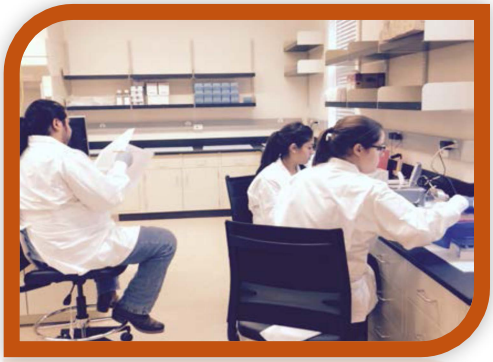
The University of Texas Rio Grande Valley
School of Medicine

South Texas Diabetes & Obesity Institute



Advancing Human Health in the Rio Grande Valley and Beyond through Cutting-Edge Research on Diabetes, Obesity, and Related Disorders

STDOI: Report of Progress During the First 18 Months of Operation

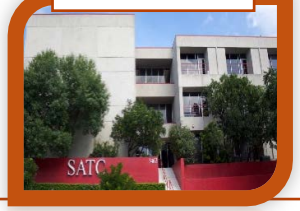
Brownsville

Luis Colom Building

Edinburg

Research Education Building

San Antonio



S. A. Technology Center

This Progress Report is distributed by the South Texas Diabetes and Obesity Institute, School of Medicine, The University of Texas Rio Grande Valley

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UTRGV

I. EXECUTIVE SUMMARY

In October of 2014, I became the Founding Director of an exciting new research organization, the South Texas Institute for Diabetes and Obesity (STDOI), that would become part of the new University of Texas Rio Grande Valley. The STDOI provided myself and my colleagues a once in a lifetime opportunity to join the leadership of a new emerging medical school and to focus research in a way that could have a very positive impact on the local community in addition to having an impact on some of the most pressing public health issues facing the nation today. 21 faculty and staff joined the STDOI in early 2015 to initiate the research efforts that ultimately will improve health in the South Texas region.

The Rio Grande Valley is an area where diabetes, obesity, and associated health issues are reaching crisis levels. That makes it an ideal location for the types of long-term, multidisciplinary studies in which STDOI investigators specialize. Basic research takes many years to achieve the ultimate goal of developing new ways for preventing and treating disease. However, discoveries that occur during the research process can improve clinical care and can improve our understanding of the biological pathways that underlie the major public health problems of diabetes and obesity prior to the identification of new treatments and preventions.

The focus of the researchers working in the STDOI is on genetic studies of diabetes and obesity. Through studies of large extended families we can identify genes in the causal pathways that influence disease. This opens up a window into the pathophysiology of the disease. There are many points at which one can interrupt the causal pathway of a given disease, and knowledge of the genes involved in the pathway may provide novel targets for drug development.

Over the past 25 years, STDOI researchers have worked with large extended Mexican American families in San Antonio to find genes influencing heart disease, diabetes, obesity, and psychiatric disease in the population. Mexican Americans have a lifetime risk of diabetes of between 25 and 30%, which was the reason we focused our work on Mexican American families. Many of the 3500 participants who have collaborated with us in our studies have relatives in the Rio Grande Valley. Now we have a great opportunity to expand our study families through new recruiting efforts here. One of the very attractive possibilities associated with the move of our research group to the Valley was the chance to develop intervention studies that will allow us to look at the genetic basis of differential response to treatment, be it behavioral or pharmacological, while simultaneously providing health improving interventions.

The STDOI team includes individuals who have been pioneers in the whole genome sequencing studies that are the great hope for genetic research today. STDOI scientists have extensive experience in translational work focused on drug target development, work that we hope ultimately will lead to new, more efficacious drugs for treating diabetes, obesity, and related disorders. The research team possesses the full range of skills required for long-term genetic population studies – from direct sampling of the individuals who participate in our studies, to medical assessments, to whole genome sequencing, to complex statistical analysis, to characterizing the actual functional consequences of the genes that we discover. The group also includes leaders in advanced imaging genomics. Our investigators were trailblazers in using MRI and other imaging modalities to identify genetic risk factors for depression and even for obesity-related behaviors and food preferences.

The research group has been highly successful in generating funding. STDOI investigators collectively generated over $200 million in funding from the National Institutes of Health between 2000 and 2014, and have brought over $13.8 million in NIH funding to UTRGV or one of its legacy institutions during the first 18 months of operation.

With the support of the UT system and UTRGV, our research team has established a number of unique resources that have dramatically improved research capabilities in the region. We developed a major high performance computing system with 11,000 processors dedicated to genetic analysis; it is one of the largest clusters devoted to human genetic analysis in the world. We developed state of the art high throughput whole genome sequencing capabilities in the STDOI laboratories in Brownsville. We also established a major program in stem cell biology – our techniques using adult stem cells derived from blood cells allow us to generate specialized cells such as brain cells, liver cells, and heart cells. The approaches allow us to conduct studies that cannot be done on a large scale on cells from biopsies collected from study volunteers. Additionally, we have established a breeding and research colony of laboratory opossums, *Monodelphis domestica*. These animals are uniquely suited for research on skin cancer, correlates of heart disease, fatty liver disease, and spinal cord injury, and will facilitate collaborations with investigators from diverse disciplines.

The STDOI research programs provide numerous opportunities for collaboration with students, postdoctoral scientists, and investigators from across the institutions represented in the Valley, especially UTRGV and the UTHealth School of Public Health. The global nature of science is evident at the STDOI, which collaborates with over 70 institutions throughout the world. Working with UTRGV, we are building a world-class research center focused on diabetes and related disorders in Mexican Americans, as well as on health disparities and minority health issues.

UTRGV President Guy Bailey noted that "as UTRGV and the School of Medicine come to fruition, we are focusing on connecting science and research with the South Texas community. We are working to create the best possible outcomes for our patients." The mission of the STDOI, to advance the health of South Texas and the world through cutting edge research on diabetes, obesity, and related disorders, is well aligned with Dr. Bailey's vision. The work being conducted at STDOI holds promise for development of novel drugs and treatments that can positively impact the lives and health of the people in this region, and of countless others around the world who struggle with diabetes, obesity and heart disease.

This first report from the STDOI presents the great progress achieved during the first 18 months after the Institute became staffed and operational in January 2015. I thank the School of Medicine's Founding Dean Francisco Fernandez, UTRGV President Bailey, UTRGV Provost Havidán Rodriguez, former Chancellor Francisco Cigarroa and UT System for the amazing opportunity to lead the South Texas Diabetes and Obesity Institute. On behalf of the STDOI, I express our thanks to UTRGV, UT System, our colleagues at the UTHealth School of Public Health, H-E-B, the McAllen Medical Center Foundation, Dr. Jean MacCluer, Dr. Bennett Dyke, and all the other donors, colleagues and friends who have supported the Institute throughout its initial phase of development. In addition, we are grateful to Dr. Steven Lieberman, new Interim Dean of the School of Medicine, for his continuing support of the STDOI. We remain committed to improving the health of the region through research and achieving even greater things in the years to come.

Sincerely,

Sarah Williams-Blangero, Ph.D.
Director, South Texas Diabetes and Obesity Institute
Director, Edinburg Regional Academic Health Center

II. History of South Texas Diabetes and Obesity Institute

The South Texas Diabetes and Obesity Institute (STDOI) was established on October 13, 2014, to be a focus for world-class biomedical research programs at the new UTRGV School of Medicine. With diverse research efforts in the areas of diabetes, obesity, heart disease, ocular health, psychiatric disease, osteoporosis, and infectious diseases of border health relevance such as Chagas disease and tuberculosis, the STDOI is working to address the public health problems of the region. The STDOI research team is increasing the amount of NIH funded research at the university and thereby helping UTRGV to achieve its goal of becoming an Emerging Research University.

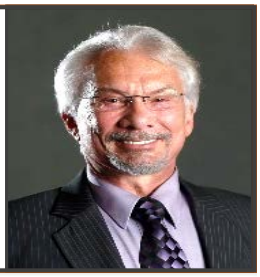
The STDOI administratively was placed in the University of Texas Health Science Center at San Antonio while the UTRGV and its School of Medicine were in development. UTRGV was created in September 2015. Upon the establishment of UTRGV, the STDOI became part of the UTRGV School of Medicine.

III. STDOI HIGHLIGHTS

STDOI scientists established research operations in the Luis Colom Building on the UTRGV Brownsville campus, at the Edinburg Regional Academic Health Center (now Edinburg Research Education Building) on the UTRGV Edinburg campus, and in the San Antonio Technology Center in San Antonio, Texas.

State-of-the-art molecular genetics laboratories and computing facilities were established on the UTRGV Brownsville campus. Dr. John Blangero, Director of STDOI Genomics Computing Center, led the installation of an 11,000-processor high-performance computer cluster named Medusa.

John VandeBerg, Ph.D., coordinated the successful transfer of 1,000 laboratory (*Monodelphis domestica*) opossums to a newly-established laboratory animal facility on the University of Texas Rio Grande Valley Brownsville campus. Dr. VandeBerg leads research using the laboratory opossum model to identify specific genes and environmental factors that influence physiological characteristics in this important animal model.



John Blangero, Ph.D., Director of the STDOI Genomics Computing Center, received the first grant awarded to STDOI at UTRGV, a $4 million award from the National Institute of Mental Health for a four-year project to search for genes influencing psychiatric disorders including schizophrenia, bipolar disorder and major depression. Other STDOI scientists working on the project include molecular geneticist Dr. Joanne Curran, statistical geneticist Dr. Laura Almasy, and bioinformatician Dr. Marcio Almeida.

STDOI celebrated its formation with Founding Dean Francisco Fernandez.

South Texas Diabetes and Obesity Institute in the News

UTRGV recruits 22-person research team to establish South Texas Diabetes & Obesity Institute

The University of Texas Rio Grande Valley and its medical school are poised to become an internationally recognized epicenter for the research and treatment of diabetes and obesity. UTRGV leaders announced Monday morning that renowned genetics and infectious diseases expert Sarah Williams-Blangero, Ph.D., has been appointed director of the South Texas Diabetes & Obesity Institute, a new research, clinical and education program being established at UTRGV. Williams-Blangero, former chair of the Department of Genetics at the Texas Biomedical Research Institute in San Antonio, is bringing 21 additional researchers and support staff with her to the Rio Grande Valley. "With the recruitment of Dr. Williams-Blangero and her team, UTRGV will achieve instantaneous national and international recognition for our health sciences research capabilities," said Francisco Fernandez, M.D., dean of the UTRGV School of Medicine. "These researchers have worked with over 15,000 study volunteers from San Antonio to Nepal, and their work spans the spectrum of medicine from diabetes and obesity to heart disease, osteoporosis, psychiatric disease, cancer and infectious diseases." The South Texas Diabetes & Obesity Institute is being established to advance research of diabetes and obesity, develop better treatments and ultimately improve the health of residents in South Texas and beyond. "As UTRGV and the school of medicine come to fruition, we are focusing on connecting science and research with the South Texas community," UTRGV President Guy Bailey, Ph.D., said. "We are working to create the best possible outcomes for our patients through research, clinical care and education."

Source: https://www.utsystem.edu/news/2014/10/13/utrgv-recruits-22-person-research-team-establish-south-texas-diabetes-obesity-instit

First Director of South Texas Diabetes and Obesity Institute Announced for UTRGV

Leaders from The University of Texas System and The University of Texas Rio Grande Valley announced the creation of the South Texas Diabetes and Obesity Institute at a press conference Monday, Oct. 13 at Main at The University of Texas at Brownsville. "We want everyone to achieve their full potential by connecting science with the health of the community and our promise is to make that possible today," said Dr. Francisco Fernandez, Dean of the UTRGV School of Medicine. "Today, we take another step towards making that a reality." The institute starting Thursday, Oct. 16 will be led by Dr. Sarah Williams-Blangero, formerly the Chair of the Department of Genetics at the Texas Biomedical Research Institute in San Antonio. Williams-Blangero has a master's degree and doctoral degree in biological anthropology from Case Western Reserve University in Cleveland, Ohio. Williams-Blangero will bring more than 20 doctoral-level researchers and support staff to work throughout the Rio Grande Valley on many facets of diabetes and obesity. The group has attracted more than $200 million in National Institutes of Health funding since 2000. "My colleagues and I, we are delighted that we are moving to this new stage of our careers and this new stage of our research program," said Williams-Blangero. "We feel this really is a once in a lifetime opportunity to become involved with a brand new medical school and to focus our research on the local community in addition to having an impact on some of the most pressing public health problems facing our whole nation today."

Source: http://www.utb.edu/newsinfo/archives/2014/Pages/First-Director-of-South-Texas-Diabetes-and-Obesity-Institute-Announced-for-UTRGV.aspx

EDITORIAL: New UT-RGV diabetes, obesity institute
Oct 15, 2014

For over a year now, we have been touting with excitement how important the new University of Texas-Rio Grande Valley medical school will be for our region. This past week, however, it was put into more tangible terms as it was announced that a nationally renowned genetics and infectious disease expert and her team of 22 stellar researchers were affiliating with the university and opening a new research and clinical education program here.

In simpler terms, UT-RGV President Guy Bailey told a meeting with The Monitor's editorial board on Monday that he "felt like he had just won the lottery," in being able to hire Dr. Sarah Williams-Blangero as director of the South Texas Diabetes & Obesity Institute.

Bailey said that since his recruitment of Williams-Blangero and her team, he has received numerous written requests from other prestigious and prominent researchers who are interested in employment here.

That can only bode well for our region. And this is happening before the first medical class matriculates. Already there is national, and possibly an international buzz by experts and top professionals who want to come here and be a part of this new institution.

Williams-Blangero is formerly the chair of the Department of Genetics at the Texas Biomedical Research Institute in San Antonio. She will start here on Oct. 16 and she tells us that she has already found a house to live in and "wants to waste no time" in getting acclimated and familiar with our area and our cultures.

She said her institute will work closely with our community and will solicit large families to help participate in studies on obesity and diabetes and other conditions. She said that medical school students will help analyze data that will be integrated into their studies.

"There are great training opportunities for students through these resources," Williams-Blangero says.

Indeed there are great opportunities for our community to learn how genetics and culture and family traits might contribute to these conditions. Such research could ultimately help our Rio Grande Valley to get fitter and healthier and to shed its image as one of the most obese places in America.

Information will be shared with 120 other institutions that Williams-Blangero works with nationwide.

"We want everyone to achieve their full potential by connecting science with the health of the community and our promise is to make that possible today," Dr. Francisco Fernandez, dean of the UT-RGV School of Medicine, has promised.

"Soon South Texas will be known as the international epicenter for the research and treatment of this complex disease," a news release from the University of Texas System stated.

We hope that will one day be true.

MONITOR EDITORALS

Editorials reflect The Monitor's majority opinion.
http://www.themonitor.com/opinion/editorials/editorial-new-ut-rgv-diabetes-obesity-institute/article_78b0c3c2-5489-11e4-8a74-0017a43b2370.html

At UTRGV, STDOI biomedical research scientist is studying genetic risk factors in diabetes-related eye disease

A trip to Nepal for most people is a once-in-a-lifetime mountain climbing adventure. For Dr. Matthew Johnson, however, it is a twice-a-year excursion for his research into diabetes-related eye diseases. Johnson, a Texas transplant from Queensland, Australia, is an associate professor and biomedical research scientist at The University of Texas Rio Grande Valley South Texas Diabetes and Obesity Institute (STDOI). Along with a 21-person team of researchers led by STDOI Founding Director Dr. Sarah Williams-Blangero, formerly chair of the Department of Genetics at the Texas Biomedical Research Institute in San Antonio, Johnson relocated with other TBOI personnel to the Valley earlier this year with the newly created research institute. "We are looking at genetic risk factors influencing eye health and disease, and are fortunate that Dr. Williams-Blangero and another key member of the STDOI research group, Dr. John Blangero, have established two strong subject cohorts," Johnson said. "One is a population of large Mexican-American families in the San Antonio area, and the other, the one I primarily work with, is in Jiri, Nepal." Jiri is a region that lies about 120 miles east of the Nepalese capital of Kathmandu, and consists of eight villages. Of the 4,500 – 5,000 residents of the area, more than 2,500 have participated in past research projects lead by Dr. Williams-Blangero, and all individuals can be linked into one large family pedigree. "This provides us with a goldmine of information for genetics research," Johnson said. "Understanding the genetics 'behind' blindness and poor vision in understudied populations from the developing world – like the Jirels of Nepal – is an important step in the fight against reducing the global burden associated with poor eye health and disease." Johnson also is leading the effort to establish a long-term project on eye health and disease in Mexican-American families of San Antonio.

Source:http://www.utrgv.edu/en-us/about-utrgv/news/press-releases/2015/november-16-at-utrgv-stdoi-biomedical-research-scientist-is-studying-genetic-risk-factors-in-diabetes-related-eye-disease/

Keeping an eye on prevention

Three May 2015 graduates of The University of Texas Rio Grande Valley's legacy institutions have their sights set on public health, medicine and epidemiology. Cecilia Colom, Samantha Gomez and Johnathon Waggoner have landed gap-year jobs as research associates in the laboratory of Dr. Matthew Johnson, a researcher and associate professor at the UTRGV South Texas Diabetes and Obesity Institute, and principal investigator on STDOI's diabetes-related eye disease research program, with a primary focus on diabetic retinopathy. The three research associates will have a key role in Johnson's research program – processing blood samples collected from populations in the South Texas region and Nepal – to identify and understand the genetic mechanisms that influence eye diseases of major public health importance. The future might be anyone's guess, but if I had to make a calculated prediction about what looms ahead, it is all about Samantha, Cecilia, and Johnathon throughout the first decade of their career as scientists, starting here in the Valley," he said. "Helping promote their core identity as scientists, while creating the scientific home-base for the School of Medicine, is the preeminent professional challenge of the next decade for our programs. STDOI is leading the way in developing a unique range of expertise to train both scientists and physicians meet RGV's health challenges, and that of the world. This is what inspires and excites us every minute of every day about UTRGV and the School of Medicine."

