A Measure of Excellence

A systematic approach to academic advancement
Welcome to BCM Quarterly.

Baylor College of Medicine is a health sciences university that creates and applies science and discoveries to further education, healthcare and community service locally and globally.

This is the mission statement for BCM. But, by itself it is just words. Translating it into action requires dedicated, passionate, intelligent people. We are fortunate to have many such individuals at BCM who creatively and collaboratively fulfill our mission.

Just in the twelve pages of this issue of BCMQ there are ample examples of such individuals. These include a student who trekked 2,100 miles by bicycle (page 1), our donors and development team who helped BCM earn a coveted award for outstanding philanthropy (page 3), the educators throughout BCM who strive for excellence in all their work (page 4), the physicians and staff of the Baylor International Pediatric AIDS Initiative (page 8), and all the BCM researchers whose contributions are adding to our understanding of health and disease (page 10).

Without these people and their colleagues our mission would be just a statement. With them, it is a promise.

I hope their stories inspire you, as they do me, and I look forward to sharing many more with you in the future.

Best regards.

Paul Klotman, MD
President and CEO, Baylor College of Medicine
John E. and Clara B. Whitmore Chair for the President

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Baylor College of Medicine is the academic center around which the Texas Medical Center, the world’s largest health science complex, evolved.

Within the four schools of BCM our faculty creates, implements and shares new knowledge, new systems and new technologies that improve the lives of our patients, our community, our nation and our world.

**Baylor College of Medicine** – Consistently ranked as one of the leading research-intensive medical schools in *U.S. News & World Report* and ranked fourth in the nation by StudentDoc.com, BCM is the least expensive private medical school in the U.S.

**BCM Graduate School of Biomedical Sciences** – BCM’s extensive research portfolio combined with faculty who are world leaders in their fields support 12 programs, which are ranked among the top 10 percent of graduate programs in biological sciences

**BCM School of Allied Health Sciences** – Drawing highly regarded applicants from throughout the region and the nation, the programs of BCM School of Allied Health Sciences consistently rank among the best in the country.

**BCM National School of Tropical Medicine** – This is the only school in the nation dedicated exclusively to patient care, research and education related to neglected tropical diseases, the most common infections of the world’s poorest people.

At BCM, leading research and technology translate to exceptional patient care. Our doctors and staff provide a patient-centered, multidisciplinary team approach to healthcare with a focus on quality and safety. More than 2,000 BCM doctors care for patients in over 40 locations across the Houston area, including:

**Baylor Clinic**  
**Lee and Joe Jamail Specialty Care Center**  
**Affiliated Hospitals**  
- Children’s Hospital of San Antonio  
- Harris Health System Ben Taub Hospital and health centers  
- Michael E. DeBakey Veterans Affairs Medical Center  
- St. Luke’s Medical Center  
- Texas Children’s Hospital  
- TIRR Memorial Hermann  
- The Menninger Clinic  
- Houston Methodist  
- The University of Texas MD Anderson Cancer Center
ORTHOTICS PROGRAM STUDENT RIDES IN TO BEGIN CLASSES

To begin classes at Baylor College of Medicine’s new Master of Science in Orthotics and Program, student Jared Rammell chose a different path.

The path was filled with adventure as he bicycled from Boise, Idaho, to Houston. That’s over 2100 miles. And the welcome on a Sunday afternoon was more than he expected. Dozens of his future colleagues greeted him at the front of Baylor to celebrate his achievement.

The new master’s program in the School of Allied Health Sciences at Baylor College of Medicine trains orthotists and prosthetists who design, build, fit and maintain prosthetic limbs and custom orthopedic appliances for individuals with limb loss and physical impairments.

Rammell, an avid cyclist who has competed in road races and endurance mountain biking events, decided to go into the field of orthotics and prosthetics at the encouragement of his wife, who is a nurse. He went back to school to complete his prerequisites and was selected for the Baylor program for his training.

His trip was also a fundraiser and he collected $3,760 for the Range of Motion Project, an organization that provides orthotic and prosthetic care to those in Guatemala who do not have access to these services.

