I’m delighted to welcome you to our 2017 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 has continued its accelerated growth and remains the No. 1 ranked genetics department in the country based on total National Institutes of Health (NIH) funding and awarded grants. Our research and clinical initiatives—whether taking place in medicine, pediatrics, obstetrics or one of our 14 specialty clinics—are completely integrated within one department, and the results speak for themselves. Our graduate and residency programs continue to attract the most highly qualified candidates, and we launched our Masters in Genetic Counseling Program. Finally, our clinical genetics program, the largest in the country, continues to offer patients unparalleled, single-source genetic testing and services.

In the past year, we continued to grow our diagnostic laboratory joint venture, Baylor Genetics, with the launch of PreSeek, the first cell-free noninvasive prenatal sequencing panel. In planning for the future of genetic medicine, we continue to develop Consultagene, the Department’s virtual platform for delivery of genetic services including direct-to-patient tele-genetic counseling, peer-to-peer consultation and genomic data interpretation services, which we have field tested locally in Hong Kong and New York. In support of our global expansion, we held our first Baylor College of Medicine and the Chinese University of Hong Kong Center International Symposium on Clinical Genetics in Hong Kong. Most importantly, we continue to make great research discoveries as this constitutes the core foundation of our various missions.

As we take measure of the past year, let us also look forward. The future holds much promise—the talent and dedication of our renowned faculty and trainees, who together advance this Department, will carry it into the next year and beyond. 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

Message from the Chair
Table of Contents

Initiatives and Partnerships — 6
Research and Patient Care — 9
Graduate and Continuing Education — 19
Community Outreach — 24
Faculty — 25
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. Through 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 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.
Vice Chair, Diagnostic Laboratory Affairs

Christine Eng, M.D.
Vice Chair, Medical Educational Affairs

Lorraine Potocki, M.D.
Vice Chair, Medical Educational Affairs

We have more than 550 FACULTY, TRAINEES AND STAFF who occupy 180,000 SQUARE FEET OF SPACE. Faculty includes:

- **4** members of the National Academy of Sciences
- **8** members of the National Academy of Medicine
- **2** Howard Hughes Medical Institute Investigators
- **9** Fellows of the American Association for the Advancement of Science
- **1** Howard Hughes Medical Institute Faculty Scholar

Gad Shaulsky, Ph.D.
Vice Chair, Graduate Educational Affairs

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

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

Department Leadership

We have more than 550 FACULTY, TRAINEES AND STAFF who occupy 180,000 SQUARE FEET OF SPACE. Faculty includes:

- **4** members of the National Academy of Sciences
- **8** members of the National Academy of Medicine
- **2** Howard Hughes Medical Institute Investigators
- **9** Fellows of the American Association for the Advancement of Science
- **1** Howard Hughes Medical Institute Faculty Scholar

Gad Shaulsky, Ph.D.
Vice Chair, Graduate Educational Affairs

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

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

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

Laura Rosales, Ed.D., M.B.A.
Vice Chair, Diagnostic Laboratory Affairs

Christine Eng, M.D.
Vice Chair, Medical Educational Affairs

Lorraine Potocki, M.D.
Vice Chair, Medical Educational Affairs

We have more than 550 FACULTY, TRAINEES AND STAFF who occupy 180,000 SQUARE FEET OF SPACE. Faculty includes:

- **4** members of the National Academy of Sciences
- **8** members of the National Academy of Medicine
- **2** Howard Hughes Medical Institute Investigators
- **9** Fellows of the American Association for the Advancement of Science
- **1** Howard Hughes Medical Institute Faculty Scholar

Gad Shaulsky, Ph.D.
Vice Chair, Graduate Educational Affairs

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

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
When Baylor College of Medicine, together with Baylor Genetics, launched Consultagene in 2016, the vision was to create a personalized web and app-based platform that serves 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, Consultagene has been refining its platform and undergoing pilot testing to integrate new educational and counseling modalities.

The Consultagene platform centers on four modules, which are highly interchangeable and evolve to uniquely serve each patient throughout their Consultagene experience. These four modules include the client engagement module, video module, health history and pedigree module, and the tele-genetic counseling module. Consultagene, importantly, guides the client through the entire process.

“We’ve been testing the Consultagene video modules and have been met with very positive feedback on their efficacy and value of information,” said Dr. Brendan Lee.

“These videos are focused on education, particularly about whole exome sequencing, and enhance the overall user experience and expand on what the platform already offers. We look forward to wrapping up development on these elements and incorporating them into the fully integrated digital platform, which we are aiming to launch in May 2018.”

Consultagene operates through a three-party interaction system, with information flowing to and from the referrer, client and provider in a guided experience. Most commonly, the referrer is a doctor or a genetics lab who is ordering a test for a patient. The client is usually the patient who is seeking genetic testing or counseling, and the provider can be a Baylor genetics physician, a genetic counselor or a clinical diagnostic molecular geneticist.

Consultagene’s platform can be generalized for other specialties in the future, providing this type of seamless experience for practices of all kinds to ultimately improve patient experience and satisfaction, and serve as a platform to advance precision medicine.

“We are also working with some companies and providers to adopt Consultagene as a whole, or package out specific elements to employ just those services that fill a need in their service line,” Lee said.

Consultagene is made possible by the unique environment within the Department of Molecular and Human Genetics at Baylor College of Medicine, which allows for the spirit of innovation to combine with the expertise of the Department’s faculty, whose areas of focus span research, education, medical care, genetic counseling and diagnostic laboratory interpretation.
Baylor Genetics

Baylor Genetics, a joint venture of Baylor College of Medicine and Miraca Holdings, Inc., is a premier health science laboratory with a mission to deliver the world’s highest quality genetics and genomics services in the industry by maximizing academic and commercial synergies.

In order to advance the comprehensive approach Baylor Genetics takes to cancer genomics, it launched a next-generation somatic tumor sequencing test called ClariFind, which correlates genomic results with specific drug therapies and clinical trial options.