Source:http://www.utrgv.edu/en-us/about-utrgv/news/press-releases/2015/october-19-keeping-an-eye-on-prevention/

McAllen Medical Center Foundation gives UTRGV $100,000 to start endowed professorship at South Texas Diabetes and Obesity Institute

The University of Texas Rio Grande Valley and the South Texas Diabetes and Obesity Institute have gained another partner in the fight against diabetes. The McAllen Medical Center Foundation, a nonprofit wing of the McAllen Medical Center, today announced that it is funding a $100,000 professorship for the institute, a UTRGV research, clinical and education program that will conduct advanced research on diabetes and obesity and, ultimately, help develop better treatments. During a press conference Monday at McAllen Medical Center, UTRGV Founding President Guy Bailey thanked the foundation for its support and said partnerships with area hospitals are key to the success of UTRGV and its School of Medicine. The partnership established with this endowment will allow UTRGV to tackle an increasingly pervasive health issue for the Valley and the nation. The professorship endowment will be funded over two years: $60,000 for the first year and $40,000 for the second year. The gift may also be eligible for additional funding from The University of Texas System's Research Incentive Program for the Comprehensive Universities (UTrip-CU) funding at 30 percent, or $30,000. "When you think about the benefits of having this partnership of the McAllen Medical Center Foundation with the STDOI team, we are looking at a permanent relationship that allows us to think about how best to plan for the future," Fernandez said. "The professorship actually ensures us that this can happen. And we will make the most of it, with new and innovative programs that will eventually improve the health of our citizens. "It not only helps the science, it helps the educational mission. And, of course, if you make the impact that you hope to make in patient care, it helps all three missions: clinical, education and research."
Source:http://www.utrgv.edu/en-us/about-utrgv/news/press-releases/2015/april-27-mcallen-medical-center-foundation-donation/

Thanks a million! H-E-B donates $1 million to UTRGV South Texas Diabetes & Obesity Institute

Christmas came early to The University of Texas Rio Grande Valley with a $1 million gift from H-E-B. H-E-B's generous gift will assist UT Rio Grande Valley in establishing an endowed Chair benefitting the South Texas Diabetes & Obesity Institute (STDOI). With this gift, UTRGV gained an ally in the fight against diabetes and obesity and the promotion of health. The crowd cheered when Dr. Guy Bailey, UTRGV President, and H-E-B representatives opened a large present with the check inside. Laura Gump, group vice president and general manager of H-E-B's Border Region, said H-E-B is proud to partner with UTRGV to embark on advancing education and health care in the Valley. "The opening of UTRGV and the School of Medicine will be monumental for our region and will be at the epicenter of the national education conversation," Gump said. "The positive impact to our families, our students, and our future generations are immeasurable. This is a shining moment in the Rio Grande Valley's history, and H-E-B is profoundly honored and proud to be a partner in the historic transformation taking place in our great South Texas region." Dr. Francisco Fernandez, Inaugural Dean of the UTRGV School of Medicine, said H-E-B's gift will allow UTRGV and the institute to connect its innovations in scientific and medical research to the community, especially diabetes, which is prevalent in the Valley. The H-E-B gift is eligible for up to $250,000 from The University of Texas System Research Incentive Program for the Comprehensive Universities (UTRIP-CU).Source: http://www.utrgv.edu/en-us/about-utrgv/news/press-releases/2014/december-18-heb-1-million-gift/

South Texas Diabetes and Obesity Institute Awarded First NIH Grant

The South Texas Diabetes and Obesity Institute (STDOI) at The University of Texas Rio Grande Valley has received its first major grant award from the National Institutes of Health. The South Texas Diabetes and Obesity Institute was established as a major research center to advance research of diabetes and obesity, develop better treatments and ultimately improve the health of residents in South Texas and beyond. "This exciting and generous grant award from the National Institutes of Health exemplifies the significant funding that UTRGV's cutting-edge research programs will generate as the university strives for Tier 1 status," said Dr. Francisco Fernandez, inaugural dean of the UTRGV School of Medicine. "Support of this caliber also catalyzes our mission at the School of Medicine, allowing us to focus our research on addressing the critical health care needs of the Rio Grande Valley." Dr. John Blangero, director of the Genomics Computing Center at the STDOI and interim director of neurosciences at the UTRGV School of Medicine, has received an award of $4 million from the National Institute of Mental Health for a four-year project to search for genes influencing psychiatric disorders including schizophrenia, bipolar disorder and major depression. The project involves sequencing the entire genomes of more than 2,000 individuals in families in which these disorders are present. The families were sampled from around the world and include participants from South Texas, Pennsylvania, Australia, Costa Rica and Scotland. After obtaining the vast sequence data containing information on the more than 3 billion DNA base pairs for each individual, Blangero and colleagues at the STDOI will employ advanced statistical methods that they have developed to exhaustively search the genomes for genes that can explain the variation in disease risk within and between families. These intensive analyses will be performed on the new 10,000-processor, high-performance computer cluster named Medusa. Other STDOI scientists working on the project include molecular geneticist Dr. Joanne Curran, statistical geneticist Dr. Laura Almasy, and Dr. Marcio Almeida, a bioinformatician.

Source: http://www.utrgv.edu/en-us/about-utrgv/news/press-releases/2015/may-7-diabetes-institute-nih-grant/

Top genetics researcher joins UTRGV's South Texas Diabetes and Obesity Institute

Another top scientist has joined the UTRGV South Texas Diabetes and Obesity Institute's team of world-class researchers. Dr. Srinivas Mummidi became a research associate professor in the UTRGV School of Medicine early this year and already has been awarded a prestigious $1.47 million R01 grant from the National Institutes of Health (NIH). The competitive award is from the National Institute of Allergy and Infectious Diseases (award number R01 AI119131) and will support Mummidi's innovative research on gene expression and its relationship to disease progression. "Dr. Mummidi has a demonstrated record of successful and productive research in several projects in the area of gene regulation, genetic epidemiology and mechanisms related to genetic variants," said Dr. Francisco Fernandez, inaugural dean of the UTRGV School of Medicine. Identifying the genetic determinants of risk for diabetes and for disease progression will generate information that can help us improve the quality of healthcare, as well as the quality of life, in the Rio Grande Valley. We are so pleased to have Dr. Mummidi on board." Mummidi said he was attracted to the STDOI by the great research opportunities available in the Rio Grande Valley and the UTRGV School of Medicine. "This is an exciting move for me, a wonderful chance to work with an outstanding group of investigators, many of whom I collaborated with in San Antonio," he said. "These investigators are world leaders in complex disease genetics, especially in the area of diabetes and obesity. The STDOI provides an excellent environment for the successful conduct of my proposed studies." Source:http://www.utrgv.edu/en-us/about-utrgv/news/press-releases/2016/april-25-top-genetics-researcher-joins-utrgvs-south-texas-diabetes-and-obesity-institute/

COMMENTARY: Diabetes and eye problems
Dr. Matthew Johnson | Guest Columnist Nov 15, 2015

Scientists at the South Texas Diabetes and Obesity Institute (STDOI) are seeking to understand the genetic determinants of risk for eye diseases so that they can contribute to the development of new treatments for eye problems such as cataract, glaucoma and diabetic retinopathy. This work is of particular relevance during this National Diabetes Month, because people with diabetes are at an increased risk of developing vision problems, including blindness.

Diabetes-related vision problems are collectively known as diabetic eye diseases and include diabetic retinopathy, diabetic macular edema, cataract and glaucoma. In diabetic retinopathy the blood vessels that feed the retina, the light sensitive area at the back of the inner eye, change in a manner that allows blood and other fluid to leak onto the retina or into the gel-like substance (vitreous humor) within the eyeball. This leakage results in impaired vision. Diabetic macular edema is a consequence of diabetic retinopathy and causes swelling of a specialized area of the retina known as the macula. The macula is responsible for central vision. Individuals suffering from diabetic macular edema are likely to have difficulty reading or recognizing faces.

Adults with diabetes are at a higher risk of developing cataract than normal adults, and also are more likely to develop cataract at a much younger age than most people. In individuals with cataract, the lens of the eye is cloudy, and so the light that enters a lens with a cataract is distorted. The distortion prevents a sharp, clear image being formed and results in blurred vision. Adults with diabetes are approximately two times more likely to develop glaucoma than adults without diabetes. Glaucoma causes damage to the optic nerve, which is the bundle of nerve fibers connecting the eye to the brain. Individuals with glaucoma suffer loss of peripheral vision; that is, they experience tunnel vision.

Diabetic eye diseases may go unnoticed until significant vision loss starts to occur. In some situations, such as untreated glaucoma, the vision loss cannot be restored. In other situations, such as is the case for diabetic retinopathy, medical therapy can treat or prevent the risk of further loss of eyesight.

It is well known that an individual's risk of developing diabetic eye diseases is also influenced by genetic factors. STDOI scientists are trying to identify the genetic mechanisms that influence risk for diabetic eye diseases. The discovery of these genetic mechanisms can provide a basis for developing tests for early detection of risk prior to any vision loss. In addition, such discoveries may trigger the development of new medical treatments to prevent further vision loss or to restore lost vision. STDOI scientists are currently working on studies of diabetic eye disease in Mexican-Americans from San Antonio and in a population living in eastern Nepal.

The STDOI's mission is to advance the health of South Texas and the world through cutting edge research on diabetes and obesity-related disorders. Researchers at the STDOI are now working to establish studies in the Rio Grande Valley that will recruit volunteers from the region to participate in research on the genetic mechanisms contributing to diabetic eye disease. Through this research, STDOI scientists hope that ultimately they can contribute to improving vision health in the Valley. Preventive measures like maintaining healthy blood sugar levels and early detection of vision problems are effective ways to protect against vision loss associated with diabetes. Individuals with diabetes should get an annual dilated eye exam to allow for close monitoring of vision health.

Dr. Matthew Johnson is an associate professor in the South Texas Diabetes and Obesity Institute. He is a molecular geneticist who has conducted research aimed at understanding the genetic determinants of risk for hypertension in pregnancy. He is now focused on understanding the role of genetics in risk for eye diseases, particularly vision loss associated with diabetes.

Source:http://www.themonitor.com/opinion/commentary-diabetes-and-eye-problems/article_8b65fb10-8a32-11e5-9f2b-773658565212.html

South Texas Diabetes and Obesity Institute
Faculty and Staff

IV. South Texas Diabetes & Obesity Institute Faculty & Staff

Name	Title
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John Blangero, Ph.D.	Professor
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Ravindranath Duggirala, Ph.D.	Professor
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Marcelo J.F. Leandro, M.S.	Research Assoc. III
Liza D. Morales Smith, M.S.	Research Assoc. III
Roy G. Resendez, M.A.	Research Assoc. III
Katherine Truax, B.S.	Research Assoc. II
Jose A. Hernandez, Ph. Cand.	Research Assoc. II
Cecilia M. Colom, B.S.	Research Assoc. I
Samantha M. Gomez, B.S.	Research Assoc. I
Johnathon M. Waggoner, B.S.	Research Assoc. I
Susan M. Mahaney, B.S.	Laboratory Manager
Azaneth Arellanes, B.S.	Lab Technician
Computing Staff	
Gerry W. Vest, M.S.	Bus. Sys. & Tech. Mgr.
Juan M. Peralta, M.Sc.	Prog. Analyst IV
Cristina R. Molina, B.S.	Syst. Analyst III
Max D. Torres, B.S.	Syst. Analyst III
Animal Care Staff	
Alejandro Reyes Canchola, B.S.	Animal Care Tech. II
Gabriel E. Lopez, B.S.	Animal Care Tech. II
Oscar Garza Salinas, B.S.	Animal Care Tech. I
Rodolfo (Rudy) Gomez	Student Assistant
Reyna G. Garcia	Student Assistant
Administrative Staff	
Lora A. Boyd, B.S.	Associate Director
Robert L. Gomez, M.B.A.	Financial Mgr.
Wanda Wiley	Admin. Asst. II
Andy Zuniga, B.A.S.	Office Asst. III

South Texas Diabetes and Obesity Institute
Faculty Profiles

V. SOUTH TEXAS DIABETES & OBESITY INSTITUTE FACULTY PROFILES

LAURA ALMASY, PH.D.

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EDUCATION:

1996	Ph.D.	Genetics Yale University, New Haven, CT
1992	M.Phil.	Genetics Yale University, New Haven, CT

RESEARCH FOCUS:

The central focus of Dr. Almasy's research is the development, testing, and application of statistical genetic methods for identifying and characterizing genes influencing complex traits and related quantitative risk factors. Much of her applied work is in the area of psychiatric genetics where she is involved in genetic studies of schizophrenia, alcoholism and other addictions, and normal variation in brain structure and function. She is one of the principal investigators of the multi-site Multiplex Multigenerational Investigation of Schizophrenia (MGI) study and collaborates on several consortium studies combining the MGI with the COGS and PAARTNERS schizophrenia studies. Further, she leads the Texas site of the multi-site Collaborative Study on the Genetics of Alcoholism (COGA) and directs the Genetic Analysis Workshop (GAW)--an international forum for development and testing of statistical genetic methods started by Dr. Jean MacCluer in 1982. GAW19 was held in 2014 and concentrated on analysis of human sequence and gene expression data. Dr. Almasy is assisting in the preparation for GAW20 which will be held in early spring of 2017.

BOOK(S):

Duggirala R, **Almasy L**, Williams-Blangero S, Paul S, Kole C (eds). Genome Mapping and Genomics in Human and Non-Human Primates, Springer-Verlag Berlin Heidelberg. 2015.

BOOK CHAPTERS:

1. **Almasy L**, Kos MZ, Blangero J. Linkage mapping: Localizing the genes that shape human variation. Pp. 33-52. In: Genome Mapping and Genomics in Human and Non-Human Primates. R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.
2. Arya R, Puppala S, Farook VS, Chittoor G, Jenkinson CP, Blangero J, Hale DE, Duggirala R, **Almasy L**. Mapping of susceptibility genes for obesity, type 2 diabetes, and the metabolic syndrome in human populations. Pp. 181-246. In: Genome Mapping and Genomics in Human and Non-Human Primates, R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.
3. Curran JE, Bellis C, **Almasy L**, Blangero J. Genomic studies of human populations: Resequencing approaches to the identification of human quantitative loci. Pp. 289-299. In: Genome Mapping and Genomics in Human and Non-Human Primates, R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.

PUBLICATIONS 01/2015 – 06/2016:

1. Nicholson AM, Finch NA, Almeida M, Perkerson RB, van Blitterswijk M, Wojtas A, Cenik B, Rotondo S, Inskeep V, **Almasy L**, Dyer T, Peralta J, Jun G, Wood AR, Frayling TM, Fuchsberger C, Fowler S, Teslovich TM, Manning AK, Kumar S, Curran J, Lehman D, Abecasis G, Duggirala R, Pottier C, Zahir HA, Crook JE, Karydas A, Mitic L, Sun Y, Dickson DW, Bu G, Herz J, Yu G, Miller BL, Ferguson S, Petersen RC, Graff-Radford N, Blangero J, Rademakers R. Prosaposin is a regulator of progranulin levels and oligomerization. Nat Commun. 2016 Jun 30;7:11992. doi: 10.1038/ncomms11992. PubMed PMID: 27356620.

2. Hodgson K, **Almasy L**, Knowles EE, Kent JW, Curran JE, Dyer TD, Göring HH, Olvera RL, Fox PT, Pearlson GD, Krystal JH, Duggirala R, Blangero J, Glahn DC. Genome-wide significant loci for addiction and anxiety. Eur Psychiatry. 2016 Jun 15;36:47-54. doi: 10.1016/j.eurpsy.2016.03.004. [Epub ahead of print] PubMed PMID: 27318301.

3. Hanson RL, Leti F, Tsinajinnie D, Kobes S, Puppala S, Curran JE, **Almasy L**, Lehman DM, Blangero J, Duggirala R, DiStefano JK. The Arg59Trp variant in ANGPTL8 (betatrophin) is associated with total and HDL-cholesterol in American Indians and Mexican Americans and differentially affects cleavage of ANGPTL3. Mol Genet Metab. 2016 Jun;118(2):128-37. doi: 10.1016/j.ymgme.2016.04.007. Epub 2016 Apr 19. PubMed PMID: 27117576; PubMed Central PMCID: PMC4880492.

4. Chartier KG, Dick DM, **Almasy L**, Chan G, Aliev F, Schuckit MA, Scott DM, Kramer J, Bucholz KK, Bierut LJ, Nurnberger J Jr, Porjesz B, Hesselbrock VM. Interactions between alcohol metabolism genes and religious involvement in association with maximum drinks and alcohol dependence symptoms. J Stud Alcohol Drugs. 2016 May;77(3):393-404. PubMed PMID: 27172571; PubMed Central PMCID: PMC4869897.

5. Mamtani M, Kulkarni H, Wong G, Weir JM, Barlow CK, Dyer TD, **Almasy L**, Mahaney MC, Comuzzie AG, Glahn DC, Magliano DJ, Zimmet P, Shaw J, Williams-Blangero S, Duggirala R, Blangero J, Meikle PJ, Curran JE. Lipidomic risk score independently and cost-effectively predicts risk of future type 2 diabetes: Results from diverse cohorts. Lipids Health Dis. 2016 Apr 4;15(1):67. doi: 10.1186/s12944-016-0234-3. PubMed PMID: 27044508; PubMed Central PMCID: PMC4820916.

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10. Mamtani M, Kulkarni H, Dyer TD, Göring HH, Neary JL, Cole SA, Kent JW, Kumar S, Glahn DC, Mahaney MC, Comuzzie AG, **Almasy L**, Curran JE, Duggirala R, Blangero J, Carless MA. Genome- and epigenome-wide association study of hypertriglyceridemic waist in Mexican American families. Clin Epigenetics. 2016 Jan 20;8:6. doi: 10.1186/s13148-016-0173-x. eCollection 2016. PubMed PMID: 26798409; PubMed Central PMCID: PMC4721061.

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Kent JW Jr, Kochunov P, Kwok JB, Lawrie SM, Liu X, Longo DL, McMahon KL, Meisenzahl E, Melle I, Mohnke S, Montgomery GW, Mostert JC, Mühleisen TW, Nalls MA, Nichols TE, Nilsson LG, Nöthen MM, Ohi K, Olvera RL, Perez-Iglesias R, Pike GB, Potkin SG, Reinvang I, Reppermund S, Rietschel M, Romanczuk-Seiferth N, Rosen GD, Rujescu D, Schnell K, Schofield PR, Smith C, Steen VM, Sussmann JE, Thalamuthu A, Toga AW, Traynor BJ, Troncoso J, Turner JA, Valdés Hernández MC, van 't Ent D, van der Brug M, van der Wee NJ, van Tol MJ, Veltman DJ, Wassink TH, Westman E, Zielke RH, Zonderman AB, Ashbrook DG, Hager R, Lu L, McMahon FJ, Morris DW, Williams RW, Brunner HG, Buckner RL, Buitelaar JK, Cahn W, Calhoun VD, Cavalleri GL, Crespo-Facorro B, Dale AM, Davies GE, Delanty N, Depondt C, Djurovic S, Drevets WC, Espeseth T, Gollub RL, Ho BC, Hoffmann W, Hosten N, Kahn RS, Le Hellard S, Meyer-Lindenberg A, Müller-Myhsok B, Nauck M, Nyberg L, Pandolfo M, Penninx BW, Roffman JL, Sisodiya SM, Smoller JW, van Bokhoven H, van Haren NE, Völzke H, Walter H, Weiner MW, Wen W, White T, Agartz I, Andreassen OA, Blangero J, Boomsma DI, Brouwer RM, Cannon DM, Cookson MR, de Geus EJ, Deary IJ, Donohoe G, Fernández G, Fisher SE, Francks C, Glahn DC, Grabe HJ, Gruber O, Hardy J, Hashimoto R, Hulshoff Pol HE, Jönsson EG, Kloszewska I, Lovestone S, Mattay VS, Mecocci P, McDonald C, McIntosh AM, Ophoff RA, Paus T, Pausova Z, Ryten M, Sachdev PS, Saykin AJ, Simmons A, Singleton A, Soininen H, Wardlaw JM, Weale ME, Weinberger DR, Adams HH, Launer LJ, Seiler S, Schmidt R, Chauhan G, Satizabal CL, Becker JT, Yanek L, van der Lee SJ, Ebling M, Fischl B, Longstreth WT Jr, Greve D, Schmidt H, Nyquist P, Vinke LN, van Duijn CM, Xue L, Mazoyer B, Bis JC, Gudnason V, Seshadri S, Ikram MA; Alzheimer's Disease Neuroimaging Initiative; CHARGE Consortium; EPIGEN; IMAGEN; SYS, Martin NG, Wright MJ, Schumann G, Franke B, Thompson PM, Medland SE. Common genetic variants influence human subcortical brain structures. Nature. 2015 Apr 9;520(7546):224-9. doi: 10.1038/nature14101. Epub 2015 Jan 21. PubMed PMID: 25607358; PubMed Central PMCID: PMC4393366.

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GRANT REVIEW:

Health Research Board (Ireland): Research Training Fellowship for Healthcare Professionals

EXTRAMURAL APPOINTMENTS:

Member, Genomics and Computational Biology Editorial Advisory Board

Member, Genes Brain and Behavior Editorial Board

Review Editor, Frontiers in Applied Genetic Epidemiology

Member, International Society of Psychiatric Genetics Finance Committee

Judge, World Congress on Psychiatric Genetics Early Career Investigator Program Oral and Poster Presentation Awards

Voting Member, Collaborative Study on the Genetics of Alcoholism Steering Committee

Member, Collaborative Study on the Genetics of Alcoholism Genetic Analysis Committee

Chair, Genetic Analysis Workshop Advisory Committee

Chair, NHGRI Center for Inherited Disease Research Access Committee

Chair, UTHSCSA Institute for Integration of Medicine and Science, GCCRI and TBRI Pilot Project proposals

Member, Special Emphasis Panel ZRG1 IMST-D (55) for New Computational Methods for Understanding the Functional Role of DNA Variants that are Associated with Mental Disorders

Member, Special Emphasis Panel ZMH1 ERB - C (03) "Unveiling the Genome: Genetic Architecture of Several Mental Disorders Revealed"

Member, Special Emphasis Panel for NHLBI PAR-13-316: NHLBI Program Project Applications

COLLABORATING INSTITUTIONS:

Icahn School of Med. at Mt. Sinai	University of California at San Diego	Virginia Commonwealth Univ.
Indiana University	University of Connecticut	Washington University
Rutgers University	University of Iowa	Yale University
SUNY Downstate	University of Pennsylvania	
Texas Biomedical Research Institute	University of Pittsburgh	

GRANT SUPPORT:

					Awarded from 01/01/2015 – 06/30/2016		
Role	Project Title	Grant Period	Funding Agency	Grant #	Direct Costs	Indirect Costs	Total
P.I.	Neurodevelopment: Genes, Environment and their Interactions	08/01/2015 - 05/31/2018	NIH/ NIMH	R01 MH107248	299,976	172,439	472,415
P.I.	A Neurobehavioral Family Study of Schizophrenia	01/01/2015 - 12/31/2018	NIH/ NIMH	R01 MH061622	640,237	265,032	905,269
P.I.	Genetic Analysis of Common Diseases: An Evaluation	08/01/2013 - 07/31/2017	NIH/ NIGMS	R01 GM031575	699,637	316,936	1,016,573
P.I. (UT-RGV)	Collaborative Study on the Genetics of Alcoholism	09/01/2014 - 08/31/2019	NIH/ NIAAA (Res. Found. State U. of N.Y., SUNY)	U10 AA008401	187,049	88,076	275,125
Co-Inv.	Whole Genome Sequencing to Identify Causal Genetic Variants Influencing CVD Risk	04/15/2012 - 03/31/2017	NIH/ NHLBI	R01 HL113323	- - -	- - -	- - -
Co-Inv.	2/3: Pedigree-Based Whole Genome Sequencing of Affective and Psychotic Disorders	03/01/2015 - 02/28/2019	NIH/ NIMH	U01 MH105632	- - -	- - -	- - -

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EDUCATION:

1986	Ph.D.	Population Genetics Case Western Reserve University, Cleveland, Ohio
1982	M.A.	Biological Anthropology Case Western Reserve University, Cleveland, Ohio

RESEARCH FOCUS:

The genetic underpinnings of common diseases like diabetes and heart disease have proven to be complex. Dr. Blangero has focused his research on the development and application of sophisticated statistical methods to analyze the vast quantities of genetic data generated in large family studies. Dr. Blangero is developing novel approaches to integrate bioinformatic data mining, gene expression analysis, epigenomic, metabolomic, functional genomic and association analysis into methods that will significantly reduce the time and cost of gene discovery.