COLLEGE OPENS EIGHTH TEEN HEALTH CLINIC

Baylor College of Medicine recently opened its eighth Teen Health Clinic, with Baylor President Dr. Paul Klotman and county officials on hand to praise the efforts of Director Dr. Peggy Smith to establish this new facility.

The new clinic is located at the Tejano Center for Community Concerns. It will offer free or low-cost comprehensive healthcare and education to students ages 13 and older enrolled at the Tejano Center’s Raul Yzaguirre School for Success, an open enrollment charter school.
It is one of the nation’s largest providers of indigent teen healthcare, offering comprehensive medical services, including reproductive care, health education, case management, counseling and support and social services, to its patients.

**JOHNSON NAMED DEAN OF THE GRADUATE SCHOOL OF BIOMEDICAL SCIENCES**

After a long career at the University of Southern California Keck School of Medicine, Dr. Deborah Johnson has accepted the position of dean of the Baylor College of Medicine Graduate School of Biomedical Sciences.

“Dr. Johnson has an excellent track record as an educator and academic leader,” said Dr. Paul Klotman, BCM President and CEO. “She will provide visionary direction for the Graduate School, building on its excellent programs to meet the needs of the academic science community of the future.”

She succeeds Dr. Hiram “Gil” Gilbert, who will transition to a leading role in establishing the College’s accreditation process, as well as developing enterprise and information technology solutions for academic issues.

Johnson, who holds the William R. Brinkley BRASS Chair, is excited to work with new colleagues in Houston, and described the College as having “an exceptionally strong research environment and commitment to graduate education and post-doctoral training.”

She has served on the faculty of USC since 1985. Active in academic affairs throughout her career, Johnson has most recently focused on developing mentoring programs. Other positions she has held at USC include professor of biochemistry and molecular biology, director for the Program in Biomedical and Biological Sciences, vice chair for graduate education in the Department of Biochemistry and Molecular Biology and professor of molecular pharmacology and toxicology. She has been a member of the USC Norris Comprehensive Cancer Center since 1985.

Her research program is defining the molecular events that lead cells to undergo oncogenic transformation.

She received a bachelor’s of science degree in biochemistry from Albright College, a doctorate in chemistry from Georgetown University and was a post-doctoral fellow in molecular biophysics and biochemistry at Yale University.

**NEW BLOG CONTINUES TO BUILD “MOMENTUM”**

Baylor College of Medicine’s new blog, “Momentum” is becoming a favorite read for a growing audience.

Whether news about the College missions, Throwback Thursdays which celebrates the past through classic photos, health tips, videos and the popular Infographics, Momentum continues to build, with more than 60,000 views.

Since Momentum’s launch in March, readership has increased steadily. Statistics show that the amount that readers share posts on social media continues to rise, and the College has received ample positive feedback. Momentum inspires conversation on the blog, dialogue on social media and in-person discussions.

Momentum bridges together the many facets of the Baylor community by providing a forum to share our opinions, memories and ideas for staying up-to-date with the present and continuing to create an innovative future together. Check it out at http://momentumblog bcm.edu/.
“Congratulation to Dr. Bert O’Malley, chair of molecular and cellular biology at BCM and recipient of the 2007 National Medal of Science, on his most recent honor, the 2013 Endocrine Regulation Prize from the foundation of the IPSEN company. O’Malley joins many of his faculty and trainee colleagues at BCM in receiving well deserved recognition for their contributions to science and medicine in recent months.”

Dr. Paul Klotman, president and CEO
Every good academic department chair wrestles with the best way to help his or faculty with academic promotions. Traditionally, this has meant a focus on publications in high impact journals and receipt of grant funding. But, what of the faculty member whose driving passion rests not primarily with research, but with teaching?

Dr. Michael Coburn, the Russell and Mary Hugh Scott Chair of Urology at Baylor College of Medicine, is grateful that over a decade ago, before he became chair, he, Drs. Joan Friedland, associate professor of medicine, and James Lomax, the Brown Foundation Chair of Psychoanalysis, and their BCM colleagues developed a clear pathway that chairs can now direct their faculty members to which they prepare for academic advancement.

