Molecular and Human Genetics
Annual Report 2012

BCM
Baylor College of Medicine
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In 2012, we celebrated the 25th anniversary of the Baylor College of Medicine Department of Molecular and Human Genetics. This milestone provided an opportunity for all those connected to the department—faculty, staff, trainees, alumni—to reflect back on the many ways in which we have all contributed not only to the development of the department, but the revolution in human genetics that is transforming both science and medicine.

We marked this event with a scientific symposium and gala. A video celebrating the department’s history, which was prepared for the event, is available on our website at http://www.bcm.edu/genetics/history.cfm.

While you are on the site watching the video, I encourage you to explore the rest of our site to learn about the latest developments in the department.

Over the past 25 years the BCM Department of Molecular and Genetics has helped transform medical genetics from a specialty with a narrow focus on relatively rare genetic disorders to an integral component of the study and care of a wide variety of common diseases. In this report we explore some of the progress made in just the last year and provide an overview of the diverse fronts on which we are currently working.

Best regards.
Arthur L. Beaudet, M.D.
Henry and Emma Meyer Chair in Molecular Genetics
Professor, Department of Molecular and Human Genetics
In January, the department hosted its 25th anniversary scientific retreat at the Moody Gardens in Galveston, Texas. Former faculty, postdocs, and students were invited to be part of this special occasion.

A gala was held at the Houston Museum of Natural Science and afterwards in Galveston, there were three days of seminars, poster sessions, symposiums, and meetings with some of the top scientists in the world.

The Alumni Scientific Symposium featured Drs. Allan Bradley, Edward McCabe, Robert Nussbaum, and Andrea Ballabio just to name a few along with keynote speakers Drs. Arthur Beaudet and C. Thomas Caskey, the visionaries responsible for creating the Molecular and Human Genetics Department and ensuring its success.

Close to 400 people attended and participated in showcasing their expertise in various fields of interests. In addition, they enjoyed watching the annual skit presentations and attending various events. On the last day, Dr. Beaudet gave his “State of the Department” address along with an awards ceremony.
History of the Department

To view the video celebrating the department’s many historic contributions, visit http://www.bcm.edu/genetics/history.cfm.

The Institute for Molecular Genetics was created in 1985 and renamed Department of Molecular and Human Genetics in 1994. The research interests of the more than 50 primary faculty members span such important areas as functional genomics, genome instability, genome sequencing, mammalian development, the metabolic bases for inherited human disease, gene therapy, genomic disorders, gene structure and expression, mechanisms of replication and repair, DNA recombination, somatic cell genetics, mouse molecular genetics, cytogenetics, bioinformatics, bacterial genetics, yeast genetics, and Drosophila genetics.

The facilities in the Department are equipped with state-of-the-art instrumentation for research in molecular, cellular, and biochemical genetics. In addition, there are several facilities for specialized techniques to support the research efforts. Among these are core laboratories for microarray analysis, next generation sequencing, production of transgenic and knockout mice, mouse phenotyping, microscopy and imaging, confocal microscopy, high throughput screening, siRNA screening and other techniques. The Department's various research, clinical, and administrative activities currently occupy over 150,000 square feet of space throughout the Texas Medical Center.

The faculty continuously receives numerous National Institutes of Health, National Science Foundation, March of Dimes, and other competitive research grants. They have also received national recognition and support from the Pew Foundation and the Howard Hughes Medical Institute.

The Department continues to rank high in both the number of grants and total funding from the National Institutes of Health among genetics departments at U.S. medical schools.
The National Institutes of Health (NIH) is an agency of the United States Department of Health and Human Services and is the primary agency of the United States government responsible for biomedical and health-related research. NIH is made up of 27 Institutes and Centers, each with a specific research agenda, often focusing on particular diseases or body systems. The ability to continuously obtain NIH grants, through a competitive peer review process, demonstrates the strength of the Department’s research and training programs.

The BCM Department of Molecular and Human Genetics received more funds from the NIH in 2012 than any genetics department at a U.S. medical school.

Other sponsors of our research and training programs which include the American Heart Association, Angelman Syndrome Foundation, Autism Speaks, Brown Foundation, Burroughs Wellcome Fund, Cancer Prevention and Research Institute of Texas (CPRIT), CHDI Foundation, Department of Defense, Doris Duke Foundation, Howard Hughes Medical Institute, Huffington Foundation, International RETT Syndrome Foundation, Lustgarten Foundation, March of Dimes, Marshall Heritage Foundation, McNair Foundation, Mizutani Foundation, Muscular Dystrophy Association, National Alliance for Research on Schizophrenia and Depression, National Science Foundation, Osteogenesis Imperfecta Foundation, Rett Syndrome Research Trust, Simons Foundation, Team Sanfilippo Foundation, Texas Department of State Health Services, and other various industry and individual sponsors.

The ability to continuously obtain NIH grants, through a competitive peer review process, demonstrates the strength of the Department’s research and training programs.
The work of the BCM department of Molecular & Human Genetics attracts attention from the scientific and lay media around the world. In addition, department faculty are frequently sought as experts on a wide range of issues as illustrated by this sampling of media coverage in 2012.

**Dr. James Lupski** and genetics research led by a team at BCM were featured on the nationally televised CBS show “The Doctors.” The segment aired Tuesday, March 6 and can be seen in two parts on the show’s website Finding a Cure Through Gene Mapping - Part 1/Gene Mapping - Part 2

**Dr. Brendan Lee** was featured on ABC News in a segment on Jonathan Oliphint, 17, who has a rare condition known as argininosuccinic aciduria, or ASA, which meant his body could not break down certain toxins. The segment can be found at [http://abcnews.go.com/Health/boy-toxic-buildup-breakthrough-treatment/story?id=16213994](http://abcnews.go.com/Health/boy-toxic-buildup-breakthrough-treatment/story?id=16213994).


*The New England Journal of Medicine*

The Medical Genetics Laboratories (MGL) at Baylor College of Medicine has been dedicated to providing the medical genetics community with high quality comprehensive diagnostic services for over 30 years. By building on our institution’s strengths in research and discovery, we aim to provide quality genetic testing services relevant to 21st century medicine. The MGL is comprised of four laboratories.

The Biochemical Genetics Laboratory performs specific testing for the purpose of diagnosing and monitoring patients with inborn errors of metabolism. The laboratory provides testing for the quantitative determination of a broad array of analytes. In addition, over 30 enzyme assays are available for specific disorders. Professionals in human genetics provide reports with interpretations and telephone consultations are available 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 is leading the field in the use of CMA in clinical diagnostics including prenatal...
The Cancer Genetics Laboratory at Baylor College of Medicine now offers the Cancer Exome Sequencing test, which uses next-generation sequencing to identify acquired changes in the DNA of a patient’s tumor.

“Cancer exome sequencing is poised to change the current paradigm of genetic testing for cancer patients,” said Dr. Federico Monzon, director of molecular pathology at the Cancer Genetics Laboratory at BCM. “Rather than testing a single gene or panel of genes, cancer exome sequencing will provide a comprehensive profile of acquired mutations in tumor tissue.”

The term exome refers to the portion of the human genome that contains the DNA sequence that directs protein synthesis. These functionally important regions of DNA are referred to as exons. The 22,000 known genes are comprised of approximately 180,000 exons and represent about 3 percent of the genome. Most errors in DNA sequence that lead to altered protein function in tumors are located in the exons; therefore, exome sequencing is an efficient method for tumor DNA sequence analysis to uncover genetic causes for tumor behavior.

Some of these acquired mutations can be used to predict tumor aggressiveness or determine the likelihood of response/resistance to targeted agents or other forms of cancer therapy.

“We are entering a new era in individualized cancer diagnosis and treatment in which molecular profiling of the cancer as well as the patient will determine the optimal therapeutic approach for a given patient,” said Dr. C. Kent Osborne, director of the Lester and Sue Smith Breast Center and the NCI-designated Dan L. Duncan Cancer Center at BCM.

Developed jointly by the Cancer Genetics Laboratory and the Whole Genome Laboratory at BCM, this test is the result of collaboration between genomic and clinical laboratory scientists, pathologists, geneticists and oncologists to provide reliable cancer genome-wide analyses that are carefully annotated and interpreted for clinical significance by cancer geneticists and molecular pathologists.
Cancer Genetics Laboratory
The Cancer Genetics Laboratory (CGL) at Baylor College of Medicine is a combined effort of the Department of Molecular and Human Genetics, Department of Medicine Division of Hematology/Oncology, Texas Children’s Pathology Laboratory, and the Dan L. Duncan Cancer Center.