In addition to serving as Director of the STDOI Genomics Computing Center in the UTRGV School of Medicine, Dr. Blangero is the director of the San Antonio Family Heart Study which continues to be an invaluable resource for exploring many genetic questions. His research group is utilizing data generated by this project to search for genes that influence risk of cardiovascular disease and diabetes. He also has a major program searching for genes that regulate brain structure and function. The San Antonio Family Heart Study was one of the first major studies to incorporate genomic imaging--an emerging field that combines genetic analysis with magnetic resonance imaging to obtain highly detailed information on both anatomic variation and function in the heart and in the brain.

Dr. Blangero led the establishment of state-of-the-art molecular genetics laboratories and computing facilities on the UTRGV Brownsville campus and orchestrated the installation of an 11,000-processor high-performance computer cluster named MEDUSA which serves as one of the world's largest parallel computing cluster devoted to genetic research. Dr. Blangero's group also added new genome sequencers to enable deep sequencing techniques to more quickly find gene variants, measure their output, and accelerate the pace of translational research leading to better diagnostics and eventual treatments for improved human health. Most recently, Dr. Blangero has been involved in the development of a large-scale program for the routine production of stem cells from blood for use in functional genomic studies. His laboratory is pursuing high dimensional phenotyping of different cell types that are derived from such stem cells to better understand the function of human sequence variation.

Dr. Blangero has major multi-year multi-million dollar research grants from the National Institute of Mental Health, the National Institute of Diabetes and Digestive Disorders, and the National Heart Lung and Blood Institute to utilize whole genome sequencing to search for genes influencing psychiatric disorders, diabetes, and cardiovascular disease respectively.

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Kiukaanniemi SM, Kiemeney LA, Knekt P, Kooner JS, Kooperberg C, Kovacs P, Kraja AT, Kumari M, Kuusisto J, Lakka TA, Langenberg C, Le Marchand L, Lehtimäki T, Lyssenko V, Männistö S, Marette A, Matise TC, McKenzie CA, McKnight B, Moll FL, Morris AD, Morris AP, Murray JC, Nelis M, Ohlsson C, Oldehinkel AJ, Ong KK, Madden PA, Pasterkamp G, Peden JF, Peters A, Postma DS, Pramstaller PP, Price JF, Qi L, Raitakari OT, Rankinen T, Rao DC, Rice TK, Ridker PM, Rioux JD, Ritchie MD, Rudan I, Salomaa V, Samani NJ, Saramies J, Sarzynski MA, Schunkert H, Schwarz PE, Sever P, Shuldiner AR, Sinisalo J, Stolk RP, Strauch K, Tönjes A, Trégouët DA, Tremblay A, Tremoli E, Virtamo J, Vohl MC, Völker U, Waeber G, Willemsen G, Witteman JC, Zillikens MC, Adair LS, Amouyel P, Asselbergs FW, Assimes TL, Bochud M, Boehm BO, Boerwinkle E, Bornstein SR, Bottinger EP, Bouchard C, Cauchi S, Chambers JC, Chanock SJ, Cooper RS, de Bakker PI, Dedoussis G, Ferrucci L, Franks PW, Froguel P, Groop LC, Haiman CA, Hamsten A, Hui J, Hunter DJ, Hveem K, Kaplan RC, Kivimaki M, Kuh D, Laakso M, Liu Y, Martin NG, März W, Melbye M, Metspalu A, Moebus S, Munroe PB, Njølstad I, Oostra BA, Palmer CN, Pedersen NL, Perola M, Pérusse L, Peters U, Power C, Quertermous T, Rauramaa R, Rivadeneira F, Saaristo TE, Saleheen D, Sattar N, Schadt EE, Schlessinger D, Slagboom PE, Snieder H, Spector TD, Thorsteinsdottir U, Stumvoll M, Tuomilehto J, Uitterlinden AG, Uusitupa M, van der Harst P, Walker M, Wallaschofski H, Wareham NJ, Watkins H, Weir DR, Wichmann HE, Wilson JF, Zanen P, Borecki IB, Deloukas P, Fox CS, Heid IM, O'Connell JR, Strachan DP, Stefansson K, van Duijn CM, Abecasis GR, Franke L, Frayling TM, McCarthy MI, Visscher PM, Scherag A, Willer CJ, Boehnke M, Mohlke KL, Lindgren CM, Beckmann JS, Barroso I, North KE, Ingelsson E, Hirschhorn JN, Loos RJ, Speliotes EK. Genetic studies of body mass index yield new insights for obesity biology. Nature. 2015 Feb 12;518(7538):197-206. doi: 10.1038/nature14177. PubMed PMID: 25673413; PubMed Central PMCID: PMC4382211.

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MANUSCRIPT REVIEW:

American Journal of Human Genetics
Biological Psychiatry
Nature Genetics

INTRAMURAL APPOINTMENTS:

Director, Genomics Computing Center, STDOI, UTRGV School of Medicine
Interim Director, Neurosciences, UTRGV School of Medicine
Member, Strategic Planning Committee, University of Texas Rio Grande Valley
Member, Search Committee for Chair, UTRGV School of Medicine Department of Biomedical Sciences

EXTRAMURAL APPOINTMENTS:

Member, Editorial Advisory Board, Genomics and Computational Biology
Member, American Diabetes Association Grant Panel
Member, NIMH Grant Review Committee for RFA “Gene Networks: Solving the Molecular Puzzle of Psychiatric Disorders”

COLLABORATING INSTITUTIONS:

Menzies Research Institute	University of Pennsylvania	Yale University
National Institute for Mental Health	University of Pittsburgh	
University of Costa Rica	University of Tasmania, Australia	
University of Edinburgh	University of Western Australia	

GRANT SUPPORT:

Role	Project Title	Grant Period	Funding Agency	Grant #	Awarded from 01/01/2015 – 06/30/2016 Direct Costs	Indirect Costs	Total
P.I. (MPI)	Whole Genome Sequencing to Identify Causal Genetic Variants Influencing CVD Risk	04/15/2012 - 03/31/2017	NIH/ NHLBI	R01 HL113323	4,038,105	787,190	4,825,295
P.I.	2/3: Pedigree-Based Whole Genome Sequencing of Affective and Psychotic Disorders	03/01/2015 - 02/28/2019	NIH/ NIMH	U01 MH105632	1,294,264	636,216	1,930,480
P.I.	Gene Networks Influencing Psychotic Dysconnectivity in African Americans (Year 01 – 2014; Year 02 - 2015)	12/10/2014 - 11/30/2019	NIH/ NIMH (Yale)	R01 MH106324	128,131	58,043	186,174
P.I. (MPI)	Examination of Sequence Variants in Large Human Pedigrees to Identify Novel Genetics Variants Involved in Cardiac (Patho) Physiology	11/12/2015 - 11/12/2018	Eli Lilly	Contract	281,300	127,428	408,728
P.I. (MPI)	Discovery of Functional Variants in Type 2 Diabetes Genes in Mexican Americans	05/01/2015 - 04/30/2019	NIH/ NIDDK	U01 DK085524	669,282	333,224	1,002,506
Co-Inv.	Neurodevelopment: Genes, Environment and their Interactions	08/01/2015 - 05/31/2018	NIH/ NIMH	R01 MH107248	- - -	- - -	- - -
Co-Inv.	A Neurobehavioral Family Study of Schizophrenia	01/01/2015 - 12/31/2018	NIH/ NIMH	R01 MH061622	- - -	- - -	- - -
Co-Inv.	A Genetic Study of Blood Metabolites and Their Relationship to Diabetes Risk	09/01/2013 - 06/30/2017	NIH/ NIDDK	R01 DK099051	- - -	- - -	- - -
Co-Inv.	Genetic Epidemiology of Ocular Health and Disease	06/01/2014 - 05/31/2019	NIH/NEI	R01 EY024384	- - -	- - -	- - -
Co-Inv.	Genetics of Type 2 Diabetes in Indian Populations: US-India Collaboration Project	04/01/2016 - 03/31/2018	NIH/ NIDDK	R21 DK105913	- - -	- - -	- - -
Co-Inv. (UT-RGV)	A Joint Linkage/Association Strategy to Interrogate AMD Genetic Susceptibility	09/01/2015 - 08/31/2016	NIH/NEI (OHSU)	R01 EY021532	- - -	- - -	- - -

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RESEARCH FOCUS:

Dr. Curran's research focuses on identifying and characterizing susceptibility genes for disease conditions such as type 2 diabetes, obesity, cardiovascular disease and related complications in the general population, with the ultimate objective of gaining an insight into the biological pathways involved in disease pathogenesis. Most recently, her research efforts have been directed towards assessment of the human lipidome and its relationship to disease. The human plasma lipidome consists of many thousands of individual lipid species, and Dr. Curran and colleagues have previously measured hundreds of these species in a large study of Mexican American families. Her research results have demonstrated a role for lipid species in hypertension, metabolic syndrome, obesity, prediction of type 2 diabetes development, and cardiovascular disease outcomes. Using these lipids and available whole genome sequencing data, she now is identifying novel genes and functional variants that influence both lipid metabolism and disease development.

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INTRAMURAL APPOINTMENTS:

Member, Admissions Committee, UTRGV School of Medicine

Discipline Coordinator for Genetics, Central Curriculum Authority Committee, UTRGV School of Medicine

EXTRAMURAL APPOINTMENTS:

Member, NIH-NIDDK Diabetes, Endocrinology and Metabolic Diseases B Committee (2011-2017)

Adjunct Research Fellow, Baker IDI Heart and Diabetes Institute

Adjunct Research Fellow, Griffith Institute for Health and Medical Research, Griffith University

COLLABORATING INSTITUTIONS:

Baker IDI Heart & Diabetes Institute	Oregon Health and Science University	Wake Forest University School of Medicine
Indiana University School of Medicine	Stanford University	Wright State University
Institute of Health & Biomedical Innovation, Queensland University of Technology	Universidad de Costa Rica	Yale University
Menzies Research Institute, University of Tasmania, TAS Australia	UT Health Science Center at San Antonio	
National Institute of Diabetes and Digestive and Kidney Diseases	UT Health Science Center at Houston School of Public Health	

GRANT SUPPORT:

Role	Project Title	Grant Period	Funding Agency	Grant #	Awarded from 01/01/2015 – 06/30/2016 Direct Costs	Indirect Costs	Total
P.I. (MPI)	Whole Genome Sequencing to Identify Causal Genetic Variants Influencing CVD Risk	04/15/2012 - 03/31/2017	NIH/ NHLBI	R01 HL113323	4,038,105	787,190	4,825,295
P.I. (MPI)	Examination of Sequence Variants in Large Human Pedigrees to Identify Novel Genetics Variants Involved in Cardiac (Patho) Physiology	11/12/2015 - 11/12/2018	Eli Lilly	Contract	281,300	127,428	408,728
Co-Inv.	2/3: Pedigree-Based Whole Genome Sequencing of Affective and Psychotic Disorders	03/01/2015 - 02/28/2019	NIH/ NIMH	U01 MH105632	- - -	- - -	- - -
Co-Inv.	A Neurobehavioral Family Study of Schizophrenia	01/01/2015 - 12/31/2018	NIH/ NIMH	R01 MH061622	- - -	- - -	- - -
Co-Inv.	Discovery of Functional Variants in Type 2 Diabetes Genes in Mexican Americans	05/01/2015 - 04/30/2019	NIH/ NIDDK	U01 DK085524	- - -	- - -	- - -
Co-Inv.	A Genetic Study of Blood Metabolites and Their Relationship to Diabetes Risk	09/01/2013 - 06/30/2017	NIH/ NIDDK	R01 DK099051	- - -	- - -	- - -

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EDUCATION:

1995	Ph.D.	Anthropology/Biological Anthropology University of Kansas, Lawrence, Kansas, USA
1991	M.A.	Anthropology University of Montana, Missoula, Montana, USA
1978	M.Sc.	Human Genetics and Physical Anthropology Andhra University, Visakhapatnam, Andhra Pradesh, India

RESEARCH FOCUS:

With dual interests in anthropological genetics and genetic epidemiology, Dr. Duggirala's research group pursues a wide range of human population studies around the world. Past, present and future collaborative projects include investigations of genetic and cultural influences on lipids in Mennonites from Kansas and Nebraska; localization and identification of genes that influence susceptibility to complex diseases including obesity, type 2 diabetes mellitus (T2DM), metabolic syndrome (MS), cardiovascular disease, diabetic nephropathy, and gallbladder disease (GBD) in Mexican Americans; genetic studies of T2DM in native populations in India; and genetic studies of tuberculosis in Mexican populations in Chihuahua, Mexico.

The growing epidemic of childhood obesity and its complications prompted Dr. Duggirala's research group to conduct a unique study of nondiabetic children and youth aged 6-17 years from large predominantly lower-income Mexican American families known to be at increased risk for diabetes. The San Antonio Family Assessment of Metabolic Risk Indicators in Youth (SAFARI) study has generated data showing that many of the children are headed early toward T2DM-related health problems including obesity, pre-diabetes, and metabolic syndrome. Critical findings generated by the project suggest that if screening of high-risk children could be performed by age six, it would provide an improved window of opportunity for interventions (e.g., diet and physical activity) that could delay or prevent the development of major health issues later in life. Dr. Duggirala is now seeking to implement culturally-sensitive and family-based lifestyle interventions for Mexican American children and adolescents to prevent the development of future serious health problems.

Dr. Duggirala's research group is strongly committed to major collaborative efforts such as the T2D-GENES Consortium. The Consortium is an effort to confirm and characterize the genetic associations for T2DM identified from recent genome-wide association studies as well as to identify additional novel T2DM susceptibility genes/variants in the Mexican American population. The T2D-GENES project examines similarities and differences in the distribution of T2DM susceptibility variants among five ancestry groups: Europeans, East Asians, South Asians, Hispanic Americans, and African Americans. Dr. Duggirala's research group is now part of the newly formed Accelerating Medicines Partnership (AMP) T2D-GENES consortium that will build on and expand the research activities of the T2D-GENES consortium.

At the international level, the research group has established an NIH-funded Collaborative Research Partnership (CRP) between two US institutions (including the University of Texas Rio Grande Valley) and three institutions in India to evaluate genetic determinants of T2DM among Indian populations. Another collaborative project aims to use genome-metabolome technologies to identify novel genetically-driven small molecule biomarkers to predict diabetes risk linked to early onset of T2DM at lower obesity thresholds, using data from Punjabi/Sikh families from India, and to explore their relevance to multi-ethnic populations of the US. Dr. Duggirala also has led collaborative interactions between U.S. and Mexican researchers that have resulted in a growing program on the genetics of susceptibility to tuberculosis and a paper on the epidemiology of TB in Ciudad Juarez, Mexico.

BOOK(S):

Duggirala R, Almasy L, Williams-Blangero S, Paul S, Kole C (eds). Genome Mapping and Genomics in Human and Non-Human Primates, Springer-Verlag Berlin Heidelberg. 2015.

BOOK CHAPTER(S):

Arya R, Puppala S, Farook VS, Chittoor G, Jenkinson CP, Blangero J, Hale DE, **Duggirala R**, Almasy L. Mapping of susceptibility genes for obesity, type 2 diabetes, and the metabolic syndrome in human populations. Pp. 181-246. In: Genome Mapping and Genomics in Human and Non-Human Primates, R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.

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15. Kim YJ, Lee J, Kim BJ; **T2D-Genes Consortium**, Park T. A new strategy for enhancing imputation quality of rare variants from next-generation sequencing data via combining SNP and exome chip data. BMC Genomics. 2015 Dec 29;16:1109. doi: 10.1186/s12864-015-2192-y. PubMed PMID: 26715385; PubMed Central PMCID: PMC4696174.

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18. Rubicz R, Yolken R, Drigalenko E, Carless MA, Dyer TD, Kent J Jr, Curran JE, Johnson MP, Cole SA, Fowler SP, Arya R, Puppala S, Almasy L, Moses EK, Kraig E, **Duggirala R**, Blangero J, Leach CT, Göring HH. Genome-wide genetic investigation of serological measures of common infections. Eur J Hum Genet. 2015; Nov; 23(11):1544-8. doi: 10.1038/ejhg.2015.24. Epub 2015 Mar 11. PubMed PMID: 25758998; PubMed Central PMCID: PMC4613484.

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MANUSCRIPT REVIEW:

Annals of Human Genetics

Diabetic Medicine

Translational Research

INTRAMURAL APPOINTMENTS:

Member, Admissions Committee, UTRGV School of Medicine

Member, Institutional Review Board (IRB), University of Texas Rio Grande Valley

EXTRAMURAL APPOINTMENTS:

Adjunct Professor, Division of Diabetes, Department of Medicine, UT Health Science Center at San Antonio

Member, Steering Committee, Type 2 Diabetes (T2D) Genetic Exploration by Next-Generation Sequencing in Ethnic Samples (T2D-GENES) Consortium (NIDDK)

Member, Steering Committee, Accelerating Medicines Partnership (AMP) T2D-GENES Consortium (NIDDK)

Member, AMP T2D GENES Data Working Group

Member, U.S. Consortium Steering Committee, International Consortium for the Study of Tuberculosis (ICST), University of Kansas, Lawrence, KS

Review Editor, Frontiers in Applied Genetic Epidemiology Journal

Reviewer, Institute for Integration of Medicine and Science (IIMS)/Clinical and Translational Science Award (CTSA), Pilot Projects, University of Texas Health Science Center at San Antonio

Member, VA SPLM (MVP beta-test projects) Review Committee (Teleconference Participant)

Member (IRB-3), Institutional Review Board-3, UT Health Science Center at San Antonio (until 09/15)

Alternate Member (IRB-1), Institutional Review Board-1, UT Health Science Center at San Antonio (until 09/15)

Alternate Member (IRB-2), Institutional Review Board-2, UT Health Science Center at San Antonio (until 09/15)

Alternate Member (IRB-E), Institutional Review Board-E, UT Health Science Center at San Antonio (until 09/15)

COLLABORATING INSTITUTIONS:

Arizona State University	Rajasthan University of Health Sciences	University of Maryland School of Medicine
Hospital General de Mexico Dr Eduardo Liceaga	Southwest Research Institute	University of North Carolina
Kuwait University	Sri Ramachandra University	University of Oklahoma HSC
Narayana Medical College & Hospital	Texas Biomedical Research Institute	UTHSC/H School of Public Health
National Jewish Health	Texas Department of State Health Services	University of Texas Health Science Center at San Antonio
National Institute for Diabetes, Digestive and Kidney Disease, NIH	Texas Tech University HSC	University of Veracruz
New Mexico Department of Health	University of California at Los Angeles	Washington U. School of Medicine
Penn State University	University of Kansas	Yale University School of Medicine
Phoenix Epidemiology and Clinical Research Branch, NIDDK, NIH	University of Chihuahua	

GRANT SUPPORT:

					Awarded from 01/01/2015 – 06/30/2016		
Role	Project Title	Grant Period	Funding Agency	Grant #	Direct Costs	Indirect Costs	Total
P.I. (MPI)	Discovery of Functional Variants in Type 2 Diabetes Genes in Mexican Americans	05/01/2015 - 04/30/2019	NIH/ NIDDK	U01 DK085524	669,282	333,224	1,002,506
P.I.	Genetics of Type 2 Diabetes in Indian Populations: US-India Collaboration Project	04/08/2016 - 03/31/2018	NIH/ NIDDK	R21 DK105913	48,681	33,377	82,058
Co-Inv.	Whole Genome Sequencing to Identify Causal Genetic Variants Influencing CVD Risk	04/15/2012 - 03/31/2017	NIH/ NHLBI	R01 HL113323	- - -	- - -	- - -

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EDUCATION:

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1993	M.A.	Molecular Genetics Columbia University, New York, NY

RESEARCH FOCUS:

To what degree do we inherit predisposition to disease, life expectancy, and personal skills, predilections, and behaviors? What are the specific genetic factors involved in shaping these characteristics? To address these questions, Dr. Göring's research group works on two aspects of genetic epidemiology, the development of statistical methods for genetic analysis and the application of those methods to well-designed human datasets.