ClariFind, developed together with Baylor College of Medicine, is designed to allow the patient and clinician to navigate through the results and corresponding treatment options easily. Each report created through ClariFind is reviewed by board-certified clinical experts and provides explanatory genomic findings, relevant therapies, available clinical trials and a detailed summary.

“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 at Baylor Genetics and vice chair of research affairs in molecular and human genetics at Baylor.

In addition to the introduction of Clarifind, Baylor Genetics is expanding its reach in clinical genomics through joint symposiums in Asia. Most recently, Baylor Genetics co-hosted its first U.S.-Japan Clinical Cancer Genomics and Personalized Medicine Symposium.

The symposium not only provided an educational forum on the implementation of cancer genetic testing in Japan, but also fostered a continued relationship as Baylor Genetics develops a diagnostic testing roadmap catered for use in Japan in the future.

“This is an exciting time, as the Japanese government is leading implementation and support for cancer testing in its healthcare system. The advent of genetic testing may improve detection, diagnosis, choice of therapies and monitoring,” said Dr. Brendan Lee.
First-ever BCM-CUHK Joint Symposium in Clinical Genetics

In May 2017, the Department of Molecular and Human Genetics at Baylor College of Medicine, in partnership with the Department of Obstetrics and Gynecology and the Department of Pediatrics at the Chinese University of Hong Kong, held its first-ever Joint Symposium in Clinical Genetics. In its inaugural year, the symposium took place at the Postgraduate Education Centre in the School of Public Health at the Prince of Wales Hospital in Hong Kong. The goal of the symposium, which will be held annually, is to educate and update clinicians and scientists on the application of clinical genetics to genomic medicine and to highlight cutting-edge technologies and scientific discoveries in clinical genetics and genomics.

The symposium connected experts and leaders in the field from Baylor with those in Hong Kong and across Asia. In addition to the symposium programming, attendees and organizers celebrated the establishment of the joint Baylor-CUHK Center of Medical Genetics, which aims to promote high-quality training and conduct state-of-the-art research in medical genetics.

The symposium played host to key speakers in the field, which included Baylor’s Dr. Igna Van Den Veyver, Dr. Richard Gibbs, Dr. Art Beaudet, Dr. James Lupski and Dr. Brendan Lee, and CUHK’s Dr. Dennis Lo, Dr. Rossa Chiu, Dr. Tak Yeung Leung and Dr. Richard Choy.

The topics of the oral presentations ranged from non-invasive prenatal genetic diagnosis, genomic technologies and the role genetics plays in neurological and cardiovascular diseases to inborn errors of metabolism, genetic screening and counseling and skeletal dysplasia, among others.
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.

Researchers map human genome in 4-D as it folds

For decades, researchers have speculated that when a human cell responds to a stimulus, DNA elements that lie far apart in the genome quickly find one another, forming loops along the chromosome. By rearranging these DNA elements in space, the cell is able to change which genes are active.

In 2014, a multi-institutional team of scientists that included members of the Aiden Laboratory at Baylor College of Medicine showed it was possible to map loops. But the first maps were static, and as a result, scientists lacked the ability to watch the loops change.

In 2017, this same team of scientists created the first high resolution 4-D map of genome folding, tracking an entire human genome as it folds over time. The report detailing this work appeared in the September 2017 issue of *Cell*.

First author, Suhas Rao, described the current approach as “more like making a movie; we can watch folds as they disappear and reappear.”

However, in some cases, loops did the exact opposite of what the researchers anticipated. “As we watched thousands of loops across the genome get weaker, we noticed a funny pattern,” said assistant professor and McNair Scholar Dr. Erez Lieberman Aiden. “There were a few odd loops that were actually becoming stronger. Then, as we put cohesin back, most loops recovered fully—but these odd loops again did the opposite—they disappeared!”

By analyzing how the maps changed over time, the team realized that extrusion was not the only mechanism that brought DNA elements together. A second mechanism, called compartmentalization, which did not involve cohesion, also played a role. “The second mechanism we observed is quite different from extrusion,” explained Rao. “Extrusion tends to bring DNA elements together two at a time, and only if they lie on the same chromosome. This other mechanism can connect big groups of elements to one another, even if they lie on different chromosomes. And it seems to be just as fast as extrusion.”

Study shows accurately transcribing DNA overrides DNA repair

A groundbreaking and surprising discovery provides a major conceptual change of what is most important to cells: the fidelity of the DNA transcription process—accurately copying the DNA message into RNA, the precursor to proteins—or DNA repair, which saves broken chromosomes from being lost. As reported in the October 2017 issue of the journal *Nature*, researchers found that in the model organism *E. coli*, the fidelity of transcribing DNA comes at the expense of DNA repair.

It is well known that DNA breaks are troublesome for cells because, if not repaired correctly, they can cause major instability in the cell’s genes or cell death. In contrast, errors during transcription are generally considered less important because the transcript is temporary, and if one is defective, cells can make another one. For these reasons, most researchers consider that DNA break repair would outweigh transcription to protect DNA integrity, and keep cells from losing their chromosomes.

The laboratory of Dr. Christophe Herman has been studying the fidelity of transcription for the past 12 years. “We showed years ago that transcription errors can lead to heritable changes,” said Herman, an associate professor...
of molecular and human genetics and member of the Dan L. Duncan Comprehensive Cancer Center at Baylor. “That made us think that transcription fidelity might be more important than we had originally thought.”

In the study, the research team wanted to investigate the consequences of removing GreA, a factor that helps ensure the fidelity of the transcription process on the bacterium, *E. coli*, on DNA break repair.

“After removing GreA, bacteria were hundreds of times more efficient at repairing DNA damage caused by drugs that mimic radiation,” said first author Dr. Priya Sivaramakrishnan, a Ph.D. student in the Herman lab during the development of this project. “Bacteria can repair DNA breaks much faster when GreA is absent.”

Using a novel whole genome sequencing method named eXOnucleases sequencing (XO-seq) along with other methods, the researchers determined the molecular mechanism by which loss of GreA promotes DNA repair.