The CGL is positioned to provide clinical diagnostic testing in the field of cancer. By incorporating existing cancer centers and laboratories in the Texas Medical Center, the CGL optimizes the current testing menus of various institutions into one offering through the CGL.

The CGL is able to provide gene sequencing testing, deletion/duplication testing, chromosome analysis, FISH testing, and Chromosomal Microarray Analysis (CMA). CGL can provide cancer diagnostic testing services for both pediatric and adult cancer markets.

The CGL offers customers distinct advantages through its breadth of tests offered, rapid turnaround times, comprehensive data interpretation by faculty and laboratory directors skilled in cancer genetics, competitive pricing and access to insurance coverage through all major commercial insurance carriers and most small carriers as well as Medicare.

Whole Genome Laboratory
The development and clinical implementation of the Whole Exome Sequencing test derives from a joint effort by Baylor’s Human Genome Sequencing Center and the Medical Genetics Laboratories of the Department of Molecular and Human Genetics to establish a clinical laboratory dedicated to state-of-the-art next generation sequencing. The collaboration between these groups brings together genomic scientists, clinical laboratory scientists, and clinicians to provide reliable genome-wide analyses that are carefully annotated and interpreted for clinical significance by medical geneticists. Whole Exome Sequencing is the first test to be offered by the WGL and is focused on the evaluation of underlying genetic causes of disease. In the near future, the WGL will implement additional clinical tests, including Whole Genome Sequencing (WGS) that will bring this technology to other aspects of medical care and treatment.

The Whole Exome Sequencing for the Evaluation of Mendelian Disorders applies the power of next generation sequencing technology to clinical genetics in a Clinical Laboratory Improvement Amendment (CLIA) approved setting with clinical interpretation of the sequence information. Whole Exome Sequencing (WES) is poised to change the current paradigm of genetic testing for Mendelian disorders, pharmacogenetic traits, and potentially complex traits. Rather than limiting testing to a single gene or panel of genes and incurring diagnostic delays and escalating costs, the Whole Exome Sequencing test will sequence nucleotide by nucleotide, the human exome to the depth of coverage required to achieve a consensus sequence with high accuracy. Point mutations, insertions, deletions, inversions, and rearrangements of the exome are potentially discoverable and could be considered pathologic depending on the defect. The reporting of the Whole Exome Sequencing test will focus on known or predicted deleterious mutations in genes known to be associated with human disorders, however, significant potentially medically actionable findings in other genes of interest will also be communicated for future reference.
ADULT GENETICS

Shweta Dhar, M.D.
Director, Adult Genetics

Tanya Eble, CGC
Manager

General
The Adult Genetics Clinic provides genetic evaluation and counseling for adult patients with a variety of inherited disorders such as skeletal dysplasias, connective tissue, neuromuscular, and metabolic disorders, as well as other genetic disorders. Patients with a variety of inherited disorders may be referred for genetic counseling and further diagnostic testing.

Cancer Genetics
The Adult Cancer Genetics Clinic provides genetic counseling for adults who have been diagnosed or have a significant family history of breast, ovarian, colon, thyroid and other cancers. Molecular diagnostic testing is offered as indicated. The Cancer Genetics Clinic is staffed by physicians in the Division of Hematology-Oncology with board certification in medical genetics.

Cardiovascular Genetics
The Cardiovascular Genetics Clinic, established in 1996 as a team effort between the Division of Cardiology and the Department of Molecular and Human Genetics, serves patients and families with familial disorder of cardiac muscle and conduction, and children with extra cardiac anomalies associated with their heart disease. Additionally, individuals and families with vascular disorders such as Marfan syndrome and Ehlers Danlos syndrome are served by this clinic.
Metabolic and Genetic Disorders of Bone (CMGB)
The Clinic for CMGB is a multidisciplinary clinic that offers a full complement of medical services to adult patients with a variety of bone disorders like osteoporosis, osteomalacia, rickets, metabolic bone diseases due to abnormalities in calcium and phosphorus metabolism, genetic disorders of bone such as brittle bone disease, inherited defects of collagen, and developmental abnormalities of bone.

The CMGB is especially geared for the care of patients with family history of bone disease, early onset osteoporosis, and osteoporosis with suboptimal responses to conventional therapy. The clinic brings together the expertise of geneticists with experience in bone and mineral research, internists, dieticians, physical therapists and pain management specialists. The clinic renders state-of-the-art diagnostic evaluations including metabolic bone tests, densitometry, quantitative CT for volumetric analysis and testing for genetic variations that are known to be important in bone metabolism.

PEDiATRIC GeneTICS
Carlos Bacino, M.D.
TCH Chief of Service
Pilar Magoulas, CGC
Manager

General
The Pediatric Genetics Clinic, a joint effort of the Department of Molecular and Human Genetics at Baylor College of Medicine and Texas Children’s Hospital, is dedicated to providing comprehensive medical genetic consultation and services to patients and their families in issues dealing with heredity.

Patients are generally referred through their primary care physicians or other specialist. However, appropriate self-referrals are accepted.

The clinic is staffed by board-certified medical geneticists. The clinical staff renders appropriate screening, diagnosis, evaluation, counseling, and treatment for patients with a family history or a presumed diagnosis of a genetic condition. In addition to laboratory services provided by Texas Children’s, we also utilize our Department’s own state-of-the-art biochemical, cytogenetic, DNA diagnostic and mitochondrial laboratories located at BCM.

The Pediatric Genetics Clinic serves patients nationally and from around the world. The clinic constantly strives to maintain top professionals in the field and continues to conduct genetic research. Through research, we are able to make an impact in the field of genetics by providing new and exciting techniques and information which are recognized and adapted internationally.

Metabolic
The Metabolic Clinic, a joint effort of the Department of Molecular and Human Genetics at Baylor College of Medicine and
Texas Children’s Hospital, offers optimal care, follow-up and diagnosis to patients with inherited diseases that deal with defects of inborn errors of metabolism.

The clinic is staffed by board-certified medical geneticists. This clinic functions with input from the nutritionist to assist patients with proper diet management as it pertains to their disorder. Continual management of diet as well as monitoring of patient’s metabolites is an essential component of this clinic. Additionally, this clinic is diligent in the ongoing counseling of patients and their families in regards to the genetic diagnosis.

Genetic conditions evaluated in the clinic:
- Biotin defects, congenital lactic acidosis, fatty acid metabolism, galactosemia, Gaucher disease, glycogen storage disease, glycolytic enzymopathies, homocystinuria, maple syrup urine disease
- Hyperphenylalaninemia: Biopterin Deficiency; Hyperphe; Phenylketonuria (PKU); Maternal PKU Syndrome
- Hyperornithinemia
- Non-Ketotic Hyperglycinemia
- Urea Cycle Disorders: Argininemia; Argininosuccinic Aciduria; Citrullinemia; Ornithine Transcarbamylase Deficiency (OTC)
- Organic Aciduria: Branched Chain Organic Aciduria; Methylmalonic Acidemia; Malonyl-CoA Dehydrogenase Deficiency; Propionic Aciduria; Straight-Chain Organic Aciduria (MCAD, SCAD, Glutaric Aciduria); Unknown Disorder

Neurofibromatosis
The Neurofibromatosis Clinic, a joint effort of the Department of Molecular and Human Genetics at Baylor College of Medicine and Texas Children’s Hospital, is a multidisciplinary clinic in which patients are seen by physicians in the subspecialties of genetics, ophthalmology, dermatology, neurology, and neurosurgery. Patients are seen by the necessary subspecialist regarding any change in status and then referred on to a particular subspecialist for additional treatment as required.

Patients with a diagnosis of neurofibromatosis I, neurofibromatosis II, Von Hippel Landau, and tuberous sclerosis are all followed in the Neurofibromatosis Clinic. Case management and ongoing counseling are provided as well as new information concerning advances in treatment and testing for these disorders.