On the methodological front, Dr. Göring's research group is working on ways to localize and identify rare genomic variants with strong phenotypic impact on complex traits in extended pedigree samples. Specifically, his research group is taking advantage of genotyped rare variants from whole genome sequence data to phase and impute missing genotype data, and, they are developing methods for assessing the phenotypic effect of unique chromosome segments (found only in a single pedigree founder and his/her descendants) on disease and disease-related quantitative risk factors.

On the applied front, Dr. Göring's research group is involved in several studies that integrate genotypic data and other "omics" technology-generated data to unravel trait etiology. One project involves the search for differences in gene expression between schizophrenic individuals and controls. His research group is examining the expression patterns of case and control lymphoblastoid cell lines (from both European ancestry and African American individuals) before and after stimulation with the neurotransmitter dopamine, with the hope that the identified differences reveal information about the etiology of schizophrenia.

Dr. Göring's group is also seeking to identify early metabolomic biomarkers for type 2 diabetes. They are screening prospectively collected blood plasma samples in a large cohort of Mexican American family members using a highly sensitive separation methodology (untargeted two-dimensional gas chromatography approach coupled to time-of-flight mass spectrometry) to identify specific molecules or chemical signatures that predict elevated risk of disease onset. In addition, he is assessing the genetic architecture that regulates the levels of specific individual compounds (or their ratios) in the blood stream.

BOOK CHAPTER(S):

Göring HH. Gene expression studies and complex diseases. Pp. 67-83. In: Genome Mapping and Genomics in Human and Non-Human Primates. R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg, 2015.

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MANUSCRIPT REVIEW:

Genetic Analysis Workshop 19

Journal of the American Medical Association (JAMA)

PLoS Genetics

Scientific Report

EXTRAMURAL APPOINTMENTS:

Member, Special Emphasis Panel/Scientific Review Group ZRG1 PSE-R (90) Cancer, Cardiovascular and Sleep Epidemiology Panel B; San Francisco, CA (9-10 Feb. 2015)

Member, Special Emphasis Panel/Scientific Review Group 2015/10 ZRG1 PSE-U (90) S Cancer, Cardiovascular and Sleep Epidemiology Panel B; Bethesda, MD (18-19 June 2015)

Member, Special Emphasis Panel/Scientific Review Group 2016/01 ZRG1 PSE-U (90) Cancer, Cardiovascular and Sleep Epidemiology Panel B; Washington, DC (19-20 Oct. 2015)

COLLABORATING INSTITUTIONS:

Baylor College of Medicine	Texas Biomedical Research Institute	UT Health Science Center San Antonio
Southwest Research Institute	University of Pennsylvania	Yale University

GRANT SUPPORT:

Role	Project Title	Grant Period	Funding Agency	Grant #	Awarded from 01/01/2015 – 06/30/2016 Direct Costs	Indirect Costs	Total
P.I.	A Genetic Study of Blood Metabolites and Their Relationship to Diabetes Risk	09/01/2013 - 06/30/2017	NIH/ NIDDK	R01 DK099051	623,763	293,889	917,652
P.I.	2/2 - An Integrative Genetic Investigation of Schizophrenia	07/21/2011 - 04/30/2016 (N/C ext. to 04/30/2017)	NIH/ NIMH	R01 MH094116	34,837	27,106	61,943
P.I. (UT-RGV)	Gene Expression in an African American Schizophrenia Dataset	07/12/2012 - 06/30/2016	NIH/ NIMH (Northshore)	R01 MH098059	78,276	35,459	113,735
Co-Inv.	Whole Genome Sequencing to Identify Causal Genetic Variants Influencing CVD Risk	04/15/2012 - 03/31/2017	NIH/ NHLBI	R01 HL113323	- - -	- - -	- - -
Co-Inv.	Discovery of Functional Variants in Type 2 Diabetes Genes in Mexican Americans	05/01/2015 - 04/30/2019	NIH/ NIDDK	U01 DK085524	- - -	- - -	- - -

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EDUCATION:

1993	Ph.D.	Biochemistry University of Otago, Dunedin, New Zealand
1986	M.Sc.	Biochemistry University of Otago, Dunedin, New Zealand

RESEARCH FOCUS:

Dr. Jenkinson's research broadly encompasses biochemistry and molecular biology and currently molecular genetics of complex multifactorial diseases including obesity, type 2 diabetes (T2D), insulin resistance, metabolic syndrome (MS) and related metabolic disorders. The primary focus of his research is the identification of the molecular factors underlying these complex multifactorial diseases. These represent a vast frontier of genetic research and have generally proven refractory to most of our current cutting-edge methods. Despite enormous progress over the past decade in defining DNA variants and genes which are associated with these disorders, there remains a perplexing problem where combined DNA variation across dozens of genes derived from large population samples has failed to account for a significant fraction of phenotypic heritability. To date, examination of rare variants using specialized variant arrays and whole genome sequencing has not resolved this problem. It appears increasingly likely that much of the missing information may lie in regulatory regions contained in non-coding regions of the genome that comprise 98.5% of the genome, and which are widely transcribed.

In a recently published review, Dr. Jenkinson proposed association of genome-wide gene expression or transcriptomics may provide a more useful technique than other genome-wide approaches and should provide a valuable and necessary bridge between the genotype and the phenotype. Dr. Jenkinson's own recent transcriptomic studies analyzed gene expression simultaneously in multiple human tissues from lean and obese Mexican American subjects (skeletal muscle, adipose tissue and peripheral blood) and examined association with multiple deep metabolic phenotypes derived from euglycemic hyperinsulinemic clamps and oral glucose tolerance tests. These studies were funded by his grants "Express" (VA) and "Discover" (NIH). Participants were selected from the family-based VAGES study (VA and NIH). These investigations identified a candidate gene, alcohol dehydrogenase 1B (ADH1B), whose expression is strongly associated with measures of obesity and insulin resistance in human abdominal subcutaneous adipose tissue. This finding has since been replicated in two other large studies in different ancestry groups: (1) The NIDDK Pima Indian study in Arizona; and (2) Europeans from the TwinsUK study. Preliminary investigations in Dr. Jenkinson's lab suggest that this gene, which is differentially expressed in obese and lean individuals, may metabolize endogenous ethanol produced by the gut microbiome, thereby influencing energy regulation in adipose tissue.

A second major focus of Dr. Jenkinson's work is the development of versatile human cell-based systems for testing the causative role and mechanistic action of genetic variants discovered using the other association-based approaches mentioned above. This work is key to translating correlation into causation and currently lags far behind variant discovery. For example, the ADH1B investigations have been extended to human subcutaneous adipose primary cell cultures. This will ultimately provide a valuable pipeline for mechanistic functional analysis of adipose-specific genes and variants obtained from association studies. As noted above, for the vast majority of such variants and genes, strong associations remain to be rigorously evaluated for functional relevance. To further these activities, in 2015 Dr. Jenkinson completed the establishment of a fully operational molecular genetics laboratory at the ERAHC for the study of obesity, T2D and related complex genetic disorders. This lab is operated in close coordination with colleagues, Drs. Duggirala and Mummidi, and intersects with their research projects.

BOOK CHAPTERS:

1. Arya R, Puppala S, Farook VS, Chittoor G, **Jenkinson CP**, Blangero J, Hale DE, Duggirala R, Almasy L. Mapping of susceptibility genes for obesity, type 2 diabetes, and the metabolic syndrome in human populations. Pp. 181-246. In: Genome Mapping and Genomics in Human and Non-Human Primates, R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.

2. T2D-GENES web portal and knowledgebase for results from human genetic association studies of type 2 diabetes and related traits, built as part of the **T2D-GENES consortium** with funding and support from the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK). http://www.type2diabetesgenetics.org/

3. **Jenkinson CP**, Genome Expression Omnibus (GEO). A public functional genomics data repository supporting MIAME-compliant data submissions. http://www.ncbi.nlm.nih.gov/geo/ Accession ID: GSE64567 Transcriptomic identification of genes for obesity and insulin resistance in human adipose tissue in a Mexican American population sample drawn from the Veterans Administration Genetic Epidemiology Study (VAGES)

PUBLICATIONS 01/2015 – 06/2016:

1. **Jenkinson CP**, Göring HH, Arya R, Blangero J, Duggirala R, DeFronzo RA. Transcriptomics in type 2 diabetes: Bridging the gap between genotype and phenotype. Genom Data. 2015 Dec 17;8:25-36. doi: 10.1016/j.gdata.2015.12.001. eCollection 2016 Jun. PubMed PMID: 27114903; PubMed Central PMCID: PMC4832048.

2. Arya R, del Rincon I, Farook VS, Restrepo JF, Winnier DA, Fourcaudot MJ, Battafarano DF, Almeida M, Kumar S, Curran JE, **Jenkinson CP**, Blangero J, Duggirala R, Escalante A. Genetic variants influencing joint damage in Mexican Americans and European Americans with rheumatoid arthritis. Genet Epidemiol. 2015 Dec;39(8):678-88. doi: 10.1002/gepi.21938. Epub 2015 Oct 26. PubMed PMID: 26498133.

3. Farook VS, Reddivari L, Chittoor G, Puppala S, Arya R, Fowler SP, Hunt KJ, Curran JE, Comuzzie AG, Lehman DM, **Jenkinson CP**, Lynch JL, DeFronzo RA, Blangero J, Hale DE, Duggirala R, Vanamala J. Metabolites as novel biomarkers for childhood obesity-related traits in Mexican-American children. Pediatr Obes. 2015 Aug;10(4):320-7. doi: 10.1111/ijpo.270. Epub 2014 Nov 18. PubMed PMID: 25405847; PubMed Central PMCID: PMC4436034.

4. Curran JE, Kumar S, Cromack DT, Hu SL, Coletta DK, Dyer TD, Arya R, Carless M, Tripathy D, Blangero J, Duggirala R, Göring HH, DeFronzo RA, **Jenkinson CP**. Transcriptomic identification of ADH1B as a novel candidate gene for obesity and insulin resistance in human adipose tissue in Mexican Americans from the Veterans Administration Genetic Epidemiology Study (VAGES). (2015) World Biomedical Frontiers, Diabetes and Obesity Section ISSN; published online. ISSN: 2328-0166. http://biomedfrontiers.org/diabetes-obesity-2015-9-11/

5. Winnier DA, Fourcaudot M, Norton L, Abdul-Ghani MA, Hu SL, Farook VS, Coletta DK, Kumar S, Puppala S, Chittoor G, Dyer TD, Arya R, Carless M, Lehman DM, Curran JE, Cromack DT, Tripathy D, Blangero J, Duggirala R, Göring HH, DeFronzo RA, **Jenkinson CP**. Transcriptomic identification of ADH1B as a novel candidate gene for obesity and insulin resistance in human adipose tissue in Mexican Americans from the Veterans Administration Genetic Epidemiology Study (VAGES). PLoS One. 2015 Apr 1;10(4):e0119941. doi:10.1371/journal.pone.0119941. eCollection 2015. PubMed PMID: 25830378; PubMed Central PMCID: PMC4382323.

GRANT REVIEW:

Diabetes UK

UTHSCSA: IIMS/CTSA Pilot Project Scientific Review Panel

EXTRAMURAL APPOINTMENTS:

Ad hoc Member, Steering Committee, Type 2 Diabetes (T2D) Genetic Exploration by Next-Generation Sequencing in Ethnic Samples (T2D-GENES) Consortium (NIDDK)

Ad hoc Member, Steering Committee, Accelerating Medicines Partnership (AMP) T2D-GENES Consortium (NIDDK)

Ad hoc Member, AMP T2D GENES Annotation Data Working Group

Ad Hoc Member of Tuberculosis Consortium (for investigation of TB and TB-T2D interactions in Mexican American cohorts in the Texas/Mexican border region).

COLLABORATING INSTITUTIONS:

Harvard University | Texas Biomedical Research Institute | UT Health Science Center San Antonio

National Institute for Diabetes and Digestive & Kidney Disorders, NIH

GRANT SUPPORT:

Role	Project Title	Grant Period	Funding Agency	Grant #	Awarded from 01/01/2015 – 06/30/2016 Direct Costs	Indirect Costs	Total
Co-Inv.	Discovery of Functional Variants in Type 2 Diabetes Genes in Mexican Americans	05/01/2015 - 04/30/2019	NIH/ NIDDK	U01 DK085524	- - -	- - -	- - -

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EDUCATION:

1984	Ph.D.	Biological Anthropology/Population Genetics The Ohio State University, Columbus, Ohio
1979	M.A.	Anthropology The Ohio State University, Columbus, Ohio

RESEARCH FOCUS:

Dr. Mahaney possesses a broad educational background in genetics, human physiological and anatomical variation, and comparative primate biology, as well as over 30 years of post-graduate research experience in quantitative biology and statistical genetics. His research since the late 1980s has focused on dissection of the genetic architecture of complex traits related to human growth, development, aging and variation in susceptibility, severity, and progression of common diseases – especially cardiovascular disease and osteoporosis – in humans and nonhuman primates. Characterized succinctly, Dr. Mahaney's research focus is on identifying pleiotropic networks of disease risk factors, i.e., multiple traits affected by the same gene or set of genes, and gene co-expression networks, sets of genes exhibiting coordinated expression and effects on diseases or their risk factors.

During 2015, most of Dr. Mahaney's research-related efforts were directed toward analyses of data collected in three major, NIH-funded studies: 1) a study of the effects of an atherogenic diet, high in cholesterol and fat, on patterns of gene expression in endothelial cells and the effects of these genes on atherosclerosis and other cardiovascular disease endpoints in pedigreed baboons; 2) the effects of genes on co-variation between biomarkers of calcification and atherosclerosis risk factors in Mexican American families from San Antonio, Texas; and 3) a study of the effects of genes on patterns of co-variation among measures of bone quality and biomarkers of calcium metabolism and bone formation/turnover in members of the Jirel population of eastern Nepal.

Dr. Mahaney has collaborated with investigators at several institutions in studies of biomedically relevant traits with implications for understanding the genetics of development and the evolution of our species. One study used evidence of pleiotropic effects of genes on tooth size and shape to infer mechanisms of development from which we can infer patterns of evolution in primates. Another study found evidence for genes that influenced patterns of brain folding. Both studies used data from a pedigreed population of baboons.

PUBLICATIONS 01/2015 – 06/2016:

1. Mamtani M, Kulkarni H, Wong G, Weir JM, Barlow CK, Dyer TD, Almasy L, **Mahaney MC**, Comuzzie AG, Glahn DC, Magliano DJ, Zimmet P, Shaw J, Williams-Blangero S, Duggirala R, Blangero J, Meikle PJ, Curran JE. Lipidomic risk score independently and cost-effectively predicts risk of future type 2 diabetes: Results from diverse cohorts. Lipids Health Dis. 2016 Apr 4;15(1):67. doi: 10.1186/s12944-016-0234-3. PubMed PMID: 27044508; PubMed Central PMCID: PMC4820916.

2. Chittoor G, Kent JW Jr, Almeida M, Puppala S, Farook VS, Cole SA, Haack K, Göring HH, MacCluer JW, Curran JE, Carless MA, Johnson MP, Moses EK, Almasy L, **Mahaney MC**, Lehman DM, Duggirala R, Comuzzie AG, Blangero J, Voruganti VS. GWAS and transcriptional analysis prioritize ITPR1 and CNTN4 for a serum uric acid 3p26 QTL in Mexican Americans. BMC Genomics. 2016 Apr 2;17(1):276. doi: 10.1186/s12864-016-2594-5. PubMed PMID: 27039371; PubMed Central PMCID: PMC4818944.

3. Mamtani M, Kulkarni H, Dyer TD, Göring HH, Neary JL, Cole SA, Kent JW, Kumar S, Glahn DC, **Mahaney MC**, Comuzzie AG, Almasy L, Curran JE, Duggirala R, Blangero J, Carless MA. Genome- and epigenome-wide association study of hypertriglyceridemic waist in Mexican American families. Clin Epigenetics. 2016 Jan 20;8:6. doi: 10.1186/s13148-016-0173-x. eCollection 2016. PubMed PMID: 26798409; PubMed Central PMCID: PMC4721061.

4. Kulkarni H, Mamtani M, Peralta J, Almeida M, Dyer TD, Göring HH, Johnson MP, Duggirala R, **Mahaney MC,** Olvera RL, Almasy L, Glahn DC, Williams-Blangero S, Curran JE, Blangero J. Soluble forms of intercellular and vascular cell adhesion molecules independently predict progression to type 2 diabetes in Mexican American families. PLoS One. 2016 Mar 23;11(3):e0151177. doi: 10.1371/journal.pone.0151177. eCollection 2016. PubMed PMID: 27007680; PubMed Central PMCID: PMC4805238.

5. Kulkarni H, Kos MZ, Neary J, Dyer TD, Kent JW Jr, Göring HH, Cole SA, Comuzzie AG, Almasy L, **Mahaney MC**, Curran JE, Blangero J, Carless MA. Novel epigenetic determinants of type 2 diabetes in Mexican-American families. Hum Mol Genet. 2015 Sep 15;24(18):5330-44. doi: 10.1093/hmg/ddv232. Epub 2015 Jun 22. PubMed PMID: 26101197; PubMed Central PMCID: PMC4550817.

6. Blackburn A, Almeida M, Dean A, Curran JE, Johnson MP, Moses EK, Abraham LJ, Carless MA, Dyer TD, Kumar S, Almasy L, **Mahaney MC**, Comuzzie A, Williams-Blangero S, Blangero J, Lehman DM, Göring HH. Effects of copy number variable regions on local gene expression in white blood cells of Mexican Americans. Eur J Hum Genet. 2015 Sep;23(9):1229-35. doi: 10.1038/ejhg.2014.280. Epub 2015 Jan 14. PubMed PMID: 25585699; PubMed Central PMCID: PMC4538210.

7. Williams KD, Blangero J, **Mahaney MC**, Subedi J, Jha B, Williams-Blangero S, Towne B. Axial quantitative ultrasound assessment of pediatric bone quality in eastern Nepal. Osteoporos Int. 2015 Sep;26(9):2319-28. doi: 10.1007/s00198-015-3115-0. Epub 2015 Apr 11. PubMed PMID: 25862355.

8. Atkinson EG, Rogers J, **Mahaney MC**, Cox LA, Cheverud JM. Cortical folding of the primate brain: An interdisciplinary examination of the genetic architecture, modularity, and evolvability of a significant neurological trait in pedigreed baboons (Genus *Papio*). Genetics. 2015 Jun;200(2):651-65. doi: 10.1534/genetics.114.173443. Epub 2015 Apr 14. PubMed PMID: 25873632; PubMed Central PMCID: PMC4492386.

9. Johnson Z, Brent L, Alvarenga JC, Comuzzie AG, Shelledy W, Ramirez S, Cox L, **Mahaney MC**, Huang YY, Mann JJ, Kaplan JR, Rogers J. Genetic influences on response to novel objects and dimensions of personality in *Papio* baboons. Behav Genet. 2015 Mar;45(2):215-27. doi: 10.1007/s10519-014-9702-6. Epub 2015 Jan 21. PubMed PMID: 25604451; PubMed Central PMCID: PMC4349218.

