I’m delighted to welcome you to our 2015 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 #1-ranked genetics department in the country based on total 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.

I’m also pleased to report the ongoing success of our recent partnership with Miraca Holdings, Inc., which established the Baylor Miraca Genetics Laboratories. This jointly governed entity combines operational excellence with innovation and is a major cornerstone of our success. The genetics laboratories fully support the Department’s academic mission and promise to extend the impact of genetic diagnostic testing to patients worldwide.

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.
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The Department of Molecular and Human Genetics has made incredible progress since its fledgling beginning in the early 1970s. 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.

Genetic research at Baylor College of Medicine began in 1971 when Drs. C. Thomas Caskey and, soon thereafter, Arthur Beaudet were recruited from the National Institutes of Health (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 cadre 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 coalesce 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, Ph.D., 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. As 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 multi-functional diagnostic services to the broader medical genetics community. The Baylor Miraca Genetics Laboratories – which includes DNA Diagnostics, Cytogenetic Testing, and Biochemical Analysis – offers efficient, full-service genetic testing services to practitioners worldwide.

The past 40 years have been an exciting time of growth and change. From the Department’s incipient beginnings in the 1970s to the present, Baylor faculty and staff have transformed the practice and science of genetics. We can’t wait to make more history!
Joining Forces
Baylor and Miraca Holdings Partner to Expand Genetic Diagnostic Testing, Commercialize Biotechnology

In February 2015, after months of behind-the-scenes planning, Baylor College of Medicine and Miraca Holdings, Inc., a Japanese-based diagnostic laboratory, formalized a joint venture that promises to accelerate genetic diagnostic innovation and commercialize research into usable technology.

The $300 million deal provided Baylor’s Medical Genetics Laboratories (now the Baylor Miraca Genetics Laboratories, or BMGL) the platform needed to develop and market its genetic and genomic testing protocols to patients around the world.

“The goal is to really accelerate growth,” said Dr. Brendan Lee, chair of molecular and human genetics at Baylor and a member of the BMGL board. “We want to turn this into a $150 million to $200 million company in the next three to four years.”

The joint venture represents a drastic change in the way physicians approach health. Instead of over-the-counter remedies, doctors can offer personalized cocktails of medication based on patients’ own genomic makeup.

“The ability to provide people information regarding their healthcare [allows patients] to manage their health better,” said Gary Huff, CEO of BMGL. “We can have a bigger impact on healthcare as a whole.”

BMGL remains headquartered in Houston and employs approximately 225 people. Under the agreement, Baylor faculty continue to conduct independent genetic diagnostic research while also providing diagnostic services as part of BMGL.

Baylor and Miraca representatives unite for a successful joint venture
The College’s educational training programs in genetics and genomic sequencing operate through a formal academic affiliation with the joint Baylor-Miraca venture. Due to the partnership’s global reach, BMGL researchers and trainees can diagnose samples from around the world and, in the process, strengthen their laboratory skills.

The joint venture also helps transform Baylor’s pioneering biotechnology research into commercially viable, patient-ready products and therapies. By combining genetic diagnostics with practical medical applications, the partnership positions Baylor, and the broader Houston community, as the nation’s preeminent medical research and commercialization hub.

“It is an exciting time at BMGL,” said Mr. Huff. “We are working diligently to achieve our vision of being the elite genetics and genomics gold standard that empowers precision medicine.”

SPOTLIGHT

Critical Trio WES

When life-threatening diseases can strike without warning, speed matters. In years past, physicians had to wait 2 to 3 months to learn the results of genetic sequencing tests.

Now, with Baylor Miraca Genetics Laboratories’ launch of an enhanced clinical Whole Exome Sequencing test (Critical WES), that delivery time has been shortened to 2-3 weeks.

“The enhanced test is definitely a game changer,” said Dr. Carlos Bacino, a medical director of the BMGL. “We work with rare disorders and usually deal with very sick children. While some diagnoses can wait 3 to 4 months, others [require] a quick diagnosis [to] help with treatment and management.”

Technological advances and optimized processes and informatics make this rapid turnaround time possible. This has proven particularly useful in neonatal intensive care settings, where rapid results are critical to patient care.

“We have listened to physicians, patients, and families who need a [more] rapid molecular diagnosis,” said Dr. Christine Eng, vice president and executive laboratory director at BMGL.

“The enhanced test is definitely a game changer...”

—Dr. Carlos Bacino, medical director of the Baylor Miraca Cytogenetics Laboratory

Other conditions for which Critical WES proves valuable include intractable seizures, congenital heart disease, and suspected inborn errors of metabolism.

