

Genetics of the Acadian People



March 10, 2001

9:00 am - 4:00 pm

Nicholls State University, Talbot Hall Auditorium

Thibodaux, LA

A community event organized by

Louisiana Louisiana State University Health Sciences Center,
The Louisiana Area Health Education Centers, and Nicholls State University.

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Moderator: John P. Doucet, Ph.D.

- 9:00 am - 9:30 am Welcome
Donald J. Ayo, Ph.D., President, Nicholls State University
William L. Jenkins, Ph.D., President, Louisiana State University System
Raychel Bartek, Staff member with Congressman Billy Tauzin, Wahington, DC
Bronya J. B. Keats, Ph.D., Chair, Department of Genetics,
Louisiana State University Health Sciences Center, New Orleans
- 9:30 am - 10:30 am “Histories of Genes; Histories of Populations”
Charles R. Scriver, M.D., Professor of Pediatrics,
Biology and Genetics, McGill University, Montreal, Quebec
- 10:30 am – 10:45 am Move to rooms for breakout sessions
- 10:45 am - 11:45 am Breakout Sessions
- 11:45 am - 1:00 pm Lunch
- 1:00 pm - 2:00 pm Breakout Sessions
- 2:00 pm - 2:15 pm Move to auditorium
- 2:15 pm - 3:15 pm “Genetic Factors in Obesity and Diabetes”
Claude Bouchard, Ph.D., Executive Director,
Pennington Biomedical Research Center, Baton Rouge
- 3:15 pm - 3:45 pm “The Importance of Louisiana Acadians to Modern Medical Science”
John P. Doucet, Ph.D., Assistant Professor of Biological Sciences,
Nicholls State University, Thibodaux
- 3:45 pm - 4:00 pm Closing Remarks

We're All In This Together

by Congressman Billy Tauzin

It's been nearly 250 years since the first Acadians arrived in Louisiana after being exiled by the British from their homes in Canada, and wandering for years along the Atlantic Coast. Their homes were burned, their crops destroyed, children and parents separated. The havoc wreaked upon these peace-loving people is hard to grasp from the perspective of the 21st century but this much we know...by maintaining their language, marriage patterns, religion and way of life, the Acadians were able to survive in America. Over the centuries, these hard-working men and women have given so much to this great country of ours and asked for so little in return.

Today we know that the Acadians and their descendants, the Cajuns, are a jewel in the crown of life. Mention "Cajun" and chances are you'll hear stories about good people, good food, good music - the good life. But go deeper into a family's history and you'll also learn of the personal challenges of genetic diseases like Friedreich's ataxia and Usher syndrome.

I never fully realized the personal impact of hereditary health problems until my longtime assistant, Raychel Bartek, made me aware that her son Keith had been diagnosed with Friedreich's ataxia. Keith's illness put in motion a series of events that ultimately led to federal funding to create the Center for Acadiana Genetics and Hereditary Health Care at LSU Health Sciences Center. I'm proud to have spearheaded this effort in Congress, and I'm proud for its support by the entire Louisiana delegation and the late Dr. Merv Trail, Chancellor of LSU Health Sciences Center. I'm pleased to report that we have received approximately \$3 million to date.

The Center's primary mission is to increase both education and clinical counseling in genetics for the people of Acadiana. It is therefore altogether fitting that *Genetics of the Acadian People* should take place in Thibodaux, in the heart of Cajun country, and at Nicholls State University, my alma mater. I want to thank Dr. Donald Ayo, President of Nicholls State University, for hosting this important community event as part of the University's third annual *Jubilee—A Festival of Arts and Humanities*. *Jubilee* was created three years ago as a way to celebrate the 50th anniversary of Nicholls State University. Since then, dozens of writers, scholars, scientists and performers have shared their talent and left their mark. And among the hundred events of Jubilee 2001, Nicholls hosts this important genetics event. I'm especially delighted that so many in the genetics community of Louisiana have come together to share and to care.

In 1948 when Nicholls was founded, the structure of the genetic material, DNA, was still unknown. Fifty-three years hence, with the entire structure of the human genome within our grasp, we are on the threshold of understanding, treating and curing diseases that have afflicted mankind since the dawn of time. That is an amazing achievement in such a short span of time. What is equally amazing is that the Acadians of Louisiana are in a unique position to not only learn from these great advances in knowledge but also to contribute to the knowledge that can save lives around the world.

Cajuns and non-Cajuns alike are realizing that we are all in this together, and together we are all making a difference.



Billy Tauzin, "the Cajun Ambassador to Congress," and Keith Andrus, at the Congressional hearings to establish the Center for Acadiana Genetics and Hereditary Health Care at LSU Health Sciences Center.