INTRAMURAL APPOINTMENTS:

Member, Institutional Animal Care and Use Committee, University of Texas Rio Grande Valley

Member, Admissions Committee, UTRGV School of Medicine

EXTRAMURAL APPOINTMENTS:

Member, Neurological, Aging, and Musculoskeletal Epidemiology (NAME) Study Section, (term: 2014-18).

Member, TransOmics Precision Medicine Whole Genome Sequencing (NHLBI TOPMed WGS) Consortium, National Heart, Lung, and Blood Institute

Member, Phenotype Harmonization Committee, National Heart, Lung, and Blood Institute

Member, Ethical, Legal, and Social Issues Committee (ELSI), National Heart, Lung, and Blood Institute

COLLABORATING INSTITUTIONS:

Harvard University	Stony Brook University, NY	University of California--Berkeley
South Dakota State University	Texas Biomedical Research Institute	

GRANT SUPPORT:

					Awarded from 01/01/2015 – 06/30/2016		
Role	Project Title	Grant Period	Funding Agency	Grant #	Direct Costs	Indirect Costs	Total
Co-Inv.	Whole Genome Sequencing to Identify Causal Genetic Variants Influencing CVD Risk	04/15/2012 - 03/31/2017	NIH/ NHLBI	R01 HL113323	- - -	- - -	- - -

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EDUCATION:

1975	Ph.D.	Genetics Macquarie University, Sydney, Australia
1970	B.Sc.(Hons)	Genetics La Trobe University, Melbourne, Australia

RESEARCH FOCUS:

Genes and environment both have profound effects on the behavioral and physiological characteristics of individuals. The premise of one of the foci of Dr. VandeBerg's research is that by identifying specific genes and environmental factors that influence physiological characteristics, and understanding the mechanisms by which they exert their individual and collective effects, we can develop new strategies for preventing and treating human diseases. Currently, this research focus is centered on the laboratory opossum model of diet-induced hypercholesterolemia. Some individuals are genetically susceptible and others are genetically resistant to this condition, which causes atherosclerosis as well as non-alcoholic fatty liver disease. We have identified a mutant gene (allele 1 of ABCB4), which when present in two copies (homozygous) in an individual, inhibits cholesterol secretion from the liver into the bile, causing hypercholesterolemia. We are now working toward identifying an alternative pathway for eliminating cholesterol from the body, and variant genes that make some ABCB4 1/1 homozygotes but not others able to evade hypercholesterolemia. A major accomplishment during 2015 was the physical transfer of approximately 1,000 laboratory opossums from San Antonio to University of Texas Rio Grande Valley (Brownsville campus) and the adaptation of the breeding stocks to their new home.

Another focus of Dr. VandeBerg's research is Chagas disease, a parasitic disease that is common in some parts of Latin America, and is naturally present in Texas and other southern states. There is no vaccine and no highly effective drug for Chagas disease which is manifested as heart disease that often leads to death. One current project involves testing a novel vaccine in monkeys for efficacy in preventing infection after experimental infection by inoculation of the parasites. Another project involves testing three drug regimens for treating monkeys that became naturally infected while living in outdoor housing in San Antonio and identifying biomarkers that indicate whether an individual has been cleared of all parasites by the drug treatment. This project is expected to lead to improvements in assessing the efficacy of novel drugs in clinical trials with people.

PUBLICATIONS 01/2015 – 06/2016:

1. Nair HB, Baker R, Owston MA, Escalona R, Dick EJ, **VandeBerg JL**, Nickisch KJ. An efficient model of human endometriosis by induced unopposed estrogenicity in baboons. Oncotarget. 2016 Mar 8;7(10):10857-69. doi: 10.18632/oncotarget.7516. PubMed PMID: 26908459.

2. Sathler-Avelar R, Vitelli-Avelar DM, Mattoso-Barbosa AM, Perdigão-de-Oliveira M, Costa RP, Elói-Santos SM, Gomes Mde S, Amaral LR, Teixeira-Carvalho A, Martins-Filho OA, Dick EJ Jr, Hubbard GB, VandeBerg JF, **VandeBerg JL**. Phenotypic features of circulating leukocytes from non-human primates naturally infected with *Trypanosoma cruzi* resemble the major immunological findings observed in human chagas disease. PLoS Negl Trop Dis. 2016 Jan 25;10(1):e0004302. doi: 10.1371/journal.pntd.0004302. eCollection 2016 Jan. PubMed PMID: 26808481; PubMed Central PMCID: PMC4726540.

3. Cura CI, Duffy T, Lucero RH, Bisio M, Péneau J, Jimenez-Coello M, Calabuig E, Gimenez MJ, Valencia Ayala E, Kjos SA, Santalla J, Mahaney SM, Cayo NM, Nagel C, Barcán L, Málaga Machaca ES, Acosta Viana KY, Brutus L, Ocampo SB, Aznar C, Cuba Cuba CA, Gürtler RE, Ramsey JM, Ribeiro I, **VandeBerg JL**, Yadon ZE, Osuna A, Schijman AG. Multiplex real-time PCR assay using TaqMan probes for the identification of *Trypanosoma cruzi* DTUs in biological and clinical samples. PLoS Negl Trop Dis. 2015 May 19;9(5):e0003765. doi: 10.1371/journal.pntd.0003765. eCollection 2015 May. PubMed PMID: 25993316; PubMed Central PMCID: PMC4437652.

4. Porto A, Sebastião H, Pavan SE, **VandeBerg JL**, Marroig G, Cheverud JM. Rate of evolutionary change in cranial morphology of the marsupial genus *Monodelphis* is constrained by the availability of additive genetic variation. J Evol Biol. 2015 Apr;28(4):973-85. doi: 10.1111/jeb.12628. Epub 2015 Apr 17. PubMed PMID: 25818173; PubMed Central PMCID: PMC4405477.

5. Shi Q, **VandeBerg JL**. Experimental approaches to derive CD34+ progenitors from human and nonhuman primate embryonic stem cells. Am J Stem Cells. 2015 Mar 15;4(1):32-7. eCollection 2015. Review. PubMed PMID: 25973329; PubMed Central PMCID: PMC4396158.

INTRAMURAL APPOINTMENTS:

Member, Faculty Senate, University of Texas Rio Grande Valley

Member, Promotion, Tenure, and Appointment Committee, UTRGV School of Medicine

Member, Graduate Curriculum Committee, University of Texas Rio Grande Valley

EXTRAMURAL APPOINTMENTS:

Chair, External Advisory Board, Caribbean Primate Research Center

Chief Scientific Director, Better Life Better Research Foundation

Affiliate Scientist, Southwest National Primate Research Center, Texas Biomedical Research Institute

COLLABORATING INSTITUTIONS:

Drugs for Neglected Diseases, Geneva Switzerland	Loyola University	University of Georgia
Federal University of Mato Grosso do Sul, Campo Grande, Brazil	Texas Biomedical Research Institute	
Fundação Oswaldo Cruz - FIOCRUZ, Belo Horizonte, Brazil	University of Buenos Aires, Buenos Aires	

GRANT SUPPORT:

					Awarded from 01/01/2015 – 06/30/2016		
Role	Project Title	Grant Period	Funding Agency	Grant #	Direct Costs	Indirect Costs	Total
P.I.	Evaluation of PCR and Other Biomarkers for Assessing Parsitological Cure in Chagas Disease	2012 - 2016	Wellcome Trust (DNDi)	Contract	- - -	- - -	- - -
P.I.	Novel Vaccine for Chagas Disease: Efficacy Testing in Baboons	2014 - 2016	Robert J. Kleberg, Jr. and Helen C. Kleberg Found.	Grant	- - -	- - -	- - -

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Email: sarah.williams-blangero@utrgv.edu

EDUCATION:

1987	Ph.D.	Biological Anthropology Case Western Reserve University, Cleveland, Ohio
1984	M.A.	Biological Anthropology Case Western Reserve University, Cleveland, Ohio

RESEARCH FOCUS:

Dr. Williams-Blangero's research program focuses on the genetic epidemiology of common complex diseases in minority populations. She has overseen the development of a number of major human population cohorts for long-term genetic research. In Nepal, she has worked with a single pedigree including more than 2,600 members of a small ethnic group, the Jirel population. This population has been the center of collaborative research projects on parasitic disease, childhood growth and development, osteoporosis and bone-related traits, dental traits, and psychiatric disease. The genetic determinants of ocular health and disease, including diabetic retinopathy, are currently under study in the Jirel population. Dr. Williams-Blangero also works with a study population of approximately 2000 residents from a rural area of Brazil that has very high rates of Trypansoma cruzi infection. *T. cruzi* is the parasitic cause of Chagas disease, which is associated with progressive cardiomyopathy. Chagas disease is found throughout Latin America and is a border health issue relevant to the Rio Grande Valley. As the Director of the South Texas Diabetes and Obesity Institute, Dr. Williams-Blangero is now expanding her research on the genetics determinants of diabetes and obesity and related disorders in Mexican Americans.

BOOK(S):

Duggirala R, Almasy L, **Williams-Blangero S**, Paul S, Kole C (eds). Genome Mapping and Genomics in Human and Non-Human Primates, Springer-Verlag Berlin Heidelberg. 2015.

BOOK CHAPTER(S):

Williams-Blangero S, Blangero J. The utility of genomics for studying primate biology. Pp. 1-6. In: Genome Mapping and Genomics in Human and Non-Human Primates. R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.

PUBLICATIONS 01/2015 – 06/2016:

1. Mamtani M, Kulkarni H, Wong G, Weir JM, Barlow CK, Dyer TD, Almasy L, Mahaney MC, Comuzzie AG, Glahn DC, Magliano DJ, Zimmet P, Shaw J, **Williams-Blangero S**, Duggirala R, Blangero J, Meikle PJ, Curran JE. Lipidomic risk score independently and cost-effectively predicts risk of future type 2 diabetes: Results from diverse cohorts. Lipids Health Dis. 2016 Apr 4;15(1):67. doi: 10.1186/s12944-016-0234-3. PubMed PMID: 27044508; PubMed Central PMCID: PMC4820916.

2. Williams KD, Subedi J, Jha B, Blangero J, **Williams-Blangero S**, Towne B. Quantitative physical activity assessment of children and adolescents in a rural population from Eastern Nepal. Am J Hum Biol. 2016 Jan-Feb;28(1):129-37. doi: 10.1002/ajhb.22762. Epub 2015 Jul 16. PubMed PMID: 26179444.

3. Kulkarni H, Mamtani M, Peralta J, Almeida M, Dyer TD, Göring HH, Johnson MP, Duggirala R, Mahaney MC, Olvera RL, Almasy L, Glahn DC, **Williams-Blangero S**, Curran JE, Blangero J. Soluble forms of intercellular and

vascular cell adhesion molecules independently predict progression to type 2 diabetes in Mexican American families. PLoS One. 2016 Mar 23;11(3):e0151177. doi: 10.1371/journal.pone.0151177. eCollection 2016. PubMed PMID: 27007680; PubMed Central PMCID: PMC4805238.

4. Peters MJ, Joehanes R, Pilling LC, Schurmann C, Conneely KN, Powell J, Reinmaa E, Sutphin GL, Zhernakova A, Schramm K, Wilson YA, Kobes S, Tukiainen T; NABEC/UKBEC Consortium, Ramos YF, Göring HH, Fornage M, Liu Y, Gharib SA, Stranger BE, De Jager PL, Aviv A, Levy D, Murabito JM, Munson PJ, Huan T, Hofman A, Uitterlinden AG, Rivadeneira F, van Rooij J, Stolk L, Broer L, Verbiest MM, Jhamai M, Arp P, Metspalu A, Tserel L, Milani L, Samani NJ, Peterson P, Kasela S, Codd V, Peters A, Ward-Caviness CK, Herder C, Waldenberger M, Roden M, Singmann P, Zeilinger S, Illig T, Homuth G, Grabe HJ, Völzke H, Steil L, Kocher T, Murray A, Melzer D, Yaghootkar H, Bandinelli S, Moses EK, Kent JW, Curran JE, Johnson MP, **Williams-Blangero S**, Westra HJ, McRae AF, Smith JA, Kardia SL, Hovatta I, Perola M, Ripatti S, Salomaa V, Henders AK, Martin NG, Smith AK, Mehta D, Binder EB, Nylocks KM, Kennedy EM, Klengel T, Ding J, Suchy-Dicey AM, Enquobahrie DA, Brody J, Rotter JI, Chen YD, Houwing-Duistermaat J, Kloppenburg M, Slagboom PE, Helmer Q, den Hollander W, Bean S, Raj T, Bakhshi N, Wang QP, Oyston LJ, Psaty BM, Tracy RP, Montgomery GW, Turner ST, Blangero J, Meulenbelt I, Ressler KJ, Yang J, Franke L, Kettunen J, Visscher PM, Neely GG, Korstanje R, Hanson RL, Prokisch H, Ferrucci L, Esko T, Teumer A, van Meurs JB, Johnson AD. The transcriptional landscape of age in human peripheral blood. Nat Commun. 2015 Oct 22;6:8570. doi: 10.1038/ncomms9570. PubMed PMID: 26490707; PubMed Central PMCID: PMC4639797.

5. Blackburn A, Almeida M, Dean A, Curran JE, Johnson MP, Moses EK, Abraham LJ, Carless MA, Dyer TD, Kumar S, Almasy L, Mahaney MC, Comuzzie A, **Williams-Blangero S**, Blangero J, Lehman DM, Göring HH. Effects of copy number variable regions on local gene expression in white blood cells of Mexican Americans. Eur J Hum Genet. 2015 Sep;23(9):1229-35. doi: 10.1038/ejhg.2014.280. Epub 2015 Jan 14. PubMed PMID: 25585699; PubMed Central PMCID: PMC4538210.

6. Williams KD, Blangero J, Mahaney MC, Subedi J, Jha B, **Williams-Blangero S**, Towne B. Axial quantitative ultrasound assessment of pediatric bone quality in eastern Nepal. Osteoporos Int. 2015 Sep;26(9):2319-28. doi: 10.1007/s00198-015-3115-0. Epub 2015 Apr 11. PubMed PMID: 25862355.

INTRAMURAL APPOINTMENTS:

Member, Promotion, Tenure, and Appointment Committee, UTRGV School of Medicine

Member, Research Deans Council, University of Texas Rio Grande Valley

Discipline Coordinator for Research, UTRGV School of Medicine

Member, Search Committee for Population Health Faculty, UTRGV School of Medicine

Chair, Search Committee for Chair of Department of Internal Medicine, UTRGV School of Medicine

EXTRAMURAL APPOINTMENTS:

Adjunct Professor, School of Public Health, University of Texas Health Science Center at Houston

Member, Editorial Board, International Journal of Human Genetics

Member, Medical Integration Working Group, Unidos Contra Diabetes

Member, UT Women's Senior Leadership Network

Member, University of Texas Diabetes Blue Ribbon Panel

COLLABORATING INSTITUTIONS:

Tilganga Institute of Ophthalmology, Nepal	Fundação Oswaldo Cruz - FIOCRUZ, Belo Horizonte, Brazil	Wright State University, Dayton, OH
Caribbean Primate Research Center, Puerto Rico	Miami University, Oxford, OH	
Chitwan Medical College, Nepal	UTHealth School of Public Health	

GRANT SUPPORT:

Role	Project Title	Grant Period	Funding Agency	Grant #	Awarded from 01/01/2015 – 06/30/2016 Direct Costs	Indirect Costs	Total
P.I. (MPI)	Genetic Epidemiology of Ocular Health and Disease	06/01/2014 - 05/31/2019	NIH/NEI	R01 EY024384	641,778	358,406	1,000,184
P.I. (UT-RGV)	Establishment and Maintenance of a Closed CPRC SPF Colony	05/01/2011 - 04/30/2016	NIH/OD (Univ. of Puerto Rico)	U42 OD011128	13,290	6,146	19,436
P.I. (UT-RGV)	Caribbean Primate Research Center	01/15/2016 - 11/30/2020	NIH/OD (Univ. of Puerto Rico)	P40 OD012217	23,462	10,629	34,091

MATTHEW JOHNSON, PH.D.

Associate Professor
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University of Texas Rio Grande Valley
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2700 E. Jackson Street
Brownsville, TX. 78520
Phone: 956-882-7504
Email: matthew.johnson@utrgv.edu

EDUCATION:

2005	Ph.D.	Molecular Genetics Griffith University, Australia
1999	Grad. Dipl.	Biotechnology Queensland University of Technology, Australia

RESEARCH FOCUS:

Dr. Johnson's research program centers on the identification of genetic mechanisms influencing normal and diseased ocular biology. Blindness and poor vision are major public health concerns across the globe. It is estimated that approximately 10% of the world's population suffer from some degree of visual impairment, and the costs associated with treating or managing all eye-related disorders is estimated to run in the billions of dollars. Poor ocular health significantly impacts independence and quality of life. The major causes of visual impairment – refractive error, cataract, age-related macular degeneration (AMD), retinopathy/macular edema, and glaucoma – progressively worsen with age if left undetected. Additionally, diabetes significantly influences the development of retinopathy/macular edema, and is known to influence cataract and glaucoma risk. The negative impact that visual impairment places on society is likely to increase given an aging global population in addition to an increase in diabetes incidence.

Dr. Johnson is involved in a project that aims to identify novel AMD genetic susceptibility loci and refine knowledge of existing loci in a cohort of Caucasian families. This project is in collaboration with Dr. Michael Klein at the Casey Eye Institute in Portland, Oregon, and involves linkage and association analyses of data from 1,200 individuals belonging to 150 families using a dense set of genome-wide genetic markers (3.5 million SNPs). The research team processed genome sequence data from approximately 100 individuals, all with AMD, from 20 families to objectively prioritize rare genetic variation with a major effect on AMD susceptibility.

Dr. Johnson led a pilot study to document the prevalence of and characterize the genetic mechanisms influencing age- and diabetes-related ocular disorders in a cohort of Mexican American families. The San Antonio Family Eye Study is conducted in collaboration with Dr. Kent L. Anderson, of the Department of Ophthalmology, School of Medicine, University of Texas Health Science Center at San Antonio. The families for this study are being drawn from the longitudinal San Antonio Family Studies. To date, the 63 individuals who have been recruited and have undergone a comprehensive eye exam, detailed medical history questionnaire, and provided a blood sample for biochemical assessment of lipid levels, and metabolic, inflammatory and immune-related biomarkers. All recruited individuals have existing whole-genome sequence data that is providing the framework for preliminary genetic analyses against the newly ascertained ocular traits. A notable feature observed early in the study is that the families have a high incidence of glaucoma suspect (~65%).

Dr. Johnson leads, with Sarah Williams-Blangero, an NIH/NEI funded project that is characterizing the genetic mechanisms of normal ocular biology, refractive error, and cataract, in the Jirel ethnic group of Nepal, the Jiri Eye Study. The project is conducted in collaboration with Dr. Suman S. Thapa, at the Tilganga Institute of Ophthalmology in Kathmandu, Nepal, and involves the recruitment of 2,000 individuals who belong to one complex pedigree. All individuals are undergoing a comprehensive eye exam, completing a medical history questionnaire, and providing a blood sample for DNA sequencing (exome/genome), RNA sequencing, and biochemical assessment of lipid levels.

PUBLICATIONS 01/2015 – 06/2016:

1. Kaartokallio T, Lokki AI, Peterson H, Kivinen K, Hiltunen L, Salmela E, Lappalainen T, Maanselkä P, Heino S, Knuutila S, Sayed A, Poston L, Brennecke SP, **Johnson MP**, Morgan L, Moses EK, Kere J, Laivuori H. Preeclampsia does not share common risk alleles in 9p21 with coronary artery disease and type 2 diabetes. Ann Med. 2016 Apr 25:1-7. [Epub ahead of print] PubMed PMID: 27111527.

2. Chittoor G, Kent JW Jr, Almeida M, Puppala S, Farook VS, Cole SA, Haack K, Göring HH, MacCluer JW, Curran JE, Carless MA, **Johnson MP**, Moses EK, Almasy L, Mahaney MC, Lehman DM, Duggirala R, Comuzzie AG, Blangero J, Voruganti VS. GWAS and transcriptional analysis prioritize ITPR1 and CNTN4 for a serum uric acid 3p26 QTL in Mexican Americans. BMC Genomics. 2016 Apr 2;17(1):276. doi: 10.1186/s12864-016-2594-5. PubMed PMID: 27039371; PubMed Central PMCID: PMC4818944.