Critical WES is a trio analysis, meaning parental samples must be included. By sequencing the family trio, de novo mutations in disease and novel genes can be rapidly detected. This helps with both discovering the underlying genetic cause of the patient’s phenotype, as well as new gene discovery.
Baylor Miraca Genetics Laboratories

Continuing the Department’s 40-year tradition of excellence, the Baylor Miraca Genetics Laboratories (BMGL) has developed innovative testing approaches – including Chromosomal Microarray Analysis and Whole Exome Sequencing – that will carry the laboratory, and Baylor College of Medicine, well into the 21st century. BMGL has more than 200 employees, offers more than 3,000 diagnostic tests, and services clients from all 50 states and 16 countries worldwide. BMGL’s six laboratories are highlighted below:

The Biochemical Genetics Laboratory tests, diagnoses, and monitors patients with inborn metabolic disorders. The laboratory tests for a broad array of analytes, offers more than 30 enzyme assays, and provides medical reports and consultations with the Director and Medical Geneticist.

The Cytogenetics Laboratory offers comprehensive diagnostic services, including high-resolution chromosome analysis, fluorescence in situ hybridization (FISH), and Chromosomal Microarray Analysis (CMA). The laboratory leads the field in the use of CMA, particularly as a prenatal diagnostic tool.
The DNA Diagnostic Laboratory provides DNA-based testing for the molecular diagnosis, carrier testing, and prenatal testing of hereditary disorders. The laboratory provides DNA tests for more than 40 genetic diseases, and continues to develop and implement improved testing methodologies.

The Mitochondrial Laboratory offers comprehensive molecular DNA analysis of mitochondrial disorders, such as common point mutations and deletions in the mitochondrial genome. It further provides whole mitochondrial genome sequence analysis and DNA sequence analysis.

The Cancer Genetics Laboratory provides gene sequencing testing, deletion/duplication testing, chromosome analysis, as well as FISH and CMA tests. The lab also offers Cancer DNA Sequencing, which uses next-generation sequencing techniques to identify acquired changes in the DNA of patients’ tumors.

The Whole Genome Laboratory brings together genomic scientists, clinical laboratory scientists, and clinicians to provide reliable, genome-wide analyses and state-of-the-art, Whole Exome Sequencing. ⚫
Every day, millions of people suffer from diseases that have never been identified. In years past, they would embark on prolonged medical odysseys to find the source of their physical and developmental problems – often to no avail.

Now, with the recent creation of the Undiagnosed Disease Network (UDN), Baylor College of Medicine geneticists and researchers hope to diagnose – and ultimately cure – the type of rare, genetic disorders that previously went untreated.

The multi-state Network is comprised of seven clinics and research centers, each of which contributes local medical expertise. Baylor, which is home to the Human Genome Sequencing Center, will serve as one of two DNA sequencing cores for the UDN.

“This Network brings experts together from across the country to help solve the most difficult medical cases,” said Dr. Brendan Lee, chair of molecular and human genetics at Baylor and the lead investigator at the College. “We hope to play an important role by bringing together our renowned experts in genetics, pediatrics, and neurology.”

Collecting and comparing patient data is crucial to the UDN’s success. Network physicians share clinical and laboratory data – including genomic
information, clinical observations, and documentation of environmental exposures. If two or more patients share similar symptoms and genetic variations, researchers can establish causal links that may previously have gone undetected.

“A big part of the program will be sequencing the patient’s whole genome,” said Dr. Lee. “That information will then be collected and integrated by a team of experts in different areas – pediatrics, medicine, neurology, genetics, and others – so we can determine what are the most likely diagnoses.”

The UDN’s goals are ambitious. In addition to pinpointing the genetic causes of mystery diseases, the Network hopes to make new discoveries that will broadly impact the world of medicine.

“We are focused on helping the patient, but the patient also helps us to advance medicine broadly,” said Dr. Lee. “By focusing on undiagnosed and difficult-to-diagnose diseases, we may actually highlight something that no one has previously observed. These types of discoveries are the ones that can transform medicine and give us a more refined, higher-resolution picture of the human body.”

How do genes become diseased? For Dr. Christian Schaaf, an assistant professor of molecular and human genetics, the question isn’t merely academic. Thirty-nine million people worldwide are afflicted with rare diseases – and identifying the genetic mutations that cause their existence is the first step in combating them.

Recently, Dr. Schaaf and his colleagues identified how mutations in the gene USP7 lead to (previously undiagnosed) developmental disorders.

These mutations begin with a trio of genes – MAGEL2, TRIM27, and USP7 – that affect the recycling of proteins within cells. If the proteins associated with these genes fail to regulate each other, the genes become diseased.