Coburn notes a recent example. Dr. Richard Link, associate professor of urology, is preparing for his next promotion to full professor with tenure. He is a skilled and talented surgeon and conducts research to advance his field. But, in recent years education has been a driving passion for Link. The many hats he wears as an educator include director of the Endourology and Minimally Invasive Surgery Fellowship Program, resident teaching, lecturer to trainees, nurses and practicing surgeons, and producer of videos and book chapters used by medical educators outside of BCM.

Coburn notes, “Traditionally teaching awards were popularity contests.” While Link is certainly popular, having won multiple awards from residents and nurses for his teaching prowess, such recognition is far from sufficient evidence of education scholarship for the appointments and promotions committee.

So, Coburn suggested that Link apply for one or more Fulbright & Jaworski L.L.P. Faculty Excellence Awards (F&J awards). The F&J Awards began at the same time that Fincher et al. issued a challenge to all medical schools to create the “infrastructure needed to foster, assess, and reward scholarship in teaching and other activities supportive of learning.” In a 2002 paper, a group of BCM faculty, including Drs. Coburn and Nancy Searle, former director of what is now called
BCM’s Office of Faculty Development, stated that for a system for recognition of education scholarship to contribute to faculty academic advancement “it must gain a reputation of merit. To those involved, it must look and feel comparable to the traditional peer-reviewed process used in selecting grants for funding or manuscripts for publication.” This is exactly the reputation the F&J Awards have attained. According to Dr. David Tweardy, chief of infectious disease, the M.D. Anderson Foundation Chair, and chair of the BCM Appointments and Promotions Committee, “The F&J awards allows us to codify and make clear to all parties what they need to accomplish for promotion based on education scholarship—clear for applicant and for reviewer.”

“In developing the criteria for the award we focused on the definition of the scholarship of teaching put forward by Ernest I. Boyer when he was president of the Carnegie Foundation for the Advancement of Teaching,” said Searle. The developers of the awards program also incorporated into the requirements the work of Glassick et al in defining six criteria for promoting quality of all types of scholarship to define the requirements for the award in all categories.

Once the concept for the Awards was developed, funds donated by the law firm of Fulbright & Jaworski L.P. were used to support them.

**THE PROCESS**

Link took his chair’s advice and began the process of applying for an F&J Awards.

Faculty must nominate themselves for the award in any of four categories: teaching/evaluation, development of enduring materials, educational leadership, or educational research. For each category, a faculty member creates a mini-portfolio documenting both the quantity and quality of his/her educational activities.

Link decided to apply for two awards in the categories of teaching/evaluation and enduring materials. After attending a workshop on how to prepare an educational portfolio for the F&J, he completed portfolios as per the detailed instructions on the BCM website, following specific examples also included on the website.

Link noted his 57-page portfolio for the enduring materials category required extensive research to track the impact of many of the print and digital materials to which he has contributed. Evidence for quality of work in his portfolios included reviews, awards, letters of support, and repeat invitations to further his involvement in a program, project or committee.

As with all portfolios for F&J Awards, when the review panel received Link’s they were assigned a primary and secondary reviewer, the same approach used by study sections for grants from the National Institutes of Health. Reviewers rate each submission with a maximum of 100 points using weighted criteria, judging them against the examples published on the F&J Awards website. Portfolios may be assigned a maximum of 50 points for quality judged using Glassick’s criteria of scholarship, 40 points for quantity and 10 points for breadth.

The primary and secondary reviewer present their findings to the review panel, which includes BCM faculty as well as members from outside the College, and all panel members assign an overall score to the portfolio. All portfolios that receive an
average score greater than or equal to 80, and are given a score greater than or equal to 80 from at least 75 percent of the panelists qualify for the award. The awards are norm referenced, i.e., judged against a published standard rather than against others submitted in a given cycle, so there is no limit to the number of awards conferred.

“Since we started the program, around 80 percent of the applications have met the criteria for award,” said Searle. She attributes the seemingly high success rate to the extensive preparation faculty members receive, which ensures most do not apply until they are confident they have what is expected of them. At the 2013 spring review panel meeting, 71 portfolios were deemed qualified to earn an award, including both of Link’s submissions.

Link also discovered an unexpected benefit through membership in the BCM Academy of Distinguished Educators, to which every recipient of an F&J Award earns membership for five years. As a member, Link became eligible to apply for grants for education research. “This spurred me to think about new projects I likely would not have pursued without this funding source,” Link said.

