I’m delighted to welcome you to our 2016 Annual Report. As we begin a new year, I’d like to briefly reflect on last year’s accomplishments. And what a year it’s been!

The Department of Molecular and Human Genetics 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 our clinical genetics program (the largest in the country), offers patients unparalleled, single-source genetic testing and services.

In this past year, we continued to grow our diagnostic laboratory joint venture, now rebranded as Baylor Genetics, with our partner Miraca Holdings, Inc. In planning for the future of genetic medicine, we launched Consultagene, the Department’s virtual platform for delivery of genetic services including direct-to-patient telegenetic counseling, peer-to-peer consultation, and genomic data interpretation services. To broaden the reach of the Department, we established a joint Baylor College of Medicine and The Chinese University of Hong Kong Center for Medical Genetics in Hong Kong.

As we take measure of the past year, let us also look forward. The future holds much promise – and 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 part of this exciting and vital effort.

Warm regards,

Brendan Lee, M.D., Ph.D.
Robert and Janice McNair Endowed Chair in Molecular and Human Genetics
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Genetic research at Baylor College of Medicine began 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 clinical investigators in genomics and biomedical research. As the research team grew in size, scope, and ambition, a centralized organization was needed to fuse disparate lines of effort through one cohesive department.

Thus, the Institute of Molecular Genetics was created in 1985, putting Baylor on the map as a genetics powerhouse and leveraging its ability to recruit the best and brightest physicians and scientists. 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 joint venture, Baylor Genetics. Baylor Genetics offers an expansive menu of genetic tests and provides efficient 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.
Consultagene
Dr. Brendan Lee debuts virtual integrative genetic services platform at 2016 American Society of Human Genetics Conference

Consultagene is 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.

A product of Baylor College of Medicine, Consultagene leverages the intellectual capital of the faculty in the Department of Molecular and Human Genetics, which includes research faculty, physician medical geneticists, genetic counselors, and diagnostic lab directors, allowing for a synergy and level of expertise that is unique to the environment at Baylor.

The Consultagene platform 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.

“The Consultagene platform centers on four modules, which are highly interchangeable and evolve to uniquely serve each patient throughout their Consultagene experience,” said Dr. Brendan Lee, the Robert and Janice McNair Endowed Chair in Molecular and Human Genetics at Baylor. “These four modules include the client engagement module, video module, health history and pedigree module, and the tele-counseling module. Consultagene, importantly, guides the client through the entire process.”

During the client engagement module, the referrer makes a request for a service for a specific client, such as genetic testing for a given condition. The referring indication directs the content and mix of modules experienced by the client. The client then receives a “token” via email to join Consultagene, upon which a patient record is established within the platform. During this engagement module, a consent for contact registry is completed, allowing for longitudinal contact for relevant research studies, new research developments or revised test results.

Moving through to the video education module, the client is directed by the system to view a relevant set of proprietary videos, explaining any preparation or logistical information the client may need for their respective service. These videos cover a range of topics: clinical genetics evaluations, indications and reasons for a genetics appointment, carrier screening, whole exome sequencing and BRCA 1/2 genetic testing. Video education allows the client to re-access learning experiences and improves the uptake of information.

“In our world, so much feels confusing, especially in the realm of genetics. After watching the videos, I have a better understanding of the whole exome sequencing process and feel confident I could explain its purpose to family and friends in laymen’s terms. It takes something so complex and makes it seem pretty simple,” said Renee Jones, a Consultagene client.

(Continued on next page)
After viewing relevant educational videos, the client moves into the health history and pedigree module, which assembles family, health, medication and social history information into a personalized client pedigree and allows for document transfer and sharing of medical records and test reports with the provider.

Finally, the client can then move into the tele-counseling module. During this module, the system will prompt the client to schedule a telecounseling session to review the services requested and their client pedigree. The referrer engages with the client and provider to receive the lab interpretations and accurately communicate the results to the client.

Depending on the referring indication, specific combinations of these modules are ordered by Consultagene and experienced by the client.

“Baylor Genetics generates the data, but there needs to be genetic services to interpret and counsel families so they can best utilize that data,” said Lee. “Consultagene turns the information into medical action, increases clinical efficiency and improves the patient experience.”

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.

(Continued from previous page)

Building on its history of genetic innovation, Baylor Genetics has launched the first clinical noninvasive prenatal multi-gene sequencing screen, PreSeek™, for the commercial market. Current noninvasive prenatal screening tests can detect abnormal numbers of chromosomes and microdeletions, but PreSeek goes further by sequencing cell-free fetal DNA for disease-causing, or pathogenic, variants associated with select single-gene conditions. PreSeek is a complementary test to traditional prenatal screening tests, allowing patients to have a more complete picture of the risk of their pregnancy being affected by a genetic disorder.

The PreSeek team carefully selected genes through a curation process focused on the detection of de novo variants in single-gene disorders affecting the skeletal, cardiac and neurological systems. PreSeek is the first noninvasive test to detect disorders that become more prevalent with advanced paternal age.

Many of the disorders detected by PreSeek are not typically associated with abnormal prenatal ultrasound findings. Since PreSeek is recommended during the late first trimester or early second trimester, knowing whether or not a fetus has one of these significant, and often devastating, genetic disorders can help families and their physicians make important reproductive and medical management decisions.

“Our clients trust us to be ready with answers, and PreSeek delves deeper than other noninvasive prenatal tests on the market,” said Dr. Shashikant Kulkarni, chief scientific officer at Baylor Genetics and professor and vice chair for research of molecular and human genetics at Baylor.
Baylor Miraca Genetics Laboratories, a joint venture of Baylor College of Medicine and Miraca Holdings, Inc., has rebranded as Baylor Genetics.

“Baylor Genetics 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. Through continuous investments in innovation and new products, Baylor Genetics strives to be the elite gold standard that empowers precision medicine,” said Kengo Takishima, president and CEO at Baylor Genetics.