3. Fritsche LG, Igl W, Bailey JN, Grassmann F, Sengupta S, Bragg-Gresham JL, Burdon KP, Hebbring SJ, Wen C, Gorski M, Kim IK, Cho D, Zack D, Souied E, Scholl HP, Bala E, Lee KE, Hunter DJ, Sardell RJ, Mitchell P, Merriam JE, Cipriani V, Hoffman JD, Schick T, Lechanteur YT, Guymer RH, **Johnson MP**, Jiang Y, Stanton CM, Buitendijk GH, Zhan X, Kwong AM, Boleda A, Brooks M, Gieser L, Ratnapriya R, Branham KE, Foerster JR, Heckenlively JR, Othman MI, Vote BJ, Liang HH, Souzeau E, McAllister IL, Isaacs T, Hall J, Lake S, Mackey DA, Constable IJ, Craig JE, Kitchner TE, Yang Z, Su Z, Luo H, Chen D, Ouyang H, Flagg K, Lin D, Mao G, Ferreyra H, Stark K, von Strachwitz CN, Wolf A, Brandl C, Rudolph G, Olden M, Morrison MA, Morgan DJ, Schu M, Ahn J, Silvestri G, Tsironi EE, Park KH, Farrer LA, Orlin A, Brucker A, Li M, Curcio CA, Mohand-Saïd S, Sahel JA, Audo I, Benchaboune M, Cree AJ, Rennie CA, Goverdhan SV, Grunin M, Hagbi-Levi S, Campochiaro P, Katsanis N, Holz FG, Blond F, Blanché H, Deleuze JF, Igo RP Jr, Truitt B, Peachey NS, Meuer SM, Myers CE, Moore EL, Klein R, Hauser MA, Postel EA, Courtenay MD, Schwartz SG, Kovach JL, Scott WK, Liew G, Tan AG, Gopinath B, Merriam JC, Smith RT, Khan JC, Shahid H, Moore AT, McGrath JA, Laux R, Brantley MA Jr, Agarwal A, Ersoy L, Caramoy A, Langmann T, Saksens NT, de Jong EK, Hoyng CB, Cain MS, Richardson AJ, Martin TM, Blangero J, Weeks DE, Dhillon B, van Duijn CM, Doheny KF, Romm J, Klaver CC, Hayward C, Gorin MB, Klein ML, Baird PN, den Hollander AI, Fauser S, Yates JR, Allikmets R, Wang JJ, Schaumberg DA, Klein BE, Hagstrom SA, Chowers I, Lotery AJ, Léveillard T, Zhang K, Brilliant MH, Hewitt AW, Swaroop A, Chew EY, Pericak-Vance MA, DeAngelis M, Stambolian D, Haines JL, Iyengar SK, Weber BH, Abecasis GR, Heid IM. A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nat Genet. 2016 Feb;48(2):134-43. doi: 10.1038/ng.3448. Epub 2015 Dec 21. PubMed PMID: 26691988; PubMed Central PMCID: PMC4745342.

4. Zhou Y, Zhu G, Charlesworth JC, Simpson S Jr, Rubicz R, Göring HH, Patsopoulos NA, Laverty C, Wu F, Henders A, Ellis JJ, van der Mei I, Montgomery GW, Blangero J, Curran JE, **Johnson MP**, Martin NG, Nyholt DR, Taylor BV; ANZgene consortium. Genetic loci for Epstein-Barr virus nuclear antigen-1 are associated with risk of multiple sclerosis. Mult Scler. 2016 Jan 27. pii: 1352458515626598. [Epub ahead of print] PubMed PMID: 26819262.

5. Kulkarni H, Mamtani M, Peralta J, Almeida M, Dyer TD, Göring HH, **Johnson MP**, Duggirala R, Mahaney MC, Olvera RL, Almasy L, Glahn DC, Williams-Blangero S, Curran JE, Blangero J. Soluble forms of intercellular and vascular cell adhesion molecules independently predict progression to type 2 diabetes in Mexican American families. PLoS One. 2016 Mar 23;11(3):e0151177. doi: 10.1371/journal.pone.0151177. eCollection 2016. PubMed PMID: 27007680; PubMed Central PMCID: PMC4805238.

6. Rubicz R, Yolken R, Drigalenko E, Carless MA, Dyer TD, Kent J Jr, Curran JE, **Johnson MP**, Cole SA, Fowler SP, Arya R, Puppala S, Almasy L, Moses EK, Kraig E, Duggirala R, Blangero J, Leach CT, Göring HH. Genome-wide genetic investigation of serological measures of common infections. Eur J Hum Genet. 2015 Nov;23(11):1544-8. doi: 10.1038/ejhg.2015.24. Epub 2015 Mar 11. PubMed PMID: 25758998; PubMed Central PMCID: PMC4613484.

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MANUSCRIPT REVIEW:

PLoS One

INTRAMURAL APPOINTMENTS:

Member, Institutional Biosafety Committee, University of Texas Rio Grande Valley

Member, Translational Research Seminar Series Committee, UT Houston School of Public Health/University of Texas Rio Grande Valley

EXTRAMURAL APPOINTMENTS:

Undergraduate Mentor, UT Health Science Center at Houston School of Public Health summer research program.

Associate Professor (Adjoint), Department of Ophthalmology, School of Medicine, University of Texas Health Science Center at San Antonio

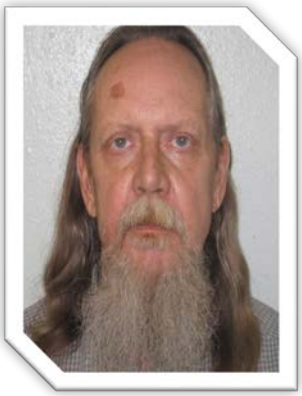
Member, International Age-related Macular Degeneration Genomics Consortium

COLLABORATING INSTITUTIONS:

Oregon Health & Sciences University	Tilganga Institute of Ophthalmology, Kathmandu	Wright State University, Dayton, OH
Miami University, Oxford, OH	University of Texas Health Science Center at San Antonio	

GRANT SUPPORT:

					Awarded from 01/01/2015 – 06/30/2016		
Role	Project Title	Grant Period	Funding Agency	Grant #	Direct Costs	Indirect Costs	Total
P.I. (MPI)	Genetic Epidemiology of Ocular Health and Disease	06/01/2014 - 05/31/2019	NIH/NEI	R01 EY024384	641,778	358,406	1,000,184
P.I.	Defining the Genetic Architecture of Macular Degeneration in Mexican American Families	03/17/2015 - 08/31/2015	Max and Minnie Tomerlin Voelcker Fund	Grant (Young Investigator Award)	171,117	17,112	188,229
P.I. (UT-RGV)	A Joint Linkage/Association Strategy to Interrogate AMD Genetic Susceptibility	09/01/2015 - 08/31/2016	NIH/NEI (OHSU)	R01 EY021532	49,677	22,504	72,181



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EDUCATION:

2002	Ph.D.	Computer Science University of Texas at San Antonio
1986	M.S.	Computer Science University of Texas at San Antonio

RESEARCH FOCUS:

The focus of Dr. Dyer's research is on computational methods for disease-related gene detection and gene identification. STDOI investigators developed SOLAR, a genetic analysis package that has over 5000 users. Dr. Dyer's work focuses in part on refinement and further development of algorithms that will enhance the utility of SOLAR for genetic research. Another area of long-term research interest is the use of computer simulation approaches for testing statistical genetic methods.

PUBLICATIONS 01/2015 – 06/2016:

1. Hodgson K, Almasy L, Knowles EE, Kent JW, Curran JE, **Dyer TD**, Göring HH, Olvera RL, Fox PT, Pearlson GD, Krystal JH, Duggirala R, Blangero J, Glahn DC. Genome-wide significant loci for addiction and anxiety. Eur Psychiatry. 2016 Jun 15;36:47-54. doi: 10.1016/j.eurpsy.2016.03.004. [Epub ahead of print] PubMed PMID: 27318301.

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3. Franke B, Stein JL, Ripke S, Anttila V, Hibar DP, van Hulzen KJ, Arias-Vasquez A, Smoller JW, Nichols TE, Neale MC, McIntosh AM, Lee P, McMahon FJ, Meyer-Lindenberg A, Mattheisen M, Andreassen OA, Gruber O, Sachdev PS, Roiz-Santiañez R, Saykin AJ, Ehrlich S, Mather KA, Turner JA, Schwarz E, Thalamuthu A, Yao Y, Ho YY, Martin NG, Wright MJ; Schizophrenia Working Group of the Psychiatric Genomics Consortium; Psychosis Endophenotypes International Consortium; Wellcome Trust Case Control Consortium 2; **Enigma Consortium**, O'Donovan MC, Thompson PM, Neale BM, Medland SE, Sullivan PF. Genetic influences on schizophrenia and subcortical brain volumes: Large-scale proof of concept. Nat Neurosci. 2016 Mar;19(3):420-31. doi: 10.1038/nn.4228. Epub 2016 Feb 1. PubMed PMID: 26854805; PubMed Central PMCID: PMC4852730.

4. Knowles EE, Kent JW Jr, McKay DR, Sprooten E, Mathias SR, Curran JE, Carless MA, Almeida MA, Göring HH, **Dyer TD**, Olvera RL, Fox PT, Duggirala R, Almasy L, Blangero J, Glahn DC. Genome-wide linkage on chromosome 10q26 for a dimensional scale of major depression. J Affect Disord. 2016 Feb;191:123-31. doi: 10.1016/j.jad.2015.11.012. Epub 2015 Nov 17. PubMed PMID: 26655122; PubMed Central PMCID: PMC4715913.

5. Clark MM, Blangero J, **Dyer TD**, Sobel EM, Sinsheimer JS. The Quantitative-MFG Test: A linear mixed effect model to detect maternal-offspring gene interactions. Ann Hum Genet. 2016 Jan;80(1):63-80. doi: 10.1111/ahg.12137. Epub 2015 Nov 15. PubMed PMID: 26567478; PubMed Central PMCID: PMC4715557.

6. Mamtani M, Kulkarni H, **Dyer TD**, Göring HH, Neary JL, Cole SA, Kent JW, Kumar S, Glahn DC, Mahaney MC, Comuzzie AG, Almasy L, Curran JE, Duggirala R, Blangero J, Carless MA. Genome- and epigenome-wide association

study of hypertriglyceridemic waist in Mexican American families. Clin Epigenetics. 2016 Jan 20;8:6. doi: 10.1186/s13148-016-0173-x. eCollection 2016. PubMed PMID: 26798409; PubMed Central PMCID: PMC4721061.

7. Mathias SR, Knowles EE, Kent JW Jr, McKay DR, Curran JE, Almeida MA, **Dyer TD**, Göring HH, Olvera RL, Duggirala R, Fox PT, Almasy L, Blangero J, Glahn DC. Recurrent major depression and right hippocampal volume: A bivariate linkage and association study. Hum Brain Mapp. 2016 Jan;37(1):191-202. doi: 10.1002/hbm.23025. Epub 2015 Oct 20. PubMed PMID: 26485182.

8. Kulkarni H, Mamtani M, Peralta J, Almeida M, **Dyer TD**, Göring HH, Johnson MP, Duggirala R, Mahaney MC, Olvera RL, Almasy L, Glahn DC, Williams-Blangero S, Curran JE, Blangero J. Soluble forms of intercellular and vascular cell adhesion molecules independently predict progression to type 2 diabetes in Mexican American families. PLoS One. 2016 Mar 23;11(3):e0151177. doi: 10.1371/journal.pone.0151177. eCollection 2016. PubMed PMID: 27007680; PubMed Central PMCID: PMC4805238.

9. Sprooten E, Gupta CN, Knowles EE, McKay DR, Mathias SR, Curran JE, Kent JW Jr, Carless MA, Almeida MA, **Dyer TD**, Göring HH, Olvera RL, Kochunov P, Fox PT, Duggirala R, Almasy L, Calhoun VD, Blangero J, Turner JA, Glahn DC. Genome-wide significant linkage of schizophrenia-related neuroanatomical trait to 12q24. Am J Med Genet B Neuropsychiatr Genet. 2015 Dec;168(8):678-86. doi: 10.1002/ajmg.b.32360. Epub 2015 Oct 5. PubMed PMID: 26440917; PubMed Central PMCID: PMC4639444.

10. Kim YJ, Lee J, Kim BJ; **T2D-Genes Consortium**, Park T. A new strategy for enhancing imputation quality of rare variants from next-generation sequencing data via combining SNP and exome chip data. BMC Genomics. 2015 Dec 29;16:1109. doi: 10.1186/s12864-015-2192-y. PubMed PMID: 26715385; PubMed Central PMCID: PMC4696174.

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12. Kulkarni H, Kos MZ, Neary J, **Dyer TD**, Kent JW Jr, Göring HH, Cole SA, Comuzzie AG, Almasy L, Mahaney MC, Curran JE, Blangero J, Carless MA. Novel epigenetic determinants of type 2 diabetes in Mexican-American families. Hum Mol Genet. 2015 Sep 15;24(18):5330-44. doi: 10.1093/hmg/ddv232. Epub 2015 Jun 22. PubMed PMID: 26101197; PubMed Central PMCID: PMC4550817.

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14. Huang YS, Ramensky V, Service SK, Jasinska AJ, Jung Y, Choi OW, Cantor RM, Juretic N, Wasserscheid J, Kaplan JR, Jorgensen MJ, **Dyer TD**, Dewar K, Blangero J, Wilson RK, Warren W, Weinstock GM, Freimer NB. Sequencing strategies and characterization of 721 vervet monkey genomes for future genetic analyses of medically relevant traits. BMC Biol. 2015 Jun 20;13:41. doi: 10.1186/s12915-015-0152-2. PubMed PMID: 26092298; PubMed Central PMCID: PMC4494155.

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16. Williams AL, Genovese G, **Dyer T**, Altemose N, Truax K, Jun G, Patterson N, Myers SR, Curran JE, Duggirala R, Blangero J, Reich D, Przeworski M; T2D-GENES Consortium. Non-crossover gene conversions show strong GC bias and unexpected clustering in humans. Elife. 2015 Mar 25;4. doi: 10.7554/eLife.04637. PubMed PMID: 25806687; PubMed Central PMCID: PMC4404656.

17. Knowles EE, McKay DR, Kent JW Jr, Sprooten E, Carless MA, Curran JE, Almeida MA, **Dyer TD**, Göring HH, Olvera RL, Duggirala R, Fox PT, Almasy L, Blangero J, Glahn DC. Pleiotropic locus for emotion recognition and amygdala volume identified using univariate and bivariate linkage. Am J Psychiatry. 2015 Feb 1;172(2):190-9. doi: 10.1176/appi.ajp.2014.14030311. Epub 2014 Oct 31. PubMed PMID: 25322361; PubMed Central PMCID: PMC4314438.

18. Glahn DC, Williams JT, McKay DR, Knowles EE, Sprooten E, Mathias SR, Curran JE, Kent JW Jr, Carless MA, Göring HH, **Dyer TD**, Woolsey MD, Winkler AM, Olvera RL, Kochunov P, Fox PT, Duggirala R, Almasy L, Blangero J. Discovering schizophrenia endophenotypes in randomly ascertained pedigrees. Biol Psychiatry. 2015 Jan 1;77(1):75-83. doi: 10.1016/j.biopsych.2014.06.027. Epub 2014 Jul 21. PubMed PMID: 25168609; PubMed Central PMCID: PMC4261014.

19. Dager AD, McKay DR, Kent JW Jr, Curran JE, Knowles E, Sprooten E, Göring HH, **Dyer TD**, Pearlson GD, Olvera RL, Fox PT, Lovallo WR, Duggirala R, Almasy L, Blangero J, Glahn DC. Shared genetic factors influence amygdala volumes and risk for alcoholism. Neuropsychopharmacology. 2015 Jan;40(2):412-20. doi: 10.1038/npp.2014.187. Epub 2014 Jul 31. PubMed PMID: 25079289; PubMed Central PMCID: PMC4443955.

20. Curran JE, Kumar S, Cromack DT, Hu SL, Coletta DK, **Dyer TD**, Arya R, Carless M, Tripathy D, Blangero J, Duggirala R, Göring HH, DeFronzo RA, Jenkinson CP. Transcriptomic identification of ADH1B as a novel candidate gene for obesity and insulin resistance in human adipose tissue in Mexican Americans from the Veterans Administration Genetic Epidemiology Study (VAGES). (2015) World Biomedical Frontiers, Diabetes and Obesity Section ISSN; published online. ISSN: 2328-0166. http://biomedfrontiers.org/diabetes-obesity-2015-9-11/

21. Winnier DA, Fourcaudot M, Norton L, Abdul-Ghani MA, Hu SL, Farook VS, Coletta DK, Kumar S, Puppala S, Chittoor G, **Dyer TD**, Arya R, Carless M, Lehman DM, Curran JE, Cromack DT, Tripathy D, Blangero J, Duggirala R, Göring HH, DeFronzo RA, Jenkinson CP. Transcriptomic identification of ADH1B as a novel candidate gene for obesity and insulin resistance in human adipose tissue in Mexican Americans from the Veterans Administration Genetic Epidemiology Study (VAGES). PLoS One. 2015 Apr 1;10(4):e0119941. doi: 10.1371/journal.pone.0119941. eCollection 2015. PubMed PMID: 25830378; PubMed Central PMCID: PMC4382323.

MANUSCRIPT REVIEW:

Circulation: Cardiovascular Genetics

EXTRAMURAL APPOINTMENTS:

Member, T2D-GENES (Type 2 Diabetes Genetic Exploration by Next-generation sequencing in multi-Ethnic Samples) consortium

Member, ENIGMA (Enhancing NeuroImaging Genetics through Meta-Analysis) Consortium

Member, CHARGE (Cohorts for Heart and Aging Research in Genetic Epidemiology) Consortium

COLLABORATING INSTITUTIONS:

University of California Los Angeles University of Maryland University of Tasmania

GRANT SUPPORT:

					Awarded from 01/01/2015 – 06/30/2016		
Role	Project Title	Grant Period	Funding Agency	Grant #	Direct Costs	Indirect Costs	Total
Co-Inv.	Whole Genome Sequencing to Identify Causal Genetic Variants Influencing CVD	04/15/2012 - 03/31/2017	NIH/ NHLBI	R01 HL113323	- - -	- - -	- - -
Co-Inv.	Genetic Analysis of Common Diseases: An Evaluation	04/01/1983 - 07/31/2017	NIH/ NIGMS	R01 GM031575	- - -	- - -	- - -

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EDUCATION:

1986	M.B.B.S.	Medicine and Surgery Nagpur University, Nagpur, India
1989	M.D. (PSM)	Preventive and Social Medicine Nagpur University, Nagpur, India

RESEARCH FOCUS:

Dr. Kulkarni's research focus is on robust biostatistical analysis of high-volume, big data emerging from genomic, epigenomic, transcriptomic and lipidomic studies of complex diseases. His primary research activities involve identification of genetic variants and other -omic factors associated with hypertension, dyslipidemia, type 2 diabetes and metabolic syndrome in Mexican Americans. Currently, he also is working on the importance of type 2 diabetes for critical care in hospital settings. He is collaborating with investigators from Washington University, St Louis (who have outcome data on over 800,000 patients) and with researchers at Flinders University, Adelaide, Australia (who have outcome data for over a million patients) to determine the effects of type 2 diabetes on outcomes for patients in intensive care. Dr. Kulkarni also is working to establish a study of the genetics of type 2 diabetes in Sindhi families in India.

PUBLICATIONS 01/2015 – 06/2016:

1. Mamtani M, **Kulkarni H**, Wong G, Weir JM, Barlow CK, Dyer TD, Almasy L, Mahaney MC, Comuzzie AG, Glahn DC, Magliano DJ, Zimmet P, Shaw J, Williams-Blangero S, Duggirala R, Blangero J, Meikle PJ, Curran JE. Lipidomic risk score independently and cost-effectively predicts risk of future type 2 diabetes: Results from diverse cohorts. Lipids Health Dis. 2016 Apr 4;15(1):67. doi: 10.1186/s12944-016-0234-3. PubMed PMID: 27044508; PubMed Central PMCID: PMC4820916.