As Coburn prepares to present Link for promotion in the near future, he is certain the Appointments and Promotions Committee will weigh these awards heavily in Link’s favor. Coburn, who has served both as chair of the F&J Review Panel and as a member of the Appointments and Promotions Committee, has seen first hand the impact of these awards on the promotions process. As one of the first recipients of an F&J Award, he also has personal experience of the impact on his own career trajectory.

**WIDER IMPACT**

“One of our goals in creating these awards was to create a culture that truly values and supports educational scholarship at Baylor,” recalled Dr. Stephen Greenberg, dean of medical education and the Herman Brown Teaching Professor of Medicine at BCM. “From my perspective as dean, these awards have succeeded in supporting a cadre of individuals who are committed to education and has facilitated retention of good educators.”

The impact of the program has also resonated far beyond the BCM campus. Over her years at BCM, Searle, who recently retired, received multiple calls a month about the F&J Awards from colleagues at other institutions. She worked with several schools to help them implement similar programs using the portfolio examples developed at BCM. At least two national organizations, the American Association of Colleges of Osteopathic Medicine and the Council of Emergency Medicine Residency Directors have also developed programs based on the BCM model as a basis for selecting members of their education academies.

For a full listing of recent F&J Award recipients and more information on the Awards and other programs designed to support a culture of commitment to educational scholarship at Baylor College of Medicine, visit the Office of Faculty Development at bcm.edu/facultydevelopment. For the citations list for this article, please see the inside back cover of this edition of BCMQ.
In June, BIPAI celebrated that decade-old battle in Botswana at the site of the Botswana-Baylor Children’s Clinical Center of Excellence in Gaborone. Since 2003, BIPAI has grown, extending its reach to many more countries in sub-Saharan Africa and opened centers in Angola, Ethiopia, Kenya, Lesotho, Liberia, Libya, Malawi, Tanzania (two), Mozambique, Swaziland and Uganda.

Attending the special ceremonies were Dr. Paul Klotman, president and CEO of BCM; Michael Mizwa, CEO of BIPAI; Nancy R. Calles, Senior Vice President of BIPAI; Lamberto Andreotti, CEO of Bristol-Myers Squibb; John Damonti, President of Bristol-Myers Squibb Foundation, and the Hon. Rev Dr. John Seakgosing, Minister of Health of Botswana.

“We are so proud of the successes of the BIPAI program and the incredible physicians, staff, past and present, who have developed this model of improving access to life-saving care for children and families around the world,” said Klotman.

BIPAI began as the dream of Dr. Mark Kline, now chair of pediatrics at Baylor College.
of Medicine and physician-in-chief at Texas Children’s. In 1996, he was a pediatric infectious disease expert on a Romanian fact-finding trip sponsored by Texas Children’s Hospital when he was struck by the lack of treatment available to children with HIV in the resource-poor setting of the eastern European nation. It took five year before he opened BIPAI’s first clinic in Constanta, Romania, where HIV/AIDS had taken its greatest toll on children in Europe.

Yet the biggest challenge remained: Sub-Saharan Africa where infections rates were as high as 39 percent and treatment opportunities nearly nil.

“I saw the results of what we were doing in Romania and knew it needed to be replicated in Africa,” said Kline, who holds the Ralph D. Feigin Chair for Physician-in-Chief. He persuaded Bristol-Myers Squibb Foundation to sign on to the program, and that group provided the funds to build the first makeshift clinic and then the shiny Clinical Center of Excellence. Other centers have followed, along with the sophisticated *HIV Curriculum for the Health Professionals*, now in its fourth edition. BIPAI experts have partnered with primary care clinics and professionals to provide HIV care and treatment to the families they treat.

At the end of June 2013, 79,561 children and family members were enrolled at Centers of Excellence and satellite clinics operated by BIPAI. A total of 190,157 children and family members were enrolled at outreach sites.

In the late 1990s, after effective antiretroviral drugs became available, some in the public health community said treating children with HIV/AIDS in Africa would be impossible. What they saw as insurmountable, Kline saw as a series of small peaks. He built the Centers of Excellence and recruited good local physicians to help operate them. He recruited idealistic young physicians from the United States to become part of the BIPAI Pediatric AIDS Corps, reversing the typical African brain drain of educated professionals.