The finding that GreA prevents DNA repair represents a major paradigm shift in the DNA world because it implies that ensuring proper transcription fidelity comes at the cost of lowering the cell’s ability to repair DNA. “That was completely unexpected,” Herman said.

“To have a process that helps transcribe DNA into high-quality RNA that will produce high-quality proteins, bacteria are paying a hundred-fold price in DNA repair efficiency,” said co-author Dr. Susan Rosenberg, Ben F. Love Chair in Cancer Research and professor of molecular and human genetics at Baylor.

“The conservation of the basic biology of nucleic acids from bacteria to humans is tremendous,” said Rosenberg, who is also the leader of the Cancer Evolvability Program in the Dan L. Duncan Comprehensive Cancer Center. “We hypothesize that this mechanism discovered in *E. coli* might also be present in other cells, which would have implications in a number of fields, from cancer to evolution.”

**Study reveals benefit of exome sequencing for infants in ICU**

When infants are admitted into intensive care units, physicians and care teams must move quickly to make a diagnosis and begin the best course of treatment. However, in infants suspected to have a genetic disorder, testing can take months, losing valuable time and causing stress to the patient and family. A team of physicians and researchers from Baylor College of Medicine, Texas Children’s Hospital and Baylor Genetics designed a study to determine the efficacy of exome sequencing in ICUs. The results appeared in the December 2017 issue of *JAMA Pediatrics*.

“In our study, we were looking to answer two key questions: can we appropriately diagnose infants with genetic disorders in critical care units using exome sequencing, and will our findings result in changes to their medical care,” said Dr. Seema Lalani, associate professor of molecular and human genetics at Baylor.

“There are times when we see babies who have various problems, but we aren’t sure what the underlying diagnosis is. Through this research, we were hoping to address these diagnostic barriers through whole exome sequencing,” added Dr. Mohan Pammi, associate professor of pediatrics – neonatology at Baylor.

Exome sequencing is a genomic technique for sequencing all of the protein-coding genes in a genome, known as the exome. The exome, which consists of about 20,000 genes, is where the majority of known defects occur.

The research team performed clinical exome sequencing for 278 unrelated infants who were 100 days old or younger. “Infants were selected for exome sequencing based on several characteristics we know to be associated with genetic disorders. Many of them had serious birth defects due to genetic changes that could not be easily diagnosed by clinical exam,” Lalani said.

These characteristics included neuromuscular diseases, syndromic congenital cardiovascular malformations, hypertrophic cardiomyopathy, skeletal malformations, neonatal cholestasis and liver failure, lung disease and metabolic disorders.

Of the 278 infants, a molecular diagnosis was found in 102 cases through clinical exome sequencing. Diagnosis resulted in altered medical management for 53 of these infants ranging from redirecting
their care, bringing in a new subspecialist, making dietary changes and even furthering life-saving procedures in some cases.

Sixty-three of the patients were tested using critical trio exome sequencing specifically, resulting in a diagnosis for 32 infants in this group. Of these 32 infants, diagnosis altered clinical care in 23 cases.

“Clinical exome sequencing is a powerful tool for physicians, especially in caring for critically ill pediatric patients with congenital diseases. It provided definitive diagnosis to about half of the patients in our cohort at a very early age and has played an important role in facilitating decision-making in the medical management of these patients,” said Dr. Linyan Meng, assistant professor of molecular and human genetics at Baylor and co-first author of the study.

Gut bacteria might one day help slow down aging process

Slowing down the aging process might be possible one day with supplements derived from gut bacteria. Scientists at Baylor College of Medicine and the University of Texas Health Science Center at Houston have identified bacterial genes and compounds that extend the life of and also slow down the progression of tumors and the accumulation of amyloid-beta, a compound associated with Alzheimer’s disease, in the laboratory worm *C. elegans*. The study appeared in the journal *Cell*.

“The scientific community is increasingly aware that our body’s interactions with the millions of microbes in our bodies, the microbiome, can influence many of our functions, such as cognitive and metabolic activities and aging,” said corresponding author Dr. Meng Wang, associate professor of molecular and human genetics at Baylor and the Huffington Center on Aging. “In this work, we investigated whether the genetic composition of the microbiome might also be important for longevity.”

This question is difficult to explore in mammals due to technical challenges, so the researchers turned to the laboratory worm *C. elegans*. To study the effect of individual bacterial genes on the lifespan of *C. elegans*, Wang joined efforts with Dr. Christophe Herman, associate professor of molecular and human genetics at Baylor, and other colleagues who are experts in bacterial genetics. They employed a complete gene-deletion library of bacterium *E. coli*; a collection of *E. coli*, each lacking one of close to 4,000 genes.

“We fed *C. elegans* each individual mutant bacteria and then looked at the worms’ life span,” Wang said. “Of the nearly 4,000 bacterial genes we tested, 29, when deleted, increased the worms’ lifespan. Twelve of these bacterial mutants also protected the worms from tumor growth and accumulation of amyloid-beta, a characteristic of Alzheimer’s disease in humans.”

Further experiments showed that some of the bacterial mutants increased longevity by acting on some of the worm’s known processes linked to aging. Other mutants encouraged longevity by over-producing the polysaccharide colanic acid. When the scientists provided purified colanic acid to *C. elegans*, the worms also lived longer. Additionally, colanic acid showed similar effects in the laboratory fruit fly and in mammalian cells cultured in the lab.

The researchers propose that, based on these results, it might be possible in the future to design preparations of bacteria or their compounds that could help slow down the aging process.

Interestingly, the scientists found that colanic acid regulates the fusion-fission dynamics of mitochondria, the structures that provide the energy for the cell’s functions.