The Importance Of Louisiana Acadians To Modern Medical Science

John P. Doucet, Ph.D.

From the moment of their arrival in the New World in the early seventeenth century, Acadians were a people apart. Initially reacting to rival cultures and the harsh climate of maritime Canada, Acadians have developed a social cohesiveness strengthened by centuries of cultural uniqueness. In eighteenth and nineteenth century Louisiana, as in their historical homeland *Acadie*, Acadians lived in geographic isolation compounded by barriers of language, religion, and lifestyle. Like other isolated populations around the world, the Acadians have an increased prevalence of certain rare diseases, such as Friedreich's ataxia and Usher syndrome.

Genetic diseases are found in all populations, and genes play a role in most of the common diseases that affect many of us, such as cancer, heart disease, diabetes, and psychiatric illness. In the effort of modern medicine to use knowledge about the human genome to create new drugs and therapies, it is the Acadian people, together with the family records maintained by the Catholic Church, that are making an important contribution to mankind's understanding of genetic diseases and potential cures.



Distinguished Speaker:

Charles R. Scriver, M.D.

Dr. Scriver is Professor of Pediatrics, Biology (Honorary) and Human Genetics in the Faculties of Medicine and Science at McGill University in Montreal, Canada, and Alva Professor of Human Genetics. His B.A. and M.D.C.M. degrees (cum laude) are from McGill University, and he holds four honorary degrees (D.Sc.) as well as civil honors from Canada (CC) and Quebec (GOQ). He has been President of the Society of Pediatric Research, the Canadian Society for Clinical Investigation, the American Society for Human Genetics, the Society for the Study of Inherited Metabolic Diseases, and the American Pediatric Society. Dr. Scriver's current research is about human genetic variation and the informatics involved in the Human Genome Organization (HUGO) Mutation Database initiative. This work links

histories of populations with histories of genes. Dr. Scriver has over 500 publications and is the senior editor of "The Metabolic and Molecular Bases of Inherited Disease", now in its 8th edition.

History Of Genes; History Of Populations

The evolutionary origins of *Homo sapiens* (us) are identifiable in the milestones of the very long journey of Life on Earth (3.5 billion years). The genome project (often called the Human Genome Project) is certainly the biggest scientific project in biology at the end of the 20th century and it is telling us about the journey. What we learn from the genes of bacteria, yeast, worms and mice, for example, all contribute to the knowledge of our own genetic makeup.

There is "unity in diversity". Unity is the organism *Homo sapiens* (our human species); diversity is our individuality. Diversity in our genetic makeup is, in part, the explanation of individuality. We each have our own "self"; our particular set of parents scrambled our genes in particular ways when we began life; our parents came from particular communities and populations. If we know our history, we begin to understand why we have both individual and collective identities.

Two points of view follow. First, if human individuality is something we treasure, why would we want to clone people, a procedure that attempts to minimize individuality. Second, biological individuality is a feature of modern diseases. At the beginning of the century, environmental diseases (infectious disease, epidemics, poor environmental standards, inadequate nutrition, etc.) were important explanations for human disease and, when combined with accidents, life could be nasty and short. Vaccination, immunization, nutrition and better quality of life have each improved human health and longevity in the 20th century. Nonetheless, disease continues. A biologist would recognize that modern disease is increasingly reflecting biological individuality; the geneticist will recognize that human disease has increased its heritability (where the genetic causes compared to environmental causes are now relatively more important). The modern physician will be asking the question: "Why does this patient have this disease now?" and will find biological (genetic) causes and susceptibilities highly relevant.

Genetics in medicine is already a beneficial resource in health care as illustrated here by three stories: i) Stanley discovered a relative in another part of the world by using his own rare mutation causing beta-thalassemia (a hereditary anemia). ii) New knowledge about a form of mental retardation led to universal newborn screening for early diagnosis and treatment to prevent the disease (phenylketonuria) and maintain health. (In this particular example, we will visit the most widely practiced genetic test in the world). iii) A story about a family dear to me that was affected by cystic fibrosis. Knowledge of DNA, mutation, history and genetics in the medical context was helpful in each of these human stories; it is an encouraging message.

Distinguished Speaker

Claude Bouchard, Ph.D.