“This [self-regulatory] process keeps everything in balance,” says Dr. Schaaf. “If you lose the function of MAGEL2, the disease called Schaaf-Yang results. If you lose the function of USP7, it may cause a disease that looks very similar – molecularly and clinically.”

Dr. Schaaf’s discovery has given hope to the children and families suffering from this disease. By understanding the causal factors that lead to genetic disorders, we just might be able to cure them.
Gene Mutations Found to Cause Lung Disease, Arthritis

Researchers led by those at Baylor College of Medicine recently identified the genetic mutation that causes hereditary autoimmune-mediated lung disease and arthritis. By sequencing the exome of five unrelated families with similar symptoms, the multi-institutional team was able to pinpoint the mutated gene that causes the disease.

The gene involved, called coater subunit alpha (or COPA), is an autosomal dominant genetic condition – meaning if a person inherits the mutated gene, he or she has the disease.

Baylor researchers, Drs. James Lupski and Jordan Orange, along with Dr. Anthony Shum of UCSF, led the study. By comparing the whole exomes of the patients to those of their disease-free siblings, the team could identify the mutated gene.

Going forward, Dr. Lupski and his team hope to apply the results from this study to better understand – and eventually treat – COPA syndrome and other genetic diseases.

“We are excited to learn how variants of this disease might be more broadly applicable,” says Dr. Orange. “[They] might be instructive to our overall understanding and treatment of arthritis.”

Genetics Researchers Perform Surgery on Human Genome, Changing How it is Folded Inside the Cell Nucleus

In a pioneering study that promises to upend the genetics field, a team of researchers led by those at Baylor’s Center for Genome Architecture recently performed the first successful genome surgery – changing, in the process, how the genome is folded inside the nucleus. The advance may lead to new methods of understanding and overcoming genetic diseases.

Dr. Erez Aiden, director of the Center, demonstrated that when the 2-meter long human genome folds up inside the nucleus of a cell, it forms roughly 10,000 loops. These loops turn genes on and off, and control how long stretches of the genome are packed. Anomalies in this folding process can lead to disease.

Dr. Aiden and his team also discovered a DNA codeword, or “motif,” that lies at both ends of nearly all loops: a string of fewer than 20 genetic letters that causes the DNA to bind a protein called CTCF. By manipulating these motifs, Dr. Aiden has demonstrated that it’s possible to destroy, move, and create new loops in the genome.

“We were able to use our insights into how loops form in nature in order to engineer genome loops artificially,” said Dr. Aiden. “This means that it is possible, at least in principle, to fix errors in genome folding by modifying a handful of genetic letters without disturbing the surrounding DNA.”
When a cell divides, it must split its DNA into two strands and then faithfully duplicate each strand to give the new cells the genetic material they need to thrive.

If the DNA breaks during that replication, the broken strands must be repaired. Most often, breaks occur when DNA is split into single strands during replication. Previous work suggested that such breaks are repaired by a process called break-induced replication – an error-prone mechanism that can cause loss of genetic information, and thus give rise to diseases such as cancer.

However, Baylor College of Medicine researchers – led by Dr. Greg Ira – have recently uncovered two primary ways in which cells limit such mutagenic repair. First, a high-fidelity replication fork quickly approaches the site of DNA damage and limits the error-prone repair. Second, an enzyme called Mus81 acts as molecular DNA scissors and converts unstable and mutagenic intermediates of repair to a stable fork structure.

“In the absence of Mus81, repair is significantly more prone to mutations,” says Ryan Mayle, a Baylor graduate student participating in Dr. Ira’s lab.

Mus81 is particularly important when a replication fork breaks in the vicinity of sequences that are repeated many times in the genome. By limiting the jump from one repeat to another during repair, Mus81 protects the human genome from unwanted chromosomal rearrangements. Because more than half of the human genome consists of repetitive sequences, Mus81 plays an important role in preserving genetic information and in limiting the likelihood of genetic diseases.

Baylor researchers recently discovered that altering brain cells’ lipid metabolism promotes the formation of lipid droplets that presage the loss of neurons. By using fruit fly mutants to dissect the molecular mechanisms that underlie neurons’ demise, genetic scientists demonstrated that elevated reactive oxygen species (ROS) in the neurons promotes lipid synthesis, which causes lipid droplets to form in brain support cells.

Lipid droplets are organelles that serve as energy storage depots. They accumulate in the brain support cells when defects in the mitochondria of neurons lead to elevated levels of ROS.