Most of all, he realized it was a battle that would be won one child at a time. Children who came to the Centers dull-eyed and emaciated underwent a metamorphosis into healthy youngsters with a future. BIPAI could not solve all the problems of Africa, but it could treat the disease shortening their lives.
LESS MAY BE MORE FOR NEURODEGENERATIVE DISORDERS

A little less mutated protein goes a long way toward helping animals with a devastating inherited condition called spinocerebellar ataxia 1 (SCA1) and might have effects in other diseases as well, researchers from Baylor College of Medicine, the Jan and Dan Duncan Neurological Research Institute at Texas Children’s Hospital and the University of Minnesota reported in the journal *Nature*.

“If you can only decrease the levels of ataxin-1 (the protein involved in SCA1) by 20 percent, you can reduce many symptoms of the disease,” said Dr. Huda Zoghbi, professor of molecular and human genetics and pediatrics at BCM and director of the Neurological Research Institute. She is also a Howard Hughes Medical Institute Investigator. She and her collaborators, Dr. Juan Botas, also of BCM and the Neurological Researcher Institute, Dr. Thomas Westbrook, assistant professor of molecular and human genetics at BCM, and Dr. Harry Orr at Minnesota identified a molecular pathway in the cell (RAS/MAPK/MSK1) with components that can be modulated slightly to reduce the levels of defective ataxin-1, the protein that causes disease in patients with the disorder. In mice, that reduction in protein improved symptoms.

“Now that we know that it works with ataxin-1, we can revisit many proteins whose levels drive neurodegeneration in sporadic and inherited diseases such as Alzheimer’s, Parkinson’s, Huntington’s and other neurological disorders,” said Zoghbi. “This is a pilot study and the results from it are as important as a new pathway in neurodegenerative disease research.”

*Nature. 2013 Jun 20;498(7454):325-31. doi: 10.1038/nature12204. Epub 2013 May 29. Funding for this work came from the Howard Hughes Medical Institute Collaborative Innovation Award and from the Ralph D. Feigin, M.D. Endowed Chair at BCM.*

WHOLE GENOME SEQUENCED OF DEADLY ELEPHANT HERPES VIRUS

Researchers looking for a vaccine against the most prevalent form of a deadly elephant herpesvirus gained a new tool earlier this year when teams from Baylor College of Medicine and Johns Hopkins School of Medicine completed the sequencing of the complete genome elephant endotheliotropic herpesviruses 1A. The finding published in the online journal *Genome Announcements* notes that the virus contains unique elements that could warrant establishing it as a new herpesvirus subfamily category.

“This, virus, EEVH1A, has been found to be the most prevalent when it comes to the hemorrhagic disease found in Asian elephants, which is why we focused on this subspecies,” said Dr. Paul Ling, associate professor of molecular virology and microbiology at BCM. The completed genome sequence of EEHV1A revealed that 60 genes are unique to this form of the virus.

EEHV affects both Asian and African elephants but is most often fatal in Asian elephants. First identified in 1995, it wasn’t until 2010 that a real-time PCR test was developed through a collaboration between BCM and the Houston Zoo. The test diagnoses the virus before elephants begin showing symptoms. Once symptoms are seen, it is usually too late to save the animal.

*Genome Announc. 2013 Apr 11;1(2):e0010613. doi: 10.1128/genomeA.00106-13. This study was funded by Houston Zoo, Inc. and from the National Institutes of Health.*
FIVE DEBAKEY AWARD WINNERS SHOWCASE BAYLOR COLLEGE OF MEDICINE RESEARCH

Research on the brain, reproduction and genetics was the focus of work for which five Baylor College of Medicine scientists received the 2013 Michael E. DeBakey Excellence in Research Award, named in honor of the College’s first president.

Dr. Mauro Costa-Mattioli, assistant professor of neuroscience, whose work was spotlighted in *Cell*, *Nature Neuroscience* and *PNAS*, focuses on memory and how it is stored, which has important implications for cognition.