Dr. Bouchard is the Executive Director of the Pennington Biomedical Research Center and the George A. Bray Chair in Nutrition. He holds a B.Ped. (Laval), a M.Sc. (University of Oregon, Eugene) in exercise physiology and a Ph.D. (University of Texas, Austin) in population genetics. His research deals with the genetics of adaptation to exercise and to nutritional interventions as well as the genetics of obesity and its co-morbidities. He has authored and coauthored several books and more than 600 scientific papers. Among other awards, he was the recipient of the Honor Award from the Canadian Association of Sport Sciences in 1988, a Citation Award from the American College of Sports Medicine in 1992, the Benjamin Delessert Award in nutrition from France in 1993, the Willendorf Award from the International Association for the Study of Obesity in 1994, the Sandoz Award from the Canadian Atherosclerosis Society in 1996, the Albert Creff Award in Nutrition of the National Academy of Medicine of France in 1997, the TOPS award from the North American Association for the Study of Obesity in 1998, the W. Henry Sebrell Award from the Weight Watchers Foundation in 1999, and of a Honoris Causa Doctorate in Science from the Katholieke Universiteit Leuven in 1998. In 2001, he became a member of the Order of Canada.



Dr. Bouchard is past president of the North American Association for the Study of Obesity and the president-elect of the International Association for the Study of Obesity. Prior to coming to Pennington, he held the Donald B. Brown Research Chair on Obesity (a chair funded by the Medical Research Council of Canada) at Laval University. He receives funding from various agencies but mainly the National Institutes of Health and the Medical Research Council of Canada.

Genetic Factors in Obesity and Diabetes

The prevalence of obesity has increased dramatically in the United States over the last 40 years or so. At present, 55 percent of the U.S. adults are overweight or obese and the situation is even worse in Louisiana. What is even more worrisome is the fact that the prevalence of obesity is rising alarmingly in children and adolescents as well. This situation is disturbing particularly because obesity is associated with a variety of common, chronic diseases especially type 2 diabetes. Indeed, in all countries of the Western world, and even among developing countries, the increase in the prevalence of diabetes parallels that of obesity. This has enormous public health and economical implications. Experts are still debating whether the current epidemic of obesity and the increasing rate of type 2 diabetes (almost 16 million Americans are affected) are caused by recent changes in our environment and lifestyle or if they are fueled primarily by genetic factors. It is true that a small fraction of severely obese individuals carry mutations in genes that exert major detrimental effects on food intake or energy expenditure. The same applies to diabetes, as there is a minority of cases that can be accounted for by mutations in one or a few genes. However, the contribution of genetic individuality to the risk of obesity or diabetes does not generally come in the form of genes causing the disease. Rather, genetic individuality manifests itself in the form of a slight predisposition to become obese or to become diabetic in the presence of obesity. This predisposition is thought to arise as a result of mutations at several genes, each mutation having only a minor effect when taken individually. As a result of the advances in the sequencing of the human genome and of model organisms, and because of the progress made in human genomic technologies, efforts are underway in many laboratories around the world to identify all genes and mutations responsible for the predisposition to obesity and diabetes. When such data are available, it will be possible to identify early in life the individuals at risk for these diseases and to develop more targeted preventive or therapeutic measures. In the meantime, maintaining a normal body weight and being physically active are the most useful preventive measures.

Breakout Sessions

In each session, the facilitators will briefly introduce themselves and then answer any questions you may wish to ask. The facilitators in each room and the topics about which they have experience are as follows:

1. Ron Bartek, Dr. William Fisher, Dr. Gloria Giarratano, Dr. Mary Kay Pelias
 - **Ethical and legislative issues** in medical genetics
 - **Establishing a non-profit support organization** for genetic disease research
 - Improved **quality measurement in health care**
 - **Nursing care** for families with genetic disorders
 - Genetics of eye disorders and **Usher syndrome**

Room Monitor: Nathan Markward

2. Dr. Claude Bouchard, Dr. Christine Pollock, Dr. George Karabatsos, Dr. Bronya Keats
 - Genetics of **Obesity** and **Diabetes**
 - **Self-management strategies** in Diabetes
 - Medical decision making
 - **Friedreich ataxia** and **Usher syndrome**

Room Monitor: Deiadra Garrett

3. Dr. Juan M. Acuña, Dr. Carlos Garcia, Dr. Jay Hunt, Dr. Yves Lacassie
 - **Genetic disorders** in children and adults
 - Neurogenetic disorders in Acadiana, in particular, **Charcot-Marie-Tooth disease**
 - Prevalance of **birth defects**
 - Lung and Prostate **Cancer**

Room Monitors: Luisa Florez, San San Ng

4. Dr. Hans Andersson, Amy Cunningham, Kelly Jackson, Dr. Gabriella Pridjian
 - Epidemiology of **PKU** in all areas of Louisiana from 1985 - 2000
 - Management of **maternal PKU** in several cases in Louisiana
 - **Dietary management** of PKU in children and adults
 - Current **newborn screening** and follow-up of PKU in Louisiana
 - **Resources** for PKU patients in Louisiana