The rebranding process was intensive and collaborative, including input from both clients and prospects, which allowed the organization to more openly consider improvements. This research also identified a need to develop messaging that spoke more to the core of the services and goals at Baylor Genetics, and the tagline “Ready With Answers” resulted.

To fit this motto, Baylor Genetics has turned its focus to operational improvements to shorten turnaround times, improve customer service, create a user-friendly billing system and evolve genetic counseling services. This new representation of Baylor Genetics shows that it is evolving by expanding new test offerings, forging into new markets, and differentiates its leadership position in the genetics and genomic industry. Miraca Holdings, the majority shareholder, will continue to provide guidance in executing operational improvements and running a stronger, more efficient business.

On the patient level, Baylor Genetics and its highly respected geneticists are prepared to provide answers to complex genetic questions through a consultative approach, extensive database of patient cases and comprehensive data interpretation, which results in fewer retests and lower costs. This high quality of data and services better equips Baylor Genetics clients to deliver accurate results to their patients and healthcare providers.

“Baylor Genetics offers a comprehensive test menu that provides a one-stop-solution in the fields of maternal-fetal and pediatric genetics, and through the development of a new laboratory information ecosystem and automation capabilities, we forecast aggressive growth over the next several years,” said Takishima.
The Chinese University of Hong Kong-Baylor College of Medicine Joint Center for Medical Genetics

Baylor and The Chinese University of Hong Kong join forces to advance genetic research and training

In 2016, Baylor College of Medicine and The Chinese University of Hong Kong (CUHK) signed a memorandum of understanding to establish The Chinese University of Hong Kong-Baylor College of Medicine Joint Center for Medical Genetics in Hong Kong with a vision to create a platform for training in clinical genetics, expert services for genetic disorders and collaborative research with cutting-edge genetic and genomic technology.

Dr. Brendan Lee and Dr. Tak Yeung Leung, head of maternal and fetal medicine in the department of obstetrics and gynecology at The Chinese University of Hong Kong, serve as co-directors of the Center. Its aims include designing, establishing and conducting training in clinical genetics and genetic counseling to fit the increasing need in China and the Asia region, establishing a leading referral center in Asia for prenatal and postnatal diagnosis and treatment for patients and families affected by genetic disorders, conducting cutting-edge, interdisciplinary research that will lead to advances in screening, diagnosis and therapy of genetic disorders as well as new discovery of the underlying genetic mechanism of diseases, and hosting an annual pan-Asian symposium on state-of-the art clinical genetics care and research.

In May 2017, the Center will host its first-ever Joint Symposium in Clinical Genetics at the Postgraduate Education Centre in the School of Public Health at the Prince of Wales Hospital in Hong Kong. The goals of the symposia are 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.

This year’s three day symposium will play host to key speakers in the field, including Dr. Igna Van Den Veyver, Dr. Richard Gibbs, Dr. Arthur Beaudet, Dr. James Lupski, and Dr. Brendan Lee, all with Baylor, and Dr. Dennis Lo, Dr. Rossa Chiu, Dr. Tak Yeung Leung and Dr. Richard Choy, with The Chinese University of Hong Kong.
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.

**Negative feedback loops help maintain function of mutated proteins**

New findings about the role of negative feedback loops were outlined in research published in *Physical Review Letters*, a leading physics journal. Negative feedback is a universal control mechanism that lets a system’s output throttle its input. In biology, genes under negative regulatory feedback increase (or decrease) their expression in response to falling (or rising) concentrations of the gene product.

The new study unites mathematics with gene regulation experiments and demonstrates a new role for negative feedback: a shock absorber to buffer the damage of mutations. As a result, genes under negative self-regulatory feedback are freer to mutate, possibly feeding into a wellspring for evolution.

“This study pins down a process by which some mutations may foster an organism’s long-term adaptation while putting its immediate fitness at less risk,” said Dr. Olivier Lichtarge, The Cullen Foundation Endowed Chair at Baylor, in whose lab the work was done, in collaboration with Dr. Christophe Herman, associate professor of molecular and human genetics.

Researchers found that mutational tolerance and feedback sensitivity are coupled. A gene circuit that strongly responds to perturbations maintains the function of a mutated protein product better than a less responsive gene circuit.

“This is of particular importance,” said Dr. David Marciano, postdoctoral fellow in the Lichtarge lab, “because mutations deliver the heritable variation upon which natural selection acts. This suggests the ability of gene networks to reshape the mutational landscape of a protein could significantly influence the course of evolution.”

**Research opens the possibility of new, noninvasive genetic prenatal testing**

Researchers at Baylor College of Medicine, Drexel University College of Medicine, Texas Children’s Hospital and RareCyte, Inc. has determined that it is feasible to develop a prenatal, noninvasive genetic test based on rare fetal cells that are present in the mother’s blood. The study appeared in the October 2016 issue of *Prenatal Diagnosis*.

Although other noninvasive methods for genetic testing are currently available, they have their limitations. For instance, cell-free DNA testing cannot reliably detect very small changes in the fetal genome, in particular, gene deletions that can lead to devastating diseases such as Angelman syndrome. According to senior author, Dr. Arthur Beaudet, if the fetal cell-based test can become routine practice in current and future forms, it could be transformative for prenatal diagnosis, offering comparable information to that which can be obtained by amniocentesis and chorionic villus sampling.

The main obstacle to achieving this goal is that there are very few fetal cells in the mother’s blood, and they are very fragile.

“About two tablespoons of blood has hundreds of billions of maternal red blood cells and hundreds of millions of white blood cells but only 20 to 40 fetal cells,” said Beaudet. “We showed that we frequently can recover 3 to 10 or more fetal cells and analyze them in various ways, including next-generation DNA sequencing. At present the group can only process 5 to 10 samples per week on a research basis, and they are focused on increasing this number so that the test could be offered as a routine clinical test.

