing. The study found that beginning with RNA sequencing could increase the diagnostic yield by 17%, bringing the overall diagnosis rate to approximately 50%.

Functional Significance of Gene Mutations in Autism (2022)

A multi-institutional study led by Dr. Shinya Yamamoto, Dr. Hugo Bellen, and Dr. Michael Wangler applied genetic strategies in fruit flies to determine the functional consequences of *de novo* variants associated with autism spectrum disorder (ASD). The research, published in *Nature Genetics*, identified 30 ASD-linked variants with functional differences and discovered that the gene *GLRA2* causes a spectrum of neurodevelopmental phenotypes beyond ASD.

Study Finds Ancestry-Driven Disparities in Pathogenic Variation (2024)

In a study published in *Communications Biology* and led by Baylor's Human Genome Sequencing Center, researchers performed an analysis of nearly 100,000 *All of Us* participants and revealed differences in the frequency of pathogenic genetic variants among different ancestry groups.

Fossils of Ancient Chromosomes Discovered (2024)

In a study that appeared in *Cell*, a multi-institutional team of researchers, including those from the Center for Genomic Architecture, discovered well-preserved chromosomes in a 52,000-year-old woolly mammoth. These fossil chromosomes provide a unique opportunity to study ancient DNA structures and compare them with modern species.

Molecular Genetics

Negative Feedback Loops in Biology (2016)

Scientists in the Herman and Lichtarge labs found that negative feedback loops act as a built-in control system to maintain balance and stability in biological systems. These loops can buffer the damage caused by mutated proteins, allowing systems to function smoothly despite changes. This adaptability might aid in the long-term evolution of organisms. The study was published in *Physical Review Letters*.

Research Improves Understanding of Cell to Cell Communication (2019)

A group of scientists that included Dr. Aleksander Milosavljevic, improved their knowledge of extracellular RNA (exRNA) communication, a system that delivers messages throughout the body. The NIH Common Fund's Extracellular RNA Communication Consortium created the exRNA Atlas, the first detailed catalog of human exRNAs in bodily fluids. This study, published in *Cell*, contributes to understanding the potential roles exRNA plays in health and disease. The researchers used computational tools to deconvolute complex experimental data, revealing six major types of exRNA cargo and their carriers.

Drug Decelerates Antibiotic Resistance (2023)

A team led by Dr. Susan M. Rosenberg discovered that the drug dequalinium chloride (DEQ) significantly reduces the ability of bacteria to develop antibiotic resistance. This proof-of-concept study, published in *Science Advances*, shows that DEQ slows the emergence of resistance mutations in bacteria, potentially prolonging the effectiveness of antibiotics.

Cardiovascular Genetics

HeartCare Study (2020)

Researchers from Baylor's Human Genome Sequencing Center worked with Baylor cardiologists to determine genetic risk factors for cardiovascular disease. The study analyzed 158 genes and tested for various cardiovascular conditions, providing participants with personalized care plans based on their genetic risk. The research was published in *Genetics in Medicine*.

Circadian Clock in Heart Failure (2022)

A study published in *Circulation* investigated the role of the protein Rev-erbα/β, a key component of the circadian clock, in heart disease development. The research team, led by Dr. Zheng Sun and Dr. Lilei Zhang, found that disrupting the circadian rhythm in cardiomyocytes leads to progressive dilated cardiomyopathy and lethal heart failure. They also discovered that correcting the metabolic defect can improve the condition, emphasizing the importance of chronotherapy.

Neurogenetics

Gut Bacteria and Aging (2017)

Scientists from Baylor College of Medicine and the University of Texas Health Science Center at Houston identified bacterial genes and compounds that extend the lifespan of the laboratory worm *C. elegans* and slow down the progression of tumors and the accumulation of amyloid-beta, a compound associated with Alzheimer's disease. The study, published in *Cell*, suggests that it might be possible to design bacterial preparations or compounds to slow down the aging process in the future.

Reduced Inhibition of Hippocampal Neurons in Rett Syndrome (2022)

A study published in *Neuron* by Dr. Huda Zoghbi's lab found that boosting the activity of specific brain cells in the hippocampus, called somatostatin-expressing neurons, helped to restore memory recall in mice with Rett syndrome. This discovery could lead to new treatments for cognitive symptoms in Rett syndrome.