Skeletal Dysplasia
The Skeletal Dysplasia Clinic is a multispecialty clinic established to provide diagnosis, treatment, and follow up care for patients from birth to adult who have abnormalities of skeletal growth and strength. Patients are seen for routine, chronic and acute care. The clinic is staffed by board-certified geneticists, orthopedists,
and endocrinologists. Patients are evaluated by each specialty during their visit. Diagnostic imaging services are afforded the patients as required.

PRENATAL GENETICS
Ignatia Van den Veyver, M.D., Ph.D.
Director
Melissa Strassberg, CGC
Manager

General
The BCM Prenatal Genetics Clinic is a joint effort between the Department of Molecular and Human Genetics and the Department of Obstetrics and Gynecology. These two Departments integrate the delivery of high quality, state-of-the-art prenatal diagnostic services through their laboratories and clinics. Together, they offer a full range of coordinated genetic services for the diagnosis, treatment, counseling, and prevention of birth defects. Our professional staff members are world-renown in genetic and obstetric services now located at Texas Children’s Hospital Women’s Pavillion.

Prenatal Diagnostic and Genetic Services Include:
• Genetic counseling and reproductive genetics (M.D.) consultation—advanced maternal age; abnormal maternal serum screening; abnormal prenatal testing; ultrasound anomalies; exposure to medications, infections, alcohol, drugs, chemicals, or radiation during pregnancy; previous child with a birth defect or genetic condition; patient with genetic condition or carrier of a genetic condition; family history concerns; history of multiple miscarriages
• High resolution ultrasound study of fetus
• Prenatal diagnostic tests: Amniocentesis; Chorionic Villus Sampling (CVS); fetal blood sampling
• Multi-fetal reduction
• Laboratory studies available: DNA testing; biochemical testing; chromosomal analysis
• Maternal serum screening: (1) First Trimester Screening (FIRST) - calculates the risk for Down syndrome and trisomy 18 between 11 weeks 1 day and 13 weeks 6 days gestation based on biochemical analysis, ultra-sound of the uncial translucency, and maternal age; (2) Quad Screen - calculates the risk for Down syndrome, trisomy 18, and open neural tube defect in the second trimester by measuring MSAFP, chg., uE3, and Papp-A.
• Preimplantation genetic diagnosis

Clinical Genetics Patient Volume (Prenatal)
Education

A broad range of training opportunities are available, including basic laboratory research and clinical training. There are over 150 trainees in various settings of the training faculty, most of whom are pursuing research training.

GRADUATE PROGRAM

Gad Shaulsky, Ph.D.
Director

Judi Coleman
Program Coordinator

The Graduate Program in Molecular and Human Genetics provides outstanding educational opportunities for Ph.D. candidates who wish to pursue a career in research, education, and service in this field. Students in the program obtain rigorous training in modern biology with a special emphasis on genetics. They also participate in cutting edge research on a variety of topics and publish their work in some of the best peer-reviewed journals in the world. The unique environment of a large medical center provides students with an opportunity to obtain education and practical experience in both basic and applied research.

In order to encourage our students to fulfill their potential and to excel in their work, we provide one of the most competitive stipends in the country in a city with relatively low cost of living. We also cover the cost of tuition and medical insurance. Students who obtain
funding through individual competitive fellowships receive an additional $3,000 bonus from the Dean of the Graduate School.

The requirements for graduation include the successful completion of 30 credit hours of required courses and electives, the successful completion of the Qualifying Examination, the conduct of an original research project, and the submission and defense of a doctoral dissertation. For the didactic phase of graduate training, students participate in a set of core courses developed as a joint graduate school effort during the first three terms. Through these courses the students obtain a broad, coherent background in basic aspects of genetics, molecular biology, bioinformatics, biochemistry, and cell biology. This material is supplemented with journal clubs and more advanced courses in molecular and classical genetics and modern applications, including genomics and gene therapy during the later terms.

Students may take relevant elective courses offered by other programs at Baylor College of Medicine, Rice University, The University of Texas Health Science Center-Houston, or the University of Houston at any time during their graduate school tenure.

We concentrate course work in the first year to enable students to progress relatively quickly to full-time laboratory research efforts. All students participate in a minimum of three laboratory rotations in their first year. Through these rotations, students obtain valuable hands-on experience in laboratory techniques and become acquainted with a variety of research topics before selecting a major thesis advisor. The research interests of the Department span a very broad range. We are studying the basic principles of DNA replication and repair, DNA recombination, cell cycle control, aging, differentiation, and development in a variety of model organisms from *E. coli* through yeast and *Dictyostelium* to flies and mice. Studies in model organisms are tightly integrated with studies on the genetic basis of the human condition. We have an extensive program that addresses a variety of genetic diseases. Students who participate in the program can obtain experience in both the basic and the applied aspects of the research.
In the first term of the second year of study, the students write a detailed research proposal on a topic of their choice. They defend the proposal to a qualifying examination committee composed of faculty from the Department. Upon successful completion of the examination and course work, the student is admitted to candidacy to pursue a thesis research project under the direction of the major advisor and a thesis advisory committee. Following admission to candidacy, students receive a travel grant from the Department to initiate their participation in national meetings. Students are strongly encouraged to publish their work in international, peer-reviewed journals and to participate in national and international meetings.

Publications by our students are often in the top ranked journals in the world, and many of our students have been the recipients of prestigious awards for their work. The final step to completion of the Ph.D. is the preparation of a thesis and presentation of the thesis research work at a formal seminar, followed by a dissertation defense to the thesis committee.

Throughout the tenure of the graduate students at BCM, attendance at Departmental seminars and journal clubs is strongly encouraged. Several excellent seminar programs exist in the Department of Molecular and Human Genetics, as well as in the other departments. The Department also sponsors an annual two-day research retreat where faculty, students, and postdoctoral trainees present and discuss their research in an informal interactive atmosphere.

### 2012 GRADUATE PROGRAM (GRADuates)

<table>
<thead>
<tr>
<th>Student Name</th>
<th>Mentor</th>
<th>Current Position</th>
</tr>
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<tbody>
<tr>
<td>Cowin, Randi-Michelle</td>
<td>Dr. Paylor</td>
<td>Scientist / Project Manager at SeqWright</td>
</tr>
<tr>
<td>Heney Lacaria, Melanie</td>
<td>Dr. Lupski</td>
<td>Postdoc at Ottawa Hospital Res. Inst.</td>
</tr>
<tr>
<td>Kuo, Tsung-Han</td>
<td>Dr. Pletcher</td>
<td>Postdoc Fellow, Stower Lab, Scripps</td>
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<tr>
<td>Liu, Pengfei</td>
<td>Dr. Lupski</td>
<td>Postdoc at BCM</td>
</tr>
<tr>
<td>Price, Brandee</td>
<td>Dr. Wilson</td>
<td>Postdoc at MDACC</td>
</tr>
<tr>
<td>Veeraragavan, Surabi</td>
<td>Dr. Paylor</td>
<td>Postdoc at BCM</td>
</tr>
<tr>
<td>Wat, Margaret</td>
<td>Dr. D. Scott</td>
<td>Returned to BCM Medical School</td>
</tr>
</tbody>
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With the rapid development of genetics and genomics, more diseases are now recognized to have a genetic basis, including the traditional Mendelian diseases, congenital malformations, autism, cancers, degenerative disorders, and metabolic dysfunction. The demand for genetic screening and diagnosis has extended from postnatal period to prenatal period, posing a challenge for prenatal diagnosis. To strengthen the training of clinical geneticists, The Chinese University of Hong Kong signed a memorandum of understanding on September 14, 2012, with Baylor College of Medicine for collaboration to provide clinical genetic training. BCM faculty members such as Dr. Carlos Bacino, Dr. Lorraine Potocki, Dr. Fernando Scaglia, and Dr. V. Reid Sutton will regularly visit Hong Kong to conduct seminars and clinical training.
MEDICAL GENETICS RESIDENCY PROGRAM

V. Reid Sutton, M.D.
Director, ABMG Residency & Fellowship Programs

Maria Martinez
Program Coordinator

The American Board of Medical Genetics (ABMG) Medical Genetics Residency Program is accredited by the Residency Review Committee (RRC) of the Accreditation Council for Graduate Medical Education (ACGME). We are currently approved for a total of 12 residents. Available training pathways include: a four-year residency in medical genetics (individuals enter this program directly from medical school); a two-year residency in medical genetics (individuals enter this program after at least two years of other residency training); and a five-year combined pediatrics/medical genetics residency program. In all of these pathways, genetics clinical time is divided between rotations on the inpatient consultation service, outpatient general adult and pediatric clinics, subspecialty clinics, and diagnostic laboratories as well as attending conferences and didactic teaching sessions. The clinical experience is broad and intensive because of the availability of large clinical services and clinical faculty; the comprehensive diagnostic laboratories in cytogenetics, biochemical genetics, and molecular genetics; the active prenatal diagnosis program; and a number of medically relevant research projects. Graduate coursework in genetics is required. Most graduates of the program stay additional years in a mentored faculty position developing independent research programs. Trainees are strongly encouraged to seek individual fellowships and NIH career development awards to broaden their experience.