This work has moved a new prenatal genetic test closer to the clinic,” said Beaudet. “We estimate that the test might be available to the public in one to two years.”
Scientists can now better diagnose diseases with multiple genetic causes

Scientists at Baylor College of Medicine, Baylor Genetics, the University of Texas Health Science Center at Houston and Texas Children's Hospital have combined descriptions of patients' clinical features with their complex genetic information in a unified effort to obtain more precise diagnoses of complex diseases, particularly those that involve more than one gene, leading to more effective treatments and counseling.

In the study that was published in the New England Journal of Medicine, the researchers used whole exome sequencing to analyze all the genes in the genomes of nearly 7,400 unrelated patients with the goal of identifying the genetic cause of their conditions. They found a genetic cause in 28 percent of the patients. Among these patients, approximately 5 percent had two or more disease genes involved. If an individual has multiple defective genes, he or she may present with a complex set of clinical features that may lead to an imprecise diagnosis.

"Clinically, multiple genetic causes can be missed because a patient may present with characteristics that overlap those of two conditions, so the patient can be diagnosed with one or the other," said co-first author, Dr. Jennifer Posey, assistant professor of molecular and human genetics at Baylor. “Alternatively, a patient’s clinical characteristics may not match those of any described condition, so they may be diagnosed with what is thought to be a new condition.”

"In these situations, we, as physicians, have to think of the possibility that more than one gene might be involved in the disease," said senior author, Dr. James R. Lupski, The Cullen Foundation Endowed Chair in Molecular Genetics at Baylor. “Our study shows the limitations of defining a disease according to what we see in the clinic alone, the need to consider that a patient may have two or more genetic diseases, and to send for a genomic test to help sort out the condition and causes of it.”

“Helping patients and their physicians discover the underlying genetic mechanism of their condition is our most important priority,” said co-author Dr. Christine M. Eng, professor of molecular and human genetics at Baylor and chief quality officer and chief medical officer at Baylor Genetics. “This data compels us to continue the search for genetic contributions to a patient’s overall clinical presentation.”

Center for Genome Architecture explores 3-D structure of DNA

In a set of papers published in Cell Systems, Dr. Erez Lieberman Aiden, assistant professor of molecular and human genetics, McNair Scholar and director of the Center for Genome Architecture (TC4GA) at Baylor, and his colleagues introduced Juicer, an open-source tool used in three-dimensional genome sequencing (Hi-C) processes.

Juicer is a fully-automated pipeline that allows users with little to no computational background to transform raw sequencing data into genome-wide maps of looping with a single click. Juicer produces the Hi-C file with loops and contact domains automatically annotated, which facilitates the visualization and analysis of the map and its structural features.

As a demonstration of the power of the new tool, Aiden and his colleagues created the deepest 3-D maps of the genome to date, spanning over three terabytes of data drawn from a single experimental condition. Edico’s DRAGEN platform accelerated the analysis of the massive data sets derived from this study of 3-D structures of DNA by nearly 20 fold, a dramatic speedup from all of other systems tested.

DRAGEN generates accelerated implementations of genome pipeline algorithms using a field-programmable gate array (FPGA). The platform is reconfigurable and flexible through remote downloads, allowing users to create custom algorithms and refine existing pipelines.

Aiden, who also is a faculty member at Rice University, commented on the experiment, saying, “The partnership between TC4GA and Edico Genome is a game-changer. The results that are possible using DRAGEN are more than a one-off exercise: they are a strong indicator of the future of the 3-D genomics field as a whole. We are confident that our collaboration will lead to a great deal of innovation both within the Texas Medical Center community, and beyond.”
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 continues to distinguish itself.

For six years running, the Department has remained the No. 1 ranked U.S. genetics department, as measured by the number of NIH-awarded grants and total funding received. As of Jan. 1, 2017, the total in funding dollars from active NIH awards was $95 million.

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.

Ranked #1 in NIH Funding

The 2012-2016 data reflected in these charts were collected from Blue Ridge Institute for Medical Research based on NIH fiscal year awards (Oct. 1 - Sept. 30). The data collected on Jan. 1, 2017, is from NIH RePORTER.
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

$27.9 million from NIH to support Baylor’s Knockout Mouse Project

Nearly $28 million has been awarded to Baylor College of Medicine’s Knockout Mouse Project led by Dr. Arthur Beaudet, the Henry and Emma Meyer Chair and Professor of Molecular and Human Genetics at Baylor, and Dr. Mary Dickinson, the Kyle and Josephine Morrow Endowed Chair and Professor of Molecular Physiology and Biophysics and Molecular and Human Genetics at Baylor.

The grant, to be used over five years, was awarded by the NIH as a renewal of a previous grant to generate modified mice and establish primary phenotype analysis as a resource for researchers worldwide. Baylor will generate and phenotype lines for 1,000 new genes using Cas9/CRISPR technology.

“Since the project began, 5,000 mouse gene lines have been produced, and we hope to continue,” said Beaudet.

“Being able to knockout or delete genes in animal models is a way to understand how genes work

and what functions they are responsible for,” said Dickinson. “This support not only provides mice and phenotype data to the community but allows for a systematic understanding of gene function in a mammalian model system.”

Baylor is one of three sites with renewed funding from the NIH for Knockout Mouse Projects, all of which are part of the International Mouse Phenotyping Consortium (IMPC). The goal of the IMPC is to discover functional insight for every gene by generating and systematically phenotyping 20,000 knockout mouse strains.

This grant is a collaborative effort between Baylor and the Mary Lyon Centre in Harwell, U.K. Much of the work at Baylor is carried out in the Advanced Core Labs, which are state-of-the-art facilities that provide instrumentation and technologies to support a wide array of research. Those cores used for the Knockout Mouse Project include Optical Imaging and Vital Microscopy, Cytometry and Cell Sorting, Genetically Engineered Mouse, Mouse Embryonic Stem Cell and Mouse Phenotyping.
The Human Genome Sequencing Center (HGSC) has been operational for more than 20 years, gaining international recognition as a large-scale DNA sequencing and analysis center, and is currently a Center for Complex Disease Genomics supported by the NIH and the National Human Genome Research Institute (NHGRI). Led by Dr. Richard Gibbs, the HGSC was originally established in 1996 to participate in (and eventually help complete) the Human Genome Project. The Center has since expanded its research focus into exciting new areas.