DHX9 Variations and Neurodevelopmental Disorders (2023)

An international team of researchers from the GREGoR Research Center at Baylor College of Medicine, the Chinese University of Hong Kong, the German Mouse Clinic, and other collaborating institutions identified mutations in the *DHX9* gene in 20 patients with undiagnosed neurodevelopmental disorders. These mutations disrupt the gene's normal function, leading to a wide spectrum of conditions ranging from severe intellectual disability to nerve degeneration and neuropathy. This study, published in the *American Journal of Human Genetics*, marks the first time *DHX9* has been associated with human disease.

MRTFB Gene and Neurodevelopmental Disorder (2023)

Researchers at Baylor College of Medicine have identified specific variants in the *MRTFB* gene that are associated with a novel neurodevelopmental disorder. The study, published in *Genetics in Medicine*, found that mutations in *MRTFB* disrupt the protein's ability to regulate gene

expression. This dysregulation affects hundreds of genes involved in cellular structure and communication between brain cells, contributing to the observed neurodevelopmental symptoms.

Transdifferentiation with RNA Sequencing Aids Diagnosis of Genetic Disorders (2024)

By converting skin fibroblasts to neurons and using RNA sequencing, researchers improved the diagnostic yield for neurological disorders. This method enhances the ability to identify disease-causing mutations. The study, published in the *American Journal of Medical Genetics*, was led by Dr. Pengfei Liu and Dr. Shenglan Li.

Reproductive Genetics

Prenatal Genetic Test Based on Rare Fetal Cells (2016)

Researchers at Baylor College of Medicine, Drexel University College of Medicine, Texas Children's Hospital, and RareCyte, Inc. developed a prenatal, noninvasive genetic test based on rare fetal cells present in the mother's blood. This test aims to provide comparable information to amniocentesis and chorionic villus sampling. The study was published in *Prenatal Diagnosis.*

Aspirin and Preeclampsia (2021)

A joint study with the Chinese University of Hong Kong found that aspirin treatment can reduce the risk of preeclampsia by decelerating the metabolic clock of gestation. The study demonstrated that aspirin significantly slowed down metabolic gestational age, suggesting its potential to prevent preeclampsia. The research was published in *Hypertension.*

Endometriosis (2021)

Research by Dr. Jeffrey Rogers at Baylor, Dr. Krina T. Zondervan at the University of Oxford, the University of Wisconsin-Madison, and Bayer AG identified the *NPSR1* gene as a genetic cause of endometriosis. This discovery points to a potential new nonhormonal drug target for treating the condition. The research was published in *Science Translational Medicine*.

Immunogenetics

Gene Mutations and Disease (2015)

Researchers from the Lupski Lab and the Cardiovascular Research Institute at the University of California, San Francisco, identified genetic mutations causing hereditary autoimmune-mediated lung disease and arthritis by sequencing the exome of five unrelated families with similar symptoms. This discovery is expected to lead to a better understanding and treatment of COPA syndrome and other genetic diseases. The research was published in *Nature Genetics*.

Biomarker and Therapy for MS (2023)

Researchers in the Bellen Lab at the Jan and Dan Duncan Research Neurological Institute and Baylor College of Medicine found that myelin breakdown in multiple sclerosis (MS) leads to the accumulation of very long-chain fatty acids (VLCFA), triggering an autoimmune response that damages brain cells. The study, published in *Cell Metabolism*, suggests that reducing VLCFA levels with drugs bezafibrate and fingolimod could improve MS treatment.

Musculoskeletal Genetics

Targeting TGF-Beta for Osteogenesis Imperfecta Treatment (2022)

A study led by Dr. Brendan Lee, published in the *Journal of Clinical Investigation*, identified the upregulation of the protein TGF-β in human bone samples from osteogenesis imperfecta (OI) patients. Through the Brittle Bone Disorders Consortium, a part of the Rare Diseases Clinical Network, researchers

tested a monoclonal antibody therapy, fresolimumab, which neutralizes TGF-β, in a Phase 1 clinical trial. The treatment showed a significant increase in bone density in patients with moderate forms of OI.

Cancer Genetics

Bacteria Help Discover Human Cancer-Causing Proteins (2019)

A team led by Dr. Susan M. Rosenberg and Dr. Christophe Herman used bacteria to discover human proteins that can lead to DNA damage and promote cancer. By overexpressing genes in *E. coli*, the researchers identified proteins that cause DNA damage. They found that many of these proteins are not directly connected to DNA processing but are involved in other cellular functions. The study, published in *Cell*, provides new insights into the mechanisms by which proteins can cause DNA damage and cancer.

