I am delighted to welcome you to our 2018 Annual Report. As we begin a new year, I’d like to briefly reflect on last year’s accomplishments.

The Department of Molecular and Human Genetics at Baylor College of Medicine remains the No. 1 ranked genetics department in the country based on total National Institutes of Health funding and awarded grants. Other firsts this past year include the inaugural class of the new masters in genetic counseling program beginning their studies and the official full launch of the latest version of our web-based genetic services platform, Consultagene.

Our research, clinical and diagnostic activities uphold the Department’s overall mission of transforming the practice and science of genetics while expanding its role in medicine. We remain committed to the growth of Baylor Genetics, our diagnostic laboratory venture with Miraca Holdings, Inc. This jointly governed entity continues to support the academic mission and innovation of the Department while promising to extend the impact of genetic diagnostic testing worldwide.

Our department continues to grow and expand globally. We held our second Baylor College of Medicine and the Chinese University of Hong Kong Center Joint Symposium on Clinical Genetics in Hong Kong and, along with the Dan L. Duncan Comprehensive Cancer Center, hosted the first U.S.-Japan Clinical Cancer Genomics and Personalized Medicine Symposium.

In addition, during these past few years, new and continuing consortia with the NIH and industry are leading to new gene discoveries and advancements in the implementation of genetics and genomics.

As we take measure of the past year, let us also look forward. The future holds much promise due to the talent and dedication of our renowned faculty and trainees. 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 and Professor of Molecular and Human Genetics
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Research in genetics began at Baylor College of Medicine in 1971 when Dr. C. Thomas Caskey, professor of molecular and human genetics, and, soon thereafter, Dr. Arthur Beaudet, the Henry and Emma Meyer Chair and Professor of Molecular and Human Genetics at Baylor, were recruited from the NIH to lead Baylor’s entry into that field. Operating initially within the Departments of Internal Medicine and Pediatrics, the pair created a clinical training program in 1976 to educate and train a group of top investigators in genomics and biomedical research.

As the research team grew in size, scope and ambition, a centralized organization was needed to fuse together the disparate lines of effort. For that reason, in 1985, the Institute of Molecular Genetics was created, thereby placing Baylor on the map as a genetics powerhouse. By leveraging its ability to recruit the best and brightest physicians and scientists in the field, the Institute grew substantially and in 1994, the decision was made to make the Institute a full department.

The Department’s success reached new heights with the creation of the Human Genome Sequencing Center in 1996. The Center, led by Dr. Richard Gibbs, the Wofford Cain Chair and Professor of Molecular and Human Genetics at Baylor, was one of three sites (out of six pilot programs) to complete the Human Genome Project. In 2000, scientists triumphantly announced they had deciphered the human genome—the blueprint for human life.

In recent years, the Department has successfully provided comprehensive clinical care to patients worldwide. By assembling the largest clinical genetics program in the country, Baylor offers patients timely and expert assistance, as well as unparalleled treatment and counseling options through 14 specialized clinics.

In addition, the Department has expanded its reach to provide diagnostic genetic testing services to the broader medical genetics community through its laboratory, Baylor Genetics, a joint venture with Miraca Holdings. Baylor Genetics offers an expansive menu of genetic tests and provides leading service to practitioners worldwide.

The past 40-plus years have been an exciting time of growth and change. Initially focused on medical and pediatric genetics, the Department has since expanded its reach into diverse areas that include functional genomics, genome sequencing, cancer genetics and more. In the process, it has become 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

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

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

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

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

Shashikant Kulkarni, M.S., Ph.D., F.A.C.M.G.
Vice Chair, Research Affairs (Baylor Genetics)

Susan Fernbach, R.N., B.S.N., Director, Office of Community Engagement and Diversity

We have more than 550 FACULTY, TRAINEES AND STAFF who occupy 115,000 SQUARE FEET OF SPACE. Faculty includes:

8 members of the National Academy of Medicine

8 Howard Hughes Medical Institute Investigators

9 Fellows of the American Association for the Advancement of Science

4 members of the National Academy of Sciences

2 members of the American Academy of Arts and Sciences
Consultagene Version 1.1 Launches

When Baylor College of Medicine, together with Baylor Genetics, launched Consultagene in 2016, the vision was to create a personalized, web-based platform that served to integrate research and clinical care through genetic counseling, peer-to-peer consultation, patient and provider education and diagnostic interpretation of clinical genomic data.

Since its initial launch, the members of the team behind Consultagene tested the first version of the online platform internally while bringing its services such as educational videos and consultations to the client outside of the platform. At the start of 2018, development began on a new version of Consultagene. Some of the many enhancements to the online platform were designed to meet business requirements, such as the ability to create different types of users, groups, and client experiences, also known as “Journeys”.

Consultagene’s Journeys are tailored to a patient’s specific indication and include a series of personalized modules that may range from pedigree creation and family history to the scheduling of a tele-genetic counseling session. A healthcare professional, when interacting with the web portal, may create a referral for a journey for one of their patients or they may also choose to refer themselves for a peer-to-peer consultation about a difficult case.

With Version 1.1, Consultagene now offers three types of Journeys: education only, tele-genetic counseling, and peer-to-peer consultations. The catalog of Journeys that Consultagene currently offers also expanded to include: Carrier Screening and Somatic Tumor Testing.

The development phase of version 1.1 concluded in the summer of 2018. There was a period of user testing which lasted until the end of September. At the beginning of October, Consultagene V1.1 was officially launched within our local genetics clinics. The online platform has incorporated eight educational videos and there are three more in development as well as multi-language translations. The videos’ topics range from the basics of genetics to multi-gene panel testing to whole exome sequencing.

BRCA1 & BRCA2 Journey in Consultagene

We have received high satisfaction scores from surveys that were taken by clients after watching the videos. For example, in response to whether or not the video provided the client with a good understanding of the types of results that they may receive from a whole exome sequencing test, 98% of those surveyed gave answers that ranged from “Agree” to “Strongly Agree”. Similarly, as to whether or not the video provided the client with a good understanding of the whole exome sequencing test, 97% of those surveyed responded favorably.

Consultagene services, both through the platform and by external means, have been extended to close to 400 patients since its initial launch. We are looking forward to the growth and development of Consultagene in the near future, beginning with the addition of its physical location, the Consultagene Clinic, in 2019.
Baylor Genetics Expands its Precision Medicine Portfolio with ClariFind

In March, Baylor Genetics, Baylor College of Medicine’s genetic diagnostic laboratory joint venture with Miraca Holdings, Inc. announced the launch of ClariFind, a comprehensive somatic tumor next-generation sequencing test that correlates genomic results with specific drug therapies and clinical trial options. Baylor Genetics’ experts, in collaboration with Baylor College of Medicine, developed ClariFind to augment its existing oncology test menu of hereditary cancer panels, chromosomal microarrays, single-gene assays, cytogenetics, and fluorescence in situ hybridization (FISH) assays.