Within the last year, the HGSC has been awarded highly competitive grant funding to help drive its mission. Totaling more than $400 million dollars combined, the HGSC had been directly awarded grants from the NHGRI, the NIH, the National Eye Institute, Mayo Clinic Rochester and the National Heart Lung and Blood Institute, among others, to pursue a wide range of sequencing projects to advance the understanding of conditions such as cardiomyopathy, ocular disease, pediatric cancer and atrial fibrillation, along with how they behave and develop in varying populations.

Among these grants is the National Heart Lung and Blood Institute (NHLBI) Trans-Omics for Precision Medicine (TOPMed) program, which has named the Human Genome Sequencing Center at Baylor as part of a groundbreaking half-billion dollar contract to bring to the forefront whole genome sequencing and other “omic” technologies that monitor the expression of the genome in response to the environment, such as diet or smoking.

As a whole, the contract will span five years and will accomplish six key task orders. The initial award from NHLBI supports whole genome sequencing of 20,000 genomes at the HGSC in the first year of the program.

“There is a significant need for large sample sizes; a need that goes beyond the research setting and into the clinic,” said Gibbs. “The TOPMed program will allow us to access this large sample number and obtain valuable insights into adult heart disease, sickle cell disease, atrial fibrillation and other heart, lung and hematologic disorders.”

The whole genome data made available to HGSC by TOPMed has the capability to be analyzed to provide a more comprehensive picture of what factors may lead to, or protect against, common disease development.

“The TOPMed program encourages data sharing and collaboration among institutions across the United States and will encourage an integrative analysis approach, which will be crucial to understanding the mechanisms that contribute to development of these common adult diseases,” said Ginger Metcalf, director of project development at the HGSC.

“The NHLBI contract is a huge contribution to the research community here at Baylor, and for the Texas Medical Center as a whole. The scope of whole genome research it will allow us to execute and apply to the clinical setting is groundbreaking, and I look forward to seeing what we are able to discover in the realm of adult hematological disorders as a result,” said Dr. Adam Kuspa, senior vice president and dean of research at Baylor.
Huffington Center on Aging

A core value of the Huffington Center on Aging (HCOA) 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, has made great strides in the past year. Besides Dr. Meng Wang’s accomplishments (see page 27), Dr. Melanie Samuel was awarded the NIH New Innovator Award to further support her research into understanding the processes that lead to cognitive dysfunction. The Center recruited Dr. Wei Cao, who works on neuro-immune regulation in aging and Alzheimer’s disease. In addition, to further the mission of the HCOA, the Center held a retreat through which a five-year Strategic Plan was developed.

Jan and Dan Duncan Neurological Research Institute

The Jan and Dan Duncan Neurological Research Institute (NRI) was established at Texas Children’s Hospital to foster collaboration between research and clinician scientists to better understand neurological disorders and develop therapeutic strategies. Internationally renowned scientist Dr. Huda Zoghbi, the Ralph D. Feigin, M.D., Endowed Chair, serves as the director of the Institute.

In 2016, scientists at the NRI made significant findings. The Arenkiel laboratory identified a novel mechanism governing circuits influencing feeding behavior. The Bellen laboratory identified the first mutant Drosophila allele of Friedreich’s ataxia and uncovered its mechanism of pathogenesis. The Botas and Zoghbi laboratories showed that reducing Nuak1 decreases Tau, thereby restoring normal synaptic plasticity, improving memory, and ameliorating pathology in tauopathy models, suggesting a therapeutic target for Alzheimer’s Disease. In addition to the awards received by Drs. Ballabio, Schaaf and Zoghbi (see pages 26-28), multiple NIH R01 grants were awarded to the NRI faculty.

Center for Skeletal Medicine and Biology

The Center for Skeletal Medicine and Biology (CSMB), directed by Dr. Brendan Lee, 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, along with the Rolanette and Berdon Lawrence Bone Disease Program of Texas, cultivates teamwork between clinicians, clinical researchers, and basic researchers within the Texas Medical Center by co-sponsoring monthly seminars, pilot grants, and core facilities. This year, the “T-Bone” Seminar Series was established to support TMC trainees interested in musculoskeletal biology by creating a space to present unpublished data and to attend educational talks by expert faculty. Also of note was the annual scientific retreat, as it had over 120 participants and hosted Dr. David Scadden, the co-director of the Harvard Stem Cell Institute, as its keynote speaker.
**Computational and Integrative Biomedical Research Center**

The Computational and Integrative Biomedical Research Center (CIBR), led by Dr. Olivier Lichtarge, supports computational education, infrastructure, and research activities. Last year, a CIBR class included in the Graduate School of Biomedical Sciences core curriculum received above average quality scores, and, separately, a new class that mixes seminars with a journal club was approved. CIBR data clinics and workshops were well-attended. CIBR licenses to Mathwork’s MATLAB and Wolfram Mathematica have over 600 users, at a huge cost savings. The CIBR cluster computer is used by over 30 different labs. The top 10 users have an annual research budget in excess of $16 million. An IARPA research award to BCM totaling $2.8 million is an example of research led by the CIBR. The center also helped to recruit a new Baylor faculty member, Dr. Bing Zhang, McNair Scholar and professor of molecular and human genetics.