DNA Transcription vs. DNA Repair (2017)

Researchers in the Herman and Rosenberg labs found that in E. coli, a protein called GreA, which helps keep transcription accurate, actually interferes with DNA repair. Removing GreA improves repair of DNA breaks by allowing RNA polymerase to pause more often, which unexpectedly helps trigger the repair process. This discovery implies that maintaining transcription accuracy comes at the cost of lowering the cell's ability to repair DNA. This finding could change how scientists understand cellular priorities and may have implications for diseases like cancer and cell evolution. The study was published in *Nature*.

Without Dna2, Genes Can Jump into DNA Breaks (2018)

Researchers, including Dr. Greg Ira, found that the absence of the enzyme Dna2, which participates in DNA repair, leads to frequent insertions of DNA fragments into DNA breaks. This discovery has implications for understanding genomic instability and cancer. The research was published in *Nature*.

hnRNPM, a Guardian of the Integrity of Cellular Protein Production (2024)

The protein hnRNPM was found to play a crucial role in maintaining the accuracy of protein synthesis in cells. Loss of hnRNPM in cancer cells triggers an immune response, suggesting that targeting this protein might help the immune system recognize and attack cancer. The study was led by Dr. Chonghui Cheng, and the findings were published in *Molecular Cell*.

Study Reveals New Opportunities to Develop Cancer Treatments (2024)

A comprehensive study from the lab of Dr. Bing Zhang, a McNair scholar at Baylor, published in *Cell*, identified new potential therapeutic targets for cancer by integrating proteomics, genomics and epigenomics data from multiple cancer types. This expands the possibilities for targeted cancer therapies.

Artifical Intelligence

Using AI to Improve Diagnosis of Rare Genetic Disorders (2024)

Researchers developed AI-MARRVEL, an AI system that helps prioritize genetic variants for diagnosing rare Mendelian disorders. This system significantly improves the speed and accuracy of diagnoses. The study was led by Dr. Pengfei Liu and Dr. Zhandong Liu and published in *NEJM AI*.

Using Generative AI Assistant to Interpret Pharmacogenetic Test Results (2024)

Investigators with Baylor's Human Genome Sequencing Center developed a generative AI assistant to help interpret pharmacogenetic test results, particularly for statins. This AI tool improves the accuracy and relevance of genetic test interpretations for both patients and healthcare providers. This study was published in *JAMIA*.

Clinical Service

The Department of Molecular and Human Genetics at Baylor College of Medicine offers one of the most comprehensive and expansive clinical genetics programs in the world. With a focus on adult, pediatric, reproductive and prenatal genetics, the Department delivers high-quality, patient-centered care across a wide range of specialties.