Medical Biochemical Genetics

Medical Biochemical Genetics is a newly-approved training pathway that is accredited by the ACGME RRC for Medical Genetics. This one-year training program is meant to provide additional training in the diagnosis and management of inborn errors of metabolism. Board-certification is available through the ABMG.

ABMG Diagnostic Laboratory Fellowships

We currently have ABMG-approved training programs in Clinical Biochemical Genetics, Clinical Cytogenetics and Clinical Molecular Genetics. We are among the largest academic genetics laboratories in the United States. The Baylor College of Medicine Medical Genetics Laboratories provides state-of-the-art diagnostic testing to hospitals and referring physicians within the Texas Medical Center, from all over the United States, and around the world. The Laboratories are divided into Molecular (General and Mitochondrial), Biochemical and Cytogenetics (including array Comparative Genomic Hybridization) work areas, and are an integral part of the Department of Molecular and Human Genetics.
Each lab is a national referral laboratory with a dedicated laboratory director and medical director who oversee the activities of the laboratory. Collectively, the laboratories process approximately 70,000 samples yearly. The laboratories use traditional techniques as well as newer techniques, including array Comparative Genomic Hybridization, Tandem Mass Spectrometry, Pyro-sequencing automated capillary DNA sequence analysis, and Maldi-TOF. The laboratories are housed in a modern facility in the Texas Medical Center.

Research Postdoctoral Training
The Faculty of the Department of Molecular and Human Genetics has broad expertise and have mentored hundreds of postdoctoral trainees. Faculty research projects range from seeking answers to basic science questions to those that are immediately clinically applicable. Examples of research areas include genome stability, epigenetics, chromosome dynamics, neural development and function, cardiovascular development, mitochondrial function, high-throughput genome sequencing, bioinformatics, developmental biology, mutagenesis, non-conding RNAs, epidemiology, protein structure and function, genomic rearrangements, trinucleotide repeat disorders, gene therapy, behavior, aging and gene function. In addition to human subjects research, model systems employed by our faculty include: *E. coli*, *Dictyostelium*, yeast, *Drosophila*, zebrafish and mouse. Specific research interests are outlined in profile of each faculty member. Applications for and inquiries regarding research postdoctoral training should be addressed to the specific faculty member.

During the academic year the Department hosts numerous speakers in order to advance knowledge and also to provide exposure to our trainees and others in the Department to leaders in the field. This effort also can lead to collaborations in education, research, and healthcare.
Former Molecular and Human Genetics Trainees’ Locations

**USA**

- California
- Colorado
- Connecticut
- Florida
- Georgia
- Hawaii
- Indiana
- Louisiana
- Maryland
- Massachusetts
- Michigan
- Minnesota
- Missouri
- New Jersey
- New York
- North Carolina
- Ohio
- Pennsylvania
- Tennessee
- Texas
- Utah
- Washington
- Wisconsin

**Other Countries**

- Australia
- Belgium
- Brazil
- Canada
- China
- Denmark
- England
- France
- Germany
- India
- Israel
- Italy
- Japan
- Portugal
- Saudi Arabia
- Scotland
- Singapore
- South Korea
- Spain
- Taiwan
The first McNair Symposium, featuring the work of McNair Medical Institute Scholars in the Texas Medical Center, was held at Baylor College of Medicine on Wednesday, April 25, 2012.

The McNair Scholarship Program—a recruitment initiative launched with a $100 million gift from the Robert and Janice McNair Foundation—is drafting top talent and encouraging teamwork, and approach similar to that of professional sports teams such as the Houston Texans founded by BCM Board of Trustee Bob McNair.

Dr. Benjamin Arenkiel, assistant professor of molecular and human genetics and neuroscience at BCM and an investigator at the Jan Duncan Neurological Research Institute at Texas Children’s Hospital, was one of the first McNair Scholar at BCM. One of the requirements for a McNair Scholarship is that the scientist must be willing to cooperate and share information with other scientists working on similar projects.
Dr. William Pu: (Harvard Stem Cell Institute) “Transcriptional Regulation of Heart Development and Disease”

Dr. David Raible: (University of Washington) “Mechanosensory hair cell death and regeneration”

Dr. Patrick Sung: (Yale University) “Mechanism of Eukaryotic Homologous Recombination”

Dr. Jennifer Trowbridge: (The Jackson Laboratory) “Roles of DNA methyltransferase-1 in hematopoietic and leukemia stem cell function”

Dr. Yingzi Yang: (NIH - National Human Genome Research Institute) “Wnt signaling in development and disease”

Dr. Fuli Yu: (Baylor College of Medicine) “Genetic variation analysis in large-scale NGS studies”

Dr. Ken Zaret: (University of Pennsylvania School of Medicine) “Pioneer factors and the initiation of cellular programming”

Dr. Xiang Zhang: (Baylor College of Medicine) “Metastasis seed pre-selection by primary tumor stroma – a possible solution to the metastasis progression”

Dr. Huda Zoghbi: (Baylor College of Medicine) “Understanding the Pathogenesis of an Inherited Ataxia: Relevance to Other Neurodegenerative Disorders”

The Margaret M. Alkek building for Biomedical Research
The fourteenth annual Frank Greenberg Memorial Lectureship was held in June, featuring Helen Firth, DM, FRCP, DCH Consultant Clinical Geneticist Addenbrooke's Hospital in Cambridge, United Kingdom. The title of her presentation was “Deciphering Genome Variation.”

Helen V. Firth, D.M., FRCP, is a Consultant Clinical Geneticist at Cambridge University Hospitals, Cambridge, UK. Her research interests have focused on the application of new technologies in Clinical Genetics. Dr. Firth studied Medicine at Oxford University graduating in 1981. After junior hospital jobs in pediatrics she began training in clinical genetics in Oxford in 1988. As a registrar she identified a cluster of babies born with severe limb defects following early chorion villus sampling (CVS). She subsequently undertook research on the teratogenic potential of early CVS, obtaining her Doctor of Medicine (DM) degree from Oxford University in 1998.

In 2004, Dr. Firth initiated the Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl Resources DECIPHER project and has worked with Dr. Nigel Carter and colleagues at the Wellcome Trust Sanger Institute to develop a web-based interface to display genomic copy number variants and their associated phenotypes in the Ensembl and USCS genome browsers http://decipher.sanger.ac.uk. DECIPHER enables clinical geneticists around the world to share information about very rare genomic variants to facilitate diagnosis and help to elucidate the role of genes whose function is not yet known.

Since 2010 Dr. Firth has been Clinical Lead for the Deciphering Developmental Disorders study (DDD study) www.ddduk.org. DDD is a partnership project between all 23 NHS Regional Genetics services in the UK and the Wellcome Trust Sanger Institute to undertake detailed genomic analysis of 12,000 children with severe developmental disorders. Dr. Firth is an Honorary Faculty Member of the Wellcome Trust Sanger Institute, Hinxton, UK and a Bye-Fellow of Newnham College, Cambridge. She has published more than 50 peer-reviewed manuscripts and co-authored two books: ‘Oxford Desk Reference: Clinical Genetics’ (OUP 2005) and ‘Oxford Handbook of Genetics’ (OUP 2009).
Community Outreach

**Evenings with Genetics**

The Evenings with Genetics seminar series has been developed with the goal of providing a community forum to effectively communicate current information on healthcare, education, and research regarding a variety of genetic-based diseases. The specific objectives of the series are:

- To provide current information for families of children affected with a genetic condition.
- Increase community knowledge of genetic care and research issues.
- Aid in family to family support for families impacted by a genetic disorder.
- Foster increased understanding and collaboration amongst geneticists, related medical specialists, and care providers.