“These findings are also interesting and have implications from the biological point of view in the way we understand host-microbe communication,” Wang said. “Mitochondria seem to have evolved from bacteria that millions of years ago entered primitive cells. Our finding suggests that products from bacteria today can still chime in the communication between mitochondria in our cells. We think that this type of communication is very important and here we have provided the first evidence of this. Fully understanding microbe-mitochondria communication can help us understand at a deeper level the interactions between microbes and their hosts.”
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 (NIH) 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 seven 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 2017, the amount in funding dollars from NIH awards totaled more than $64 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
Grant to compare large-scale genomic sequencing, standard clinical tests for childhood cancer patients

Baylor College of Medicine is one of six U.S. institutions to receive a grant through the National Human Genome Research Institute’s (NHGRI) Clinical Sequencing Evidence-Generating Research Consortium, or CSER2. The four-year grant, including $2.8 million for fiscal year 2017, co-funded by the National Cancer Institute, will support Baylor’s new KidsCanSeq program that compares the results of large-scale genomic testing, such as whole exome sequencing, to targeted clinical tests in childhood cancer patients at five sites across the state that serve a highly diverse patient population, including Texas Children’s Cancer Center.

In addition to Texas Children’s Cancer Center, pediatric patients will be enrolled in KidsCanSeq at the Vannie E. Cook Children’s Cancer Clinic in McAllen, the Children’s Hospital of San Antonio, The University of Texas Health Science Center at San Antonio and Cook Children’s Health Care System in Fort Worth.

KidsCanSeq follows the Baylor Advancing Sequencing in Childhood Cancer Care (BASIC3) study at Baylor and Texas Children’s Cancer Center, which developed the initial protocols for performing clinical genomic testing of pediatric cancer patients, reporting results and communicating those results to families and oncologists. BASIC3 was part of the NHGRI Clinical Sequencing Exploratory Research program, a precursor to CSER2.

“Through BASIC3 we explored broad questions, such as whether we could conduct large-scale genomic testing in a clinical setting, what kind of results it would generate, and how to communicate the results to families and physicians. KidsCanSeq is focused more on generating specific data on what tests are better or worse than standard tests in pediatric cancer patients,” said the study’s principal investigator Dr. Sharon Plon, professor of pediatrics and of molecular and human genetics at Baylor, and director of the Cancer Genetics Clinical and Research Programs at Texas Children’s Hospital.

“BASIC3 was essentially a pilot study, and now that we have a better idea of how to implement broad-scale genetic testing in the clinic, we can focus this study more specifically on determining which patients would be most likely to benefit from it or for whom it would be most likely to impact care,” said Dr. Will Parsons, co-principal investigator and associate professor of pediatrics at Baylor and Texas Children’s Cancer Center. “For example, tumor sequencing of cancer types for which kids are almost always cured at the time of diagnosis is not likely to be as useful as for high-risk and relapsed cancers.”

During the study, the comprehensive set of genomic tests will be performed by a unique collaboration between multiple diagnostic facilities with the involvement of Dr. Richard Gibbs, director of the Human Genome Sequencing Center, Drs. Christine Eng and Shashikant Kulkarni, all professors of molecular and human genetics at Baylor, and Dr. Angshumoy Roy, assistant professor of pathology & immunology at Baylor and Texas Children’s Hospital.

The program, in which about 900 patients are expected to be enrolled over four years, also will include parent and doctor surveys to determine what they found most useful from the testing as well as the development of video and other educational materials in both English and Spanish. Understanding differences among families from different ethnic or racial backgrounds as well as in different healthcare settings, including large academic medical centers versus smaller clinical settings, also is a goal of KidsCanSeq.

Dr. Amy McGuire, Leon Jaworski Professor of Biomedical Ethics and director of the Center for Medical Ethics and Health Policy at Baylor, also is co-principal investigator of the study. She will investigate the ethics and utility of genomic testing for pediatric cancer patients.

“It is important to study the clinical and psychosocial risks and benefits of any new technology in order to plan for its responsible use,” McGuire said. “We also want to make sure the infrastructure is in place so that oncologists in non-academic settings can understand, effectively communicate and appropriately manage the results of germline and tumor whole exome sequencing.”
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.

Jan and Dan Duncan Neurological Research Institute

The Jan and Dan Duncan Neurological Research Institute (NRI) at Texas Children’s Hospital is a basic research institute committed to understanding the pathogenesis of neurological disorders in order to develop effective treatments. The goal is to reduce the temporal and conceptual gap between initial gene discovery and clinical application. Cross-species genetic studies are proving to be a powerhouse for solving medical genetic puzzles.

A collaborative study in the NRI lab of Dr. Michael Wangler, assistant professor of molecular and human genetics at Baylor, published in *PLoS Genetics*, helped physicians determine a unique treatment plan for an ataxia patient carrying a toxic, gain-of-function mutation in a calcium channel gene, CACNA1A (also known as cacophony).

The NRI’s Dr. Joshua Shulman, an associate professor of molecular and human genetics at Baylor, led an international scientific team in a study that sheds new light on genetic factors associated with Parkinson’s disease (PD). Published in *Brain*, investigators found that genes previously implicated in lysosomal storage disorders were also potential major contributors to the onset and progression of PD.

A study from the lab of Dr. Juan Botas, professor of molecular and human genetics at Baylor, published by *eLife*, uncovered a potential strategy against Huntington’s disease. Using fruit fly models, the lab showed that silencing the activity of the enzyme PIP4Kγ reduced the accumulation of Huntington’s disease protein, lessening neuronal damage and improving the fly’s ability to move.

In a collaborative study published in *Cell*, the lab of Dr. Huda Zoghbi, a Howard Hughes Medical Institute investigator and professor of molecular and human genetics who holds the Ralph D. Feigin, M.D., Endowed Chair at Baylor College of Medicine and also serves as the director of the NRI, demonstrated that mutations that alter the protein levels of human PUMIL101 (PUMI) can cause a variety of neurological symptoms ranging from childhood-onset ataxia to mild adult-onset ataxia, intellectual disability, developmental delay and seizures. This is the first study to uncover a role for PUM1 in human neurological disorders, providing novel insights into how PUM1 levels regulate human brain function. The study highlights the importance of investigating the role of other RNA-binding proteins in neurodegenerative disorders such as amyotrophic lateral sclerosis (ALS) and Parkinson’s disease.