“ROS or lipid droplets alone do not lead to the rapid onset of neurodegeneration,” says Dr. Hugo Bellen, who leads the research team. “The synergism of ROS with lipid droplets is key. Reducing one or the other delays neurodegeneration.”

Dr. Bellen and his team further showed that reducing many of the components of this pathway can delay neurodegeneration. For example, treatments with a blood-brain-barrier penetrating antioxidant can delay the onset of neurodegeneration in flies and mice.
The pace at which scientific discoveries lead to improved health is often frustratingly slow. New drugs and therapies take time to reach patients, and healthcare advances often fail to keep pace with the vast amounts of data generated by biomedical researchers.

Now, the newly established Division of Clinical Research promises to change that.

The Division will provide researchers and clinical investigators with advanced facilities to enable new gene discovery, validate new biomarkers for disease, and explore new therapies for genetic disorders in robust clinical trial settings.

“Recent advances in the understanding of genetic disorders have provided unparalleled opportunities to translate basic, mechanistic findings into patient-oriented outcomes,” says Dr. Sandesh Nagamani, director of clinical research at Baylor College of Medicine. “I am very excited to be a part of this endeavor.”

The Division is already conducting clinical investigations on inborn errors of metabolism, genetic disorders of the skeleton, and mitochondrial diseases.

The Division consists of three research nurses, one project manager, and numerous research coordinators. Together they will explore ways to increase the speed and efficiency with which research discoveries are translated into advances in patient care.
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 continues to distinguish itself.

For 5 years running, we have remained the #1-ranked U.S. genetics department, as measured by the number of NIH-awarded grants and total funding received – and 2015 was our best year yet! We received NIH awards totaling $81 million.

As excited as we are to receive this funding, it’s what we do with the support that counts. And we’ve put that funding to excellent use. Whether funding the establishment of the Undiagnosed Disease Network Center or the Knockout Mouse Project, the Department is finding answers to science’s most pressing questions. In the process, we are improving the well-being of patients across the world.

Ranked #1 in NIH Funding

Other Grants/Awards

The Department is proud to receive generous funding from many agencies and foundations some of whom 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
For patients with genetic disorders who need comprehensive evaluation and treatment, Baylor College of Medicine’s genetics program offers specialized care and unparalleled services and testing. As the largest clinical genetics program in the country – with 14 clinics spanning across multiple genetics-based disciplines – we take a collaborative approach that provides patients the highest-quality, individualized care available. Our clinical activities span across several sites including Texas Children’s Hospital, Baylor St. Lukes Medical Center, the Michael E. DeBakey Veterans Affairs Medical Center, and the Harris Health System.

**Adult Genetics Clinic**

The Adult Genetics Clinic, staffed by board-certified medical geneticists, provides genetic evaluation and counseling for adult patients with a variety of genetic and inherited disorders – such as skeletal dysplasias, connective tissue, neuromuscular, and metabolic disorders. Patients with a variety of inherited disorders may be referred for genetic counseling and further diagnostic testing.

Shweta U. Dhar, M.D., *Clinic Director*

**Adult Cardiovascular Genetics Clinic**

The Adult Cardiovascular Genetics Clinic, established in 2010, services the needs of patients and families with familial heart conditions – such as cardiomyopathy, sudden cardiac death, Long QT, and congenital heart defects. The clinic also focuses on the cardiovascular complaints of patients with various genetic syndromes such as Marfan syndrome, Loey-Dietz syndrome, Duchenne Muscular Dystrophy, Williams syndrome and velocardiofacial syndrome. The clinic is staffed by a board-certified clinical geneticist and a cardiologist.

Shweta U. Dhar, M.D., *Clinic Director*

**Neurogenetics Clinic**

The Neurogenetics Clinic provides genetic evaluation and counseling for patients with a variety of inherited disorders that primarily affect the nervous system, with special emphasis on neurodegenerative and metabolic diseases. A neurologist specializing in medical genetics staffs the clinic. The Neurogenetics Clinic is fully integrated with the Adult Genetics Clinic, where internists trained in medical genetics are available to consult on medical problems outside the nervous system.

Paolo M. Moretti, M.D., *Clinic Director*
Clinic for Metabolic and Genetic Disorders of Bone

The Clinic for Metabolic and Genetic Disorders of Bone offers medical services to adult patients with a variety of bone disorders – such as osteoporosis, osteomalacia, rickets, and metabolic bone disease. An internist, a clinical geneticist specializing in genetic disorders of bone, and a certified genetic counselor, staff the clinic.

Services essential to the multidisciplinary management of bone disease – including pain management, physical therapy and rehabilitation, orthopedics, genetic testing services, and infusion center – are readily available within the Baylor Clinic system.