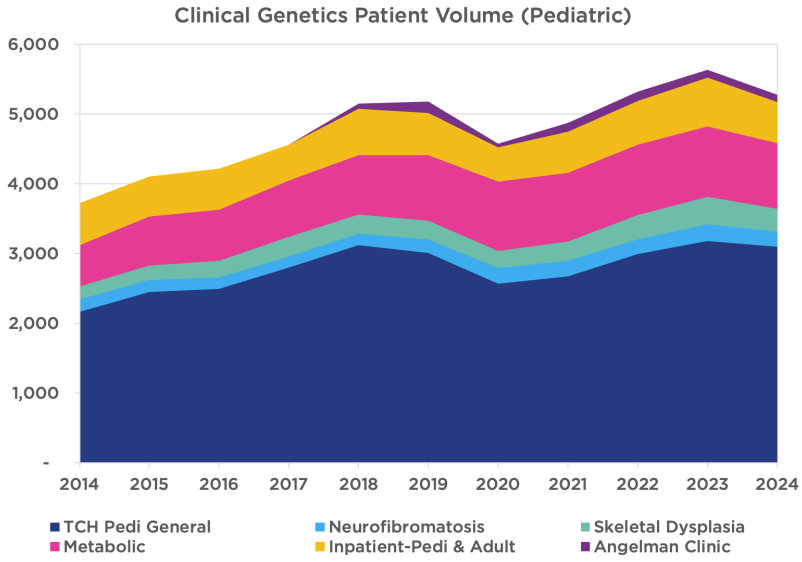
Pediatric Genetics

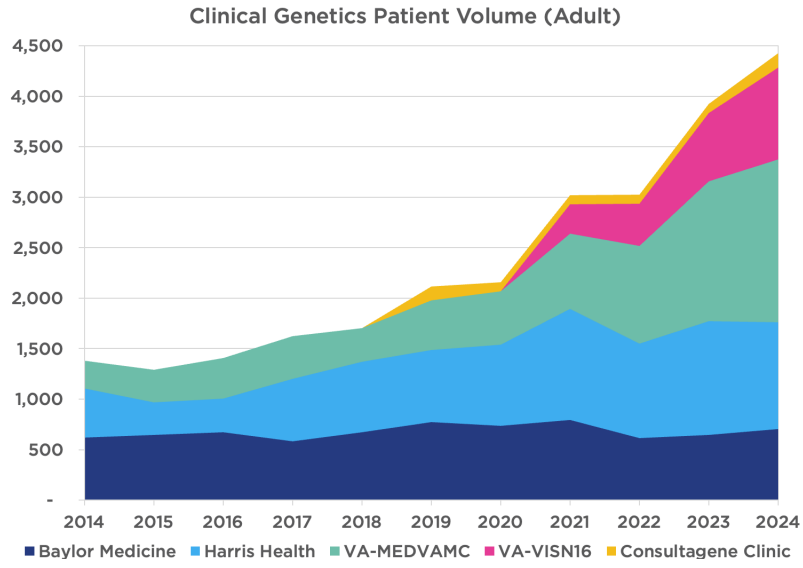
The pediatric genetics clinical service provides inpatient care at Texas Children's Hospital and affiliated sites. The outpatient clinics serve over 5,000 patients annually and conducts joint clinics with other departments including cancer genetics, otogenetics and neurogenetics/tuberous sclerosis. Specialty clinics include:

- Metabolic Clinic
- Neurofibromatosis Clinic
- Skeletal Dysplasia Clinic
- Angelman Syndrome Clinic
- Center for Genetic Disorders of Obesity
- Mitochondrial Medicine Clinic

In response to growing demand, the pediatric genetics service expanded to Texas Children's Hospital The Woodlands in 2016, establishing both inpatient and outpatient clinical genetics services.

Most recently, the Department extended its reach to Central Texas with the launch of inpatient and outpatient clinical genetics services at the new Texas Children's Hospital North Austin Campus, which opened in February 2024. This expansion was part of a broader initiative to bring specialized pediatric, fetal and OB/GYN care to Central Texas, including subspecialties like genetics.



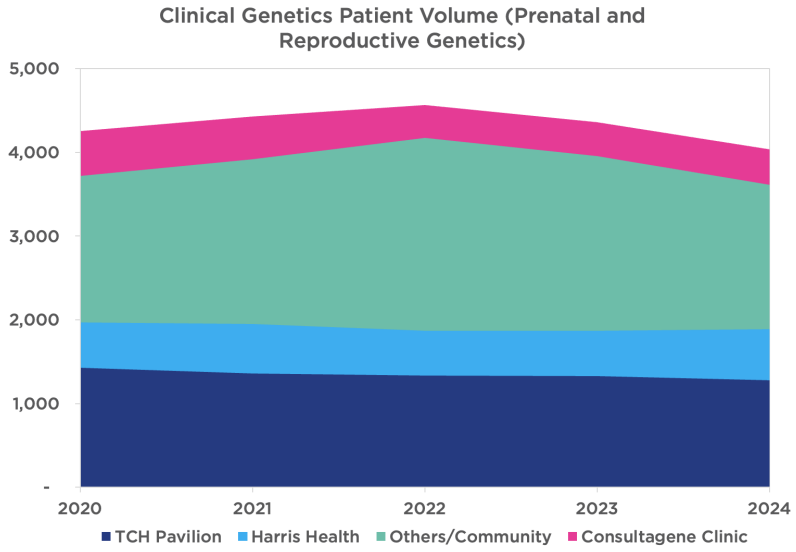


Adult Genetics

One of the largest adult genetics services in the U.S. offers inpatient and outpatient care at Baylor Medicine, Harris Health, the VA system, and virtually via Consultagene. Specialty clinics include:

- Cancer Genetics Clinic

- Cardiovascular Genetics Clinic
- Metabolic and Genetic Disorders of Bone
- Mitochondrial Medicine Clinic
- Neurogenetics Clinic



Prenatal and Reproductive Genetics

The largest prenatal genetics clinic in the U.S. is based at Texas Children's Pavilion for Women and also offers services through seven community maternal-fetal medicine clinics. The service provides advanced screening, diagnostic testing and counseling. The service is also available at Ben Taub Tower and virtually through Consultagene.

Award-Winning Faculty

Presented below is a selection of distinguished awards received by our faculty over the past decade.