Computational and Integrative Biomedical Research Center

The Computational and Integrative Biomedical Research Center (CIBR), directed by Dr. Olivier Lichtarge, who is the Cullen Chair and Professor
of molecular and human genetics at Baylor, is a resource to help students and faculty address the broad range of analytical problems posed by the complexity of high throughput biological datasets. The goal of the center is to help bridge the translational gap from data to models, and from models to drug discovery and personalized therapy by fostering collaborations and developing original quantitative approaches to biological and clinical problems.

To assist students and faculty with their research and education, the CIBR Center provides multiple resources, including consultations, seminars, computational workshops, seed grants, hardware and access to software such as MathWork’s MatLab, Wolfram Mathematica and several software applications developed by CIBR faculty members.

Human Genome Sequencing Center

Baylor’s Human Genome Sequencing Center (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 HGSC has since expanded its research focus into new and exciting areas.

Following the news last year that named Baylor’s HGSC as a participant in the National Heart Lung and Blood Institute’s (NHLBI) Trans-Omics for Precision Medicine (TOPMed) program, the HGSC has continued to put its indelible stamp on research activity and discovery. Among these activities, the HGSC played a role in sequencing the genomes of the world’s most destructive caterpillar pests of broad-acre crops and the whole genome of the common white-tailed deer. The deer genome has the potential to provide insights into bone behavior, more specifically how deer are able to regenerate and repair bone.

Huffington Center on Aging

A core value of the Huffington Center on Aging is to strive for excellence in aging and age-related disease research. The Center, directed by Dr. Hui Zheng, the Huffington Foundation Endowed Chair in Aging at Baylor, reached a new level of growth last year.

Following the remarkable accomplishments of Drs. Meng Wang and Melanie Samuel, who were recipients of the NIH Directors Pioneer Award and New Innovator Award, respectively, the Center received four new NIH R01 grants in 2017. Taking advantage of the increased Alzheimer’s disease funding from the NIH, Zheng is leading multiple efforts to begin a research program focused on Alzheimer’s disease at the institutional level.

On the publication side, multiple papers were published by the Center faculty, a highlight being Dr. Meng Wang’s paper in Cell in which she discovered that a probiotic compound secreted from bacteria can promote longevity in C. elegans. Overall, the Huffington Center on Aging is poised for continued success in the coming year (see page 11).

Intellectual and Developmental Disabilities Research Center

The Eunice Kennedy Shriver Intellectual and Developmental Disabilities Research Center (IDDRC) at Baylor College of Medicine, led by Dr. Huda Zoghbi and co-led by Drs. David Nelson and Rodney Samaco, is one of 14 research centers across the country supporting 64 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, 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 Selever and Jianrong Tang, and the Neurobehavioral Core led by Dr. Rodney Samaco with Dr. Surabi Veeraragavan.

This centralized network of services and expertise complemented by state-of-the-art equipment, allows investigators to maximize efficiency without compromising quality, leverage innovative technologies to address the complexities of human health and nurture the next generation of IDD researchers through training and mentorship opportunities provided by Center and Core Leadership.
In 2017, 50 studies were published by IDDRC investigators in the areas of genetics, neuroscience, developmental biology, cancer biology and various medical disciplines. One example is work published in *Nature Genetics* from Dr. Huda Zoghbi with contributions from Drs. Roy Sillitoe, Cecilia Ljungberg and Christian Schaaf, where it was shown that selectively deleting the cancer metastasis gene Capicua from areas of the mouse brain, including the hypothalamus, resulted in ADHD- and autism-like features in mice. These mouse model studies prompted an international search leading to the identification of Capicua mutations in people with ADHD, DD/ID, ASD and seizures.

**Center for Skeletal Medicine and Biology**

The Center for Skeletal Medicine and Biology (CSMB), co-directed by Drs. Brendan Lee and Florent Elefteriou, seeks to improve the prevention and treatment of congenital and degenerative diseases of the skeleton, including skeletal dysplasias, osteoporosis, osteoarthritis 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, UT MD Anderson Cancer Center and the University of Texas Health Science Center at Houston, to cultivate teamwork between clinicians, clinical researchers and basic researchers within the Texas Medical Center by sponsoring monthly seminars, pilot grants and core facilities.

TMC trainees interested in musculoskeletal biology also receive support from the CSMB T-Bone seminar series, where they can receive feedback on unpublished data, attend educational talks by expert faculty and learn about the latest science and methodologies in the Bone Disease Program. This year, the micro Computed Tomography (mCT) imaging facility was upgraded to accommodate the increasing demand for usage.

**Department Research Core Services**

Established this past year, the following research cores in the Department of Molecular and Human Genetics were developed to complement the Baylor College of Medicine Advanced Technology Cores (ATC) by providing services not available via the ATC.

**The Kenneth Scott cDNA Clone Collection**

Originally curated at Baylor College of Medicine by our late colleague Dr. Kenneth Scott, the cDNA collection contains over 32,000 clones. These fully sequenced clones are sourced from the Thermo-Scientific Ultimate™ ORF collection (IOH clones) and from the hORFeome V8.1 libraries (Broad clones). Each clone contains an open reading frame (ORF) sub-cloned into Gateway™ recombinational entry vector for ease of cloning into a Gateway™ destination vector that suits the experimental purpose.

**Baylor Genetics Research Core**

Baylor Genetics is the joint venture diagnostic laboratory of Baylor College of Medicine and Miraca Holdings. The diagnostic laboratory supports the academic mission of Baylor College of Medicine and provides CLIA/CAP approved clinical services for research programs.
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 span 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 3,500 families each year.

Specialty clinics within the Texas Children’s Genetics Clinic include the metabolic, neurofibromatosis, skeletal dysplasia and cancer genetics clinics. Baylor genetics physicians and counselors also staff joint clinics with other departments, such as otolaryngology (otogenetics) and neurology (neurogenetics), as well as multidisciplinary teams, such as the Craniofacial Program and the Program for Gender Medicine.