Topics for the seminars are selected based on the most common genetic diseases, requests by families, or a new understanding of the pediatric or adult condition. Medical genetics specialists are paired with a physician from a Baylor College of Medicine clinical department for each seminar. This team works together to present best clinical practice and cutting edge research information. A crucial element of the seminar is the presentation by a parent or family member impacted by the condition who shares their perspective. The seminars are held one evening each month at The Health Museum allowing attendance by families, school nurses, teachers, speech pathologists, medical translators, medical and nursing students and other academic faculty. The use of the museum facilities rather than a medical center auditorium offers a non-stressful setting for open communication and learning. Local organizations such as Early Childhood Intervention, It's My Heart, Down Syndrome
Association, Facing Our Risk of Cancer Empowered (F.O.R.C.E.) are invited to have a booth on the relevant evening.

The family speakers have been immensely powerful in conveying the process of understanding and accepting their child's unique health needs as well as sharing information about resources and hope. This program allows genetic information to be shared in a non-threatening, accessible fashion as an adjunct to clinical practice.

**Seminars**

**TOPIC:** "The many faces of 5p- Syndrome"
**SPEAKERS:** Dr. Lorraine Potocki, Professor, Department of Molecular and Human Genetics, Baylor College of Medicine; Kasey Vernon, M.A., CCC-SP, Sr. Speech-Language Pathologist, Texas Children's Hospital - West Campus

**TOPIC:** "Updates on care and research for Wolff-Parkinson-White: a heart rhythm disorder"
**SPEAKERS:** Dr. Seema Lalani, Department of Molecular and Human Genetics, Baylor College of Medicine; Dr. Santiago Valdes, Pediatrics, Division of Cardiology, Baylor College of Medicine

**TOPIC:** "Caring Communities: perspectives in caring for people with intellectual disabilities"
**SPEAKERS:** Renée Wallace, Founder of Vita-Living, Inc; Dr. Luis M. Franco, Department of Molecular and Human Genetics, Baylor College of Medicine

**TOPIC:** "Children with Autism Spectrum Disorder: new information on how to help your child"
**SPEAKERS:** Dr. Carlos Bacino, Chief, Genetics Service at Texas Children's Hospital and Dr. M. Paige Powell, Psychology Service, Lead Autism Specialist, Autism Center, Texas Children’s Hospital

This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

**TOPIC:** Seminars for Health Professionals: “Autism Spectrum Disorders: Overview and Updates”
**SPEAKERS:** Dr. M. Paige Powell, Psychology Service, Texas Children's Hospital; Lead Autism Specialist, Autism Center, Texas Children's Hospital

This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

**TOPIC:** Seminar for Health Professionals: “Children with Chromosome Disorders: Medical Issues and Therapeutic Interventions”
**SPEAKERS:** Dr. Robert Stratton, Pediatric Geneticist, Driscoll Children’s Hospital and Susan Fernbach, R.N., B.S.N., Director of Genetic Outreach, Baylor College of Medicine

This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

**TOPIC:** “Taking Care of the School Aged Child with a Genetic Condition”
**SPEAKERS:** Susan Fernbach, R.N., B.S.N., Director of Genetic Outreach, Baylor College of Medicine

**TOPIC:** “Children with Chromosomal Disorders: new tests guide management, treatment & counseling”
**SPEAKERS:** Dr. Kari Casas, Geneticist, Trinity Clinic Genetics; Dr. Luis Casas, Endocrinologist, Trinity Clinic Endocrinology

This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

**TOPIC:** “Children with Chromosomal Disorders: new tests guide management, treatment & counseling”
**SPEAKERS:** Dr. Kari Casas and Susan Fernbach, R.N., B.S.N.

This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

**TOPIC:** “Children with disabilities: topics on parenting, genetics and development”
**SPEAKERS:** Dr. Elizabeth Roeder, Associate Professor, Division of Genetics, UT Health Science Center San Antonio; Dr. Nhung Tran, Developmental-Behavioral Pediatrician, Assistant Professor, Department of Pediatrics, UT Health Science Center at San Antonio, Medical Director, Center of Hope for Child Development, CHRISTUS Santa Rosa Children’s Hospital

This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

**TOPIC:** “Children with Developmental Disabilities: Practical interventions for medical, educational & behavioral problems”
**SPEAKERS:** Dr. Nhung Tran, Developmental-Behavioral Pediatrician, Assistant Professor, Department of Pediatrics, UT Health Science Center at San Antonio, Medical Director, Center of Hope for Child Development, CHRISTUS Santa Rosa Children's Hospital

This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.
TOPIC: “Children with Chromosomal Disorders: new tests guide management, treatment & counseling”
SPEAKERS: Dr. Elizabeth Roeder, Associate Professor, Division of Genetics, UT Health Science Center San Antonio; Susan Fernbach, R.N., B.S.N., Director of Genetic Outreach, Baylor College of Medicine
This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

TOPIC: “Children with Chromosomal Disorders: medical issues and therapeutic interventions”
SPEAKERS: Dr. Carlos Bacino, Director of Genetic Services, Baylor College of Medicine; Susan Fernbach, R.N., B.S.N., Director of Genetic Outreach, Baylor College of Medicine
This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

TOPIC: “Children with Congenital Heart Defects”
SPEAKERS: Dr. John W. Belmont, Professor, Department of Molecular and Human Genetics, Baylor College of Medicine; Dr. Shaine A. Morris, Assistant Professor, Department of Pediatrics, Division of Pediatric Cardiology, Baylor College of Medicine

TOPIC: “Care for Children with Congenital Adrenal Hyperplasia: A conversation with pioneers”
SPEAKERS: Dr. Maria New, Mount Sinai School of Medicine, New York; Dr. Lawrence McCullough, Baylor College of Medicine

TOPIC: “Children with special needs and autism: new information on how to help your child”
SPEAKERS: Dr. Carlos Bacino, Director of Genetics Clinic, Texas Children’s Hospital and Dr. Robin Kochel, Department of Pediatrics, Baylor College of Medicine

TOPIC: “Updates on care & development of children with Down Syndrome”
SPEAKERS: Dr. Robert Stratton, Pediatric Genetics, Driscoll Children’s Hospital and Dr. Sherry Vinson, Developmental Pediatrician, Texas Children’s Hospital

TOPIC: “Medical Management of Children with Special Needs”
SPEAKERS: Dr. Carlos Bacino, Director of Genetic Services, Baylor College of Medicine
This seminar was brought to the community in partnership with the Children’s Disabilities Information Coalition.

TOPIC: “Dental care for children with developmental disabilities”
SPEAKERS: Dr. Elsa Echeverri and Lesley Chong, M.A., C.C.L.S.

TOPIC: “Marfan Syndrome: Care issues for adults”
SPEAKERS: Dr. Sandesh Sreenath Nagamani and Dr. Richard Lewis, Baylor College of Medicine

TOPIC: “Autism Spectrum Disorders: Update on genetics & medical care”
SPEAKERS: Dr. Golder Wilson, Genetics Clinic at University Medical Center; Dr. Karen Rogers, Developmental Pediatrician, PALS Developmental Center
This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

TOPIC: “Children with Chromosomal Disorders: New tests guide management, treatment & counseling”
SPEAKERS: Dr. Vijay Tonk, Professor of Pediatrics, Texas Tech University Health Sciences Center; Susan Fernbach, R.N., B.S.N., Instructor, Baylor College of Medicine
This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

TOPIC: “Children with Developmental Disabilities: Practical interventions for medical, educational & behavioral problems”
SPEAKER: Dr. Karen Rogers, Developmental Pediatrician, PALS Developmental Center
This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

TOPIC: “Autism Spectrum Disorders: Update on genetics & medical care”
SPEAKER: Dr. Golder Wilson, Genetics Clinic at University Medical Center
This seminar was made possible by Title V funding awarded by the Texas Department of State Health Services.

TOPIC: “Ectodermal Dysplasias: Update on treatment of eye and skin conditions”
SPEAKERS: Dr. Richard Lewis and Dr. Alanna Bree
Faculty and Staff Outreach

Since 2005, Dr. Lorraine Potocki has been the director of “Parent’s Night Out” held yearly at the Texas Children’s Hospital Clinical Care Center. It is hosted by Baylor Pediatric Student Association and more recently with the Baylor Genetics Interest Group. Parents Night Out is a community service activity where Baylor College of Medicine medical students provide an evening of care and entertainment for children and young adults with special needs. It helps families caring for children with special needs to have a night off and is also a great opportunity for children to meet new friends and for medical students to gain experience interacting with special needs children. This year, it was held on November 2, 2012, and was the largest one ever.