“Cancer is a complex genomic disease. Therefore, we need efficient tests to focus on detecting low-frequency variants to identify cancer mutations and study clonal evolution over time to better serve our cancer patients,” said Dr. Shashikant Kulkarni, Chief Scientific Officer and Senior Vice President of Baylor Genetics, “We do this by indexing individual molecules with a unique molecular identifier (UMI) before PCR [PCR, also known as Polymerase Chain Reaction, is a widely used method that allows one to make many copies of a specific DNA segment] and deep sequencing allows us to robustly and accurately detect low-frequency mutations.”

In a single next-generation sequencing assay, ClariFind assesses genomic alterations in 277 key cancer genes for both solid tumors and hematologic malignancies. Due to its unique molecular barcoding approach, ClariFind allows for confident, low-level variant detection. Additionally, ClariFind’s chemistry has been optimized to improve analysis of difficult guanine-cytosine content rich regions in genes such as CEBPA and CCND1. Furthermore, low DNA input samples will be accepted, which is crucial for small biopsy samples.

The ClariFind report has been designed to allow the patient and clinician to navigate through the results and corresponding treatment options easily. Each ClariFind report provides annotated genomic findings, associated targeted therapies, potential clinical trials when available, and a detailed interpretative summary. Each patient’s case is thoroughly reviewed by board-certified clinical experts to aid in optimizing patient care. Most importantly, our experts are available for clinical consultation to serve as partners throughout the patient’s journey.

“Baylor Genetics has always believed that a comprehensive approach is needed for treatment of genetic conditions, whether it is inherited or somatic. Therefore, we believe that ClariFind is an important component to Baylor Genetics’ existing cancer menu and will provide more support and options to the oncology community,” said Kengo Takishima, President and Chief Executive Office of Baylor Genetics.
Baylor Geneticists, Oncologists Host Symposium in Japan

On March 3, the Department of Molecular and Human Genetics and the NCI-designated Dan L. Duncan Comprehensive Cancer Center at Baylor College of Medicine hosted their first joint U.S.-Japan Clinical Cancer Genomics and Personalized Medicine Symposium in Tokyo, Japan, a meeting on state-of-the-art cancer genetics and genomics.

“This is an exciting time, as the Japanese government is leading implementation and support for cancer genetic testing in its healthcare system,” said Dr. Brendan Lee, the Robert and Janice McNair Endowed Chair and professor of molecular and human genetics at Baylor. “In 2015, there were approximately 981,000 cases of cancer affecting men and women. The advent of genetic testing may improve detection, diagnosis, choice of targeted therapies and monitoring.”

Speakers included leading genetics and cancer researchers, with presentations focused on the implementation of cancer genetic testing in Japan. Baylor was represented by Drs. Lee, Shashikant Kulkarni, David Wheeler, Matthew Ellis and Sharon Plon, all of whom presented to leading oncologists during the symposium.

The meeting was sponsored and hosted by Miraca Holdings, the joint venture partner in Baylor Genetics, Baylor’s genetic diagnostic laboratory.
Research and Discoveries

Research in the Department of Molecular and Human Genetics at Baylor College of Medicine has led to important discoveries that increase understanding of disease and guide potential new treatment. Here are four recent studies that are representative of the groundbreaking research in the department.

Undiagnosed Diseases Network finds 31 new syndromes

Identifying genes responsible for rare or unknown disorders using traditional approaches can be extremely time-consuming and typically takes several years or decades. However, the Undiagnosed Diseases Network (UDN), a consortium that includes Baylor College of Medicine, Texas Children’s Hospital, Baylor St. Luke’s Medical Center, Stanford University and other institutions, has found 31 new syndromes and has been able to make diagnoses for 132 patients within two years of the group’s inception.

In late November 2018, a study published in *New England Journal of Medicine* gave a detailed description of the inner workings of UDN. It was an in-depth analysis of the referral and acceptance patterns, diagnoses, impact rates and follow-up scientific investigations of 1,519 cases that were referred to the UDN in the last two years. Of the 1519 cases that were referred to the UDN, a total of 601 patients across the nation were accepted for evaluation.

It was reported that in less than two years, 382 out of the 601 patients have completed extensive medical and genetic evaluations. Of these, 132 have received a diagnosis, yielding an astounding diagnosis rate of 35 percent.

Based on the new findings from the UDN team, physicians were able to take quick action to better manage the patients who were diagnosed. Twenty-one percent of the patients who received a diagnosis were recommended alternative therapies, and 36 percent received variant-specific genetic counseling.

The UDN was established in 2014 with the goal of solving medical mysteries through team science. Phase I of the UDN, which is funded by the National Institutes of Health, began as a network of seven clinical sites, two sequencing cores, and a coordinating center. Since then, a central biorepository, metabolomics core and the Model Organisms Sequencing Center (MOSC) were added. Now in Phase II, the number of clinical sites for the UDN has increased from 7 to 12.

The Model Organisms Sequencing Center (MOSC), a functional study core for the UDN is located at Baylor, the Jan and Dan Duncan Neurological Research Institute (NRI) at Texas Children’s Hospital, Washington University in St. Louis and the University of Oregon. The goal of the MOSC is to help physicians and scientists understand how specific genetic changes (variants) contribute to a particular disease by studying these variants in fruit flies or zebrafish. MOSC researchers work closely with the physicians at UDN clinical sites to identify the genes responsible for rare and undiagnosed disorders. MOSC is spearheaded by NRI investigators Drs. Hugo Bellen, Shinya Yamamoto and Michael Wangler.

Baylor College of Medicine and Baylor Genetics house one of the sequencing cores, and in phase II, it is the sole sequencing core for the network. Dr. Christine Eng, professor of molecular and human genetics and chief medical officer of Baylor Genetics, is the principal investigator of this core.

Baylor, Texas Children’s Hospital and Baylor St. Luke’s Medical Center house one of the UDN clinical sites where doctors and healthcare providers from many specialties, come together to help find the cause of participants’ symptoms. Dr. Brendan Lee, chair of molecular and human genetics, leads the UDN clinical site.

As the study pointed out, the most unique feature and perhaps the biggest contributor toward UDN’s success is its model of multi-institutional collaborations. Teams of researchers and physicians from participating institutions all over the nation leverage their multidisciplinary expertise and resources to quickly find specific diagnoses for extremely challenging clinical cases, with no additional cost to the patients.

“UDN is a unique effort that brings together experts in many disciplines to tackle some of the most difficult medical mysteries,” said contributing author Dr. Shinya Yamamoto, assistant professor of molecular and human genetics at Baylor. “The team not only includes physicians, genetic counselors and human geneticists, but also model organism biologists and structural biologists who traditionally have not been involved in the diagnostic phase of clinical research.”
FDA recognizes database of clinically relevant genetic variants

In December, the database created by the Clinical Genome Resource program (ClinGen) was recognized by the U.S. Food and Drug Administration in an announcement made by FDA Commissioner Dr. Scott Gottlieb and National Institutes of Health Director Dr. Francis Collins.