Adult Genetics

The Adult Genetics Clinic provides 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 clinic, there is also a specialized Ehlers Danlos Syndrome Clinic, a Metabolic and Genetic Disorders of the Bone Clinic and a Neurogenetics Clinic.
Prenatal Genetics

The Prenatal Genetics Clinic, the largest of its kind in the United States, specializes in prenatal and reproductive genetic risk assessments, as well as 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 in Molecular and Human Genetics in the Baylor College of Medicine Graduate School of Biomedical Sciences, led by Dr. Gad Shaulsky, professor of molecular and human genetics, provides outstanding educational opportunities for students who wish to pursue a career in the broad and exciting field of genetics.

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 Shaulsky. “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.

Awards and Special Recognition for MHG Graduate Program Students

Graduate students in the Molecular and Human Genetics Graduate Program received many recognitions for their hard work in 2017. Here are some of the highlights:

Vitaliy Bondar was selected to present at the 2017 Keystone Conference in Colorado

Lois Dodson was awarded first place for her poster at the 2017 Texas Children’s Hospital Research Symposium Trainee Poster Presentation

Patrick Hunt was named the 2017 Metals Service Center Institute Rene Morrison Foundation Scholar

Kristen Meyer was awarded a Ruth L. Kirschstein National Research Service Award for an Individual Predoctoral MD/PhD F30 Fellowship (2017-2020)

Prasanna Ramachandran was an invited speaker and received the Best Abstract Award at the 2017 MD-PhD National Student Conference

Xiaofei Song was selected as the recipient of the 2017 Bravo Award by fellow students in the Molecular and Human Genetics Graduate Program

Li Wang received a travel scholarship for the 2017 Keystone Symposium on Synapses and Circuits: Formation, Function and Dysfunction, and was named the Beckman Platform Award Speaker for best platform talk among all the graduate program speakers at the 2017 Baylor Graduate Student Symposium.

Ruofan Yu received the 2017 Society of Chinese Bioscientists in America International Symposium Travel Award.
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.

Program milestones this past year have included the development and rollout of a website, application portal development and recruitment activities, such as a webinar, which attracted 42 prospective applicants. The genetic counseling program development team’s hard work paid off with the receipt of the 2017 Annual RIITE Role Model of the Year award in Teamwork. The RIITE Role Model program honors current faculty and staff whose sustained high performance and exceptional contributions to Baylor directly or indirectly advance the College’s mission and values.

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 is led by program director, Daniel Riconda, M.S., C.G.C., assistant program director, Salma Nassef, M.S., C.G.C., and medical director, Lindsay Burrage, M.D., Ph.D., all of whom are faculty in the Department of Molecular and Human Genetics. The program anticipates matriculating their first cohort of eight students in July of 2018.
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.

Locations of Former Medical Genetics Trainees

2017 Graduating Class

Mohammed Almannai, M.D.  
Erin Cooney, M.D.  
Xiaoyan Ge, Ph.D.  
Leroy Hubert, Ph.D.  
Keren Machol, M.D.  
Elie Moussallem, M.D.  
Juanita Neira Fresneda, M.D.  
Andrea Petersen, M.D.  
Teresa Sim, Ph.D.  
Mari Tokita, M.D.  
Dihong Zhou, M.D.
Frank Greenberg Memorial Lectureship

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 received his B.A. in Zoology from the University of Michigan and an M.S. from Rutgers Medical School. Greenberg obtained his M.D. from the University of Pennsylvania. After his pediatric residency and fellowship training in genetics at St. Christopher’s Hospital for Children in Philadelphia, he worked at the Birth Defects Branch of the Center for Disease Control as an Epidemiological Intelligence Officer and was a Clinical Assistant Professor in Pediatrics at Emory University in Atlanta.

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. Stefan Mundlos was the featured lecturer at the 18th annual Frank Greenberg Memorial Lectureship. The title of his presentation was “Our Genome in 3-D—How Structural Variations Influence the Folding of DNA.”

Brenda Finucane was the featured lecturer at the 18th annual Frank Greenberg Memorial Lectureship.

18th Annual Frank Greenberg Memorial Lectureship features Brenda Finucane

Brenda Finucane, MS, is a licensed genetic counselor and the associate director of the Autism & Developmental Medicine Institute at Geisinger Health System in Lewisburg, Pennsylvania. Her clinical and research activities have focused on genetic neurodevelopmental disorders. Finucane has particular expertise in the behavioral and cognitive manifestations of fragile X, Smith-Magenis, 15q duplication and other syndromes that result in complex intellectual and behavioral symptoms. Her experience with this population includes young children through adults, and she has a specific interest in research related to the natural history of behavioral phenotypes over the lifespan. A current area of inquiry at Geisinger is the study of family genomic background on phenotypic expression of neuropsychiatric symptoms in individuals with pathogenic copy number and sequence level variants. Finucane has been in leadership roles in many professional and advocacy organizations throughout her career and is a past president of the National Society of Genetic Counselors.
Dealing with the diagnosis of a developmental or genetic disorder can be a difficult and often confusing time for both parents and their children.

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 seminar series that is free and open to the public. A different rare disease or genetic condition is discussed at each session, combining the expertise of a faculty member from genetics and from a specialty area involved in care and treatment of the disease.

“Many times, parents and children, and even adult patients, have never met another person with their same condition,” said Susan Fernbach, R.N., B.S.N., director of the Office of Community Engagement and Diversity within the department and the co-founder of the series along with Dr. Arthur Beaudet. “With this outreach, we are able to connect patients and families with others similarly affected and with medical specialists.”

Between Houston and statewide sessions, Evenings with Genetics has reached over 8,000 people and more than 3,000 healthcare providers. “It’s a unique program that reaches a broad audience of parents, caregivers and even students who have access to experts they wouldn’t otherwise,” said Dr. Daryl Scott, associate professor of molecular and human genetics at Baylor. “It’s not geared to physicians and researchers. In fact, anyone can benefit from the information, even healthcare professionals.”