Since 2009, Dr. Fernando Scaglia has actively participated in the Geneticist Educator Network Alliance (GENA) project in collaboration with a Bellaire High School biology teacher. GENA is a science project designed to build a framework of long-term collaboration between educators and geneticists and a sustainable infrastructure to facilitate the meaningful outreach efforts by genetics faculty members in secondary education. This collaborative program involves teaching genetics to students, genetic counselors, metabolic nurses, dietitians, and parents of children with a genetic disorder.

It’s sponsored by the The American Society of Human Genetics, the Genetics Society of America, the National Sciences Resources Center, and the National Association of Biology Teachers as part of The Geneticist-Educator Network of Alliances Project.

Susan Fernbach, R.N., travels throughout Texas to increase community knowledge of genetics services, research issues, and foster increased collaboration among geneticists, related medical specialists, and other healthcare providers in providing care to clients with genetic conditions. She develops and implements around sixteen educational seminars that provide relevant and current information on health care, education, and research regarding genetic-based disorders. Some of these locations are Brownsville, El Paso, Houston, Lubbock, McAllen, Midland, San Antonio, and Tyler.

Since 1997, Kerri Lamance, R.N., has organized the CAMP PHEver Children’s PKU Camp that is held each summer in Burton, Texas. The camp provides children and adolescents with phenylketonuria (PKU) a typical summer camp experience where they can meet new friends, reunite with old camp buddies, and receive education about PKU. Activities include canoeing, fishing, swimming, hiking on nature trails, sports, horseback riding, campfires, dances, and much more.
PRIMARry APPOIntmEnts

Benjamin Arenkiel, Ph.D.
Assistant Professor and McNair Scholar
Molecular genetic studies to investigate mechanisms of neural circuit formation, function, and maintenance.

Carlos A. Bacino, M.D.
Professor
Clinical studies in patients with imprinting disorders (Angelman Syndrome), skeletal dysplasias, and genomic disorders.

David Bates, Ph.D.
Assistant Professor
Chromosome dynamics, molecular mechanisms of DNA replication, and cell cycle control in E. coli.

Arthur L. Beaudet, M.D.
Professor
Henry and Emma Meyer Chair in Molecular Genetics
Role of epigenetics and genomic imprinting in evolution and disease including Prader-Willi and Angelman syndromes and autism; hepatocyte gene therapy.

Hugo J. Bellen, D.V.M., Ph.D.
Professor
March of Dimes Chair in Developmental Biology Investigator, Howard Hughes Medical Institute
Genetic and molecular analysis of neurotransmitter release, neural development, and neuronal degeneration in Drosophila.

John W. Belmont, M.D., Ph.D.
Professor
Structural congenital heart defects including abnormalities in laterality and hypoplastic left heart syndrome; functional studies of Zic3; genetics of human immune responses; medical population genetics.

Penelope E. Bonnen, Ph.D.
Assistant Professor
Genomics, population genetics, and genetics of metabolic disease.

Juan Botas, Ph.D.
Professor
Comparative analysis of pathogenic mechanisms in neurodegenerative disorders. Screens for common ‘druggable’ targets.

Chester Brown, M.D., Ph.D.
Associate Professor
Genetic and genomic contributions to obesity and nutrient metabolism.

C. Thomas Caskey, M.D., F.A.C.P.
Professor
Genetic basis of schizophrenia.

Rui Chen, Ph.D.
Associate Professor
Functional genomics of visual system development and diseases; High throughput technology; Bioinformatics.

Sau Wai Cheung, Ph.D., M.B.A.
Professor
Clinical Cytogenetics; Preimplantation Genetic Screening; Chromosomal Microarray Analysis (Clinical utility of aCGH).

William J. Craigen, M.D., Ph.D.
Professor
Regulation of cellular energy metabolism; mouse models of metabolic diseases.

Herman A. Dierick, M.D.
Assistant Professor
Genetic and neurobiological mechanism of Drosophila aggression.

Christine M. Eng, M.D.
Professor
Natural history, molecular genetics, and treatment of lysosomal storage disorders; genetic testing for inherited diseases.

Richard A. Gibbs, Ph.D.
Professor, HGSC Director
Wofford Cain Chair in Molecular and Human Genetics
Genome science, human molecular evolution, and molecular basis of inherited disease.
<table>
<thead>
<tr>
<th>Name</th>
<th>Title</th>
<th>Research Area</th>
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<tbody>
<tr>
<td>Brett Graham, M.D., Ph.D.</td>
<td>Assistant Professor</td>
<td>Genetics of inborn errors of metabolism; genetic models of mitochondrial disease in <em>Drosophila</em> and mice.</td>
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<tr>
<td>Andy Groves, Ph.D.</td>
<td>Associate Professor</td>
<td>The development, evolution and regeneration of the inner ear.</td>
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<tr>
<td>Philip J. Hastings, Ph.D.</td>
<td>Professor</td>
<td>Mechanism of amplification and genome instability in <em>Escherichia coli</em>.</td>
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<tr>
<td>Xiangwei He, Ph.D.</td>
<td>Assistant Professor</td>
<td>Mechanisms of chromosome segregation: centromere, kinetochore, fission yeast.</td>
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<tr>
<td>Jason Heaney, Ph.D.</td>
<td>Assistant Professor</td>
<td>Testicular and colon cancer genetics, stem/progenitor cell maintenance, and mouse models of human diseases.</td>
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<tr>
<td>Christophe Herman, Ph.D.</td>
<td>Associate Professor</td>
<td>Epigenetic inheritance and phenotypic variability.</td>
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<tr>
<td>Gregory Ira, Ph.D.</td>
<td>Associate Professor</td>
<td>DNA repair, recombination and genome instability. Molecular mechanisms of mitotic DNA recombination.</td>
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<tr>
<td>Hamed Jafar-Nejad, Ph.D.</td>
<td>Assistant Professor</td>
<td>Contribution of protein O-glycosylation to Notch signaling, animal development and human disease.</td>
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<tr>
<td>Milan Jamrich, Ph.D.</td>
<td>Professor</td>
<td>Pattern formation in vertebrate embryos; ocular development; gene therapy.</td>
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<tr>
<td>Monica Justice, Ph.D.</td>
<td>Professor</td>
<td>Molecular genetic analysis of hematopoiesis; cancer genetics; mouse models of human disease.</td>
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<tr>
<td>Seema Lalani, M.D.</td>
<td>Assistant Professor</td>
<td>Molecular basis of CHARGE syndrome; Use of array-based comparative genomic hybridization to study the genetic basis of congenital heart defects.</td>
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<tr>
<td>Suzanne M. Leal, Ph.D.</td>
<td>Professor</td>
<td>Statistical genetics; genetic epidemiology; gene mapping and identification; nonsyndromic hearing loss.</td>
</tr>
<tr>
<td>Brendan Lee, M.D., Ph.D.</td>
<td>Professor</td>
<td>Investigator, Howard Hughes Medical Institute Developmental genetics of the skeleton and human skeletal dysplasias; development of helper-dependent adenoviral gene therapy for immune modulation, inborn errors of metabolism, and skeletal diseases; Clinical research into the diagnosis, intervention, and treatment of metabolic and skeletal diseases.</td>
</tr>
</tbody>
</table>
Marilyn M. Li, M.D.
Professor
Clinical application of microarray technologies in cancer research and diagnosis; leukemia- and lymphoma-associated genetic aberrations in healthy individuals; microdeletion syndromes.

Olivier Lichtarge, M.D., Ph.D.
Professor
The Cullen Foundation Endowed Professorship In Molecular Genetics
Structural bioinformatics studies of proteins to study and engineer pathway perturbations.

James R. Lupski, M.D., Ph.D., D.Sc. (hon.)
Professor, Vice Chair
The Cullen Endowed Chair in Molecular and Human Genetics
Molecular genetics of Charcot-Marie-Tooth disease and related inherited neuropathies; molecular mechanisms for human DNA rearrangements; genomic disorders; copy number variation (CNV) and disease.