ClinGen, a NIH-funded program, is a resource that is dedicated to establishing a central database that defines the clinical relevance of genes and variants with the intention of informing precision medicine and research. ClinGen was launched in 2013, with faculty from Baylor College of Medicine playing key roles in developing and advancing the program.

“FDA recognition of ClinGen’s process for interpreting variants, or mutations, found through genetic testing highlights for the genetics community our goal of developing authoritative sources of clinical genetics that is publicly available to them. ClinGen is filling a critical need in the substantial challenge of gathering data and connecting it with the community for use in supporting genomic medicine and research,” said Dr. Sharon Plon, principal investigator on the ClinGen project and professor of pediatrics-oncology and molecular and human genetics, and co-leader of the pediatric cancer program in the Dan L. Duncan Comprehensive Cancer Center at Baylor.

ClinGen’s recognition is the first of its kind issued through the new FDA Human Variant Database Program, which seeks to support an easier path for marketing clearance or approval for clinical gene test developers. This recognition enables ClinGen to provide a gold-standard example of an approach for interpretation of inherited genetic variation that other organizations can adopt, and underscores the importance of a consistent approach, collaboration and sharing of knowledge to improve healthcare.

Dr. Aleksandar Milosavljevic, professor of molecular and human genetics at Baylor, and his team have played a critical role in developing the informatic tools and infrastructure to support the interpretation of variants and make the process and evidence available to the public. Particularly relevant parts of the database are the ClinGen Allele Registry and the ClinGen Evidence Repository, both developed by the Bioinformatics Research Laboratory directed by Milosavljevic.

“The Allele Registry enables real-time creation of universal IDs for variants, meaning a given variant can be uniquely referenced by its ID. The Evidence Repository enables sharing of evidence for or against pathogenicity, or the disease association, of variants, along with guideline-based conclusions about their pathogenicity,” said Milosavljevic.

Early examples of ClinGen interpretation included two genes, PTEN and CDH1, which are included in many cancer genetic tests. ClinGen is working to expand the use of cancer genetic data for estimating risk of cancer or the interpretation of genetic changes found in tumors. The ClinGen database provides a resource of human variants that have been interpreted for their potential association with disease and helps to assure that clinical interpretations of genomic test results are accurate.

ClinGen has also assembled expert teams that develop the needed specifications and employ a systematic approach to this variant interpretation and provide evaluations of relevant scientific evidence. ClinGen Variant Expert Panels are international in make-up and include experts from both industry and large academic medical centers to achieve robust, optimized classifications of genomic variation.

This expert knowledge, together with the approach to variant interpretation and the evidence evaluated, is publicly disseminated and made fully accessible to other clinicians and laboratory scientists with the aim of improving the quality of genomic medicine.

Primate genomics study reveals clues into AIDS resistance

An international group of institutions led by Dr. Guido Silvestri at Emory University and Dr. Jeffrey Rogers, associate professor in the Human Genome Sequencing Center (HGSC) and Department of Molecular and Human Genetics at Baylor College of Medicine, set out to investigate the mechanisms that drive disease progression and develop new insights into the behavior of the HIV virus in humans.
The human immunodeficiency virus can be divided into two major types, HIV-1 and HIV-2. HIV-1 is related to simian immunodeficiency viruses (SIVs) found in West African chimpanzees and gorillas, while HIV-2 has been found to be closely related to the SIV found in the sooty mangabey, a species of monkey that also lives in West Africa.

“Sooty Mangabeys are natural hosts for SIV, meaning they can be infected with the virus and sustain long-term infection without ever progressing to disease. However, macaques rapidly progress into AIDS-like disease and succumb to SIV when infected,” said Rogers.

Silvestri and his team at Emory shared sooty mangabey samples with Roger and his colleagues at the HGSC to sequence and assemble its genome. Direct comparison of the new sooty-mangabey genome to the genomes of the rhesus macaque and human led to the discovery that genes related to the immune system, particularly the TLR4 and ICAM2 genes, in sooty mangabeys have significant divergence from the equivalent genes of macaques or humans. These genomic differences suggest that sooty mangabeys are able to avoid SIV-induced disease progression due to specific differences underlying their immune responses.

“This research gives us part, albeit an exciting one, of the answer as to how sooty mangabeys can avoid disease. We hope this leads to insights into new therapies and treatments in humans to fight HIV infection,” said Rogers.

This research appeared in the January 2018 issue of Nature and was supported by the National Institute of Allergy and Infectious Diseases, the National Human Genome Research Institute and the NIH Director’s Office of Research Infrastructure Programs.

**Without Dna2, genes can jump into DNA breaks**

Researchers from several institutions including those from the lab of Dr. Greg Ira, associate professor of molecular and human genetics and member of the Dan L. Duncan Comprehensive Cancer Center at Baylor College of Medicine, wanted to find out the consequences of the absence of the enzyme Dna2, which is known to participate in DNA repair, on the integrity of the genome.

“One of the interests of my lab is to understand basic mechanisms of DNA repair,” said Ira, a corresponding author on this paper. “In this project we worked with yeast and discovered for the first time a mutant that shows frequent insertions of DNA fragments into DNA breaks. This mutant lacks Dna2, an enzyme conserved among all organisms.”

What Ira and his colleagues discovered was that when DNA2 is absent, any piece of any chromosome can jump into the DNA break.

“During DNA synthesis, occasionally long single strands of DNA can form and are normally eliminated by Dna2. In mutants that lack Dna2, these oversynthesized fragments can get caught in DNA breaks, causing genomic instability, which mostly has negative effects on cells. However, these insertions also can lead to gene duplication and chromosome evolution, which may have positive consequences for cells,” said first author Dr. Yang Yu, postdoctoral associate in the Ira lab. “We propose that Dna2, whose activities include degradation of oversynthesized DNA fragments, prevents their insertion into the genome and alteration of the genetic code.”

The researchers also investigated the origin of the DNA fragments that insert themselves into chromosomal breaks. They discovered that small genes, DNA segments at the end of chromosomes called telomeres as well as other sequences of inserted DNA had originated from all over the genome.

“It’s been reported that similar insertion of DNA fragments is common in cancer,” Ira said. “We think that the mechanism we describe here for Dna2-deficient cells likely will be observed in many cellular conditions where small DNA fragments are generated and not properly degraded. We think that this is common in cancer but can also occur in patients that are deficient in some components of innate immune system responsible for elimination of foreign DNA.”