Sandesh C.S. Nagamani, M.D., Medical Director

Pediatric Genetics Clinic

The Texas Children’s Hospital (TCH) Genetics Clinic, as the largest in the country, leads the way in pediatric genetic research, while offering families the latest diagnostic and clinical resources available.

The TCH Genetics Clinic consists of 18 board-certified medical geneticists, three board-certified genetic counselors, and six genetics nurses. Clinic physicians see more than 3,000 families each year.

Children are referred to the clinic for many reasons, including developmental delays or suspected genetic conditions. The clinic also offers prospective parents in-depth counseling and genetic tests to identify pregnancy risks and reproductive options.

Specialty clinics within the TCH Genetics Clinic include the Metabolic Clinic, Neurofibromatosis clinic, Skeletal Dysplasia Clinic, and Cancer Genetics Clinic.

Carlos Bacino, M.D., Chief, Pediatrics
William J. Craigen, M.D., Chief, Neurofibromatosis Clinic
Brendan Lee, M.D., Ph.D., Chief, Skeletal Dysplasia Clinic
Pilar Magoulas, M.S., C.G.C., Chief, Genetic Counseling
Sharon E. Plon, M.D., Ph.D., Director, Cancer Genetics Clinic
Vernon R. Sutton, M.D., Director, Inborn Errors of Metabolism Service

Prenatal Genetics Clinic

The Prenatal Genetics Clinic specializes in prenatal and reproductive genetic risk assessments, as well as the latest genetic testing technologies. The clinic, as the largest of its kind in the United States, offers comprehensive clinical and research expertise to ensure that families receive the best possible care.

Ignas Van den Veyver, M.D., Clinic Director
Human Genome Sequencing Center

Baylor’s Human Genome Sequencing Center (HGSC) has been at the forefront of genomic innovation for 20 years. 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, and continues to push the boundaries of scientific discovery.

The HGSC is currently exploring individual DNA sequence variation and its association with human diseases – such as Parkinson’s disease, childhood diabetes, bipolar disorder, and others. In addition, the Center is characterizing normal genetic variation in human populations, the results of which are anticipated to change the way we understand disease and other life processes.

Since the completion of the human genome project, the HGSC has sequenced many other genomes, including the mouse, rat, fruit fly, rhesus monkey, honey bee, orangutan, and cow. Additional sequencing projects are currently under way for many other species – including the dolphin, baboon, and more than 200 different microbial organisms. By comparing these non-human genomes with ours, HGSC research scientists can more easily predict where genes are located in human chromosomes. This will lead to a better understanding of how genomes have evolved, and will further expand our knowledge of DNA, the building blocks of life.

Richard Gibbs, Ph.D., Director

Neurological Research Institute

As different as they are, all neurological disorders (Epilepsy, stroke, Alzheimer’s, traumatic brain injury, Autism) have one thing in common: they strike at the core of our identity. They are also shockingly prevalent: an estimated 1 billion people around the world suffer from some form of neurological disease.

Less than a generation ago there was little hope for treating most of these disorders, but we are now beginning to appreciate the brain’s resilience and its capacity for renewal.

Less than a generation ago there was little hope for treating most of these disorders, but we are now beginning to appreciate the brain’s resilience and its capacity for renewal. We are acquiring new tools to illuminate neurological function and dysfunction; more scientists and clinicians are studying these disorders than ever before. We are, in short, on the cusp of a new era in biomedicine.

To meet this moment of opportunity, Texas Children’s Hospital (TCH) and Baylor conceived the Neurological Research Institute (NRI). Building on our considerable strengths in genetics, neuroscience, and pediatrics, NRI is carefully designed to foster collaboration among 30-40 basic faculty and clinician-scientists with the goal of developing treatments for childhood neurological diseases.

Huda Zoghbi, M.D., Director
Huffington Center on Aging

The Huffington Center on Aging (HCOA), one of the premier centers on aging in the world, was formed in 1988 to study aging, provide needed healthcare for older people, and teach future health professionals and researchers about geriatrics and gerontology.

Today, the Center aims to translate research into improved quality-of-life outcomes for people as they age. It also educates the general public on health and social practices that can assist them to have a long, well-lived life.

The Center uses a multifaceted approach to improve the lives of aging individuals, and maintains excellence through:

- Pursuing research to understand all components of aging, and develop novel therapeutic and treatment options
- Educating future leaders in gerontology, geriatrics, and the biology of aging
- Enhancing elder quality of life by providing inpatient and outpatient care, and working with the section of Geriatrics in the Department of Medicine
- Providing resources to the general public to improve their knowledge of aging and healthcare, and offer lifestyle advice for healthy aging

HCOA facilitates and coordinates interdepartmental research and initiates its own research studies. HCOA-initiated research includes cell and molecular biology of aging, adrenal cell biology, DHEA, aging of the skin, the aging cardiovascular system, healthcare outcomes, and ethical issues in acute and long-term care settings.