Graeme Mardon, Ph.D.
Professor
Retinal cell fate determination, development, and function in Drosophila and vertebrates.

Michael L. Metzker, Ph.D.
Associate Professor
Genetic screening of juvenile diabetics; technology development of miniature DNA sequencing devices; characterization of HIV-1 drug resistance using novel reagents; fluorescent technology development for existing and novel DNA sequencing strategies.

Aleksandar Milosavljevic, Ph.D.
Associate Professor
Genomics, cancer genomics, epigenomics, and bioinformatics.

Daisuke Nakada, Ph.D.
Assistant Professor and CPRIT Scholar
Elucidate the molecular and cellular mechanisms that regulate stem cell function.

David L. Nelson, Ph.D.
Professor
The Cullen Foundation Professorship in Molecular Genetics
Human genome and disease gene analysis; fragile X syndrome; incontinentia pigmenti; complex genetics.

Philip Ng, Ph.D.
Associate Professor
Liver and lung gene therapy.

Ankita Patel, Ph.D.
Associate Professor
Clinical cytogenetics; development of new diagnostic technologies for cytogenetics.

Richard E. Paylor, Ph.D.
Professor
Behavioral analyses of mutant mouse models of developmental and neurodegenerative disorders.

Lorraine Potocki, M.D.
Professor
Clinical characterization of selected genomic disorders; medical education in genetics.
Frank Probst, M.D., Ph.D.
Assistant Professor
Generation and characterization of mouse models of human diseases; mouse models of hearing impairment; detection of small genomic inversions; familial multiple lipomatosis.

Jeffrey Rogers, Ph.D.
Associate Professor
Genetic analysis of nonhuman primate models of human disease, especially psychiatric diseases; comparative primate genomics.

Susan M. Rosenberg, Ph.D.
Professor
Ben F. Love Chair in Cancer Research
Molecular mechanisms of genome instability in evolution, antibiotic resistance, and cancer.

Marco Sardiello, Ph.D.
Assistant Professor
Lysoosomal enhancement as a therapeutic strategy for treating neuronal ceroid lipofuscinoses.

Fernando Scaglia, M.D.
Professor
Molecular bases of hepatocerebral mitochondrial DNA depletion syndromes; studies of prevalence of autism spectrum disorders in mitochondrial encephalomyopathies; stable isotope studies in MELAS syndrome to evaluate nitric oxide flux and production.

Daryl Scott, M.D., Ph.D.
Assistant Professor
Genes responsible for common birth defects and microdeletion/microduplication syndromes: congenital diaphragmatic hernia, esophageal atresia/tracheoesophageal fistula, and 1p36 deletion syndrome.

Kenneth L. Scott, Ph.D.
Assistant Professor
Cancer gene discovery, pathways governing tumor metastasis and animal models for cancer.

Gad Shaulsky, Ph.D.
Professor, Director of Graduate Program
Allorecognition, evolution of sociality and functional genomics in Dictyostelium.

Chad Shaw, Ph.D.
Assistant Professor
Devising new statistical methods for genome scale data using a systems biology approach.

Joshua M. Shulman, M.D., Ph.D.
Assistant Professor

Pawel Stankiewicz, M.D., Ph.D.
Associate Professor
Molecular mechanisms and clinical consequences of genomic rearrangements.

V. Reid Sutton, M.D.
Associate Professor
Aicardi syndrome, Focal Dermal Hypoplasia (Goltz syndrome), skeletal dysplasias (Osteogenesis Imperfecta), and inborn errors of metabolism.

Meng Wang, Ph.D.
Assistant Professor
Systemic studies of endocrine and metabolic signaling in promoting healthy aging.

Thomas (Trey) Westbrook, Ph.D.
Assistant Professor
RNAi-based strategies to cancer gene discovery; REST tumor suppressor pathway.

Lisa D. White, Ph.D.
Associate Professor
Genomic and RNA profiling using microarray and high throughput sequencing technologies to answer questions in cancer.

Lee-Jun C. Wong, Ph.D.
Professor
Mitochondrial genetics and function in human diseases, cancer, and aging.

Fuli Yu, Ph.D.
Assistant Professor
High throughput DNA technologies; Next-Generation Sequencing (NGS) data analysis; 1000 Genomes Project.

Hui Zheng, Ph.D.
Professor
Huffington Foundation Endowed Chair in Aging
Pathophysiological studies of Alzheimer’s disease pathways using mouse models.

Huda Y. Zoghbi, M.D.
Professor
Marvin Fishman Chair in Pediatric Neurology Research
Ralph D. Feigin, M.D. Endowed Chair Investigator, Howard Hughes Medical Institute
Molecular basis of neurodegenerative and neurodevelopmental disorders; nervous system development.
Research Interests

In my laboratory we use mouse genetics and genomics to identify genes and pathways involved in the neoplastic transformation of stem/progenitor cells. Our overall goal is to utilize knowledge gained from our mouse models to understand the causes of tumor initiation in humans and to provide new targets for the early diagnosis and treatment of cancer.

Ongoing research is focused on two questions:

What genes and developmental pathways contribute to testicular germ cell tumor (TGCT) initiation? Male germ cell development in the 129 family of inbred mice is an important in vivo experimental model system for studying fundamental questions about maintenance of pluripotency and induction of differentiation. Germ cells arise during embryogenesis as pluripotent primordial germ cells (PGCs) that differentiate into mature gametes and ultimately the cells and tissues of an adult organism. Defects during male germ cell development can lead to the formation of testicular germ cell tumors (TGCTs). In 129 mice, TGCTs arise during embryonic days (E)13.5-15.5 as foci of pluripotent embryonal carcinoma cells (EC cells), which differentiate to form teratomas. At E13.5, male germ cells normally enter mitotic arrest until after birth and female germ cells initiate the meiotic program, both of which are accompanied by down-regulation of pluripotency. We recently identified a defect in this developmental switch as the cause of TGCT initiation. In TGCT susceptible gonads, germ cells fail to enter mitotic arrest, retain pluripotency (Nanog and Prdm14 expression), and prematurely express genes associated with meiotic differentiation (cyclin D1 and Strah). Continued expression of both pluripotency and differentiation-associated genes through E15.5 is directly related with germ cell transformation into EC cells. Ongoing studies are testing how germ cell differentiation-associated and pluripotency-related genes influence 1) mouse germ cell developmental defects, 2) the mouse male germ cell transcriptome, 3) TGCT initiation in mice, and 4) human EC cell tumorigenic potential and cisplatin resistance. We are also using chromosome substitution strains to identify new genetic modifiers of TGCT risk.

How does IL-33-mediated signaling contribute to intestinal cancer susceptibility? We recently started testing whether genetic modifiers of TGCTs also influence adenoma risk in the ApcMin mouse model of colorectal cancer. We have found that one TGCT modifier, the Chr19MOLF/Ei chromosome substitution, increases intestinal polyp burden in APCMin mice. Using segregating crosses and congenic mice, we are identifying regions of chromosome 19 that harbor modifiers of adenoma risk. One candidate modifier, interleukin 33 (Il33), is a pro-inflammatory cytokine produced by intestinal epithelial cells (IECs) in response to infection and tissue damage. IL-33 signals through the IL1RL1 receptor in a variety of cell types to stimulate pro-inflammatory cytokine release in the intestine. The resulting inflammation stimulates a tissue repair response in IECs, which includes increased proliferation. Importantly, induction of IEC proliferation by inflammation is proposed to promote the neoplastic transformation of IECs in mice and humans genetically pre-disposed to intestinal cancer. We recently discovered that adenoma-associated IECs in APCMin mice express high levels of IL33. We also discovered that intestinal subepithelial myofibroblasts (ISEMFs), which secrete factors into the intestinal stem cell niche to activate IEC tissue repair responses, and mast cells, which produce pro-inflammatory cytokines that promote IEC proliferation, express IL1RL1 at the sites of adenoma growth in APCMin mice. Importantly, an antagonist antibody against IL1RL1 significantly decreased APCMin adenoma burden, reduced pro-inflammatory cytokine production at the site of adenoma formation, and inhibited adenoma-associated mast cell activation, suggesting that IL-33 signaling mediates intestinal cancer risk through inflammatory mediators. Ongoing studies are testing whether 1) IL-33 signaling through mast cells or ISEMFs influences adenoma risk, 2) IL-33 functions as a transcriptional regulator in IECs, and 3) IL-33 mediates inflammation and colorectal cancer risk associated with high fat diets.
Research Interests