Dr. Matthew Rasband, the Vivian L. Smith Endowed Chair in Neuroscienc, was cited for work “characterizing the molecular architecture of the axon initial segment and nodes of Ranvier...Nodes are essential for conduction of nerve impulses throughout the nervous system, and their disruption contributes to the pathophysiology of many disorders including multiple sclerosis, peripheral neuropathies, and other white matter diseases.”

Dr. Jeffrey Noebels, the Cullen Trust for Health Care Endowed Chair, has pioneered efforts to understand how a mutant ion channel gene can lead to an epileptic brain. His experiment showing that deletion of the tau protein—usually associated with dementia in a form of Alzheimer’s disease—can prevent epilepsy and its sequelae in both mouse and fly models strengthens understanding of the link between epilepsy and some forms of inherited Alzheimer’s disease.

Dr. Martin Matzuk, the Stuart A. Wallace Chair in Pathology and director of the new Center for Drug Discovery, was cited for a voluminous body of work including one paper in *Cell* in which Matzuk and his colleagues identified a small molecular inhibitor JQ1 that targets a testis-specific protein called BRDT. The small molecule has a reversible, contraceptive effect on male mice.

Dr. Thomas Trey Westbrook, professor of biochemistry and molecular biology and molecular and human genetics, has broken new ground in understanding triple negative breast cancer with identification of the role of the tumor suppressor gene PTPN12 tyrosine phosphatase. He has also identified genes essential to survival of cancer genes driven by the oncogene Myc. He has also developed a new system (pINDUCER) that enables researchers to turn a gene on and off.

The award is sponsored by the DeBakey Medical Foundation and consists of a commemorative medallion and funds to support further research.

REDUNDANT BUILDING MECHANISM PROTECTS BOOSTER SITES FOR CNS COMMUNICATION

Booster stations called the nodes of Ranvier speed electrical signals from neuron to neuron along the exons of nerve cells. Now researchers at Baylor College of Medicine describe the three partially redundant mechanisms needed to make up and assemble those booster sites.

All depend on the myelin sheath that surrounds the axon, said Dr. Matthew Rasband, professor of neuroscience at Baylor College of Medicine, and corresponding author of the report in the journal *Neuron*.

“Even if you eliminate one of the mechanisms, the other two compensate and nodes of Ranvier still form. However, if two are eliminated, the booster stations fail to assemble,” said Rasband.

“All of these processes depend on interaction with the myelinating glial cell,” said Rasband. “There are some disorders, like multiple sclerosis and spinal cord injury, where the overlying myelin sheath is lost, and you lose the ability to maintain the nodes of Ranvier. You effectively lose the booster stations and the electrical signal dies out before reaching its destination. Therefore, any therapeutic strategy aimed at nervous system repair or regeneration must consider how nodes are built.”

*Neuron*. 2013 May 8;78(3):469-82. doi: 10.1016/j.neuron.2013.03.005. Funding for this research was from: the National Institutes of Health; the Dr. Miriam and Sheldon Adelson Medical Research Foundation; the Ministry of Education, Culture, Sports Science, the Vivian L. Smith Endowed Chair in Neuroscience at BCM, and Technology of Japan 24107516; the U.S.–Israel Binational Science Foundation.

FEMALE MICE OVERFED AS INFANTS BECOME COUCH POTATOES FOR LIFE

Overfed baby mice become indolent and obese adults, said researchers from Baylor College of Medicine in a report that appears online in the journal *Diabetes*.

“We have known for decades that when mice are overfed during the newborn period they tend to stay fatter for their entire lives, but we did not know why,” said Dr. Robert Waterland, associate professor of pediatrics - nutrition at BCM and a scientist in the USDA/ARS Children’s Nutrition Research Center at Baylor College of Medicine and Texas Children’s Hospital. “It was generally thought that animals that are over-nourished in infancy just maintain a higher food intake throughout life.”
However, his studies show that these overfed mice remained fatter, even when they received the same amount of food as their skinnier counterpart. The difference was they were less physically active.

“Infancy is critical period for developmental epigenetics in the mouse hypothalamus,” said Waterland. “Overnutrition in infancy is causing persistent changes that last into adulthood. These could mediate the persistent changes in physical activity.”