Leadership and Lifetime Achievement Awards

American Society of Human Genetics Victor A. McKusick Leadership Award

- Arthur Beaudet, M.D. (2017)
- James Lupski, M.D., Ph.D. (2018)
- Huda Y. Zoghbi, M.D. (2019)
- David Nelson, Ph.D. (2022)

American Society of Human Genetics Lifetime Achievement Award

- C. Thomas Caskey, M.D. (2021)
- James Lupski, M.D., Ph.D. (2024)

American Society of Bone Mineral Research William F. Neuman Award

- Brendan Lee, M.D., Ph.D. (2022)

Colonel Harland Sanders Lifetime Achievement Award

- Arthur Beaudet, M.D. (2015)
- C. Thomas Caskey, M.D. (2016)

European Society of Human Genetics Mendel Lecturer and Gilded Pea Award

- James Lupski, M.D., Ph.D. (2024)

Foundation for Angelman Syndrome Lifetime Achievement Award

- Arthur Beaudet, M.D. (2024)

Research Excellence Awards

American Neurological Society George W. Jacoby Award

- Huda Y. Zoghbi, M.D. (2017)

American Society of Bone Mineral Research Fuller Albright Award

- Florent Elefteriou, Ph.D. (2016)

American Society of Clinical Investigation Donald Seldin-Holly Smith Award for Pioneering Research

- Christian Schaaf, M.D., Ph.D. (2016)

Breakthrough Prize in Life Sciences

- Huda Y. Zoghbi, M.D. (2016)

National Academy of Sciences Jessie Stevenson Kovalenko Medal

- Huda Y. Zoghbi, M.D. (2016)

National Institutes of Health Director's Pioneer Award

- Meng Wang, Ph.D. (2016)

- Christophe Herman, Ph.D. (2019)
- Susan Rosenberg, Ph.D. (2020)

Gairdner International Award

- Huda Y. Zoghbi, M.D. (2017)

Gilbert S. Omenn Computational Proteomics Award

- Bing Zhang, Ph.D. (2023)

Gruber Genetics Prize

- Hugo J. Bellen, D.V.M., Ph.D. (2024)

Howard Hughes Medical Institute Investigator

- Meng Wang, Ph.D. (2018)

Howard Hughes Medical Institute Faculty Scholar Award

- Meng Wang, Ph.D. (2016)

Kavli Prize Laureate in Neuroscience

- Huda Y. Zoghbi, M.D. (2022)

Louis-Jeantet Prize for Medicine

- Andrea Ballabio, M.D. (2016)

Lundbeck Brain Prize

- Huda Y. Zoghbi, M.D. (2020)

Shaw Prize in Life Science and Medicine

- Huda Y. Zoghbi, M.D. (2016)

Society for Glycobiology Significant Achievement Award

- Hamed Jafar Nejad, Ph.D. (2017)

TAMEST Edith and Peter O'Donnell Award

- Thomas F. Westbrook, Ph.D. (2015)
- Meng Wang, Ph.D. (2017)
- Benjamin Arenkiel, Ph.D. (2021)
- Erez Lieberman Aiden, Ph.D. (2023)

Michael E. DeBakey Excellence in Research Award

- Hugo J. Bellen, D.V.M., Ph.D. (2016)
- Erez Lieberman Aiden, Ph.D. (2018)
- Aleksander Milosavljevic, Ph.D. (2019)
- Marco Sardiello, Ph.D. (2019)
- Olivier Lichtarge, Ph.D. (2023)
- Daisuke Nakada, Ph.D. (2023)
- Fritz Sedlazeck, Ph.D. (2023)
- Jeffrey Rogers, Ph.D. (2023)
- Bing Zhang, Ph.D. (2024)

Innovation and Rising Star Awards

American Society for Cell Biology (ASCB) Early Career Life Scientist Award

- Meng Wang, Ph.D. (2017)

American Society for Clinical Investigation Young Physician-Scientist Award

- Lindsay C. Burrage, M.D., Ph.D. (2019)
- Jennifer E. Posey, M.D., Ph.D. (2019)

American Society of Human Genetics (ASHG) Outstanding Scientist Award

- Brendan Lee, M.D., Ph.D. (2016)

American Society of Human Genetics Early Career Award

- Jennifer Posey, M.D., Ph.D. (2023)

Child Neurology Society Philip R. Dodge Young Investigator Award

- Hsiao-Tuan Chao, M.D., Ph.D. (2020)

Levy-Longenbaugh Young Investigator:

- Steven Boeynaems, Ph.D. (2024)

National Institutes of Health Director's New Innovator Award

- Chengzhang Zong, Ph.D. (2014)
- Steven Boeynaems, Ph.D. (2024)
- Hongjie Li, Ph.D. (2024)

National Institutes of Health Director's Early Independence Award

- Hsiao-Tuan Chao, M.D., Ph.D. (2018)