**Intellectual and Developmental Disabilities Research Center**

The Intellectual and Developmental Disabilities Research Center (IDDRC), led by Dr. Huda Zoghbi, supports more than 40 investigators studying developmental disorders. During the past year, IDDRC investigators made many discoveries. Dr. Jeffrey Noebels demonstrated in the Journal of Neuroscience that an early P/Q-type calcium channel release defect remodels firing behavior and produces generalized epilepsy reminiscent of absence epilepsy in children. Dr. Christian Schaaf published several papers on the cognitive and behavioral phenotypes of individuals with copy number variants of 15q13.3 encompassing CHRNA7. Dr. James Lupski and Dr. Hugo Bellen, the March of Dimes Chair in Developmental Biology, reported in Neuron that a loss of Nardilysin, a mitochondrial chaperone, promotes neurodegeneration. Dr. Zoghbi’s Neuron paper on the properties of CA1 neurons in the Rett and MECP2 duplication models was among Spectrum’s Top 10 Notable Papers of 2016.
Pediatric Genetics

Our 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 (Texas Children’s Hospital West Campus and Texas Children’s Hospital The Woodlands). Physicians at the Texas Children’s clinic see over 3500 families each year.

Specialty clinics within the Texas Children’s Genetics Clinic include the metabolic, neurofibromatosis, skeletal dysplasia, and cancer genetics clinics. Our 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 our general genetics clinic, we also have 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.

Clinical Genetics Patient Volume (Prenatal)

TCH Genetics Expands into The Woodlands

With the demand for pediatric genetics services continuously growing, the Texas Children’s Hospital Genetics Clinic will begin its expansion into The Woodlands Texas Children’s Hospital location beginning in 2017.

“Many of our pediatric clinical genetics patients come from the northern part of town, and there has been incredible demand for genetics services as The Woodlands and surrounding areas continue to grow,” said Dr. Carlos Bacino, chief of the genetics service at Texas Children’s Hospital and professor of molecular and human genetics at Baylor College of Medicine.

The clinic at The Woodlands location will begin with offering one clinic day per week, initially focusing on general pediatric genetics and complex diagnoses, with the intention of growing from there as demand requires. Leading The Woodlands clinic is Dr. Bret Bostwick, assistant professor of molecular and human genetics at Baylor, who said the genetics services will round out the comprehensive care available at The Woodlands location and allows the patient to be treated where they live.

“With the expansion of pediatric genetics services in The Woodlands, we are providing another step of quality care to an expanding population right in their backyard. We are one step closer to reaching more of the underserved communities we know are in need of these services,” Bostwick said.

In addition to diagnostic and clinical genetics services, The Woodlands Genetics Clinic will offer genetic counseling services, as well. “Education is a large part of what we will be doing in The Woodlands. It is important to educate not only the families and patients affected by these genetic conditions, but also society as a whole so that we bring greater awareness to the treatments, risks and diagnosis of these complex conditions,” said Bacino.

In addition to the statistical graphs and charts, the text includes a description of the services offered by the Prenatal Genetics Clinic, as well as information about the expansion of the Texas Children’s Hospital Genetics Clinic into The Woodlands.

The text also emphasizes the importance of genetic counseling and the need to educate both families and society about genetic conditions and their treatments.
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 and 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 Dr. 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 best peer-reviewed scientific journals in the world.

Awards and Special Recognition for Molecular and Human Genetics Graduate Program Students in 2016

Graduate students in the Molecular and Human Genetics Graduate Program received many recognitions for their hard work in 2016, here are some of the highlights:

**Smriti Agrawal** received the 2016 MIT Outstanding Poster Award in Retinal Cell Biology at the 2016 Association of Research in Vision and Ophthalmology Meeting.

**Brittany Barreto** received the Future of Microbiologist Award from the American Society for Microbiology.

**Ivan Bochkov** received the John J. Trentin Scholarship Award.

**Paul Fullerton** was awarded the Larry Ewing Memorial Trainee Travel Fund grant to present at the 49th Annual Meeting of the Society for the Study of Reproduction.

**Maddie Gillentine** will be a selected attendee for the 2017 St. Jude National Graduate Student Symposium.

**Yi-Chen Hsieh** received a Travel Fellowship for the 2016 Alzheimer’s Association International Conference.

**Patrick Hunt** was selected as a 2016 BRASS Scholar.

**Evan Jones** was awarded a Fellowship in the National Library of Medicine Training Program in Biomedical Informatics through the Gulf Coast Consortia for 2016 and 2017.

**Angel Lopez** received a Travel Award to attend and speak at the Mechanisms of Epilepsy & Neuronal Synchronization Gordon Conference in Spain.

**Meagan Siehr** received an American Epilepsy Society Predoctoral Fellowship.

**Mingchu Xu** was a Semifinalist for the 2016 American Society of Human Genetics Charles J. Epstein Trainee Award for Excellence in Human Genetics Research.
Internal Medicine-Genetics Residency Program
Baylor College of Medicine launches new residency program in internal medicine and genetics

The departments of medicine and molecular and human genetics at Baylor College of Medicine have come together to offer a new residency program for medical students interested in pursuing adult medical genetics. The internal medicine-genetics program will enable Baylor to stay at the cutting edge of patient care and help attract top talent in the field.

“We expect this track will produce new physician-scientists to engage three growing areas in adult medical genetics: cancer, connective tissue disease and cardiovascular disease. They will then go on to populate clinics and institutions all over the country and the world,” said Dr. Brendan Lee. “We are training the future of this field and rounding out the comprehensive portfolio of genetics training and services, which is really rare to find.”

With the existing strength in pediatric genetics, the internal medicine-genetics program will also provide expert, transitional care to patients with genetic conditions as they move from pediatric care into adulthood and offer more direct access to the Undiagnosed Diseases Network for particularly rare conditions. One of only seven of its kind in the nation, the five year internal medicine-genetics program will benefit residents in all specialties. The field of adult medical genetics is small and relatively young, with only an estimated 150 certified, trained internal medicine geneticists in the country, four of whom are Baylor faculty.

“This program will better equip our trainees with the tools to crack complex adult diseases in the future. It will likely lead to significant discoveries, as these residents will not only be learning through the track, but also creating the clinical techniques and approaches for treatment that will become the standard in patient care. In the process of training, they will become natural leaders in the field,” said Dr. Ashok Balasubramanyam, professor of medicine, vice president of academic integration and senior associate dean for academic affairs at Baylor.