This study was published in the December 2018 issue of Nature and was funded by grants from the National Institutes of Health and the Cancer Prevention Research Institute of Texas.
Grant Awards Continue to Drive Progress

The Department of Molecular and Human Genetics continues to be ranked No. 1 in NIH funding

The National Institutes of Health is the primary governmental agency responsible for biomedical and health-related research in the United States. A department’s ability to consistently obtain NIH grants, which are awarded through a competitive peer review process, demonstrates the strength of its research and training programs. On that basis alone, the Department of Molecular and Human Genetics at Baylor College of Medicine continues to distinguish itself.

For eight years running, the Department remains the No. 1 ranked U.S. genetics department, as measured by the number of NIH-awarded grants and total funding received. For 2018, the amount in funding dollars from NIH awards totaled more than $71 million (source Blueridge rankings).

The Department is excited to receive this funding, and has put this support to excellent use. Through the funding of the Undiagnosed Disease Network Center, the Center for Mendelian Genomics, the Knockout Mouse Project, and many other investigator-initiated grants, the Department is finding answers to science’s most pressing questions. In the process, the Department is improving the well-being of patients across the world.

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 Doris Duke Foundation
- W. M. Keck Foundation
- The March of Dimes
- The Angelman Syndrome 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 is proud to work hand-in-hand with six research centers, each of which focuses on specialized areas of medical research. These centers are led by primary faculty of the Department and, together, advance the current boundaries of scientific knowledge.

Baylor’s HGSC named an NIH All of Us research center

The Baylor College of Medicine Human Genome Sequencing Center (Baylor HGSC), led by Dr. Richard Gibbs, has been operational for more than 20 years. Originally established in 1996 to participate in, and eventually help complete, the Human Genome Project, the HGSC has grown and achieved international recognition as a large-scale DNA sequencing and analysis center. Currently a Center for Complex Disease Genomics supported by the NIH and the National Human Genome Research Institute (NHGRI), the Baylor HGSC has since expanded its research focus into new and exciting areas.

In September 2018, the All of Us Research Program, part of the National Institutes of Health, named a consortia led by the Baylor HGSC as one of three centers responsible for generating clinical grade genomic data for the program. These centers will generate genomic data from biosamples contributed by the program’s participants. Ultimately, this information will become a critical component in the program’s precision medicine research platform, a national resource to support studies on a variety of important health questions.

In this phase of the program, the genomic DNA sequence of the genomes of the participants will be determined by genotyping and whole genome sequencing. The Baylor HGSC will lead one of three efforts to generate the sequence and to provide clinical reports for key genes relevant to each participant’s health. The work will be carried out in the Baylor HGSC’s CAP/CLIA certified clinical laboratory and will engage the Johns Hopkins Genomics Center for Inherited Disease Research (CIDR) in Baltimore, led by CIDR director Dr. Kim Doheny, and the University of Texas Health Science Center at Houston’s School of Public Health, led by Dr. Eric Boerwinkle, dean of the UT School of Public Health. As the Baylor-Hopkins Clinical Genomics Center, these three groups bring decades of genomics, clinical and disease research expertise to the All of Us Research Program.

The Baylor HGSC participation follows an intensive period of building certified clinical assays based on genome testing. “The HGSC pioneered comprehensive clinical genome testing in pediatrics here in Houston. Now with the All of Us Research Program, we will have the opportunity to work with these critical technologies in a wide range of adult participants,” said Dr. Richard Gibbs, founding director of the Baylor HGSC and principal investigator of the Baylor-Hopkins Clinical Genomics Center.

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- W. M. Keck Foundation
- The March of Dimes
- The Angelman Syndrome Foundation
- The American Heart Association
- Autism Speaks
The genotyping component of the All of Us Research Program will be the first level of genome data generated for many participants. The data communication and flow to the All of Us Data and Research Center will be a critical aspect of the new period and will draw on the group’s extensive cloud computing experience with commercial partners.

The All of Us Research Program is one of the country’s most ambitious biomedical research efforts ever undertaken. It aims to build a nationwide community of 1 million or more participants from all walks of life, including groups that have been historically underrepresented in research. Eligible adults, ages 18 and up, living in the United States are welcome to participate. As of February 2019, the program reports that more than 175,000 people are registered and more than 105,000 have completed enrollment.

These participants are sharing information through surveys, access to their electronic health records, and by providing biosamples. Over time, they will continue to share information through additional surveys, fitness trackers and more. These data, stripped of obvious identifiers, will be broadly accessible to researchers, whose findings may lead to more tailored treatments and prevention strategies in the future.

“The All of Us Research Program perfectly complements Baylor College of Medicine’s strategic plan, of which precision medicine is foundational. We are honored to be a part of this groundbreaking national program and look forward to contributing to this wonderful scientific resource,” said Dr. Adam Kuspa, senior vice president and dean of research at Baylor.

Jan and Dan Duncan Neurological Research Institute

During the past year, investigators at the Jan and Dan Duncan Neurological Research Institute (NRI) at Texas Children’s Hospital have made many discoveries that enhance our understanding the pathogenesis of neurological disorders. The following are a few examples:

Dr. Benjamin Arenkiel, an associate professor of molecular and human genetics and McNair scholar at Baylor, and his lab recreated trajectories of newly born neurons and pinpointed the genes that they turn on or off during brain circuit formation. This discovery informs strategies for restoring proper brain activity after injury. The Arenkiel lab also identified a neural circuit in the basal forebrain that when impaired, increased food intake, resulting in severe obesity. Thus, charting a path for better understanding and treatment of eating disorders and addiction.
An international team led by Dr. Hugo Bellen, a professor of molecular and human genetics and HHMI investigator at Baylor College of Medicine, and researchers from UCSF discovered that a Zika protein binds to ANKLE2 (The Bellen team previously showed that ANKLE2 mutations cause microcephaly in both humans and fruit flies). These insights open new paths to develop therapeutics for Zika and Dengue virus.

Dr. Juan Botas, a professor of molecular and human genetics at Baylor, along with Dr. Huda Zoghbi, who holds the Ralph D. Feigin, M.D. Endowed Chair at Baylor and is the NRI’s director, and Dr. Zhandong Liu, an associate professor of pediatrics-neurology at Baylor, identified six potentially druggable targets that modulate levels and toxicity of alpha-synuclein (a protein that drives Parkinson disease) in cell lines, fruit flies, mouse brain, and human neurons.

Dr. Marco Sardiello, an assistant professor of molecular and human genetics at Baylor, and his team discovered that the gene, CLN8 is essential for the biogenesis of the lysosome, which clears cellular waste. Identifying this novel mechanism solves a long-term medical mystery and will lead to a better understanding of a range of devastating disorders.

**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 at Baylor.

The CIBR Center is comprised of over 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 over 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 also 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).

**Huffington Center on Aging**

This year marked the 30th anniversary of the Huffington Center on Aging (HCOA) and the Center had much to celebrate. Some of the highlights include:

Dr. Meng Wang, the Robert C. Fyfe Endowed Chair on Aging at Baylor, was named an Investigator of the Howard Hughes Medical Institute. The Center’s overall research funding increased by more than 50 percent over the total reported for 2017. Our faculty published numerous outstanding papers in journals such as Neuron, Developmental Cell, Cell Reports, Cell Metabolism and Journal of Experimental Medicine.