Ron Konopka Memorial Junior Faculty Award

- Lilei Zhang, M.D., Ph.D. (2022)

Society of Pediatric Research Young Investigator Award

- Lindsay Burrage, M.D., Ph.D. (2020)

Notable Elections

American Academy of Arts and Sciences

- Huda Y. Zoghbi, M.D. (2018)
- Hugo J. Bellen, D.V.M., Ph.D. (2020)
- Brendan Lee, M.D., Ph.D. (2024)

American Academy of Science and Letters

- Huda Y. Zoghbi, M.D. (2024)

National Academy of Inventors

- Huda Y. Zoghbi, M.D. (2019)

National Academy of Sciences

- Hugo J. Bellen, D.V.M., Ph.D. (2020)

American Association for the Advancement of Science (AAAS) Fellow

- David Nelson, Ph.D. (2014)
- Brendan Lee, M.D., Ph.D. (2014)
- Phil Hastings, Ph.D. (2017)
- Olivier Lichtarge, Ph.D. (2019)
- Christophe Herman, Ph.D. (2021)

American Society for Clinical Investigation

- Christian Schaaf, M.D., Ph.D. (2017)
- Lilei Zhang, M.D., Ph.D. (2024)

American Society of Human Genetics President

- David Nelson, Ph.D. (2016)
- Brendan Lee, M.D., Ph.D. (2021)

Poland's Presidential Scholar Award of Full Professor

- Pawel Stankiewicz, Ph.D. (2017)

Teaching and Educational Awards

Barbara and Corbin J. Robertson, Jr. Presidential Award for Excellence in Education

- Gad Shaulsky, Ph.D. (2020)
- Hugo J. Bellen, D.V.M., Ph.D. (2021)
- Daryl Scott, M.D., Ph.D. (2024)

Norton Rose Fulbright Faculty Excellence Award

- Seema Lalani, M.D. (2014, 2019)
- Robb E. Moses, M.D. (2015)
- Benjamin Arenkiel, Ph.D. (2015)
- David Bates, Ph.D. (2015)
- Brett Graham, M.D. (2015)
- Shweta Dhar, M.D. (2015)
- Christian Schaaf, M.D., Ph.D. (2015)
- Herman Dierick, M.D. (2016)
- Sandesh C.S. Nagamani, M.D. (2017, 2021)
- Hamed Jafar-Nejad, Ph.D. (2018)
- Jason Heaney, Ph.D. (2019)
- Lindsay C. Burrage, M.D., Ph.D. (2021)
- Tanya Eble, M.S., C.G.C. (2021)
- Debra Murray, Ph.D. (2021)
- Weiwei Dang, Ph.D. (2022)
- Lilei Zhang, M.D., Ph.D. (2022)
- Benny Kaipparettu, Ph.D. (2023)
- Matthew Roth, Ph.D. (2024)
- Qin Sun, Ph.D. (2024)

Marc Dresden Excellence in Graduate Education Award

- Shinya Yamamoto, D.V.M., Ph.D. (2022)
- Herman Dierick, M.D. (2024)

Service and Professionalism Awards

Clark Faculty Service Award:

- Susan Fernbach, B.S.N. (2020)
- Shweta Dhar, M.D. (2018)
- Sandesh Nagamani, M.D. (2023)
- Seema Lalani, M.D. (2024)

Ben and Margaret Love Foundation Bobby Alford Award for Academic Clinical Professionalism

- Daryl A. Scott, M.D., Ph.D. (2024)

Master Clinician Award

- Arthur Beaudet, M.D. (2016)
- Carlos Bacino, M.D. (2017)
- V. Reid Sutton, M.D. (2021)
- William J. Craigen, M.D., Ph.D. (2022)
- Lorraine Potocki, M.D. (2022)

Rising Star Clinician Award

- Shweta Dhar, M.D. (2014)

Department Endowed Awards

Kenneth Scott Graduate Mentor Award

- Sharon Plon, M.D., Ph.D. (2018)
- Gad Shaulsky, Ph.D. (2019)
- Andy Groves, Ph.D. (2020)
- Benjamin Arenkiel, Ph.D. (2021)
- Herman Dierick, Ph.D. (2022)
- Christophe Herman, Ph.D. (2023)
- Jennifer Posey, M.D., Ph.D. (2024)