Diabetes. 2013 Aug;62(8):2773-83. doi: 10.2337/db12-1306. Epub 2013 Apr 1. Funding for this work came from the National Institute of Diabetes and Digestive and Kidney Diseases, and the U.S. Department of Agriculture (USDA). Studies were also performed in the Mouse Metabolic Research Unit at the USDA/Agricultural Research Service Children’s Nutrition Research Center at BCM and Texas Children’s Hospital, which is supported by funds from the USDA ARS.

OSTEOPOROSIS, OSTEOGENESIS IMPERFECTA
MAY START WITH WNT1 MUTATIONS

Mutations in the gene WNT1 are associated with both early-onset osteoporosis and the brittle bone disease osteogenesis imperfecta, said an international consortium of researchers led by those at Baylor College of Medicine in a report in the New England Journal of Medicine.

“What happens if you make higher levels of the protein associated with WNT1? Can you use this protein as a biomarker in blood for the status of bone formation?” said Dr. Brendan Lee, professor of molecular and human genetics at BCM and corresponding author of the report.

Lee and his colleagues found mutations in the WNT1 gene in children with osteogenesis imperfecta and, in collaboration with Finnish colleagues, they found that early-onset osteoporosis can occur when a person carries a single mutated WNT1 gene.

“When there is a mutation in only one copy of the gene, the person develops osteoporosis at an early age,” said Lee. “When both copies are affected, the person has osteogenesis imperfecta.”

The genetic evidence suggests that the strength of the bone is exquisitely sensitive to the dose of WNT1 protein, said Lee.

N Engl J Med. 2013 May 9;368(19):1809-16. doi: 10.1056/NEJMoa1215458. Funding for this work came from the Folkhälsan Research Foundation, the Academy of Finland, the Sigrid Juselius Foundation, the Foundation for Pediatric Research, the Waldemar von Frenckell Foundation, the Helsinki University Research Funds, Helsinki, Finland, the European Calcified Tissue Society Career Establishment Award, National Institutes of Health, Canadian Institutes of Health Research clinician-scientist training award, the Rolanette and Berdon Lawrence Bone Disease Program of Texas, the Robert and Janice McNair Endowed Chair in Molecular and Human Genetics at BCM, and the NIH National Research Service Award.

The architecture of the genome—its variation, its genes and the elements that control them—can define traits that affect our bodies and our health—even the levels of so-called “good cholesterol” (high density lipoprotein cholesterol or HDL-C), said a consortium of researchers including those at Baylor College of Medicine and The University of Texas Health Science Center at Houston (UTHealth) in the journal Nature Genetics.

Researchers analyzed the genome sequences of 962 people to determine the genomic determinants of levels of high density lipoprotein C (HDL-C), the so-called good cholesterol.

“This research shows that parts of the genome with no known function are influencing differences of disease risk,” said Dr. Eric Boerwinkle, associate director of BCM Human Genome Sequencing Center and director of the Human Genetics Center at The University of Texas School of Public Health.

“This study is a precursor to the application of whole genome studies of healthy people as a part of medical practice,” said Dr. Richard Gibbs, director of the BCM Human Genome Sequencing Center and a senior author of the report.

Nat Genet. 2013 Aug;45(8):899-901. doi: 10.1038/ng.2671. Epub 2013 Jun 16. Funding for this work came from Atherosclerosis Risk in Communities (ARIC) Study with National Heart, Lung, Blood Institute contracts and sponsored projects. Other funding came from the Cardiovascular Health Study (CHS) supported by NHLBI contracts, the National Institute of Neurological Disorders and Stroke, the Wofford Cain Chair in Molecular and Human Genetics at BCM, grants from from the National Institute on Aging (NIA) and the Framingham Heart Study (FHS) of the NHLBI and Boston University School of Medicine.
A MEASURE OF EXCELLENCE

Citations List


BCM BITES
The schools of Baylor College of Medicine attract a diverse group of top students from across the country and around the globe. BCM ranks #2 in the U.S. and #1 in Texas in *Hispanic Business Magazine*’s listing of best medical schools for Hispanics.

2013 ENROLLMENT STATISTICS

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* Black or African American, Hispanic/Latino, American Indian or Alaskan Native, Native Hawaiian or other Pacific Islander ethnic categories are considered to be under-represented in science and healthcare.