In October, to mark the 30th milestone, the HCOA held a one-day symposium, which included scientific lectures, poster presentations and a community outreach event in the evening that featured Arianna Huffington as the keynote speaker.

In conjunction with the symposium, a Scientific Advisory Board was established to help guide the Center in its programmatic growth.

**Intellectual and Developmental Disabilities Research Center**

The Eunice Kennedy Shriver Intellectual and Developmental Disabilities Research Center (IDDRC) at Baylor, led by Dr. Huda Zoghbi and co-led by Dr. David Nelson, the Cullen Foundation Professor of Molecular and Human Genetics at Baylor, and Dr. Rodney Samaco, an assistant professor of molecular and human genetics at Baylor, is one of 14 centers across the country supporting 57 investigators engaged in basic, translational and clinical studies of intellectual and developmental disabilities (IDDs).

Housed in the Department of Molecular and Human Genetics, the IDDRC’s Core Facilities play a pivotal role in moving novel basic science discoveries ‘at the bench’ into preclinical and eventual clinical trials in humans. The facilities include the Clinical Translational
Research Core led by Dr. Sandesh Nagamani, an associate professor of molecular and human genetics at Baylor, with Dr. Eric Storch, the Neuropathology Core led by Dr. Roy Sillitoe with Drs. Cecilia Ljungberg and Dinghui Yu, the Neuroconnectivity Core led by Dr. Benjamin Arenkiel with Drs. Jennifer Seleve and Jianrong Tang, and the Neurobehavioral Core led by Dr. Rodney Samaco with Dr. Surabi Veeraragavan, an assistant professor in molecular and human genetics at Baylor.

In 2018, 53 studies were published by IDDRC investigators in various medical disciplines. One example is work published in the *American Journal of Human Genetics* from Dr. Christian Schaaf. The study showed that mice lacking *Otud7a*, a gene affected in 15q13.3 microdeletion syndrome, recapitulated key features found in humans with the condition. Having a better understanding of the contributions of each gene in 15q13.3 microdeletion syndrome using mice as a preclinical tool is an important first step in therapy development.

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 at Baylor College of Medicine, and Dr. Florent Elefteriou, an associate professor of molecular and human genetics at Baylor College of Medicine, seeks to improve the prevention and treatment of congenital and degenerative diseases of the skeleton, including skeletal dysplasias, osteoporosis, osteoarthritis, low back pain and bone cancers. The CSMB at Baylor leverages the Rolanette and Berdon Lawrence Bone Disease Program of Texas, a collaboration of Baylor College of Medicine, University of Texas MD Anderson Cancer Center and the University of Texas Health Science Center at Houston, to cultivate teamwork between clinicians, clinical researchers and basic scientists of the Texas Medical Center by sponsoring monthly seminars, pilot grants and core facilities.

The CSMB T-Bone seminar series continues to offer trainees of the Bone Disease Program feedback and discussions on unpublished data, educational talks by the faculty and the opportunity to learn about the latest science and methodologies related to bone and cartilage research in the Texas Medical Center.

The CSMB also sponsors the visit of external speakers who meet with the faculty and trainees and present their research at the Bone Disease Program of Texas seminars. The CSMB also organizes a monthly Bone Club, a venue aimed at promoting interactions and cross-institutions collaborative research in bone and cartilage research.
Genetics Clinics

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 14 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, including Texas Children’s Hospital, Baylor St. Luke’s Medical Center, the Michael E. Debakey Veterans Affairs Medical Center and the Harris Health System.

Pediatric Genetics

The pediatric genetics service provides genetic counseling and inpatient and outpatient care to patients at Texas Children’s Hospital and several other hospitals within the Texas Medical Center and beyond, including Texas Children’s Hospital West Campus and Texas Children’s Hospital The Woodlands. Physicians at the Texas Children’s Genetics Clinic see more than 3,000 families each year.

Specialty clinics within the Texas Children’s Genetics Clinic include the metabolic, neurofibromatosis, skeletal dysplasia and cancer genetics clinics. Genetics physicians and counselors from Baylor also staff joint clinics with other departments, such as otolaryngology (oto genetics) and neurology (neuro genetics/tuberous sclerosis), as well as multidisciplinary teams, such as the Angelman, Craniofacial/Craniosynostosis Clinics and the Gender Medicine Program.

Adult Genetics

The Adult Genetics Clinic is one of the largest genetics clinics in the country providing inpatient and outpatient care and genetic counseling exclusively for adult patients. We see 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 the general genetics clinics, there is also a specialized Ehlers Danlos Syndrome Clinic, a Metabolic and Genetic Disorders of the Bone Clinic and a Cardiomyopathy clinic.
Prenatal Genetics

The Baylor Prenatal and Reproductive Genetics Clinic at Texas Children’s Pavilion for Women is the largest of its kind in the United States. The clinic is comprised of physicians and genetic counselors who specialize in prenatal and reproductive genetic risk assessment and the latest genetic testing technologies.

Genetic counseling is offered to couples who have an increased chance of having a child with a genetic condition or birth defect, women who will be over 35 years of age at the time of delivery, couples who have had multiple miscarriages, couples who are carriers of a genetic condition, and couples who have had abnormal genetic or prenatal screening tests.
Graduate Program

Rigorous Training is Essential for Tomorrow’s Genetic Discoveries

The graduate program provides outstanding educational opportunities for students who wish to pursue a career in the broad and exciting field of genetics. Currently there are 83 students enrolled in the program.

Students are trained by first-class researchers in an unmatched collaborative environment. “Collaborations between different types of researchers prepare our trainees for the challenges of modern biomedical research,” said Dr. Gad Shaulsky, professor of molecular and human genetics and the director of the program. “These collaborations are greatly facilitated by easy access to large genome sequencing and diagnostic datasets that are not available to graduate students elsewhere.”

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 the most respected peer-reviewed scientific journals in the world.

This past year has been spent making preparations to launch the new Graduate Program in Genetics and Genomics. The inaugural class will matriculate in July 2019.