Rolanette and Berdon Lawrence Family Achievement Award

- Judi Coleman (2021)
- Patrick Hunt (2021)
- Oguz Kanca (2021)
- Shinya Yamamoto, D.V.M., Ph.D. (2021)
- Carlos Bacino, M.D. (2022)
- Charlotte Cherry (2022)
- Alyssa Crowder (2022)
- Richard Lewis, M.D., M.S. (2022)
- Jessica Swanson (2022)
- Demetria Dalco (2023)
- Janel Peterson (2023)
- Seema Lalani, M.D. (2023)
- Benjamin Belfort (2024)
- Lisa Folloder (2024)
- Olga Medina-Martinez (2024)
- Sandesh Nagamani, M.D. (2024)

The Shan and Lee-Jun Wong Fellowship

- Debdeep Dutta, Ph.D. (2022)
- Yuki Kageyama, M.D., Ph.D. (2024)

Recruitment Awards

McNair Scholars

McNair Scholars are part of the McNair Medical Institute at Baylor College of Medicine established in 2007 by the Robert and Janice McNair Foundation. This program aims to recruit and support talented scientists from around the world to conduct research in the Texas Medical Center.

- Benjamin Russell Arenkiel, Ph.D. (2010)
- Chenghang Zong, Ph.D. (2013)
- Bing Zhang, Ph.D. (2016)
- Hsiao-Tuan Chao, M.D., Ph.D. (2019)

CPRIT Scholars

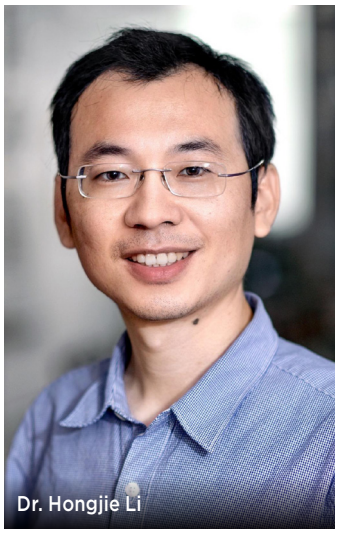
CPRIT Scholar Recruitment Awards help to attract scientific talent to Texas. The awards are funded by The Cancer Prevention and Research Institute of Texas (CPRIT), a state agency dedicated to funding cancer research and prevention programs across Texas.

- Charles Lin, Ph.D. (2015)
- Chonghui Cheng, Ph.D. (2015)
- Jihye Yun, Ph.D. (2018)
- Tao Wu, Ph.D. (2018)
- Steven Boeynaems, Ph.D. (2022)
- Graham Erwin, Ph.D. (2023)
- Vishnu Dileep, Ph.D. (2024)

New Research Faculty


Dr. Shinya Yamamoto


Dr. Lilei Zhang


Dr. Hongjie Li


Dr. Elizabeth Atkinson

2014

Dongsu Park, Ph.D.

Jointly recruited with the Center of Skeletal Medicine and Biology, Park investigates the molecular and cellular biology of mesenchymal/skeletal stem cells in tissue regeneration and cancer, aiming to develop better treatments for bone and connective tissue disorders.

2015

Florent Elefteriou, Ph.D.

Jointly recruited with the Center of Skeletal Medicine and Biology, Elefteriou studies the mechanisms of bone development, remodeling, repair and cancer cell metastasis, with a particular focus on neurofibromatosis type I (NF1) and the interaction between the autonomic nervous system and bone cells.

Charles Lin, Ph.D.

Lin's research focuses on combining molecular, computational, and chemical biology approaches to study gene control in cancer.

2016

Chonghui Cheng, M.D., Ph.D.

Cheng explores RNA regulation in cancer, particularly focusing on breast cancer metastasis driven by alternative splicing.

Shashikant Kulkarni, Ph.D.

Kulkarni's research is focused on the understanding cancer genomes by elucidating various classes of genomic alterations and discovering recurring mutations relevant for pathogenesis.

Michael Wangler, M.D.

Wangler's research uses *Drosophila* to study Mendelian disorders and their underlying genetic and developmental mechanisms.

Shinya Yamamoto, D.V.M., Ph.D.

Yamamoto focuses on the integration of *Drosophila* genetics and human genomics to discover new disease genes and develop bioinformatic tools.

Bing Zhang, Ph.D.

Zhang develops computational and statistical approaches to translate multidimensional omics data into biological and clinical insights, with a focus on cancer systems biology.

Lilei Zhang, M.D., Ph.D.

Zhang researches genetic and epigenetic regulation of cardiovascular disease, particularly circadian gene regulation in cardiac remodeling.

2017

Lindsay Burrage, M.D., Ph.D.

Burrage is interested in the genetic and biochemical basis for rare diseases with a particular focus on inborn errors of metabolism, such as urea cycle disorders.