Masters of Science Genetic Counseling Program in Development
Promoting excellence in the practice of genetic counseling

In 2016, the Department of Molecular and Human Genetics began developing a Masters of Science Genetic Counseling Program (MSGC). The mission of this future 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. We are assembling an interdisciplinary team comprised of clinical, laboratory, and research faculty members available within the Department to provide experiences that will empower graduates to become empathic professionals with effective critical thinking skills. The program will utilize over 35 genetic counselors in the Department and will provide an integrated medical genetics graduate education that will consist of access to patient care and research activities, the full service genetic diagnostic testing laboratory, Baylor Genetics, and the DNA sequencing performed as part of the NIH funded Undiagnosed Disease Network.
The Medical Genetics Residency Program, led by Dr. V. Reid Sutton, professor of molecular and human genetics and director of the inborn errors of metabolism service at Texas Children’s Hospital, prepares individuals for an accomplished academic career by providing an integrated experience in both clinical and experimental genetics. The program offers trainees extensive laboratory experience, and prepares them to care for adult and pediatric patients with cytogenetic, biochemical, and developmental diseases.

During 18 months of clinical rotations, residents divide their time among inpatient consultation services, outpatient clinics, subspecialty clinics, and various diagnostic laboratories; they also attend conferences and teaching sessions.

The clinical experience is broad and intensive. Residents enjoy easy access to teaching faculty, and have the opportunity to participate in a number of path-breaking genetics research projects.

The program enjoys preeminence in the genetics community and is approved by the Accreditation Council for Graduate Medical Education (ACGME). It is also supported by a training grant from the National Institute of General Medical Sciences (NIGMS).

In 2017, Baylor College of Medicine will be offering a new clinical fellowship program in Maternal-Fetal Medicine and Medical Genetics and Genomics. The combined training program capitalizes on Baylor College of Medicine’s strength in both areas and will be fully accredited by the American Board of Obstetrics and Gynecology and the American Board of Medical Genetics and Genomics. The program consists of 18 months of clinical medical genetics training, 18 months of clinical maternal-fetal medicine training as well as 12 months of research training.
Clinical Laboratory Fellowship Training Programs

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

Genetics fellows train at Baylor College of Medicine’s genetics diagnostic laboratories, Baylor Genetics, for 24 months. At that time, they are eligible for board certification by the American Board of Medical Genetics and Genomics (ABMGG). We are proud to offer fellowships in the following areas:

**Laboratory Genetics and Genomics**

Beginning in July 2017, Baylor Genetics will offer training in only Laboratory Genetics and Genomics (LGG) instead of separate training for cytogenetics or molecular genetics alone. LGG is a new specialty of the ABMGG that incorporates training in both molecular and cytogenetic techniques and interpretations in 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**

Trainees in the Clinical Biochemical Genetics fellowship program 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.

Locations of Former Medical Genetics Trainees
Frank Greenberg Memorial Lectureship

This lectureship was established in memory of Dr. Frank Greenberg. Dr. Greenberg was a faculty member in the Departments of Molecular and Human Genetics and Pediatrics, Baylor College of Medicine, from 1981 until his retirement in 1994. Dr. Greenberg received his B.A. in Zoology from the University of Michigan and an M.S. from Rutgers Medical School. Dr. 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 at the Center for Disease Control as an Epidemiological Intelligence Officer and was a Clinical Assistant Professor in Pediatrics at Emory University in Atlanta.

Dr. Greenberg published more than 100 articles in all areas of clinical genetics and established himself as an expert in contiguous gene deletion syndromes. Dr. Greenberg contributed to the clinical delineation of a number of congenital chromosomal abnormalities including Prader-Willi, Williams, DiGeorge, and Smith-Magenis syndromes. Dr. 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. Dr. Greenberg proposed the creation of diagnostic criteria for Williams syndrome, which allowed better assessment of the clinical phenotype.

Through his involvement in the Baylor Medical Genetics Training Program, Dr. Greenberg’s extraordinary abilities in dysmorphology and clinical evaluation have contributed to the education of numerous clinical geneticists throughout the world. Dr. 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. Dr. Greenberg will be remembered as a gifted educator, mentor, talented dysmorphologist, and 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.”

18th Annual Frank Greenberg Memorial Lectureship features Dr. Stefan Mundlos

Dr. Stefan Mundlos is currently Professor and Chairman of the Institute for Medical and Human Genetics at the Charité, Berlin’s Medical School, and head of the research group Development & Disease at the Max Planck Institute for Molecular Genetics in Berlin.

His lifelong interest is in genotype-phenotype correlations and the molecular basis of Mendelian disorders with a particular focus on congenital malformations and skeletal disease. He discovered numerous disease genes and has a special interest in understanding the developmental biology of mutations.

His current interests and research projects are focused on the function of non-coding DNA in gene regulation and disease, the functional *in vivo* analysis of structural variations, as well as the development of tools for the diagnostics of genetic disorders.
Evenings with Genetics
Baylor shares access to experts through Evenings with Genetics seminars

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 genetic outreach at Baylor, who launched the series in 2006 with Dr. Arthur Beaudet, who was the chair of the Department at the time. “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 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.”
Rare Disease Day
Houston’s first rare disease day debuts at the health museum

On the last day of February of every year, millions of people across the U.S. join the National Organization for Rare Disorders in observing Rare Disease Day. Rare Disease Day was established to raise awareness amongst the general public and policy makers about rare diseases and their impact on patients’ lives. In 2016, Houston hosted its inaugural event at The Health Museum. The local event was a joint effort by Texas Children’s Hospital and Baylor College of Medicine and featured approximately 20 informational booths from rare disease support organizations as well as seminars given by local physicians.