Awards and Special Recognition for MHG Graduate Program Students

Students in the Graduate Program in Molecular and Human Genetics received many recognitions for their hard work in 2018. Here are some of the highlights:

Catherine Bradley was named a McNair M.D./Ph.D. Student Scholar by the Robert and Janice McNair Foundation/McNair Medical Institute M.D./Ph.D. Scholars Program

Lois Dodson received a F31 Fellowship from the National Heart, Lung, and Blood Institute

Adam Hansen and Michael Khayat each received an RFA in Precision Medicine and Population Health Initiative from Baylor College of Medicine

Angad Jolly was an invited platform speaker at the American Society of Human Genetics 68th Annual Meeting

Pamela Lurie received funding on a T32 Hematology Training Program from the National Institute of Diabetes and Digestive and Kidney Diseases

Kristen Meyer received the Family Larsson-Rosenquist Foundation Trainee Travel Fund Award from the International Society for Research in Human Milk and Lactation

Saumya Sisoudiya received a Cancer Research Training Fellowship for 2018-2020 from the Cancer Prevention and Research Institute of Texas (CPRIT)

Xiaofei Song received a registration bursary for the Genomics of Rare Disease Meeting from Wellcome Genome Campus Scientific Conferences

Hui Ye received a Travel Grant Award from International Congress of Parkinson’s Disease and Movement Disorders
Genetic Counseling Program

Promoting excellence in the practice of genetic counseling

The mission of this program is to provide a genomic medicine education promoting excellence in the science of genetics and the practice of genetic counseling across the continuum of care. The interdisciplinary team of clinical, laboratory, and research faculty at Baylor College of Medicine provide experiences that empower graduates to become empathic professionals with effective critical thinking skills.

In February 2018, the Baylor College of Medicine Genetic Counseling Program received “New Program” accreditation status from the Accreditation Council for Genetic Counseling.

The program received 63 applications last year and accepted 8 students for its first class. Since beginning their studies in July 2018, the first class of students have also been participating in community outreach activities as Rare Disease Day volunteers, volunteering at Gigi’s Playhouse (Down Syndrome Achievement Centers) and many have spoken to high school or undergraduate students about the genetic counseling career.

Awards and Special Recognition for Genetic Counseling Program Students

Hannah Helber was named the president of the inaugural Genetic Counseling Program class.

Rachel Thomas was awarded the Public Health Genetics and Precision Medicine Fellowship by the National Society of Genetic Counselors.

Stacey Edwards was selected for the HudsonAlpha Genetic Counseling mini-rotation.
Medical Genetics and Genomics Residency Programs

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

The programs enjoy preeminence in the genetics community and are approved by the Accreditation Council for Graduate Medical Education. The following programs are also supported by a training grant from the National Institute of General Medical Sciences: two-year Medical Genetics and Genomics, four-year Combined Pediatrics and Medical Genetics and Genomics, and four-year Combined Internal Medicine and Medical Genetics and Genomics.

The Department also offers two fellowship programs to residents: the four-year combined fellowship in maternal-fetal medicine and medical genetics and genomics, which consists of 18 months of clinical medical genetics training, 18 months of clinical maternal-fetal medicine training and 12 months of research, and the one-year Medical Biochemical Genetics Fellowship, which is meant to provide specialized training in the diagnosis and management of inborn errors of metabolism.

Clinical Laboratory Fellowship Training Programs

The clinical laboratory fellowship programs provide postdoctoral physician-scientists opportunities to conduct and interpret laboratory analyses useful to the diagnosis and management of human genetic diseases.

Genetics fellows train at Baylor College of Medicine’s genetics diagnostic laboratory, Baylor Genetics, for 24 months. After that period, they are eligible for board certification by the American Board of Medical Genetics and Genomics. Fellowships are offered in the following areas:

LABORATORY GENETICS AND GENOMICS is a newly-designed specialty that incorporates training in both molecular and cytogenetic techniques and interpretations into a single program. The specialty will integrate training in the laboratory assessment of aneuploidies, copy number variants, single nucleotide variants, absence of heterozygosity and abnormal methylation for both constitutional disorders as well as cancers.

CLINICAL BIOCHEMICAL GENETICS is a specialty where trainees spend three months learning each of the following methods: tandem mass spectrometry, gas chromatography/mass spectrometry, high-pressure liquid chromatography (amino acid analysis) and enzyme analysis. Each day, the trainee participates in writing interpretations for all tests with one of the laboratory directors. The remainder of the training is spent developing new diagnostic tests or methodologies for the laboratory or working on a research project.
2018 Graduating Class of Residents and Fellows

Alison Tam, M.D. (Pediatrics Genetics)

Peyman Bizargity, M.D. (Medical Genetics)

Nishitha Pillai, M.D. (Medical Genetics)

Anirudh Saronwala, M.D. (Medical Genetics)

Erin Cooney, M.D. (Medical Biochemical Fellowship)

Anjali Aggarwal, M.D. (Medical Biochemical Fellowship)

Joseph Alaimo (Clinical Molecular Genetics)

Hongzheng Dai (Clinical Molecular Genetics)

Elizabeth Normand (Clinical Molecular Genetics)

Atteeq Rehman (Clinical Molecular Genetics)

Bo Yuan (Combined Clinical Cytogenetics and Clinical Molecular Genetics)

Locations of Former Medical Genetics Trainees
19th Annual Frank Greenberg Memorial Lectureship features Dr. Cynthia Morton

This lectureship was established in memory of Dr. Frank Greenberg, a faculty member in the Department of Molecular and Human Genetics and the Department of Pediatrics at Baylor College of Medicine from 1981 until his retirement in 1994.

Greenberg published more than 100 articles in all areas of clinical genetics and established himself as an expert in contiguous gene deletion syndromes. He contributed to the clinical delineation of a number of congenital chromosomal abnormalities including Prader-Willi, Williams, DiGeorge and Smith-Magenis syndromes. Greenberg was instrumental in the founding of the Williams Syndrome Professional Symposium that brought scientific presentations to the parental support organization of the Williams Syndrome Association National Convention. Greenberg proposed the creation of diagnostic criteria for Williams syndrome, which allowed better assessment of the clinical phenotype.

Through his involvement in the Medical Genetics Training Program at Baylor, Greenberg’s extraordinary abilities in dysmorphology and clinical evaluation contributed to the education of numerous clinical geneticists throughout the world. Greenberg introduced innovative teaching methods, including the use of video to capture physical features, minor anomalies and behavioral characteristics of patients seen during clinical consultations. He will be remembered as a gifted educator, mentor, talented dysmorphologist and an empathetic and caring physician.

Dr. Cynthia Morton was the featured lecturer at the 19th annual Frank Greenberg Memorial Lectureship. The title of her presentation was “A Time to Sequence in Clinical Cytogenetics: The Need for Nucleotide Level Precision.”

Morton is the William Lambert Richardson Professor of Obstetrics, Gynecology and Reproductive Biology and a professor of pathology at Harvard Medical School, Kenneth J. Ryan, M.D. Distinguished Chair in Obstetrics and Gynecology, director of cytogenetics and past director of the Biomedical Research Institute at Brigham and Women’s Hospital. She is an Institute Member of the Broad Institute of MIT and Harvard.

Dr. Morton is a past member of the Board of Directors of the American Board of Medical Genetics where she served as Secretary, Treasurer and Chair of the Accreditation Committee. Dr. Morton was a member of the Board of Directors of the American Society of Human Genetics for 12 years and served as the 2014 President. Dr. Morton is currently a member of the Counsel of Scientific Trustees of the Hearing Health Foundation and Chair of the Veteran’s Administration Genomic Medicine Program Advisory Committee. She completed a six-year tenure as Editor of The American Journal of Human Genetics and is currently Co-Editor of Human Genetics. Dr. Morton is a Fellow of the American Association for the Advancement of Science.