Rodney Samaco, Ph.D.

Samaco's research explores the molecular and behavioral mechanisms underlying neurodevelopmental disorders and using animal models to study gene function and test potential therapeutic strategies.

Jihye Yun, Ph.D.

Yun's research focuses on how sugary drinks can alter gut bacteria and how this altered gut bacteria, in turn, can contribute to colon cancer development.

2018

Tao Wu, Ph.D.

Wu focuses on cancer epigenetics, particularly the role of novel DNA methylation (6mA) in cancer therapeutic resistance.

2019

Hsiao-Tuan Chao, M.D., Ph.D.

Jointly recruited with the Department of Pediatrics, Section of Neurology, Chao investigates how genetic mutations disrupt inhibitory and excitatory neuronal development, contributing to neurodevelopmental and neuropsychiatric disorders.

2021

Hongjie Li, Ph.D.

Jointly recruited with the Huffington Center on Aging, Li investigates molecular and cellular mechanisms contributing to brain aging and age-triggered tumor initiation using *Drosophila* models.

Nicholas Tran, Ph.D.

Tran studies neurodegenerative conditions using single-cell genomic approaches, with a focus on the retina to understand blinding disorders like glaucoma.

Elizabeth Atkinson, Ph.D.

Atkinson aims to reduce disparities in genomics research across ancestries, focusing on neuropsychiatric traits and population genetics.

Anthony Zoghbi, M.D.

Jointly, recruited with the Department of Psychiatry, Zoghbi is focused on studying rare genetic variation in severe forms of schizophrenia and obsessive-compulsive disorder. Zoghbi is the Beth K. and Stuart C. Yudofsky Scholar and chief of psychiatric genetics in the Menninger Department of Psychiatry and Behavioral Sciences.

2022

Steven Boeynaems, Ph.D.

Boeynaems focuses on understanding how cells perceive and respond to stress, particularly through the study of biomolecular condensates and their role in neurodegenerative diseases and cancer.

Qian Zhu Ph.D.

Zhu focuses on developing computational methods for genomic technologies to understand tumor heterogeneity, tumor-immune interactions, and identify biomarkers predictive of clinical outcomes.

2023

Graham Erwin, Ph.D.

Erwin focuses on understanding the functional role of repetitive DNA sequences in the human genome and developing synthetic transcription elongation factors to reverse pathogenic gene expression.

Ronit Marom, M.D.

Marom is a Caroline Wiess Law Scholar and her research focuses on secretory pathway defects, which can impact various organ systems, particularly skeletal development and neurodevelopment.

Fritz Sedlazeck, Ph.D.

Recruited by the Human Genome Sequencing Center, Sedlazeck's research focuses on understanding genome instability and complex variations and their impact on evolution and disease.

2024

Vishnu Dileep, Ph.D.

Jointly recruited with the Huffington Center on Aging, Dileep investigates how genome and epigenome alterations contribute to brain aging and neurodegeneration, aiming to identify therapeutic targets to improve brain health.

In Memorium

Dr. Kenneth Scott, a dedicated cancer geneticist, trained at Baylor as a graduate student and returned as faculty in 2009 after postdoctoral training at Harvard. Scott led pioneering work in cancer genetics using advanced genomic screening technologies. In honor of his legacy, the Department established the Kenneth Scott cDNA Clone Collection and a Graduate Mentor Award to continue his spirit of innovation and graduate student mentorship.

Dr. Lee Jun Wong was a world-renowned geneticist and researcher known for her contributions to mitochondrial genetics and rare disease diagnosis. Wong left behind a legacy of groundbreaking work that advanced the understanding of mitochondrial disorders. Her work continues to inspire progress in genetic research and diagnostics.

Dr. C. Thomas Caskey, a pioneer in genetics and genomics was renowned for his groundbreaking contributions to genetic research, including defining the universality of the genetic code and discovering codon-specific proteins controlling translation termination. Caskey founded the Department of Molecular and Human Genetics at Baylor College of Medicine, which became a global leader in genetics research.

Dr. William E. O'Brien, a distinguished geneticist and biochemist was instrumental in founding the Biochemical Genetics Laboratory at Baylor College of Medicine and played a pivotal role in the development of its Medical Genetics Laboratory, which became a global leader in genetic research. O'Brien was known for his scientific expertise, mentorship, and contributions to the growth of the Department of Molecular and Human Genetics at Baylor.

Baylor College of Medicine

DEPARTMENT OF
MOLECULAR & HUMAN
GENETICS

One Baylor Plaza
Houston, Texas 77030

bcm.edu/genetics
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