Room Monitor: Tara Turley

5. Raychel Bartek, Charles Myers, Dr. Jay Rao, Dr. Charles Scriver
 - Genetic disorders and **genetic testing**
 - Neurological disorders in Acadiana, in particular, **Parkinson disease**
 - Friedreich ataxia
 - Clinical **Genetic Programs** in Acadiana

Room Monitor: Gauri Gaikwad

6. Dr. Elizabeth Fontham, Dr. Diptasri Mandal, Dr. Paul Swarzenberger, Dr. Ted Thurmon
 - Genetic disorders and **genetic testing**
 - **Cancer** in Acadiana
 - Prevalance of **lung** and **prostate cancer**
 - **Cancer** and **gene therapy**

Room Monitor: Berniece Parker

7. Dr. Kevin Brown, Dr. Elizabeth Humphrey, Dr. Walter Rayford, Dr. Jess Thoene
- Genetic disorders and **Orphan Drugs**
 - Prostate Cancer
 - **Psychological and social impact** of genetic disorders
 - **Ataxia Telangiectasia** and Cancer

Room Monitor: Miriam Martinez

8. Julie Lovell, Dr. Michael Marble, Scharalda Jeanfreau, Heidi Schumacher
- **PKU** and other disorders of metabolism
 - **Newborn screening** for metabolic disease
 - Down syndrome
 - Genetic counseling
 - Dietary tips and self-management strategies in **diabetes**

Room Monitor: Mimi Sammarco

9. Dr. Charles Berlin, Dr. John Doucet, Dr. James Gnarra, Dr. Sevtap Savas
- Hereditary **hearing loss**
 - Inherited **cancer** syndromes
 - Genetics of hearing loss and eye disorders, in particular, **Usher syndrome**

Room monitors: Deidre Arnaud, Xing Cheng

10. Regan Challinor, Dr. Prescott Deininger, Dr. Doug Scheer, Dr. Duane Superneau
- **Cancer** and Gene Therapy
 - Abnormalities of **chromosomes**
 - **Genetic counseling** and testing
 - **Diabetes** and **heart disease**

Room Monitor: Stephanie Laurent

11. Dr. Michal Jazwinski, Dr. Jakob Reiser, Dr. Tal Thomas, Dr. Raj Warrior
- **Mitochondrial disorders** in Acadiana
 - Pediatric **Cancer**
 - Genetics and **Aging**
 - **Gene Therapy** for neurological disorders

Room Monitor: Derek Ragusa

12. Jacqueline Favret, Dr. Paul Friedlander, Dr. Jay Kolls, Dr. Gary Truett
- Gene Therapy
 - Head and Neck **Cancers**
 - **Obesity, diabetes** and **heart disease**
 - Community Health **Nursing**

Room Monitor: Sharon Lee

Facilitators for Breakout Sessions

Juan M. Acuña, M.D., MSc, has clinical specialization in Obstetrics and Gynecology, and Clinical Genetics and a Masters Degree in clinical Epidemiology. He is in the Division of Reproductive Health in the Centers for Disease Control and Prevention and is working with the Louisiana Office of Public Health as the Maternal Child Health Medical Epidemiologist. His responsibilities include coordination of the Assessment and Evaluation program, and technical and scientific assistance for the Birth defects Surveillance system program for Louisiana.

Hans C. Andersson, M.D., is Associate Professor of Pediatrics in the Hayward Genetics Program at Tulane University Medical School. Dr. Andersson is board-certified in Pediatrics, Clinical Genetics and Clinical Biochemical/Molecular Genetics. As a clinician-scientist, his research activities involve clinical and pathophysiologic characterization of inborn errors of metabolism. As Associate Director of the Biochemical Genetics Laboratory in the Human Genetics Program and in genetics clinics in Acadiana, Dr Andersson cares for patients with many genetic metabolic diseases present in this region.

Raychel Bartek has served on the Washington, DC staff of Congressman Billy Tauzin for the past 21 years. Prior to moving to Washington, she served on the House Natural Resources committee staff in Baton Rouge under the chairmanship of Billy Tauzin. Raychel is a native of Shreveport and graduate of USL. She is the mother of 3 sons, Byron, Keith and Stuart Andrus - ages 17, 15, and 12, respectively. Her 15 year-old son, Keith, is afflicted with the life-shortening neurodegenerative disorder, Friedreich's Ataxia (FA), and Stuart is a carrier of the FA gene.

Ronald J. Bartek is President of FARA, the Friedreich's Ataxia Research Alliance, a non-profit organization headquartered in Washington, DC. He is a graduate of the US Military Academy (West Point) and has a master's degree from Georgetown University. In addition to his work with FARA, Ron is a business consultant, specializing in foreign and national security issues. He has served in senior staff positions with the State Department, the CIA and the US Congress. Ron is a dedicated stepdad to Byron, Keith and Stuart Andrus.