Her research interests are in molecular cytogenetics, hereditary deafness, genetics of uterine leiomyomata and human developmental disorders.
Jeanette Oshman Efron, who passed away in 2009 at the age of 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. Joan Steitz of Yale University, Richard Axel of Columbia University, Joseph Goldstein of UT Southwestern, and Susan Lindquist of MIT are some of our recent guests.

This year, Dr. George Church was the featured speaker. The title of his talk was “Reading and Editing Genomes.”

Dr. George Church is a professor of genetics in the Blavatnik Institute at Harvard Medical School and a professor of Health Sciences and Technology at Harvard and the Massachusetts Institute of Technology (MIT). He is the director of the HMS NHGRI-Center of Excellence in Genomic Science at Harvard Medical School and director of the Personal Genome Project. He was a founding member of the Wyss Harvard Institute of Biologically Inspired Engineering and an associate member of the Broad Institute.

Church leads Synthetic Biology at the Wyss Institute, where he oversees the directed evolution of molecules, polymers, and whole genomes to create new tools with applications in regenerative medicine and bio-production of chemicals. Among his recent work at the Wyss is development of a technology for synthesizing whole genes, and engineering whole genomes, far faster, more accurate, and less costly than current methods.

Church is widely recognized for his innovative contributions to genomic science and his many pioneering contributions to chemistry and biomedicine. In 1984, he developed the first direct genomic sequencing method, which resulted in the first genome sequence (the human pathogen, H. pylori). Church helped initiate the Human Genome Project in 1984 and the Personal Genome Project in 2005. He also invented the broadly applied concepts of molecular multiplexing and tags, homologous recombination methods, and array DNA synthesizers.

Church was featured in TIME Magazine’s annual list TIME 100: The Most Influential People of 2017, under the category of “Titans.”
Community Engagement and Diversity
Baylor shares access to experts through Community Outreach

Since 2006, Baylor College of Medicine’s Department of Molecular and Human Genetics and Texas Children’s Hospital have partnered to host Evenings with Genetics, a free seminar series open to the public. The expertise of a genetics faculty member paired with faculty from another specialty area plus a parent expert speaker are highlighted at each seminar. Topics covered in 2018 include Phelan-McDermid Syndrome, Primary Ciliary Dyskinesia, X-linked Adrenoleukodystrophy, Glycogen Storage Disorders and Angelman Syndrome with 562 family, caregiver and student attendees. In February, Rare Disease Day events were held at 2 locations: the TCH Auxiliary Bridge for patients and healthcare providers as well as an event open to the community at The Health Museum, where 37 rare disease organizations and scientists involved in rare disease research hosted booths. The event also held an “Ask the Expert” session and seminar on the Social Security Determination process.

Statewide genetic outreach, in collaboration with the UT Texas Center for Disability Studies and the Texas Department of State Health Services, has included half-day genetic seminars and resource fairs in seven underserved communities. These community events, in partnership with 20 local organizations, showcased 91 exhibitors and were attended by 353 people that included parents, care providers, and teachers. Subsequently, geneticists have been invited to speak at regional medical / nursing conferences. Seven genetic webinars for healthcare providers offered continuing education for physicians, nurses, social workers and early childhood intervention providers with 268 attending the live webinars and 1216 viewing the archived series.

The recruitment of underrepresented minority students for careers in genetics is active and in addition to speaking with medical students statewide, BCM hosted booths at the Society for Advancement of Chicanos/Hispanics and Native Americans in Science conference, the Annual Biomedical Research Conference for Minority Students, the University of Houston Downtown Scholars Academy STEM Graduate School & Internship Fair and the 2018 Biology Undergraduate Research Symposium at Prairie A&M University. 214 students indicated interest in genetic/genomic careers. Seven students indicated that they will be applying to graduate or medical school and two indicating interest in postdoctoral positions in genetics/genomics.
Faculty Awards and Recognitions

Dr. Lupski recognized for leadership in genetics

At its 68th annual meeting in October, The American Society of Human Genetics (ASHG) honored Dr. James Lupski, Cullen Professor of Molecular and Human Genetics and professor of pediatrics at Baylor College of Medicine, with the 2018 Victor A. McKusick Leadership Award.

This award, named in honor of the late Dr. Victor A. McKusick, recognizes individuals whose professional achievements have fostered and enriched the development of human genetics as well as its assimilation into the broader context of science, medicine and health.

“I knew Victor McKusick quite well and have had many meaningful scientific discussions with him,” said Lupski, who also is the attending medical geneticist at Texas Children’s Hospital and member of the NCI-designated Dan L Duncan Comprehensive Cancer Center at Baylor. “He was a terrific physician-scientist, visionary and true leader, and this award in his name is a tremendous honor for me.”

Lupski’s research focuses on understanding mutational mechanisms and linking specific mutations and genes to human disease. Lupski started his laboratory at Baylor in 1989, where he still resides. His most significant contributions to genomics are centered on conceptualizing and understanding the mechanisms underlying genomic disorders, which is seen through his studies of Charcot-Marie Tooth (CMT) disease – specifically, duplication of the CMT1A gene. In 1991, Lupski showed that CMT1A copy number variation and gene dosage are causes of CMT-related peripheral nerve dysfunction. In 2014, Lupski and colleagues found that the presence of three copies of CMT1A on one chromosome 17, a phenomenon known as triplication, causes a more severe form of CMT. His group was also the first to describe non-allelic homologous recombination as a mechanism for copy number variation formation and chromosomal aberrations.

Lupski received an honorary doctorate in 2011 from the Watson School of Biological Science at the Cold Spring Harbor Laboratory. Lupski has coauthored more than 700 scientific publications, including 88 in The American Journal of Human Genetics, and is a co-inventor on more than a dozen molecular diagnostic patents.
“This award is a well-deserved recognition of Dr. Lupski’s achievements as a physician-scientist and leader in human genetics research,” said Dr. David L. Nelson, professor of molecular and human genetics at Baylor College of Medicine and president of ASHG.

Dr. Lupski also was awarded with the March of Dimes Colonel Harland Sanders Award at the 2018 American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting. This award is given by the March of Dimes and American College of Medical Genetics and Genomics for lifetime achievements in the studies of birth defects.

Dr. Meng Wang named Howard Hughes Investigator

In May of 2018, Dr. Meng Wang, professor in the Huffington Center on Aging and the Department of Molecular and Human Genetics at Baylor, was named a Howard Hughes Medical Institute (HHMI) Investigator at Baylor.

“It is a big honor to be named an HHMI Investigator and join a very prestigious group of scientists,” Wang said. “Being an HHMI Investigator gives me an opportunity to interact with other outstanding scientists in the institute, and I’m really excited.”