Some topics generate a high level of interest and are addressed at Evenings with Genetics regularly, including updates on autism research, Down syndrome and new genetic testing available for children with developmental conditions. Other sessions take on much more rare diseases and conditions, such as brittle bone disease, Marfan syndrome and neurofibromatosis.

Fernbach said the goal remains to provide practical information people can leave with that will help them, and to reach as many people as possible. “What’s made this program possible is the wealth of expertise at Baylor and Texas Children’s and people who are willing to share it in an understandable format,” she said. “When you see next steps developing from these seminars, it’s very rewarding.”
Symposium

Harnessing Insights from Human Genetics — Celebrating 30 Years of the Lupski Lab

Dr. James Lupski, the Cullen Professor of Molecular and Human Genetics and professor of pediatrics, has had a game-changing impact on contemporary biomedical research. His achievements include a body of work illuminating the importance of genomic disorders—a term he coined—and the mechanisms underlying structural variant mutagenesis and rearrangements of the human genome, which explain many human diseases. He is an accomplished scientist who earned national distinction for his groundbreaking work when he identified the first gene to be associated with Charcot-Marie-Tooth (CMT) syndrome, a nerve disorder with which he is afflicted. The duplication of the gene, PMP22, is responsible for a high percentage of CMT cases.

Twenty years later, by sequencing his own genome, Lupski was able to identify the gene responsible for his own CMT disease. Though the culprit gene SH3TC2 had been previously associated with CMT, this pioneering work helped further define clinical genomics and provided a glimpse into the future of still unrealized breakthroughs stemming from the Human Genome Project.

Lupski went to New York University on a full scholarship, where he majored in chemistry and biology, and minored in mathematics and psychology. He was accepted to NYU Medical School and then to the MD/PhD program at NYU. He wrote his doctoral thesis on the \textit{rpsU-dnaG-rpoD} macromolecular synthesis operon after cloning one of the first DNA replication genes. The mapping of the gene associated with Huntington disease inspired him to search for the CMT disease gene(s). He was recruited by Baylor College of Medicine where he was given a faculty appointment and the privilege to write his own grants, while he was still a resident. In 1986, he set up his own laboratory and began his research into the genetics of CMT by studying many large families and patients in Louisiana. Eventually, Lupski patented a diagnostic test for CMT and continued his research on the disease.

Lupski is an elected member of the American Association for the Advancement of Science (1996), the American Society of Clinical Investigation (1998), the National Academy of Medicine of the National Academies of Science (2002) and the American Academy of Arts and Sciences (2013). For his work in human genomics and the elucidation of genomic disorders, Lupski was awarded a Doctor of Science degree honoris causa in 2011 from the Watson School of Biological Science at the Cold Spring Harbor Laboratory where he was given the honor to deliver the commencement speech. He has coauthored over 700 scientific publications, is a co-inventor on more than 20 patents regarding molecular diagnostics and has delivered over 500 invited lectures in 37 countries.

The symposium “Harnessing Insights into Human Genetics—Celebrating 30 Years of the Lupski Laboratory” was held on Nov. 3, 2017. The event was organized by prominent alumni from his laboratory, including Dr. James Versalovic, Pathologist in Chief at Texas Children’s Hospital and Professor in the Department of Pathology and Immunology. Major support for the symposium was generously provided by Regeneron, Lasergen, Baylor Genetics - A Miraca Company, and the Department of Pathology at Texas Children’s Hospital and the Department of Molecular and Human Genetics at Baylor College of Medicine. The keynote speaker was Dr. Matthew Hurles, who is the head of the Human Genetics Programme and senior group leader at the Wellcome Trust Sanger Institute in Cambridge, United Kingdom.
Faculty Awards and Recognitions

Beaudet receives 2017 Victor A. McKusick Leadership Award

In 2017, The American Society of Human Genetics (ASHG) honored Dr. Arthur L. Beaudet with its annual 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.

“It is an honor to accept the 2017 McKusick Award,” said Beaudet. “The American Society of Human Genetics is a prominent organization for genetics specialists all over the world, and I am proud to join the ranks of past award winners, all of whom have contributed significantly to the field.”

In the 1980s, Beaudet and colleagues were the first to document uniparental disomy, a phenomenon in which a person receives two copies of a chromosome from one parent and zero from the other. In the following years, they drew an important distinction between genetic and epigenetic diseases that both lead to altered expression of the same genes and identified ways to study these and better understand the conditions they caused. Currently, his research focuses on neuronal carnitine deficiency as a risk factor for autism; the role of genomic imprinting in diseases such as Prader-Willi syndrome, Angelman syndrome and autism; and prenatal genetic diagnosis based on fetal cells isolated from maternal blood.

In addition to his scientific leadership, ASHG also honors Beaudet’s contributions to the Society as well as the broader research community. A longtime member of the ASHG, he belonged to its Program Committee from 1984-86, its Board of Directors from 1987-90, and its Awards Committee from 2010-12, and served as President in 1998. He received the Society’s William Allan Award in 2007 and belonged to the Editorial Board of the ASHG-published *The American Journal of Human Genetics* from 1986-1989. In addition, he was awarded the Texas Genetics Society Barbara H. Bowman Award in 1999 and the March of Dimes’ Colonel Harland Sanders Award for Lifetime Achievement in Genetic Research and Education in 2002. He has published more than 350 articles in scientific literature.

“Dr. Beaudet’s outstanding leadership in human genetics has transcended all aspects of the academic mission from clinical care, education and training, to basic and translational research,” said Dr. Brendan Lee, an ASHG Executive Committee member.
Zoghbi honored with Canada International Gairdner Award

In 2017, Dr. Huda Zoghbi was honored by the Gairdner Foundation with the 2017 Canada Gairdner International Award for seminal discoveries and contributions to biomedical science. This award recognizes some of the most significant medical discoveries from around the world.