Dr. Hui Zheng, Ph.D., Director

Computational and Integrative Biomedical Research Center

The Computational and Integrative Biomedical Research Center 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.

As part of its mission the CIBR Center supports research and education through hardware and software; through seminars and computational workshops; and through consultations and seed grants that promote new research programs.

Olivier Lichtarge, M.D., Ph.D., Director

Center for Skeletal Medicine and Biology and The Rolanette and Berdon Lawrence Bone Disease Program of Texas

Baylor College of Medicine is a formal partner in the Rolanette and Berdon Lawrence Bone Disease Program of Texas (BDPT), working side-by-side with MD Anderson Cancer Center and UT Health. In 2013, Baylor leveraged its involvement in the BDPT to establish the Center for Skeletal Medicine and Biology (CSMB). This unique Center brings together diverse researchers within the clinical and basic science departments at Baylor to develop coordinated clinical, research, and training opportunities broadly focused on musculoskeletal health disorders. These disorders include osteoarthritis, tendon and ligament injury, cancer of the skeleton, and genetic disorders such as skeletal dysplasias.

Brendan Lee, M.D., Ph.D., Director

Intellectual and Developmental Disabilities Research Center

The Baylor College of Medicine Intellectual and Developmental Disabilities Research Center is one of 14 such centers funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development; it is also a member of the Association of University Centers on Disabilities.

The IDDRC is committed to advancing research in Intellectual and Developmental Disabilities in order to contribute to the resolution of many of the problems encountered by individuals with IDD and their families. The goals of the Center are to identify causes of mental retardation and disability, to prevent these disorders, and to provide interventional schemes that can improve individuals’ quality of life.

Huda Zoghbi, M.D., Director
The Graduate Program in Molecular and Human Genetics, within the Baylor College of Medicine Graduate School of Biomedical Sciences and led by Gad Shaulsky, Ph.D., 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,” says 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. In 2015, 12 deserving students were awarded one or more prestigious awards and fellowships. Join us in congratulating these present and future superstars:

Richard Chapple  
Cynthia Kim  
Peter Kundert  
Eugene Lurie  
Kristen Meyer  
Chih-Chun-Lin  
Angel Lopez  
Ninad Oak  
Seung-Yeop Han  
Meagan Siehr  
Priva Sivaramakrishnan  
Jessica Sowa

Our location within the Texas Medical Center provides students endless opportunities to obtain education and practical experience in both basic and applied research. Come join us, and let’s make history together!
The Medical Genetics Residency Program, led by Dr. V. Reid Sutton, M.D., prepares individuals for an accomplished academic career by providing an integrated experience in both clinical and experimental genetics. Our 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.

Our program enjoys preeminence in the genetics community and is approved by the Accreditation Council for Graduate Medical Education. We are also supported by a training grant from the National Institute of General Medical Sciences.
Our fellowship program provides post-doctoral 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 for 24 months. At that time, they are eligible for board certification by the American Board of Medical Genetics and Genomics. We are proud to offer fellowship training programs in the following areas:

**Clinical Biochemical Genetics.** Trainees spend 3 months learning each of the following methods: tandem mass spectrometry, gas chromatography/mass spectrometry, high-pressure liquid chromatography (amino acid analysis), and enzyme analysis.

**Clinical Cytogenetics.** Trainees receive instruction and experience on basic cytogenetic techniques as well as pioneering cytogenetic-molecular genetic diagnostic technology. Upon completing our program, individuals are well-prepared to use microarray-based diagnostic tools and interpret standard cytogenetic analyses.

**Clinical Molecular Genetics.** Our DNA laboratory is one of the oldest diagnostic and general and mitochondrial/metabolic molecular laboratories in the United States. Fellows learn a variety of molecular diagnostic techniques during their rotational period.

Medical Genetics Trainees’ Locations - United States
This lectureship was established in the 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.
Jeanette Oshman Efron, who passed away in 2009 at the age of 98, was an ardent supporter of science and the arts – and a generous friend to Baylor College of Medicine.

The Oshman Lectureship in Molecular Genetics was established at Baylor in 1989 by her daughters, Marilyn Oshman and Judy Margolis, and her grandchildren, Karen Desenberg, Gary Gerson, Jay Gerson, and Andrew Lubetkin, to honor Jeanette’s passion and commitment to the advancement of medical education and biomedical research.