Wang and her lab colleagues conduct research that focuses on healthy aging and how it interacts with metabolism. Specifically, she is interested in identifying natural metabolites that derive from metabolic reactions and how these metabolites function can be used as a new target to promote healthy aging.

Wang will have research support from the HHMI in studying metabolic cues in improving longevity and healthy aging over the course of seven years.

“I have been with Baylor for eight years now, and I have received a lot of support from my colleagues, the Huffington Center on Aging, the Department of Molecular and Human Genetics and the College. Without their support, none of this would be possible,” Wang said.

The HHMI Investigator Program supports nearly 300 investigators, located at more than 60 research institutions across the United States, who are widely known for their scientific discoveries, their innovation and their success in pushing the bounds of knowledge in biomedical research.

Wang joins Dr. Huda Zoghbi, director of the Jan and Dan Duncan Neurological Research Institute at Texas Children’s Hospital and professor of molecular and human genetics, Dr. Hugo Bellen, professor of molecular and human genetics, and Dr. Jeffrey Magee, professor of Neuroscience as HHMI Investigators at Baylor.
Dr. Huda Y. Zoghbi, professor and Howard Hughes Medical Institute Investigator at Baylor College of Medicine was elected to the American Academy of Arts and Sciences, one of the nation’s most prestigious honorary titles.

The Academy is one of the country’s oldest societies and independent policy research centers. It recognizes exceptional scholars, leaders, artists and innovators and engages them in sharing knowledge and addressing challenges facing the world. This year, Zoghbi joins along with more than 200 other individuals from a wide range of disciplines and professions as elected members of the Class of 2018.

Zoghbi, who is the director of the Jan and Dan Duncan Neurological Research Institute at Texas Children’s Hospital and professor of pediatrics, molecular and human genetics, neurology and neuroscience at Baylor, is the world’s leading expert on Rett syndrome.

Zoghbi and her research team identified mutations in the MECP2 gene as the cause for Rett Syndrome and revealed the importance of the protein, MeCP2 for the function of various neuronal subtypes. Her work in mouse models showed just how sensitive the brain is to the levels of MeCP2. Too little MeCP2 causes Rett syndrome; doubling MeCP2 levels causes progressive neurological deficits. The latter disorder is now recognized as MECP2 duplication syndrome.

Her discovery provided a straightforward diagnostic genetic test, allowing early and accurate diagnosis and also opened up a new area of research on the role of epigenetics in neuropsychiatric disorders. Her more recent work has shown that symptoms of adult mice modeling the duplication disorder can be reversed using antisense oligonucleotides that normalize MeCP2 levels. This discovery provides a potential therapeutic strategy for the MECP2 duplication syndrome and inspires similar studies for other duplication disorders.

Zoghbi and collaborators also have made many discoveries toward understanding mechanisms driving adult-onset neurodegenerative disorders and are now focused on identifying potential therapeutics for these disorders.

In January, Zoghbi was also awarded with Lebanon’s highest honor, the National Order of the Cedar, Knight grade, by Lebanese President General Michel Aoun.

Born in Beirut, Zoghbi graduated from the American University of Beirut (AUB) in Lebanon with a bachelor of science in 1976, she received her medical degree from Meharry Medical College in Nashville, Tennessee in 1979, and completed her postgraduate training at Baylor College of Medicine in Houston. She also holds honorary doctorates from Middlebury College, Meharry Medical College, and Harvard University.

Zoghbi received this award in a ceremony held on Jan. 15, 2018 at the Presidential Palace in Baabda, Lebanon.

“Dr. Huda Zoghbi is a Lebanese treasure, truly one of the greatest scientists anywhere in the world,” said the Chairman of the AUB, Mr. Philip S. Khoury, “Her devotion to her country of birth and to AUB, the university that educated her, is simply tremendous. We celebrate her as she receives Lebanon’s highest honor.”

Other accolades Zoghbi received this past year include the The Ross Prize in Molecular Medicine by The Feinstein Institute for Medical Research and an Honorary degree from the University of Massachusetts Medical School.
More Awards and Recognitions for MHG Faculty

Erez Lieberman Aiden, Ph.D., assistant professor of molecular and human genetics, was one of four recipients of the 2018 Michael E. DeBakey M.D. Award for Excellence in Research from Baylor College of Medicine.

Shweta Dhar, M.D., associate professor of molecular and human genetics, was awarded the 2018 Clark Faculty Service Award by Baylor College of Medicine.

Hamed Jafar-Nejad, M.D., associate professor of molecular and human genetics at Baylor, was invited to serve as a Standing Member on the Intercellular Interactions (ICI) NIH Study Section.

Richard Lewis, M.D., professor of molecular and human genetics at Baylor, was awarded the Marshall M. Parks Children’s Eye Care Bronze Medal by the American Association for Pediatric Ophthalmology and Strabismus at its 44th Annual Meeting in Washington D.C. He was the first non-pediatric ophthalmologist to receive this honor.

David Nelson, Ph.D., professor of molecular and human genetics at Baylor, was elected President of the American Society of Human Genetics for 2018.

Susan M Rosenberg, Ph.D., Ben F. Love Chair in Cancer Research and professor of molecular and human genetics at Baylor, served as the Honorary Lecturer for the 2018 Dean’s Distinguished Seminar at the University of Colorado School of Medicine.

V. Reid Sutton, M.D., professor of molecular and human genetics at Baylor, was elected to the American College of Graduate Medical Education (ACGME) Board of Directors.

Thomas (Trey) Westbrook, Ph.D., professor of molecular and human genetics at Baylor, was a part of a team led by Harvard’s Stephen Elledge that received a Cancer Research UK Grand Challenge Award.
New Faculty Appointments

Research Faculty

Tao Wu, Ph.D.
Assistant Professor, Tenure-Track

Hongzheng Dai, Ph.D.
Assistant Professor

Chaya Murali, Ph.D.
Instructor

Haley Streff, M.S., C.G.C.
Instructor

Diagnostic Laboratory, Genetic Counseling & Clinical Faculty

Hongzheng Dai, Ph.D.
Assistant Professor

Chaya Murali, Ph.D.
Instructor

Haley Streff, M.S., C.G.C.
Instructor

Ido Machol, Assistant Professor (Not pictured)

Promotions

INSTRUCTOR
Aleksandar Bajic, Ph.D.
Matthew Grol, Ph.D.
Brian Dawson, M.S.

ASSISTANT PROFESSOR
(Non-tenure track)
Ingo Grafe, M.D.
Nunzia Pastore, Ph.D.
Ji-Yoen Kim
Salma Nassef, M.S., C.G.C.

PROFESSOR
(Tenured)
Christophe Herman, Ph.D.
Grzegorz Ira, Ph.D.
Pawel Stankiewicz, M.D., Ph.D.
Meng Wang, Ph.D.