Zoghbi was one of five individuals given this award. She was recognized for her work surrounding the discovery of the gene responsible for Rett syndrome.

After years of treating patients, Zoghbi began to focus on finding the genetic causes of the disease, which strikes after a year of normal development and presents with developmental regression, social withdrawal, loss of hand use and compulsive wringing of the hands, seizures and a variety of neurobehavioral symptoms.

The MECP2 gene provides instructions for making a protein called MeCP2. Zoghbi identified that mutations in this gene are the cause of Rett syndrome, revealing the importance of MeCP2 for the function of various neuronal subtypes. Her work showed just how sensitive the brain is to the levels of MeCP2, and that doubling MeCP2 levels causes progressive neurological deficits. This disorder is now recognized as MECP2 Duplication Syndrome in humans.

Her recent work has shown that the symptoms of adult mice modeling the duplication disorder can be reversed using antisense oligonucleotides that normalize MeCP2 levels.

The discovery of the Rett syndrome gene provided a straightforward diagnostic genetic test, allowing early and accurate diagnosis of the syndrome. It also revealed that mutations in MECP2 also can cause a host of other neuropsychiatric features ranging from autism to juvenile onset schizophrenia. Further, it provided evidence that an autism spectrum disorder or an intellectual disability disorder can be genetic even if it is not inherited.

Her discovery has opened up a new area of research on the role of epigenetics in neuropsychiatric phenotypes. Her use of an antisense oligonucleotide to lower MeCP2 levels provides a potential therapeutic strategy for the MECP2 duplication syndrome and inspires similar studies for other duplication disorders.

Zoghbi was also the recipient of other accolades this year. She received an honorary doctorate from Harvard and was named the winner of the 2017 George W. Jacoby Award by the American Neurological Association.
More Awards and Recognitions for MHG Faculty

Benjamin Russell Arenkiel, Ph.D., a McNair Scholar, received both the Outstanding Educator Award in Neuroscience and the Michael E. DeBakey Excellence in Research Award from Baylor College of Medicine.

Carlos Bacino, M.D., was a recipient of a Master Clinician Lifetime Award from Baylor College of Medicine.

Hugo Bellen, D.V.M., Ph.D., was the Seymour Benzer Keynote Speaker at the 2017 Neurobiology of Drosophila Meeting held in Cold Spring Harbor.

Lindsay Burrage, M.D., Ph.D., received a 2017 Burroughs Wellcome Fund Career Award.

Brendan Lee, M.D., Ph.D., received the Baylor College of Medicine Alumni Association Medical School Distinguished Alumnus Award.

Charles Lin, Ph.D., was named one of five 2017 Pew-Stewart Scholars for Cancer Research.

Hamed Jafar Nejad, Ph.D., was given the 2017 Glycobiology Significant Achievement Award by the Society for Glycobiology and Oxford University Press.

Susan M Rosenberg, Ph.D., who holds the Ben F Love Chair in Cancer Research, was elected to the American Association for the Advancement of Science’s Committee on Council Affairs.

Christian Schaaf, M.D., Ph.D., elected to membership in the American Society of Clinical Investigation and was listed in Texas Monthly’s Super Doctors: Rising Stars Edition 2017.

Pawel Stankiewicz, M.D., Ph.D., was a recipient of the Polish Presidential Scholar Award of Full Professor, the highest scientific title in Poland.

Meng Wang, Ph.D., received the Early Career Life Scientist Award from the American Society for Cell Biology.

Thomas (Trey) Westbrook, Ph.D., was named a McNair Scholar at Baylor College of Medicine.
New Faculty Appointments

Research Faculty

Rodney Samaco, Ph.D.
Assistant Professor, Tenure-Track

Lindsay Burrage, M.D., Ph.D.
Assistant Professor, Tenure-Track

Jihye Yun, Ph.D.
CPRIT Scholar Assistant Professor, Tenure-Track

Claudia Soler-Alfonso, M.D., F.A.C.M.G.
Assistant Professor

Diagnostic Laboratory, Genetic Counseling & Clinical Faculty

David R. Murdock, M.D., F.A.C.M.G.
Assistant Professor

Deanna Erwin
Instructor

Sarah Huguenard, M.S., C.G.C.
Instructor

Andrea Lewis, M.S., C.G.C.
Instructor

Sureni Mullegama, Ph.D.
Assistant Professor

Andrea Harbison, M.S., C.G.C., Instructor (Not pictured)

Promotions

INSTRUCTOR
Alicia Turner
David Marciano, B.S.
Shan Chen

ASSISTANT PROFESSOR
(non-tenure track)
Eric Venner, Ph.D.
Isabelle Schrauwen, Ph.D.
Keren Machol, M.D.
Maxime W.C. Rousseaux, Ph.D.
Baiping Wang, Ph.D.

ASSOCIATE PROFESSOR
(non-tenured)
Weimin Bi, Ph.D.

ASSOCIATE PROFESSOR
(tenured)
Penelope Bonnen, Ph.D.
Herman Dierick, M.D.
Sandesh C.S. Nagamani, M.D.
Joshua Shulman, M.D., Ph.D.

PROFESSOR
(non-tenured)
Sarah Elsea, Ph.D.

PROFESSOR
(tenured)
Rui Chen, Ph.D.
Thomas Westbrook, Ph.D.
In 2017, we lost a dear friend, colleague, teacher, mentor and scientist. Dr. Ken Scott began his training at Baylor in the Department of Molecular and Human Genetics as a graduate student and returned as faculty in 2009 after postdoctoral training at Harvard. He was promoted to tenured Associate Professor in 2017 and led an innovative program in cancer genetics, applying state-of-the-art screening methodologies for identifying genetic determinants of cancer initiation and progression. In recognition of his legacy and contributions, the Department established the Kenneth Scott cDNA Clone Collection as a department research core to give a new generation of scientists access to his technologies. In addition, the Department established the Kenneth Scott Graduate Mentor Award to recognize a department faculty member for outstanding mentoring in the graduate program.