Charles I. Berlin, Ph.D., is Kenneth and Francis Barnes Bullington Professor of Hearing Science, Professor of Otolaryngology-Head and Neck Surgery and Director of the Kresge Hearing Research Laboratory of the South at LSU Health Sciences Center. His scientific interests include the early diagnosis and management of deafness in infants with hearing aids and cochlear implants, the latest technologies in hearing health care and service, and hereditary hearing loss. He has seen and/or treated many families in Acadiana for their hearing losses.

Kevin D. Brown, Ph.D., is Assistant Professor of Biochemistry and Molecular Biology and a Member of the Stanley S. Scott Cancer Center at LSU Health Sciences Center. His research interests involve the molecular pathology of the hereditary ataxias, Ataxia Telangiectasia (A-T) and Friedreich's Ataxia (FA). Specifically, he is studying cancer development in A-T patients and using modern gene therapy approaches for treatment of FA.

Regan Challinor, M.S., is a genetic counselor at Woman's Hospital in Baton Rouge and is a graduate of the human genetics program at Sarah Lawrence College in Bronxville, NY. She works with the Maternal Fetal Medicine Department to provide prenatal counseling and coordinate testing, and also provides genetic counseling to individuals and research participants in the Louisiana Breast Cancer Study.

Amy Cunningham, M.S., L.D.N., R.D., is the metabolic nutritionist with the clinical care team of the Human Genetics Program, Tulane University Medical Center. She is responsible for coordination of dietary treatment for patients who have been diagnosed with genetically inherited metabolic conditions. This includes recommending appropriate diet restrictions and metabolic formulas, monitoring the ongoing nutritional treatment for each patient, and assisting families and patients in understanding and providing for these special dietary needs.

Prescott L. Deininger, Ph.D., is Director of Basic Sciences for the Tulane Cancer Center and the Zimmerman Chair in Basic Cancer Research. He is a professor of Environmental Health Sciences, and he is also the Founding Director of the Laboratory of Molecular Genetics at Ochsner Medical Foundation. His research interests involve the basic human genetic defects underlying cancer and the potential uses of gene therapy.

John P. Doucet, Ph.D., is a descendant of the original Lafourche Doucets who arrived in Louisiana in 1785 and settled across the bayou from Nicholls State University. He is a past two-time National Institutes of Health Research Fellow in retinal biochemistry and molecular genetics, and he is currently Assistant Professor of Biological Sciences at Nicholls State University. His research is focused on gene discovery in inherited diseases of peoples indigenous to Louisiana. Dr. Doucet is also an award-winning author of twelve plays set with the historical and cultural backgrounds of Louisiana peoples.

Jacqueline O. Favret, R.N., M.P.H., is a native of Scott, Louisiana, who has maintained her ties and interest with Cajun culture. At LSU Health Sciences Center she is an assistant professor of clinical nursing. Her specialty is community health nursing, with a special interest in the legal aspects of nursing. She serves as a counselor for the R.N. to B.S.N. students. She was formerly the nursing consultant for compliance with the Board of Nursing.

William P. Fisher, Jr., Ph.D., is Professor of Research, with appointments in the Departments of Genetics, and Public Health & Preventive Medicine, at LSU Health Science Center. He is internationally recognized for his vision of how our usual ideas about surveys and tests could be transformed into new and exciting possibilities for enhanced communication. His responsibilities include roles in the Quality Council of the LSUHSC Health Services Division, on the Editorial Boards of the Journal of Applied Measurement and the Journal of Outcome Measurement, and as Chair of the Institute of Objective Measurement Advisory Board's Program Committee.

Elizabeth Fontham, Ph.D., is Professor and Head of the Department of Public Health and Preventive Medicine. For the past 20 years, Dr. Fontham has conducted epidemiological studies of cancer, particularly cancers occurring at high rates in south Louisiana. Her current research interests include the role of gene-environment interaction in cancer risk. Dr. Fontham's family was one of the first settlers of the town of Crowley.

Paul L. Friedlander, M.D., FACS, is an Assistant Professor in the Department of Otolaryngology and Biocommunications at the LSU Health Sciences Center. His clinical interests include laryngology and the treatment of cancers of the head and neck. His research interests include the use of tumor specific adenoviruses in gene therapy as well as characterization of cell surface receptors in head and neck tumors.

Carlos A. Garcia, M.D., is Professor of Clinical Neurology at Tulane University School of Medicine. He has been in charge of a referral center for muscle and nerve biopsies for the last twenty years. His main interests are the clinical diagnostic aspects of inherited and acquired neuromuscular disorders, in particular, Charcot-Marie-Tooth disease and the histological aspects of peripheral nerves and muscle. At the present time, he is the clinical director of the Muscular Dystrophy Clinic in Lafayette, Louisiana.