This lecture series brings internationally renowned scientists to Baylor to present seminars on important developments in genetics. Shirley Tilghmann of Princeton, Lucy Shapiro of Stanford, and Joan Steitz of Yale University are some of our most recent guests, as well as Richard Axel and Joe Goldstein.

To quote her daughter, Marilyn Oshman, “She helped put people through school and gave and loaned them money,” Marilyn Oshman said. “She was amazingly generous.”
DNA is the blueprint of human life. At its core, it contains sets of instructions for making proteins – which do all the work of keeping our bodies functioning. More specifically, the structure and behavior of proteins play an enormous role in determining our health.

Dr. Susan Lindquist, Ph.D., the 2015 Jeanette Oshman Efron Lecture speaker, is a pioneer in the study of protein folding – how proteins adopt the extraordinarily complex structures that allow them to carry out their various functions. She has shown that changes in protein structure can have profound effects on mechanisms of inheritance, evolutionary processes, and how we age. Because protein folding is a universal requirement for all life forms, the Lindquist lab has been able to develop a "living test tube" model in yeast to study protein folding transitions in neurodegenerative diseases such as Alzheimer’s and Parkinson’s disease.

Dr. Lindquist is a Professor of Biology at MIT, a Howard Hughes Investigator, and a member of MIT’s Whitehead Institute for Biomedical Research, which she also directed from 2001-2004. She was named one of the 50 most important women in science by Discover Magazine in 2002, and is a member of the National Academy of Sciences, the Institute of Medicine, and the Royal Society. Dr. Lindquist participated in the 2007 World Economic Forum in Davos, Switzerland, and in 2010, President Obama presented her with the National Medal of Science.

**Seminar Series**

Although geneticists’ schedules are exceedingly busy, large numbers make a point to attend the Department’s regularly scheduled seminar series. In 2015, a total of 28 presentations were made by some of the most accomplished geneticists in the country. A sampling of these speakers included Dr. Aaron Gitler from Mount Sinai Hospital who spoke about “Noonan Syndrome and related disorders: RASpy voices heard along the pathway”; Gerry Shadel from Yale School of Medicine who shared information on “Mitochondrial Stress Signaling in Disease, Aging and Immunity”; and Kat Hadjantonakis from Memorial Sloan Kettering Cancer Center who delivered information on “Guts and Gastrulation: Cell dynamics driving lineage specification and morphogenesis in the mouse embryo.”
Families who have a loved one with a genetic disorder are eager to learn more. What caused the disorder? How can we improve quality of life? What advances are on the horizon?

The broader community, too, has an interest in learning about recent advances in genomic research and medicine.

**What caused the disorder? How can we improve quality of life? What advances are on the horizon?**

To that end, the Department’s *Evenings with Genetics* seminar series is designed to provide a community forum to communicate current information on healthcare, education, and research regarding a variety of genetics-based diseases. Since its inception in 2006, the series has:

- Provided current information to families impacted by a genetic condition.
- Increased community knowledge of genetic care and research issues.
- Aided family-to-family support.
- Fostered increased collaboration amongst geneticists, related medical specialists, and care providers.

Seminar topics are based on family requests, input from healthcare providers, and new understanding of genetic conditions. For each seminar, medical geneticists pair with a Baylor College of Medicine physician, and family members impacted by the genetic condition are invited to share their unique perspectives.

“It is incredibly rewarding when parents tell us they have used the information and resources from a seminar to help their child,” says Dr. Susan Fernbach, Director of Genetic Outreach at the College. “These seminars come alive with the combination of genetics faculty providing cutting-edge information and parent-speakers providing [their] unique perspectives.”

Seminars are held one evening per month at the Children’s Museum of Houston. Many seminars are videotaped and available to view on the Baylor website. We invite all members of the community to attend and hope to see you soon!
Department Leadership and Faculty

LEADERSHIP

Brendan Lee, M.D., Ph.D.
Chair

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

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

Faculty includes:

2 Howard Hughes Medical Investigators
3 Members of the National Academy of Sciences
4 Fellows of the American Association Advancement of Science
6 Members of the National Academy of Medicine
Recent Faculty Arrivals

The recruitment and retention of great faculty propels our department forward. Here are some of the faculty hires we welcomed from 2013-2015.

Erez Aiden, Ph.D.
Assistant Professor

Dr. Aiden and his team study the processes by which genomes are folded within the nuclei of functioning cells. His lab integrates advanced molecular technologies, high-throughput DNA sequencing, as well as powerful computational and biophysical methods, to better understand the genomic folding process and its effect on other cellular pathways. In addition, the Aiden lab makes use of massive datasets to make previously intractable measurements (such as three-dimensional genome sequencing) possible.