2. Mamtani M, **Kulkarni H**, Dyer TD, Göring HH, Neary JL, Cole SA, Kent JW, Kumar S, Glahn DC, Mahaney MC, Comuzzie AG, Almasy L, Curran JE, Duggirala R, Blangero J, Carless MA. Genome- and epigenome-wide association study of hypertriglyceridemic waist in Mexican American families. Clin Epigenetics. 2016 Jan 20;8:6. doi: 10.1186/s13148-016-0173-x. eCollection 2016. PubMed PMID: 26798409; PubMed Central PMCID: PMC4721061.

3. Mamtani M, Curran JE, Blangero J, **Kulkarni H**. Association of urinary phthalates with self-reported eye affliction/retinopathy in individuals with diabetes: National Health and Nutrition Examination Survey, 2001-2010. J Diabetes Res. 2016;2016:7269896. doi: 10.1155/2016/7269896. Epub 2015 Dec 20. PubMed PMID: 26798652; PubMed Central PMCID: PMC4698956.

4. **Kulkarni H**, Mamtani M, Peralta J, Almeida M, Dyer TD, Göring HH, Johnson MP, Duggirala R, Mahaney MC, Olvera RL, Almasy L, Glahn DC, Williams-Blangero S, Curran JE, Blangero J. Soluble forms of intercellular and vascular cell adhesion molecules independently predict progression to type 2 diabetes in Mexican American families. PLoS One. 2016 Mar 23;11(3):e0151177. doi: 10.1371/journal.pone.0151177. eCollection 2016. PubMed PMID: 27007680; PubMed Central PMCID: PMC4805238.

5. **Kulkarni H**, Kos MZ, Neary J, Dyer TD, Kent JW Jr, Göring HH, Cole SA, Comuzzie AG, Almasy L, Mahaney MC, Curran JE, Blangero J, Carless MA. Novel epigenetic determinants of type 2 diabetes in Mexican-American families. Hum Mol Genet. 2015 Sep 15;24(18):5330-44. doi: 10.1093/hmg/ddv232. Epub 2015 Jun 22. PubMed PMID: 26101197; PubMed Central PMCID: PMC4550817.

MANUSCRIPT REVIEW:

Diabetologia
Journal of Interventional Cardiology
Journal of Investigative Medicine
PLoS ONE

EXTRAMURAL APPOINTMENTS:

Trustee and President, Lata Medical Research Foundation, Nagpur, India

COLLABORATING INSTITUTIONS:

Lata Medical Research Foundation, Nagpur, India
Texas Biomedical Research Institute

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Associate Professor/Research
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EDUCATION:

1992	Ph.D.	Medical Anthropology Case Western Reserve University, Cleveland, OH
1987	M.A.	Medical Anthropology Case Western Reserve University, Cleveland, OH
1972	Diploma	Nursing Idabelle Firestone School of Nursing, Akron, OH

RESEARCH FOCUS:

Dr. Laston's research efforts are focused on oversight of large-scale genetic studies conducted with human populations. With specialized expertise in medical anthropology and nursing, Dr. Laston has directed the fieldwork for studies of a number of minority populations including Alaskan Eskimos and Zuni Indians. Her current research is focused on ocular health and disease in the Jirel population of eastern Nepal. A member of the UTRGV institutional review board, Dr. Laston has strong interests in the design of human subjects research projects and cross-cultural research.

PUBLICATIONS 01/2015 – 06/2016:

1. Vazquez-Vidal I, Chittoor G, **Laston S**, Puppala S, Kayani Z, Mody K, Comuzzie AG, Cole SA, Voruganti VS. Assessment of cardiovascular disease risk factors in a genetically homogenous population of Parsi Zoroastrians in the United States: A pilot study. Am J Hum Biol. 2016 May;28(3):440-3. doi: 10.1002/ajhb.22834. Epub 2016 Jan 28. PubMed PMID: 26819065.

2. Gribble MO, Voruganti VS, Cole SA, Haack K, Balakrishnan P, **Laston SL**, Tellez-Plaza M, Francesconi KA, Goessler W, Umans JG, Thomas DC, Gilliland F, North KE, Franceschini N, Navas-Acien A. Linkage analysis of urine arsenic species patterns in the Strong Heart Family Study. Toxicol Sci. 2015 Nov;148(1):89-100. doi: 10.1093/toxsci/kfv164. Epub 2015 Jul 23. PubMed PMID: 26209557; PubMed Central PMCID: PMC4731407.

3. Voruganti VS, **Laston S**, Haack K, Mehta NR, Cole SA, Butte NF, Comuzzie AG. Serum uric acid concentrations and SLC2A9 genetic variation in Hispanic children: The Viva La Familia Study. Am J Clin Nutr. 2015 Apr;101(4):725-32. doi: 10.3945/ajcn.114.095364. Epub 2015 Jan 28. PubMed PMID: 25833971; PubMed Central PMCID: PMC4381775.

4. Tsai CW, North KE, Tin A, Haack K, Franceschini N, Saroja Voruganti V, **Laston S**, Zhang Y, Best LG, MacCluer JW, Beaty TH, Navas-Acien A, Kao WH, Howard BV. Both rare and common variants in PCSK9 influence plasma low-density lipoprotein cholesterol level in American Indians. J Clin Endocrinol Metab. 2015 Feb;100(2):E345-9. doi: 10.1210/jc.2014-3340. Epub 2014 Nov 20. PubMed PMID: 25412415; PubMed Central PMCID: PMC4318886/

5. **Laston SL**, Voruganti VS, Haack K, Shah VO, Bobelu A, Bobelu J, Ghahate D, Harford AM, Paine SS, Tentori F, Cole SA, MacCluer JW, Comuzzie AG, Zager PG. Genetics of kidney disease and related cardiometabolic phenotypes in Zuni Indians: The Zuni Kidney Project. Front Genet. 2015 Jan 30;6:6. doi: 10.3389/fgene.2015.00006. eCollection 2015. PubMed PMID: 25688259; PubMed Central PMCID: PMC4311707.

6. Ebbesson SO, Voruganti VS, Higgins PB, Fabsitz RR, Ebbesson LO, **Laston S**, Harris WS, Kennish J, Umans BD, Wang H, Devereux RB, Okin PM, Weissman NJ, MacCluer JW, Umans JG, Howard BV. Fatty acids linked to cardiovascular mortality are associated with risk factors. Int J Circumpolar Health. 2015 Aug 12;74:28055. doi: 10.3402/ijch.v74.28055. eCollection 2015. PubMed PMID: 26274054; PubMed Central PMCID: PMC4536775.

COLLABORATING INSTITUTIONS:

Baylor College of Medicine	Rollins School of Public Health, Emory University	University of North Carolina at Chapel Hill
Johns Hopkins Bloomberg School of Public Health	Texas Biomedical Research Institute	Wright State University, Dayton, OH
Miami University, Oxford, OH	Tilganga Institute of Ophthalmology	

GRANT SUPPORT:

					Awarded from 01/01/2015 – 06/30/2016		
Role	Project Title	Grant Period	Funding Agency	Grant #	Direct Costs	Indirect Costs	Total
Co-Inv.	Genetic Epidemiology of Ocular Health and Disease	06/01/2014 - 05/31/2019	NIH/NEI	R01 EY024384	- - -	- - -	- - -

SRINIVAS MUMMIDI, PH.D.

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EDUCATION:

1996	Ph.D.	Immunobiology Iowa State University, Ames, OH
1990	M.S.	Veterinary Immunology Indian Veterinary Research Institute

RESEARCH FOCUS:

Dr. Mummidi's long-term research focus is on understanding the functional mechanisms of single nucleotide polymorphisms modulate gene expression in the context of disease pathogenesis. Dr. Mummidi has conducted extensive research on the regulation of genes involved in inflammation to define gene structure, identify cis-regulatory elements and regions, and determine how polymorphisms disrupt DNA-protein interactions leading to changes in gene expression with implications for disease pathogens.

Dr. Mummidi's ongoing research examines the impact of 3'UTR polymorphisms on the stability and translatability of transcripts encoded by clinically relevant genes involved in inflammatory/immune response. These studies will use state-of-the-art technologies such as genome engineering and induced pluripotent stem cells.

PUBLICATIONS 01/2015 – 06/2016:

1. Gornalusse GG, **Mummidi S**, Gaitan AA, Jimenez F, Ramsuran V, Picton A, Rogers K, Manoharan MS, Avadhanam N, Murthy KK, Martinez H, Molano Murillo A, Chykarenko ZA, Hutt R, Daskalakis D, Shostakovich-Koretskaya L, Abdool Karim S, Martin JN, Deeks SG, Hecht F, Sinclair E, Clark RA, Okulicz J, Valentine FT, Martinson N, Tiemessen CT, Ndung'u T, Hunt PW, He W, Ahuja SK. Epigenetic mechanisms, T-cell activation, and CCR5 genetics interact to regulate T-cell expression of CCR5, the major HIV-1 coreceptor. Proc Natl Acad Sci U S A. 2015 Aug 25;112(34):E4762-71. doi: 10.1073/pnas.1423228112. Epub 2015 Aug 11. PubMed PMID: 26307764; PubMed Central PMCID: PMC4553789.
2. Somanna NK, Yariswamy M, Garagliano JM, Siebenlist U, **Mummidi S**, Valente AJ, Chandrasekar B. Aldosterone-induced cardiomyocyte growth, and fibroblast migration and proliferation are mediated by TRAF3IP2. Cell Signal. 2015 Oct;27(10):1928-38. doi: 10.1016/j.cellsig.2015.07.001. Epub 2015 Jul 4. PubMed PMID: 26148936.

EXTRAMURAL APPOINTMENTS:

Academic Editor, PLoS ONE

Member, Editorial Board, World Journal of Medical Genetics

COLLABORATING INSTITUTIONS:

University of Missouri School of Medicine	University of Texas Health Science Center at San Antonio

GRANT SUPPORT:

Role	Project Title	Grant Period	Funding Agency	Grant #	Awarded from 01/01/2015 – 06/30/2016 Direct Costs	Indirect Costs	Total
P.I.	Impact of disease associated noncoding polymorphisms on post-transcriptional gene regulation	04/01/2016 - 03/31/2020	NIH/ NIAID	R01 AI119131	223,933	112,767	336,700

JUAN CARLOS LOPEZ-ALVARENGA, M.D., DSC.

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EDUCATION:

2003	D.Sc.	Diabetes & Molecular Biology National Autonomous University of Mexico, Mexico City, Mexico
1997	M.Sc.	Diabetes National Autonomous University of Mexico, Mexico City, Mexico
1989	M.D.	Medicine Universidad Evangelica de El Salvador, San Salvador, El Salvador

RESEARCH FOCUS:

Dr. Lopez-Alvarenga's research focus is on complex diseases such as diabetes mellitus, dyslipidemia, and insulin resistance. Dr. Lopez-Alvarenga has significant experience in whole genome scanning approaches. He conducted genetic studies of Omani and Alaskan populations and participated in several studies of metabolic disease involving non-human primates. While conducting research in Mexico, he performed insulin clamps and muscle biopsies in adults and children to measure the effect of treatment to insulin resistance. Dr. Lopez-Alvarenga's latest research focuses on tuberculosis associated with type 2 diabetes mellitus and genetics of susceptibility to gallbladder stones.

GRANT REVIEW:

Consejo Nacional de Ciencia y Tecnología (CONACYT)

EXTRAMURAL APPOINTMENTS:

Member, Microbiota Consortium, Universidad Nacional Autónoma de México (UNAM)

Statistical Advisor, Clinical and Translational Investigation Editorial Board

COLLABORATING INSTITUTIONS:

Hospital General de Mexico, Mexico City	Universidad México Americana del Norte A.C., Ciudad Reynosa, Mexico (UMAN)	Universidad Nacional Autónoma de México (UNAM)

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EDUCATION:

1999	Ph.D.	Biological Anthropology (Population Genetics) University of Kansas, Lawrence, KS, USA
1990	M.Ed.	Education (Research Methodology & Educational Statistics) Annamalia University, Annamalai Nagar, India
1982	M.A.	Anthropology Andhra University, Visakhapatnam, India

RESEARCH FOCUS:

Obesity, type 2 diabetes (T2D), and metabolic syndrome (MS) are complex diseases causing considerable morbidity and mortality worldwide. These diseases and related traits have multifactorial inheritance patterns, and are influenced by multiple genes, environmental factors, and their interactions. There have been continued efforts to localize and identify genetic variants that contribute to susceptibility to these diseases, although knowledge of actual causal variants influencing these traits is extremely limited. Dr. Arya's research focuses on genetic epidemiological studies of common complex diseases including obesity/abdominal obesity, T2D, metabolic syndrome, rheumatoid arthritis, chronic kidney disease, gallbladder disease, and cardiovascular disease risk factors in Mexican Americans and Europeans. His areas of expertise include the study of genetic-environmental (i.e., dietary intake and physical activity) factors and their interactions, and the identification of disease susceptibility genes using genome-wide linkage and association analyses, candidate gene studies, and interventions studies.

BOOK CHAPTER(S):

Arya R, Puppala S, Farook VS, Chittoor G, Jenkinson CP, Blangero J, Hale DE, Duggirala R, Almasy L. Mapping of susceptibility genes for obesity, type 2 diabetes, and the metabolic syndrome in human populations. Pp. 181-246. In: Genome Mapping and Genomics in Human and Non-Human Primates, R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.

PUBLICATIONS 01/2015 – 06/2016:

1. Jenkinson CP, Göring HH, **Arya R**, Blangero J, Duggirala R, DeFronzo RA. Transcriptomics in type 2 diabetes: Bridging the gap between genotype and phenotype. Genom Data. 2015 Dec 17;8:25-36. doi: 10.1016/j.gdata.2015.12.001. eCollection 2016 Jun. PubMed PMID: 27114903; PubMed Central PMCID: PMC4832048.

2. **Arya R**, Del Rincon I, Farook VS, Restrepo JF, Winnier DA, Fourcaudot MJ, Battafarano DF, Almeida M, Kumar S, Curran JE, Jenkinson CP, Blangero J, Duggirala R, Escalante A. Genetic variants influencing joint damage in Mexican Americans and European Americans with rheumatoid arthritis. Genet Epidemiol. 2015 Dec;39(8):678-88. doi: 10.1002/gepi.21938. Epub 2015 Oct 26. PubMed PMID: 26498133.

3. Rubicz R, Yolken R, Drigalenko E, Carless MA, Dyer TD, Kent J Jr, Curran JE, Johnson MP, Cole SA, Fowler SP, **Arya R**, Puppala S, Almasy L, Moses EK, Kraig E, Duggirala R, Blangero J, Leach CT, Göring HH. Genome-wide genetic investigation of serological measures of common infections. Eur J Hum Genet. 2015 Nov;23(11):1544-8. doi: 10.1038/ejhg.2015.24. Epub 2015 Mar 11. PubMed PMID: 25758998; PubMed Central PMCID: PMC4613484.

4. Farook VS, Reddivari L, Chittoor G, Puppala S, **Arya R**, Fowler SP, Hunt KJ, Curran JE, Comuzzie AG, Lehman DM, Jenkinson CP, Lynch JL, DeFronzo RA, Blangero J, Hale DE, Duggirala R, Vanamala J. Metabolites as novel biomarkers for childhood obesity-related traits in Mexican-American children. Pediatr Obes. 2015 Aug;10(4):320-7. doi: 10.1111/ijpo.270. Epub 2014 Nov 18. PubMed PMID: 25405847; PubMed Central PMCID: PMC4436034.

5. Winnier DA, Fourcaudot M, Norton L, Abdul-Ghani MA, Hu SL, Farook VS, Coletta DK, Kumar S, Puppala S, Chittoor G, Dyer TD, **Arya R**, Carless M, Lehman DM, Curran JE, Cromack DT, Tripathy D, Blangero J, Duggirala R, Göring HH, DeFronzo RA, Jenkinson CP. Transcriptomic identification of ADH1B as a novel candidate gene for obesity and insulin resistance in human adipose tissue in Mexican Americans from the Veterans Administration Genetic Epidemiology Study (VAGES). PLoS One. 2015 Apr 1;10(4):e0119941. doi: 10.1371/journal.pone.0119941. eCollection 2015. PubMed PMID: 25830378; PubMed Central PMCID: PMC4382323.

6. Curran JE, Kumar S, Cromack DT, Hu SL, Coletta DK, Dyer TD, **Arya R**, Carless M, Tripathy D, Blangero J, Duggirala R, Göring HH, DeFronzo RA, Jenkinson CP. Transcriptomic identification of ADH1B as a novel candidate gene for obesity and insulin resistance in human adipose tissue in Mexican Americans from the Veterans Administration Genetic Epidemiology Study (VAGES). (2015) World Biomedical Frontiers, Diabetes and Obesity Section ISSN; published online. ISSN: 2328-0166. http://biomedfrontiers.org/diabetes-obesity-2015-9-11/

7. T2D-GENES web portal and knowledgebase for results from human genetic association studies of type 2 diabetes and related traits, built as part of the **T2D-GENES Consortium** with funding and support from the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK). http://www.type2diabetesgenetics.org/.

8. Horikoshi M, Pasquali L, Wiltshire S, Huyghe JR, Mahajan A, Asimit JL, Ferreira T, Locke AE, Robertson NR, Wang X, Sim X, Fujita H, Hara K, Young R, Zhang W, Choi S, Chen H, Kaur I, Takeuchi F, Fontanillas P, Thuillier D, Yengo L, Below JE, Tam CH, Wu Y, Abecasis G, Altshuler D, Bell GI, Blangero J, Burtt NP, Duggirala R, Florez JC, Hanis CL, Seielstad M, Atzmon G, Chan JC, Ma RC, Froguel P, Wilson JG, Bharadwaj D, Dupuis J, Meigs JB, Cho YS, Park T, Kooner JS, Chambers JC, Saleheen D, Kadowaki T, Tai ES, Mohlke KL, Cox NJ, Ferrer J, Zeggini E, Kato N, Teo YY, Boehnke M, McCarthy MI, Morris AP; **T2D-GENES Consortium**, et al. Transancestral fine-mapping of four type 2 diabetes susceptibility loci highlights potential causal regulatory Mechanisms (**Arya R**, collaborator). Hum Mol Genet. 2016 Feb 23. pii: ddw048. [Epub ahead of print].

EXTRAMURAL APPOINTMENTS:

Alternate Member, Institutional Review Board (IRB), IRB-1, UTHSCSA

Member, Editorial Board, Frontiers of Nutrition and Research

External Reviewer, Pennsylvania Department of Health Translational Genomics Peer Review Board, Pennsylvania Department of Health.

Member, Amp-T2D Consortium Phenotype Harmonization Working Group

COLLABORATING INSTITUTIONS:

Baylor College of Medicine	Pennsylvania State University	Universidad Veracruzana Xalapa, Veracruz, Mexico
Harvard University	University of Texas Health Science Center at San Antonio	Wright State University

GRANT SUPPORT:

					Awarded from 01/01/2015 – 06/30/2016		
Role	Project Title	Grant Period	Funding Agency	Grant #	Direct Costs	Indirect Costs	Total
Co-Inv.	Discovery of Functional Variants in Type 2 Diabetes Genes in Mexican Americans	05/01/2015 - 04/30/2016	NIH/ NIDDK	U01 DK085524	- - -	- - -	- - -
Co-Inv.	Genetics of Type 2 Diabetes in Indian Populations: US-India Collaboration Project	04/01/2016 - 03/31/2018	NIH/ NIDDK	R21 DK105913	- - -	- - -	- - -

VINCENT P. DIEGO, PH.D.

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EDUCATION:

2005	Ph.D.	Anthropology, Human Statistical Genetics State University of New York at Binghamton
2001	M.A.	Anthropology, Human Population Biology State University of New York at Binghamton

RESEARCH FOCUS:

Dr. Diego's research has been in two general areas: 1) aging in humans and 2) gene-environment interaction (GEI) analysis and theoretical development. Dr. Diego performs research on biological aging in participants of the San Antonio Family Heart Study and is currently developing approaches to study the role of entropy computed over DNA sequences and gene networks in aging and senescence. Dr. Diego also developed novel methods for the analysis of GEI in relation to multivariate environments. Using these methods to analyze data from a large study of risk for obesity in children, Dr. Diego found significant GEI in relation to dietary multivariate environments. Dr. Diego continues to work on the theoretical development of a novel approach to modeling GEI for gene networks.