Glycosylation is the most common post-translational modification of extracellular proteins and plays major roles in various aspects of cellular and organismal biology. However, the specific role of various carbohydrate residues in modifying the behavior of proteins and consequently regulating cellular processes are largely unknown. The long-term goal of our research is to understand the contribution of carbohydrates to the regulation of animal development, which is a key question in Developmental Glycobiology. Our current work focuses on the regulation of the Notch signaling pathway by “protein O-glucosylation.” A number of secreted and transmembrane proteins, including several coagulation factors and Notch proteins, harbor an O-linked glucose residue on epidermal growth factor-like (EGF) repeats with a C1-X-S-X-(P/A)-C2 motif. The O-glucose can be extended by the addition of one or two xylose residues. Several years ago, we identified the protein O-glucosyltransferase Rumi in Drosophila and showed that loss of Rumi results in temperature-dependent loss of Notch signaling in flies. Building on this initial discovery, the goal of our research is to address the following questions:

- How do O-glucose residues and their extended forms regulate Drosophila Notch signaling? Using a combination of genetic engineering, cell biology, and biochemistry, we aim to understand how O-linked glucose help maintain wild-type levels of Notch signaling despite variations in the temperature at which the flies are raised. The recent identification of the enzymes responsible for the addition of xylose to O-linked glucose has paved the way for functional analysis of the role of xylose residues in Notch signaling. Using a combination of fly genetics, cell culture and biochemical experiments, we are examining whether xylose residues play a regulatory role in Notch signaling.

- What is the role of O-linked glucosylation in the regulation of mammalian Notch signaling? Drosophila Rumi is the first protein O-glucosyltransferase (Poglut) enzyme identified in animals. Moreover, the extracellular domains of the mammalian Notch proteins have a large number of O-glucosylation motifs with an evolutionarily conserved distribution. We therefore asked whether Rumi regulates mammalian Notch signaling, and found that mouse Rumi (official name: Poglut1) is involved in the regulation of the Notch signaling and embryonic development. We are currently using mouse genetics and cell culture experiments to examine the molecular basis for the sensitivity of the mammalian Notch signaling to the level of Rumi.

- Does Rumi play a regulatory role in contexts other than the Notch pathway? Although Notch receptors harbor a large number of Rumi target motifs, a number of other transmembrane and secreted proteins also contain EGF repeats with a C1-X-S-X-(P/A)-C2 motif and might therefore be glucosylated by Rumi. Part of our efforts is devoted to the identification of other biologically-relevant targets of Rumi.

The Notch signaling pathway plays key roles in animal development and in stem cell biology. Moreover, mutations in the components of this pathway cause a variety of human diseases, including cancer, developmental disorders affecting heart, liver, skeleton and other organ systems, and adult-onset dementia. Therefore, providing insight into the role of carbohydrates in regulating the Notch signaling pathway could potentially contribute to our understanding of the pathogenesis of Notch-related diseases and could provide new therapeutic targets in these diseases.
Joshua M. Shulman, Ph.D.
Assistant Professor

Education
A.B., Harvard College, 1997
Ph.D., University of Cambridge, 2000
M.D., Harvard Medical School, 2005
Resident in Neurology, Brigham & Women’s Hospital, Massachusetts General Hospital, 2009
Movement Disorders Fellow, Brigham & Women’s Hospital, Massachusetts General Hospital, 2010

Research Interests
Recent advances have made the discovery of genetic susceptibility loci for complex human phenotypes a reality, including nervous system disorders. The critical next step will be to definitively identify the responsible genes and understand their functions in both health and disease. Our research integrates genetic investigation in human subjects and model organisms, with the goal of understanding brain function and aging, and improving the treatment of neurologic disease. We focus on Alzheimer’s disease and Parkinson’s disease, two incurable neurodegenerative disorders and experimental paradigms for the age-dependent failure of brain cognitive and motor control in humans.

Human Genetics: The clinical manifestation of neurodegenerative disease is the culmination of a multi-tiered pathogenic cascade that evolves over decades—understanding how genetic variants impact this causal chain is essential. Although 2 percent of the population over age 65 are clinically diagnosed with Parkinson’s disease, the defining pathology of disease (alpha-synuclein Lewy bodies) is discovered in 20 percent of brains from population-based autopsy studies. We are therefore investigating the impact of genomic variation on directly measured Lewy pathology, neuronal loss in the midbrain substantia nigra, and progressive motor impairment, leveraging human subject cohorts with detailed clinical and pathological data. We also participate in collaborative studies for the functional genetic dissection of Alzheimer’s disease, focusing on the responsible neuropathology, amyloid neuritic plaques andTau neurofibrillary tangles.

Drosophila Genetics: Despite the promise of current human genetic methods, such as genome-wide association studies, they often fail to identify disease susceptibility genes with certainty, instead highlighting broad genomic regions. We are taking advantage of the rapid and powerful genetics available in the fruit fly Drosophila melanogaster in order to accelerate the validation of responsible genes and an understanding of their functions in disease pathogenesis. Expression of human amyloid-beta, Tau, or alpha-synuclein proteins in the fly nervous system recapitulates many core features of Alzheimer’s disease and Parkinson’s disease pathogenesis. We are testing candidate human susceptibility genes for functional genetic interactions in these fly models of neurodegeneration. Implicated molecular pathways are probed in greater depth, using both Drosophila and human genetic approaches. Our strategy has recently identified cell adhesion converging on the cytoskeleton as likely important for Tau-mediated neurodegeneration and Alzheimer’s disease susceptibility, and we are now following up these insights to elucidate the detailed mechanisms.
Fuli Yu, Ph.D.
Assistant Professor

**Education**
Ph.D., Baylor College of Medicine, 2005
Postdoc, Harvard Medical School, 2008

**Research Interests**
My research program is centered around high throughput DNA technologies. One major area that I focus on is computational tool development for next-generation sequencing (NGS) data analysis. We have developed two software packages (Atlas2 and SNPTools) for variation analysis in both personal genomic data and population sequencing data. They have been applied in a number of large-scale projects, including the 1000 Genomes Project, the Cancer Genome Atlas Project, CHARGE-S, and Autisms. Atlas2 also powers the backend of the BCM Whole Genome Laboratory (WGL) pipeline.

My close affiliation with the BCM Human Genome Sequencing Center allows me to stay updated on technologies in both the bench side and the computational side. My group interacts with others in the Center to drive the development and dissemination of the technologies. For example, we are in the process of developing an AWS Cloud based environment to address the genomic BIG DATA challenge.

I am also interested in both population genetics and human genetics. They are very active fields because of the abundance of datasets, and technological revolution in the sequencing arena. I have a number of ongoing collaborative projects in these areas.
Faculty AWARDS & recognition

Hugo Bellen, D.V.M., Ph.D.
Gill Distinguished Award from the Indiana University’s Linda and Jack Gill Center for Biomolecular Science

John W. Belmont, M.D., Ph.D.
BCM You FIRST Program Research Faculty Winner FY13 QTR 1 from Baylor College of Medicine

Ayelet Erez, Ph.D.
William K. Bowes Award in Medical Genetics

Suzanne Leal, Ph.D.
Elected President of the International Genetics Epidemiology Society

Brendan Lee, Ph.D.
Men of Distinction Award

David Nelson, Ph.D.
Began serving as new editor of the American Journal of Human Genetics

Sharon E. Plon, M.D., Ph.D.
Barbara and Corbin J. Robertson Presidential Award for Excellence in Education from Baylor College of Medicine
The world-renowned Genetics Program at Baylor College of Medicine is an important component of what makes the Texas Medical Center a community of healing, learning, and discovery. This is truly an exciting time in the field of Genetics due to many discoveries that will advance the field in different directions. We are prepared for the daunting task of sifting through newly-discovered genes in search of those that lead to disease. These efforts will change the way we view diseases and receive personalized medical care.

The Department of Molecular and Human Genetics is enthusiastic about the future as we will be shaping diagnostics, risk assessments, prevention, and treatment strategies that will impact medicine. We look forward to sharing these experiences with you as they unfold.