Gloria Giarratano, RN, Ph.D., is an assistant professor in the School of Nursing at LSU Health Sciences Center. She teaches health care of childbearing families in the graduate and undergraduate programs. Her areas of interest include genetic factors affecting women's health, pre-conceptual health care, and the care of families throughout the childbearing cycle who have genetic concerns or problems.

James R. Gnarra, Ph.D., is Associate Professor of Biochemistry and Molecular Biology, Genetics, and Urology, LSU Health Sciences Center. His research and clinical interests include cancer biology, cancer genetics, and developmental biology. In particular, his focus is on understanding how tumors associated with von Hippel-Lindau disease, an inherited human cancer syndrome, arise and the development of therapeutic applications.

Elizabeth Humphrey, R.N., Ed.D., is Professor and Dean of the School of Nursing, LSU Health Sciences Center. She has held positions on the Louisiana Rural Health Care Authority and several professional Boards. Her interests include primary health care, care of mothers and children, and the education of nurses and nurse practitioners.

Jay D. Hunt, III, Ph.D., is an Associate Professor of Biochemistry and Molecular Biology in the Stanley S. Scott Cancer Center of LSU Health Sciences Center. His research interests include the molecular genetics of lung cancer and the development of new therapies for lung cancer and prostate cancer.

Kelly Jackson, M.S., is a certified genetic counselor in the Human Genetics Program at Tulane University School of Medicine. At weekly Tulane genetics clinics and satellite genetics clinics throughout the state, she provides genetic counseling to families and individuals who have, or may have, a genetic condition. Ms. Jackson is also part of a multi-disciplinary breast cancer clinic and provides genetic counseling to individuals with a family history of breast cancer. Her interests include chromosome abnormalities, inborn errors of metabolism, and cancer genetics.

S. Michal Jazwinski, Ph.D., is Professor of Biochemistry and Molecular Biology, and of Family Medicine at LSU Health Sciences Center in New Orleans. His research is in the area of the biology of aging with emphasis on genetics and environmental interactions, and population genetics.

Scharalda Jeanfreau, M.N., C-FNP, is an Instructor in the LSU Health Sciences Center School of Nursing Primary Care Nurse Practitioner program and Coordinator of the Disease State Management program at the Daughters of Charity Health Center in New Orleans. Her clinical practice at the Health Center involves educating patients about diabetes and helping them make changes in behavior that will increase their self-management ability. Her areas of interest include psychosocial responses to living with diabetes, especially grief or stress responses, and self-management education and strategies.

George Karabatsos, Ph.D., is Assistant Professor of Biometry and Genetics at the LSU Health Sciences Center, and is a 1998 graduate of The University of Chicago's Measurement, Evaluation, and Statistical Analysis program. His primary research interests involve psychological modeling, cultural consensus theory, fundamental measurement, order restricted inference, Markov Chain estimation theory, the analysis of measurement disturbances, and the applications of psychological models for the measurement of medical knowledge and reasoning, cultural perceptions, and medical decision making.

Bronya J. B. Keats, Ph.D., is Professor and Head of the Department of Genetics, LSU Health Sciences Center. She is the Director of the Molecular and Human Genetics Center of Excellence, one component of which is the *Center for Acadiana Genetics and Hereditary Health Care*. Her major areas of interest include the genetics of hearing impairment and neurodegenerative disorders, in particular Friedreich's Ataxia.

Jay K. Kolls, M.D., is Professor of Medicine and Pediatrics, and Director of the Gene Therapy Program at LSU Health Sciences Center. His research interests include immune responses to gene delivery, gene therapy for cystic fibrosis, muscular dystrophy, and gene-based therapies to modulate immunity against infections.

Yves Lacassie, M.D., is Professor of Clinical Pediatrics, Head of the Division of Clinical Genetics, Department of Pediatrics, LSU Health Sciences Center, and Director of Genetic Services, Children's Hospital of New Orleans. A graduate of the University of Chile, Dr. Lacassie is board certified in Clinical Genetics. His major interests are dysmorphology, the delineation of new syndromes, the nosology of genetic diseases, and the clinical use of dermatoglyphics.

Julie Lovell, M.S., is Instructor of Pediatrics at LSU Health Sciences Center and the genetic counselor for Children's Hospital in New Orleans. She provides information and counseling to families at genetic clinics in New Orleans and satellite clinics throughout Louisiana. Her areas of interest include the psychosocial aspects of genetic disease, the impact that genetic conditions can have on the family, and the education of health care professionals and the public regarding genetic conditions.