Dr. Christian Schaaf, assistant professor of molecular and human genetics at Baylor College of Medicine, and parent advocate Lace Mitchell presented a seminar on “Social Media and Rare Diseases.” They were followed by Dr. Carlos Bacino, who spoke about updates in testing for rare diseases.
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

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

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

Gad Shaulsky, Ph.D.
Vice Chair, 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)

We have more than 180 primary faculty members that occupy 180,000 square feet of space. Faculty includes:

- 3 members of the National Academy of Sciences
- 7 members of the National Academy of Medicine
- 2 Howard Hughes Medical Institute Investigators
- 4 Fellows of the American Association for the Advancement of Science
- 1 Howard Hughes Medical Institute Faculty Scholar
Faculty Awards and Recognitions

Zoghbi honored with 2017 Breakthrough Prize in Life Sciences

With scientific accomplishments including leading a team of researchers that discovered the gene that causes Rett syndrome, and most recently describing a protein that may play an important role in the development of neurological diseases such as Alzheimer's, it is no wonder that Baylor College of Medicine's Dr. Huda Zoghbi has been praised for her work focused on neurological disorders.

In 2016, Zoghbi, a Howard Hughes Medical Institute investigator who holds the Ralph D. Feigin, M.D., Endowed Chair at Baylor and also serves as director of the Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital, was recognized with three prestigious honors: the Jessie Stevenson Kovalenko Medal, the 2016 Shaw Prize in Life Science and Medicine and the Breakthrough Prize in Life Sciences.

"Dr. Zoghbi has made seminal discoveries in diverse areas of neuroscience. Her work has significantly furthered the understanding of neurodevelopmental and neurodegenerative disorders and revealed novel strategies to reverse these conditions," said Dr. Paul Klotman, president, CEO and executive dean of Baylor. "She is a superb scientist, educator and clinician. We’re fortunate she calls Baylor and Texas Children’s home."

The Jessie Stevenson Kovalenko Medal is awarded by the National Academy of Science (NAS) every two years to distinguished scientists for outstanding research in the medical sciences. Zoghbi became a member of NAS in 2004; a widely-accepted mark of excellence in science and one of the highest honors a scientist can receive.

The 2016 Shaw Prize in Life Science and Medicine was presented jointly to Zoghbi and Dr. Adrian P. Bird, Buchanan Professor of Genetics at the University of Edinburgh, for their groundbreaking discovery of the genes and encoded proteins associated with Rett syndrome, which has paved the way for the development of a diagnostic genetic test for the disorder.

The Breakthrough Prize celebrates the world's top scientists in the areas of life sciences, fundamental physics and mathematics, recognizing transformative advances toward understanding living systems and extending human life. Zoghbi was again honored for her discoveries of the genetic causes and biochemical mechanisms of spinocerebellar ataxia and Rett syndrome.

“Every award is an honor and brings with it motivation to keep moving forward in my work,” said Zoghbi. Her work has caught the eye of children and young girls who are now considering a career in science, an unexpected, but rewarding, outcome.
Dr. Meng Wang, associate professor in the Huffington Center on Aging and the Department of Molecular and Human Genetics at Baylor College of Medicine, is one of the brightest young standouts in her field. Her research focuses on aging biology and discovering clues to natural treatments that could allow the human body to age better and improve its lifespan. Through her detailed study of worms, Wang has identified genetic and molecular insights into how humans age.

“Meng Wang is truly outstanding and one of the best young scientists I have interacted with. Her research is at the forefront of genetics, metabolism and aging,” said Dr. Huda Zoghbi. “She has already demonstrated that she is a pioneer in her field, and her creativity has yielded many exciting discoveries that frame the work she hopes to accomplish.”

Honoring young scientists with bright futures, Wang received the Howard Hughes Medical Institute Faculty Scholar award, supporting her out-of-the-box project focused on developing new technologies to harness chemical imaging, functional metabolomics and genomic screens for pinpointing metabolites that occur naturally and have longevity effects on human metabolic health.

To further her work in identifying strategies to improve human health and longevity by harnessing the power of functional genomics and metabolomics, the NIH Pioneer Award supports Wang’s work to test a new idea for metabolite-directed signaling communication.

Wang also received the 2017 Edith and Peter O’Donnell Award in Medicine from The Academy of Medicine, Engineering and Science of Texas (TAMEST), which recognizes three rising star researchers in Texas whose work has set them apart in medicine, engineering or science and meets the highest standards of professional performance, creativity and resourcefulness.

“My goal is to eventually improve aging among the general population overall,” said Wang. To reach this goal, Wang has started to explore natural compounds that exist in the body and can contribute to therapies that promote healthier aging. “It’s incredible to be recognized, and it encourages me to continue to think big. I’ve had amazing support through Baylor and the senior faculty here, and I look forward to continuing to advance my research in healthy aging.”
Dr. Brendan Lee honored with American Society of Human Genetics Curt Stern Award

Dr. Brendan Lee, the Robert and Janice McNair Endowed Chair in Molecular and Human Genetics at Baylor, was awarded the American Society of Human Genetics 2016 Curt Stern Award, an honor given annually to a genetics and genomics researcher who has made significant scientific contributions in the last decade.

“It’s an honor to be recognized by the American Society of Human Genetics, an organization I’ve been involved with for many years,” said Lee. “It is humbling to join the list of Curt Stern Award winners, who have each contributed so much to the field.”

The Curt Stern Award celebrates Lee’s influential career in identifying human inborn errors of metabolism and structural birth defects of the skeleton. His internationally-recognized research continues to focus on understanding how gene mutations affect skeletal development, combining laboratory studies with clinical research, as well as nitrogen regulation.

Aside from his role as chair and professor in molecular and human genetics, Lee also is a leader and mentor across many of Baylor’s other programs, including integrative molecular and biomedical sciences, developmental biology, translational biology and molecular medicine, the Center for Skeletal Medicine and Biology, Medical School Research Track and the Rolanette and Berdon Lawrence Bone Disease Program of Texas.
More Awards and Recognitions for MHG Faculty

Andrea Ballabio, M.D., was awarded the 2016 Louis-Jeantet Prize for Medicine.