BOOK CHAPTER(S):

Diego VP, Kent JW, Blangero J, Familial studies: Genetic inferences. In: International Encyclopedia of the Social & Behavioral Sciences (Second Edition), edited by JD Wright. Elsevier, Oxford, 2015, Pages 715-724, ISBN 9780080970875, http://dx.doi.org/10.1016/B978-0-08-097086-8.82029-2.
(http://www.sciencedirect.com/science/article/pii/B9780080970868820292)

PUBLICATIONS 01/2015 – 06/2016:

Diego VP, de Chaves RN, Blangero J, de Souza MC, Santos D, Gomes TN, dos Santos FK, Garganta R, Katzmarzyk PT, Maia JA. Sex-specific genetic effects in physical activity: Results from a quantitative genetic analysis. BMC Med Genet. 2015 Aug 1;16:58. doi: 10.1186/s12881-015-0207-9. PubMed PMID: 26231751; PubMed Central PMCID: PMC4557754.

COLLABORATING INSTITUTIONS:

University of Porto, Portugal

MARK Z. KOS, PH.D.

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EDUCATION:

2008	Ph.D.	Biological Anthropology University of Kansas
2006	M.A.	Anthropology University of Kansas

RESEARCH FOCUS:

Dr. Kos' research interests are in the fields of statistical genetics and genetics of complex diseases, with a focus on psychiatric disorders. In collaboration with Dr. Laura Almasy, his research centers on two family-based studies: 1) the Multiplex-Multigenerational Genetic Investigation of Schizophrenia (MGI); and 2) the Collaborative Study on the Genetics of Alcoholism (COGA). Examining whole exome sequence data from European-American families obtained from MGI, two genes involved in AMPA receptor trafficking in the post-synapse, SYNPO and WWC1, were found to be significantly associated with neurocognitive impairment and schizophrenia risk. Analyses of the MGI exome data set revealed other schizophrenia risk loci, including ones belonging to lipid metabolism pathways, raising interesting questions regarding the role of compromised brain metabolism in psychiatric disorders.

Dr. Kos' research also examines the genetic architecture of electrical activity in the brain as it relates to alcohol dependence (AD). Using genome-wide association (GWA) data available from COGA, polygenic scores based on aggregated genotypes were found to be significant predictors of AD in independent samples, with preliminary results showing a significant enrichment of genes involved in the metabolism of arachidonic acid, a polyunsaturated fatty acid that is abundant in the brain and is believed to modulate various neuronal activities.

BOOK CHAPTER(S):

Almasy L, **Kos MZ**, Blangero J. Linkage mapping: Localizing the genes that shape human variation. Pp. 33-52. In: Genome Mapping and Genomics in Human and Non-Human Primates. R Duggirala, L Almasy, S Williams-Blangero, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.

PUBLICATIONS 01/2015 – 06/2016:

1. **Kos MZ**, Carless MA, Peralta J, Blackburn A, Almeida M, Roalf D, Pogue-Geile MF, Prasad K, Gur RC, Nimgaonkar V, Curran JE, Duggirala R, Glahn DC, Blangero J, Gur RE, Almasy L. Exome sequence data from multigenerational families implicate AMPA receptor trafficking in neurocognitive impairment and schizophrenia risk. Schizophr Bull. 2016 Mar;42(2):288-300. doi: 10.1093/schbul/sbv135. Epub 2015 Sep 24. PubMed PMID: 26405221; PubMed Central PMCID: PMC4753604.
2. Kulkarni H, **Kos MZ**, Neary J, Dyer TD, Kent JW Jr, Göring HH, Cole SA, Comuzzie AG, Almasy L, Mahaney MC, Curran JE, Blangero J, Carless MA. Novel epigenetic determinants of type 2 diabetes in Mexican-American families. Hum Mol Genet. 2015 Sep 15;24(18):5330-44. doi: 10.1093/hmg/ddv232. Epub 2015 Jun 22. PubMed PMID: 26101197; PubMed Central PMCID: PMC4550817.

COLLABORATING INSTITUTIONS:

Indiana University School of Medicine	Texas Biomedical Research Institute	Washington University School of Medicine
Rutgers University	University of Pennsylvania	Yale University School of Medicine
State University of New York	University of Pittsburgh	

GRANT SUPPORT:

Role	Project Title	Grant Period	Funding Agency	Grant #	Awarded from 01/01/2015 – 06/30/2016 Direct Costs	Indirect Costs	Total
Res. Sci.	A Neurobehavioral Family Study of Schizophrenia	04/01/2000 - 12/31/2018	NIH/ NIMH	R01 MH061622	- - -	- - -	- - -
Res. Sci.	Collaborative Study on the Genetics of Alcoholism	12/31/2014 - 08/31/2016	NIH /NIAAA [Res. Found., State Univ. of N.Y. SUNY)]	U10 AA008401	- - -	- - -	- - -

SATISH KUMAR. PH.D.

Assistant Professor/Research
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EDUCATION:

2003	Ph.D.	Anthropology University of Delhi, New Delhi, India
1996	M.Sc.	Anthropology (Specialization: Physical/Biological Anthropology) University of Delhi, New Delhi, India
1994	B.Sc.	Botany (Honours), Zoology and Chemistry University of Delhi, New Delhi, India

RESEARCH FOCUS:

Dr. Kumar's research focuses on the identification and characterization of genes involved in human common complex diseases. Specifically, he uses induced pluripotent stem cell (iPSC) based disease models for finding genes influencing human common complex diseases, particularly where assessment of genetic influence is difficult due to environmental factors and medical interventions.

Dr. Kumar utilizes a rich supply of lymphoblastoid cell lines (LCLs) established using the peripheral blood mononuclear cells (PBMCs) collected from more than 1400 Mexican American participants of our San Antonio Family Heart Study (SAFHS). Whole genome sequence data and extensive phenotypic data for common complex human diseases are available for most of these SAFHS participants. The large, well characterized LCL resource provides a unique opportunity to generate iPSCs from any of these individuals.

Dr. Kumar optimized an efficient LCL to iPSC reprogramming method, achieving 100% reprogramming success and high reprogramming efficiency (~50-200 colonies/million nucleofected cells). Using this optimized reprogramming protocol he has generated iPSC lines for a variety of ongoing studies at STDOI. In 2015, Dr. Kumar and his research colleagues standardized the iPSC validation protocols and performed validation assays on the generated iPSC lines. They also performed whole genome miRNA and mRNA analysis of six LCLs and their reprogrammed iPSCs to better understand the LCL to iPSC reprogramming process.

BOOK CHAPTER(S):

Kumar S, Kingsley C and DiStefano. The human genome project: where are we now and where are we going. Pp. 7-31. In Genome Mapping and Genomics in Human and Non-Human Primates. R Duggirala, L Almasy, S Williams-Blangero, S Paul, C. Kole (eds). Springer-Verlag Berlin Heidelberg, 2015.

PUBLICATIONS 01/2015 – 06/2016:

1. Nicholson AM, Finch NA, Almeida M, Perkerson RB, van Blitterswijk M, Wojtas A, Cenik B, Rotondo S, Inskeep V, Almasy L, Dyer T, Peralta J, Jun G, Wood AR, Frayling TM, Fuchsberger C, Fowler S, Teslovich TM, Manning AK, **Kumar S**, Curran J, Lehman D, Abecasis G, Duggirala R, Pottier C, Zahir HA, Crook JE, Karydas A, Mitic L, Sun Y, Dickson DW, Bu G, Herz J, Yu G, Miller BL, Ferguson S, Petersen RC, Graff-Radford N, Blangero J, Rademakers R. Prosaposin is a regulator of progranulin levels and oligomerization. Nat Commun. 2016 Jun 30;7:11992. doi: 10.1038/ncomms11992. PubMed PMID: 27356620.

2. **Kumar S**, Curran JE, Glahn DC, Blangero J. Utility of lymphoblastoid cell lines for induced pluripotent stem cell generation. Stem Cells Int. 2016;2016:2349261. doi: 10.1155/2016/2349261. Epub 2016 Jun 7. PubMed PMID: 27375745; PubMed Central PMCID: PMC4914736.

3. Traurig M, Hanson RL, Marinelarena A, Kobes S, Piaggi P, Cole S, Curran JE, Blangero J, Göring H, **Kumar S**, Nelson RG, Howard BV, Knowler WC, Baier LJ, Bogardus C. Analysis of SLC16A11 variants in 12,811 American Indians: Genotype-obesity interaction for Type 2 diabetes and an association with RNASEK expression. Diabetes. 2016 Feb;65(2):510-9. doi: 10.2337/db15-0571. Epub 2015 Oct 20. PubMed PMID: 26487785; PubMed Central PMCID: PMC4747458.

4. Mamtani M, Kulkarni H, Dyer TD, Göring HH, Neary JL, Cole SA, Kent JW, **Kumar S**, Glahn DC, Mahaney MC, Comuzzie AG, Almasy L, Curran JE, Duggirala R, Blangero J, Carless MA. Genome- and epigenome-wide association study of hypertriglyceridemic waist in Mexican American families. Clin Epigenetics. 2016 Jan 20;8:6. doi: 10.1186/s13148-016-0173-x. eCollection 2016. PubMed PMID: 26798409; PubMed Central PMCID: PMC4721061.

5. Arya R, Del Rincon I, Farook VS, Restrepo JF, Winnier DA, Fourcaudot MJ, Battafarano DF, Almeida M, **Kumar S**, Curran JE, Jenkinson CP, Blangero J, Duggirala R, Escalante A. Genetic variants influencing joint damage in Mexican Americans and European Americans with rheumatoid arthritis. Genet Epidemiol. 2015 Dec;39(8):678-88. doi: 10.1002/gepi.21938. Epub 2015 Oct 26. PubMed PMID: 26498133.

6. Blackburn A, Almeida M, Dean A, Curran JE, Johnson MP, Moses EK, Abraham LJ, Carless MA, Dyer TD, **Kumar S**, Almasy L, Mahaney MC, Comuzzie A, Williams-Blangero S, Blangero J, Lehman DM, Göring HH. Effects of copy number variable regions on local gene expression in white blood cells of Mexican Americans. Eur J Hum Genet. 2015 Sep;23(9):1229-35. doi: 10.1038/ejhg.2014.280. Epub 2015 Jan 14. PubMed PMID: 25585699; PubMed Central PMCID: PMC4538210.

7. Curran JE, **Kumar S**, Cromack DT, Hu SL, Coletta DK, Dyer TD, Arya R, Carless M, Tripathy D, Blangero J, Duggirala R, Göring HH, DeFronzo RA, Jenkinson CP. Transcriptomic identification of ADH1B as a novel candidate gene for obesity and insulin resistance in human adipose tissue in Mexican Americans from the Veterans Administration Genetic Epidemiology Study (VAGES). (2015) World Biomedical Frontiers, Diabetes and Obesity Section ISSN; published online. ISSN: 2328-0166. http://biomedfrontiers.org/diabetes-obesity-2015-9-11/

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RESEARCH FOCUS:

Dr. Mamtani's research interests are focused on the genetic and environmental determinants of type 2 diabetes. In particular, she has studied the value of waist circumference as a predictor of risk for type 2 diabetes based on studies in Asian Indians and Mexican Americans. She has approached this phenotype using case-control studies, family studies, genomic and epigenomic association studies. In addition, Dr. Mamtani is developing research projects in the area of gestational diabetes and is helping to establish a study of diabetes in Sindhi families in India.

PUBLICATIONS 01/2015 – 06/2016:

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EXTRAMURAL APPOINTMENTS:

Reviewer, Canadian Journal of Diabetes

Trustee, Lata Medical Research Foundation, Nagpur, India

COLLABORATING INSTITUTIONS:

Lata Medical Research Foundation, India Texas Biomedical Research Institute

VI. MASTER PUBLICATION LIST FOR STDOI INVESTIGATORS (01/2015 – 06/2016)

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3. **Curran JE**, Bellis C, **Almasy L**, **Blangero J**. Genomic studies of human populations: Resequencing approaches to the identification of human quantitative loci. Pp. 289-299. In: Genome Mapping and Genomics in Human and Non-Human Primates, **R Duggirala**, **L Almasy**, **S Williams-Blangero**, S Paul, C Kole (eds), Springer-Verlag Berlin Heidelberg. 2015.

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ONLINE DATABASES:

1. **Jenkinson CP**, Genome Expression Omnibus (GEO). A public functional genomics data repository supporting MIAME-compliant data submissions. http://www.ncbi.nlm.nih.gov/geo/ Accession ID: GSE64567 Transcriptomic identification of genes for obesity and insulin resistance in human adipose tissue in a Mexican American population sample drawn from the Veterans Administration Genetic Epidemiology Study (VAGES).

2. T2D-GENES web portal and knowledgebase for results from human genetic association studies of type 2 diabetes and related traits, built as part of the **T2D-GENES** consortium with funding and support from the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK). http://www.type2diabetesgenetics.org/.

VII. MASTER GRANT FUNDING LIST FOR STDOI INVESTIGATORS (07/2015 – 06/2016)

P.I.	Project Title	Total Funding Period	Funding Agency	Grant No.	Awarded from July 2015 – June 2016				
					Direct Costs	Indirect Costs	Total Costs (to UTRGV)	Subcontract	Total Award Funding
Almasy	Neurodevelopment: Genes, Environment and their Interactions	08/01/2015 - 05/31/2018	NIH/NIMH	R01 MH107248	299,976	172,439	472,415	393,189	865,604
Almasy (UTRGV)	Collaborative Study on the Genetics of Alcoholism	09/01/2014 - 08/31/2019	NIH/NIAAA [(Res. Found. State Univ. of N.Y., SUNY)]	U10 AA008401	187,049	88,076	275,125	- - -	275,125
Almasy	A Neurobehavioral Family Study of Schizophrenia	01/01/2015 - 12/31/2018	NIH/NIMH	R01 MH061622	640,237	265,032	905,269	- - -	905,269
Almasy	Genetic Analysis of Common Diseases: An Evaluation	08/01/2013 - 07/31/2017	NIH/ NIGMS	R01 GM031575	699,637	316,936	1,016,573	- - -	1,016,573
Blangero	2/3: Pedigree-Based Whole Genome Sequencing of Affective and Psychotic Disorders	03/01/2015 - 02/28/2019	NIH/NIMH	U01 MH105632	1,294,264	636,216	1,930,480	26,000	1,956,480
Blangero	Gene Networks Influencing Psychotic Dysconnectivity in African Americans (Year 01 – 2014; Year 02 - 2015)	12/10/2014 - 11/30/2019	NIH/NIMH (Yale)	R01 MH106324	128,131	58,043	186,174	- - -	186,174
Blangero Curran	Whole Genome Sequencing to Identify Causal Genetic Variants Influencing CVD Risk	04/15/2012 - 03/31/2017	NIH/NHLBI	R01 HL113323	4,038,105	787,190	4,825,295	111,636	4,936,931
Blangero Curran	Examination of Sequence Variants in Large Human Pedigrees to Identify Novel Genetics Variants Involved in Cardiac (Patho) Physiology	11/12/2015 - 11/12/2018	Eli Lilly	Contract	281,300	127,428	408,728	- - -	408,729
Duggirala	Genetics of Type 2 Diabetes in Indian Populations: US-India Collaboration Project	04/08/2016 - 03/31/2018	NIH/NIDDK	R21 DK105913	48,681	33,377	82,058	167,610	249,668
Duggirala Blangero	Discovery of Functional Variants in Type 2 Diabetes Genes in Mexican Americans	05/01/2015 - 04/30/2019	NIH/NIDDK	U01 DK085524	669,282	333,224	1,002,506	296,552	1,299,058
Göring	A Genetic Study of Blood Metabolites and Their Relationship to Diabetes Risk	09/01/2013 - 06/30/2017	NIH/NIDDK	R01 DK099051	623,763	293,889	917,652	164,470	1,082,122
Göring	2/2 - An Integrative Genetic Investigation of Schizophrenia	07/21/2011 - 04/30/2016 (N/C 04/2017)	NIH/NIMH	R01 MH094116	34,837	27,106	61,943	59,404	121,347
Göring (UTRGV)	Gene Expression in an African American Schizophrenia Dataset	07/12/2012 - 06/30/2016	NIH/NIMH (Northshore)	R01 MH098059	78,276	35,459	113,735	- - -	113,735
Johnson	Defining the Genetic Architecture of Macular Degeneration in Mexican American Families	03/17/2015 - 08/31/2015	Max and Minnie Tomerlin Voelcker Fund	Grant (Young Inv. Award)	171,117	17,112	188,229	- - -	188,229
Johnson (UTRGV)	A Joint Linkage/Association Strategy to Interrogate AMD Genetic Susceptibility	09/01/2015 - 08/31/2016	NIH/NEI (OHSU)	R01 EY021532	49,677	22,504	72,181	- - -	72,181
Johnson Williams-Blangero	Genetic Epidemiology of Ocular Health and Disease	06/01/2014 - 05/31/2019	NIH/NEI	R01 EY024384	641,778	358,406	1,000,184	455,638	1,455,822
Mummidi	Impact of disease associated noncoding polymorphisms on post-transcriptional gene regulation	04/01/2016 - 03/31/2020	NIH/NIAID	R01 AI119131	223,933	112,767	336,700	39,752	376,452
Williams-Blangero (UTRGV)	Establishment and Maintenance of a Closed CPRC SPF Colony	05/01/2011 - 04/30/2016	NIH/OD (University of Puerto Rico)	U42 OD011128	13,290	6,146	19,436	- - -	19,436
Williams-Blangero (UTRGV)	Caribbean Primate Research Center	01/15/2016 - 11/30/2020	NIH/OD (University of Puerto Rico)	P40 OD012217	23,462	10,629	34,091	- - -	34,091
				Total	10,146,795	3,701,979	13,848,774	1,714,251	15,653,026

VIII. COLLABORATING INSTITUTIONS – UNITED STATES

Arizona
Arizona State University
Phoenix Epidemiology and Clinical Research Branch, NIDDK, NIH

California
Stanford University
University of California Berkeley
University of California San Diego
University of California Los Angeles

Connecticut
University of Connecticut
Yale University

Georgia
University of Georgia
Rollins School of Public Health, Emory University

Illinois
Loyola University

Indiana
Indiana University
Indiana University School of Medicine

Iowa
University of Iowa

Kansas
University of Kansas

Maryland
Johns Hopkins Bloomberg School of Public Health
University of Maryland School of Medicine

Massachusetts
Harvard University

Missouri
Washington University

New Jersey
Rutgers University

New Mexico
New Mexico Department of Health

New York
Icahn School of Medicine at Mt. Sinai
SUNY Downstate
Sony Brook University

North Carolina
University of North Carolina
Wake Forest University School of Medicine

Ohio
Miami University
Wright State University

Oklahoma
University of Oklahoma Health Science Center

Oregon
Oregon Health and Sciences University

Pennsylvania
Penn State University
University of Pennsylvania
University of Pittsburgh

South Dakota
South Dakota State University

Texas
Baylor College of Medicine
Southwest Research Institute
Texas Biomedical Research Institute
Texas Department of State Health Services
Texas Tech University Health Science Center
University of Texas Health Science Center at Houston
University of Texas Health Science Center at San Antonio
University of Texas Rio Grande Valley
University of Texas School of Public Health – Brownsville

Virginia
Virginia Commonwealth University

Washington D.C.
National Institute for Mental Health
National Institute of Diabetes and Digestive and Kidney Diseases

Puerto Rico
Caribbean Primate Research Center

IX. COLLABORATING INSTITUTIONS – INTERNATIONAL

Argentina
University of Buenos Aires

Australia
Baker IDI Heart & Diabetes Institute
Institute of Health and Biomedical Innovation, Queensland University of Technology
Menzies Research Institute, University of Tasmania, Tasmania
University of Western Australia

Brazil
Federal University of Mato Grosso do Sul, Campo Grande
Fundação Oswaldo Cruz – FIOCRUZ, Belo Horizonte
Fundação Oswaldo Cruz – FIOCRUZ, Brasilia

Costa Rica
University of Costa Rica

India
Lata Medical Research Foundation, Nagpur
Narayana Medical College and Hospital
Rajasthan University of Health Sciences
Sri Ramachandra University

Kuwait
Kuwait University

Mexico
Hospital General de Mexico Dr. Eduardo Liceaga
Universidad México Americana del Norte A.C., Ciudad Reynosa Mexico
Universidad Nacional Autónoma de México
University of Chihuahua
University of Veracruz
University Veracruzana Xalapa

Nepal
Chitwan Medical College, Chitwan
Tilganga Institute of Ophthalmology, Kathmandu

Portugal
University of Porto

Switzerland
Drugs for Neglected Diseases, International

United Kingdom
University of Edinburgh