Diptasri M. Mandal, Ph.D., is Instructor of Genetics at the LSU Health Sciences Center. Her major areas of interest include investigating the properties of various methods of genetic analysis through computer simulation and studying the genetics of complex diseases such as the human cancers, in particular, lung and prostate cancer, in different populations.

Michael Marble, M.D., is Assistant Professor of Pediatrics at Children's Hospital of New Orleans/ Louisiana State University. Dr. Marble is board certified in Clinical and Biochemical Genetics. He directs the Metabolic/PKU clinic located at Children's Hospital and is co-medical director of the Children's Hospital Metabolic Laboratory. In addition to clinics in New Orleans, Dr. Marble conducts outreach genetics clinics in Thibodaux, Lafayette and other locations. His interests include metabolism, genetic syndromes and chromosomal disorders.

Charles Myers, M.S.W., received his Master of Social Work Degree from Tulane University with a specialization in maternal and child health. Since graduating, Mr. Myers has administered the Louisiana Genetic Diseases Program in the Office of Public Health, Louisiana Department of Health and Hospitals. His responsibilities include coordination of the newborn screening program for genetic diseases as well as management of regional genetics clinics throughout Louisiana.

Mary Kay Pelias, Ph.D., J.D., a native of New Orleans, is Professor of Genetics at the LSU Health Sciences Center in New Orleans, where she teaches human genetics and medical genetics to students in graduate studies, medicine, dentistry, and allied health. She is a board-certified Ph.D. Medical Geneticist with 25 years of experience in clinical genetics and in genetic research among the rural populations of southern Louisiana. She is also an attorney, admitted to the Bar, with a special focus on the legal and ethical dilemmas generated by the new genetic technologies.

Christine A. Pollock, RN, Ph.D., is an associate professor of clinical nursing at LSU Health Sciences Center. She provides theoretical and clinical instruction to students on maternity nursing, research, and transcultural nursing issues. In addition, through a private group, she also provides individual, couple and family therapy for those with counseling concerns. Her areas of interest include women's health, international health issues, and alternative and complementary therapies. Of special concern is the global problem of violence against women and the impact that this has on the health of women and families throughout the world.

Gabriella Pridjian, M.D., is Associate Professor in the Department of Obstetrics/Gynecology, where she is Section Chief of Maternal-Fetal Medicine, and a faculty member of the Hayward Genetics Center at Tulane University School of Medicine. Dr. Pridjian is a diplomate of the National Board of Obstetrics and Gynecology, has subspecialty certification in Maternal-Fetal Medicine and is board certified in Clinical Genetics. Her clinical interests include prenatal diagnosis and management of fetal abnormalities and high risk pregnancies, using ultrasonography, traditional and early amniocentesis, and chorionic villus sampling.

Jayaraman Rao, M.D., is Professor of Neurology and Anatomy at the LSU Health Sciences Center where he is also the Dr. Carl Baldrige Endowed Professor of Neurology and Neuroscience. His special clinical and research interest is Parkinson's disease and movement disorders. He is the founder of a nationally recognized center for the diagnosis and treatment of Parkinson's disease, one of only 30 such centers in America.

Walter Rayford, M.D., Ph.D., is Assistant Professor of Urology and director of urologic oncology research at LSU Health Sciences Center. His clinical and research interests are focused on effective management of prostate cancer.

Jakob Reiser, Ph.D., is Associate Professor of Medicine, and Microbiology, Immunology & Parasitology at LSU Health Sciences Center. His research interests involve the design of lentivirus-based vectors for gene therapy, vector targeting, gene therapy of CNS disorders, global protein delivery, and functional genomics.

Sevtap Savas, Ph.D., is a post-doctoral fellow in the Department of Genetics at Louisiana State University Health Sciences Center in New Orleans. She works with Dr. Bronya Keats on the Usher Syndromes. Her major research areas include mutation analysis of Usher genes, and studies on population diversity and molecular evolution.

Douglas Scheer, Ph.D., is Professor of Pathology and Scientific Director of the Molecular Pathology Laboratory at Children's Hospital. His scientific interests involve adapting molecular assays for clinical applications and risk factors of cardiovascular disease.

Heidi Schumacher, L.D.N., R.D., C.D.E., is the chief clinical dietician at Children's Hospital in New Orleans. She works with the metabolic team managing the dietary needs of infants and children with inborn errors of amino acid metabolism. In addition, she is a certified diabetes educator involved with diabetes nutrition education.

Paul Schwarzenberger, M.D., is Associate Professor of Medicine at LSU Health Sciences Center in New Orleans. He is a medical oncologist and physician scientist. His research interests involve gene therapy with emphasis on vector targeting, and cytokine biology.