Weiwei Dang, Ph.D.
Assistant Professor

Dr. Dang studies how chromatin and epigenetic pathways regulate aging and age-related diseases. His team uses budding yeast replicative aging as a model to identify how various histone modifications change during aging and how epigenetic factors regulate longevity through chromatin remodeling. In addition, the Dang lab has developed an innovative and high-throughput longevity screen method that significantly accelerates the discovery of molecular pathways of aging.

Florent Elefteriou, Ph.D.
Associate Professor

Dr. Elefteriou’s research focuses on multiple aspects of skeletal biology, including bone development, bone repair, and bone physiology. His group investigates the etiology of bone maladies associated with neurofibromatosis type 1 and other RASopathies, the biological and clinical relevance of the interaction between the autonomic nervous system and bone, and the mechanisms by which metastatic breast cancer cells disseminate into the skeleton.

Sarah Elsea, Ph.D.
Associate Professor

Dr. Elsea focuses on understanding the biochemical mechanisms and molecular pathways that underlie human neurodevelopmental disorders—particularly those disorders involving autism, intellectual disability, seizures, and behavioral phenotypes. By using a variety of model systems and diagnostic techniques, including transcriptomics and metabolomics, the Elsea lab has improved diagnoses, enhanced understanding of phenotypes, and provided insight into the molecular relationships among neurodevelopmental disorders.
Neil Hanchard, M.D., Ph.D.
Assistant Professor

Dr. Hanchard’s lab uses genomics to explore complex pediatric disease traits. The lab has a special interest in diseases of global health relevance, and is currently integrating genome sequence variation with transcriptomics, epigenomics, and population genetics to understand disease pathogenesis in severe childhood malnutrition, pediatric HIV/AIDS, and sickle cell disease. Dr. Hanchard also sees patients in the Clinical Genetics Clinic at Texas Children’s Hospital.

Charles Lin, Ph.D.
Assistant Professor

Dr. Lin studies cancer through the lens of altered gene regulation, as practical cancer therapeutic strategies require an understanding of the complex transcriptional mechanisms driving tumorigenesis. Dr. Lin and his team apply computational, molecular, and chemical biology approaches to understand how gene regulation and downstream pathways are altered in cancer. The Lin lab seeks to better understand mechanisms of transcriptional deregulation by observing whether compounds that target chromatin and transcription regulators affect oncogene transcription and tumor proliferation. As these compounds transition into clinical development, Dr. Lin will work to understand how they alter gene control in tumor cells, and their potential therapeutic benefits for use in targeting different cancers.

Sandesh Nagamani, M.B.B.S., M.D.
Assistant Professor

Dr. Nagamani translates basic science discoveries into potential therapies for various genetic disorders. More specifically, Dr. Nagamani conducts natural history studies, proof-of-concept pilot studies, and interventional clinical trials in patients with inborn errors of metabolism and skeletal dysplasias. In addition, as the Director for the Clinical Research Division, Dr. Nagamani facilitates other investigators’ human subject-related research activities.

Dongsu Park, Ph.D.
Assistant Professor

Dr. Park seeks to understand the biology of mesenchymal/skeletal stem cells in tissue regeneration and cancer. By using genetic pulse-chase models and advanced, live-animal imaging technology, Dr. Park and his team hope to demonstrate the lifespan and unexpectedly short-term recycling of osteoblasts in vivo. The Park lab also seeks to address the identity and function of mesenchymal/skeletal stem cells in the context of skeletal tissue regeneration in living animals. It is currently exploring the clinical relevance of these cells in inherited bone disorders and cancer bone metastasis.

Christian Schaaf, M.D., Ph.D.
Assistant Professor

Dr. Schaaf investigates the genetic causes of neurodevelopmental disorders, such as intellectual disabilities and autism spectrum disorder. Several new gene discoveries have been made possible by his translational research approach, which spans from the clinic to the basic science research laboratory. Dr. Schaaf and his team use mouse models and induced pluripotent stem cells to understand the molecular and functional consequences of genes that cause neuropsychiatric disease.

Chenghang (Chuck) Zong, Ph.D.
Assistant Professor

Dr. Zong’s research lies in the interface between novel single cell technologies and quantitative biology. Dr. Zong and his team pursue the development of new quantitative and high-throughput methods for characterizing genomic, epigenetic and transcriptional variations at single cell resolution. His lab’s main focus is on pancreatic cancer and the detection of early events that drive tumorigenesis as well as the early stage of tumor heterogeneity that will influence later tumor development.