Hugo Bellen, Ph.D., was selected as the Edward J. Masoro Distinguished Lecture at the UT Health Science Center in San Antonio and also received the Michael E. DeBakey M.D., Excellence in Research Award from Baylor College of Medicine.

Penelope Bonnen, Ph.D., received the 2016 Kinkaid School Weiner Fellow Award.

C. Thomas Caskey, M.D., F.A.C.P., F.A.C.M.G., F.R.S.C., received The 2016 Colonel Harland D. Sanders Lifetime Achievement Award in Genetics from the March of Dimes.

Chonghui Cheng, M.D., Ph.D., received the Cancer Prevention & Research Institute of Texas Rising Star Award.

Florent Elefteriou, Ph.D., received the 2016 American Society of Bone Mineral Research Fuller Albright Award.

Sarah Elsea, Ph.D., was selected as the 2016 Meinhard Robinow Memorial Lectureship by the Department of Pediatrics at the University of Virginia, School of Medicine.

Hamed Jafar-Nejad, M.D., received a Pilot/Feasibility Award from the Texas Medical Center Digestive Disease Center.

Richard Lewis, M.D., M.S., received an Outstanding Service Award from the National Foundation for Ectodermal Dysplasias.

David Nelson, Ph.D., who holds The Cullen Foundation Professorship in Molecular Genetics, was elected president of the American Society of Human Genetics.

Christian Schaaf, M.D., Ph.D., received the inaugural Donald Seldin-Holly Smith Award for Pioneering Research from the American Society for Clinical Investigation.

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New Arrivals

The recruitment and retention of great faculty propels our department forward. Here are some of the faculty hires we welcomed in 2016.

Research Faculty

Atul Chopra, M.D., Ph.D.
Caroline Wiess Law Scholar,
Assistant Professor

Dr. Chopra is a physician-scientist with clinical expertise in medical genetics and research expertise in the field of energy homeostasis and metabolic disease. By studying a rare genetic disorder, Neonatal Progeroid Syndrome, Dr. Chopra’s lab was the first to identify a novel hormone, asprosin, that enables hepatic glucose production to meet the needs of the organism when dietary glucose is unavailable. In the therapeutic arena, antibodies targeting asprosin are showing considerable promise in the treatment of diabetes and insulin resistance by acutely resulting in reduced blood glucose and insulin levels.

Chonghui Cheng, M.D., Ph.D.
Associate Professor

Dr. Cheng’s laboratory strives to understand the fundamental questions of how RNA regulation controls cellular processes in normal biology and in the context of cancer. Working at the interface of RNA splicing and breast cancer biology, their focus is on regulation of breast cancer metastasis driven by alternative splicing. The Cheng lab uses molecular biology, genomics and bioinformatics approaches in conjunction with genetic models and patient samples to discover rules and networks that regulate metastasis and associated processes. By working closely with physician scientists, the Cheng lab aims to translate their findings from basic research to the development of prognostic markers and therapeutics for the treatment of breast cancer.

Shashikant Kulkarni M.S., Ph.D., F.A.C.M.G.
Professor

Dr. Kulkarni’s research is focused on the following related areas: (1) understanding cancer genomes by elucidating various classes of genomic alterations and discovering recurring mutations relevant for pathogenesis, (2) developing standards and guidelines for the interpretation of sequence variants, and (3) the design and optimization of next-generation sequencing technical and informatics pipelines for clinical laboratory practice.

Michael Wangler, M.D., M.S., B.S.
Assistant Professor

The overall long-term goal of Wangler Lab is to improve our understanding of the molecular pathogenesis of Mendelian disease by merging clinical observations, genomics and studies in model organisms, particularly Drosophila melanogaster. The lab is currently using Drosophila to study Mendelian disorders and their underlying genetic and developmental mechanisms.

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

The Yamamoto laboratory is interested in classes of genes that regulate cell to cell communication in development and disease. Their aim is to identify and characterize novel genes in pathways, such as Notch signaling and Dopamine signaling, and understand their precise functions in vivo using genetic, cell/molecular biological, biochemical, and electrophysiological methodologies. The Yamamoto lab is also part of the Undiagnosed Diseases Network, a nationwide consortium that employs state-of-the-art technologies to solve medical mysteries. The goal of these projects is to provide a better understanding of the molecular mechanisms underlying diseases, allowing researchers to explore novel drug targets and potential therapies toward a cure.
**Bing Zhang, Ph.D.**  
McNair Scholar, Professor

Dr. Bing Zhang is a computational biologist whose work centers on integrating proteomic and genomic data into the study of cancer to improve overall cancer care. Analyzing the proteomic data helps to understand what exactly within the tumor needs to be targeted in order to see the most impactful results from treatment. At Baylor, Dr. Zhang will continue his work in proteogenomics and systems biology focusing on their impact in the treatment of breast cancer.

**Lilei Zhang, M.D., Ph.D.**  
Assistant Professor

The overarching theme of Dr. Zhang’s laboratory is to understand the genomic and epigenomic regulation of the cardiovascular system in health and in disease with an emphasis on heart failure and cardiomyopathies. The laboratory uses combinatorial tools of transcriptomics and cistromics in rodent models and human induced pluripotent stem cell-derived cardiomyocytes to uncover the genetic control of cardiac adaptation and pathogenic remodeling. Their ultimate goal is to design novel therapeutics through direct targeting of gene regulation.

**Diagnostic Laboratory, Genetic Counseling, and Clinical Faculty**

- **Brett Bostwick, M.D.**  
  Assistant Professor
- **Zhao Chen, Ph.D.**  
  Assistant Professor
- **Magalie Leduc, Ph.D.**  
  Assistant Professor
- **Linyan Meng, Ph.D.**  
  Assistant Professor
- **George Miles, M.D., Ph.D.**  
  Assistant Professor
- **Marcus Miller, Ph.D.**  
  Assistant Professor
- **Dan Riconda, B.S., M.S.**  
  Associate Professor
- **Xia Wang, Ph.D.**  
  Assistant Professor