Duane W. Superneau, M.D., is a board certified Clinical Geneticist and Chief of the Section of Medical Genetics for the departments of Pediatrics, Pathology, and Obstetrics/Gynecology at Ochsner Clinic in New Orleans. He also serves as Clinical Consultant to the LSU Health Sciences Center Cytogenetics Laboratory in the Pathology Department. In addition to his professional responsibilities, Dr. Superneau is on the Board of Directors of the Arc of Greater New Orleans and is President of the Board of Directors of the Arc of Louisiana.

Jess G. Thoene, M.D., is the Director of the Hayward Genetics Center and Karen Gore Professor of Pediatrics at the Tulane University Health Sciences Center. His major research interest has been clinical and biochemical investigation of cystinosis, a lethal disease of lysosomal cystine storage. He has been active in the orphan drug movement, serving as Chairman of the Board of the national organization for rare Disorders and Chair of the National Commission on Orphan Diseases.

Ioan Talfryn Thomas, M.D., is a board certified Clinical Geneticist in the Department of Pediatrics at LSU Health Sciences Center. He played viola in the Welsh National Youth Orchestra before obtaining his medical degree from the Welsh National School of Medicine. Dr. Thomas provides both general and specialty genetics clinics in New Orleans as well as Lafayette and Lake Charles. His clinical and research interests include the postnatal effects of alcohol and drug exposure, neurogenetic disorders, mitochondrial myopathies, and inborn errors of metabolism.

Theodore F. Thurmon, M.D., is Professor of Pediatrics and Director of the Medical Genetics Section at the LSU School of Medicine in Shreveport. His teaching, service and research in Louisiana spans the last 30 years. His current research is in spastic paraplegia genes, segregation analysis, laboratory genetic techniques, and clinical genetic techniques. His most recent book, *A Comprehensive Primer on Medical Genetics*, was published in 1999 by Parthenon Publishing Group, New York.

Gary E. Truett, Ph.D., is Assistant Professor in the Developmental Genetics Laboratory at Pennington Biomedical Research Center in Baton Rouge. His research interests include studies of early developmental events that affect growth and that contribute to the development of obesity and diabetes. He also studies genetic factors that contribute to the development of cerebral palsy.

Raj P. Warrier, M.D., is Professor and Vice chairman of Pediatrics, and Associate Director of the Stanley Scott Cancer Center at LSU Health Sciences Center, and Director of the Center for Cancer and Blood Disorders at Children's Hospital in New Orleans. His clinical interests include sickle cell anemia, hereditary bleeding disorders, leukemia and solid tumors in children. He provides clinics in Lake Charles, Lafayette, and Baton Rouge as well as at Children's Hospital in New Orleans.

Honoring Excellence

“As the world becomes increasingly scientific and technological, our future grows more dependent on how wisely humans use science and technology. And that, in turn, depends on the effectiveness of the education we receive.”

*George N. Nelson,
Director of Project 2061
American Association for the Advancement of Science*

Every year hundreds of junior high and senior high school students visit the campus of LSU Health Sciences Center to interact with scientists who demonstrate the latest in cutting edge research. This hands-on outreach is part of a deep commitment to educating young minds about the exciting potential of science and medicine.

This commitment has long been shared by Nicholls State University which has hosted for nearly 50 years the regional student science fairs of south central Louisiana, where elementary, junior high, and high school students present results of their science projects and compete for prizes based on their achievement. The 2001 Louisiana Region X Science Fair is being held this weekend, March 9 and 10 in Shaver Gymnasium and Peltier Auditorium.

In celebration of the efforts of these student-scientists, the Center for Acadiana Genetics and Hereditary Health Care, together with the Louisiana Area Health Education Centers (AHEC) have created the ACHIEVEMENT AWARD IN HEREDITARY SCIENCE for the student(s) showing the most promise to contribute to genetics research and hereditary health care in their future careers.

Today, during ceremonies in Peltier Auditorium, the very first Achievement Award will be presented by Mr. Brian Jakes, Chief Executive Officer of AHEC. The Louisiana AHEC system began operating in 1988 through a federal grant administered by the LSU School of Medicine. There are four centers in Louisiana and each is an independent, non-profit corporation responsible for the administration of community based education, recruitment, and training programs for health care professionals, students, and faculty. One of its most important roles is to serve as a bridge between health education institutions, health providers, and communities.

The Center for Acadiana Genetics and Hereditary Health Care in conjunction with AHEC are proud to sponsor this first award at Nicholls State University for students in the south central region of Acadiana. Future plans call for the creation of additional awards for regional science fairs across Acadiana, culminating with a top award at the annual Louisiana State Science Fair.

Merci!

We sincerely appreciate the following supporters of “Genetics of the Acadian People”:

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