Alzheimer’s Researchers, Physicians Have Strong Presence at International Conference

Leading Alzheimer’s genetics researchers with the University of Miami Miller School of Medicine’s John P. Hussman Institute for Human Genomics (HIHG) and neurologists with the University of Miami Health System presented groundbreaking research and received national media coverage at the Alzheimer’s Association International Conference (AAIC) 2019 in Los Angeles.

Faculty members, postdocs and research support staff from the Hussman Institute for Human Genomics attended the conference.
Nearly 25 faculty members, postdocs and research support staff from the HIHG attended the conference, most of whom collaborated on the Institute’s more than 20 original research presentations. The HIHG also provided updates on its latest genetic discoveries during AAIC symposiums, pre-conferences and press briefings. This included breakthrough scientific explorations into sex differences in Alzheimer’s disease and insightful findings from dementia studies that recruit people from a wide variety of ethnic backgrounds that have historically been excluded from biomedical research, such as African Americans and individuals of Hispanic/Latino heritage.

“Our diversity research at UM is critical because it addresses the problem of health disparities,” said Margaret A. Pericak-Vance, Ph.D., director of the HIHG and the Dr. John T. Macdonald Foundation Professor of Human Genetics.

“Disparities in genomic research will result in health disparities in clinical care, especially in light of the development of new potential therapies. In the age of ‘precision medicine’ it is critical that no group is left behind as these therapies and preventions are realized.”

Dr. Pericak-Vance and Jeffery Vance, M.D., Ph.D., professor of neurology and founding chair of the Dr. John T. Macdonald Foundation Department of Human Genetics, were interviewed during the July 14-18 conference by Neurology Live about the importance of including underserved and diverse groups in Alzheimer’s disease research. Dr. Jeffery Vance was interviewed on the role of genetics, specifically ApoE4 allele, in distinguishing Alzheimer’s disease risk.

“It has been known for a while that the risk for Alzheimer’s
disease due to the ApoE4 allele is much less in African carriers of the allele, even though the actual ApoE genes are the same in each population,” said Dr. Vance. “Our group recently published that this difference is due to the genetic region surrounding the ApoE4 allele. If you inherit the ApoE4 allele from an African ancestor, you have the low African risk for Alzheimer’s from the ApoE4 allele. If you inherit it from a European ancestor, you have the increased risk seen in the European population. Thus, it really matters where that ApoE4 came from in any single individual. It is a good example of how ‘precision medicine’ works for a person.”

Dr. Eden R. Martin, left, and Dr. Brian Kunkle at the conference.
Gary Beecham, Ph.D., director of bioinformatics at the HIHG and associate professor in the Dr. John T. Macdonald Foundation Department of Human Genetics, was honored with an invited symposium presentation on “Potential Contribution of Rare Variants to Neurodegenerative Brain Dementia.”

Brian Kunkle, Ph.D., M.P.H., a genetic epidemiologist and associate scientist with the HIHG, presented his high-profile findings on sex-specific genes associated with risk and resilience for Alzheimer’s disease, which could provide unique risk profiles for men and women.

The study, titled “Novel Sex-Specific Genes for Alzheimer’s Disease,” looked at whole-exome sequencing in two large datasets from the Alzheimer’s Disease Sequencing Project to identify any genes that might convey different levels of Alzheimer’s risk for men vs. women. Sex-specific associations with Alzheimer’s disease risk were found for 11 different genes, many of which have functions that may be relevant to the development of Alzheimer’s. These include genes that confer risk only in men, which are involved in endocytosis, a process critical to the development of Alzheimer’s, and genes that confer risk only in women, which play essential roles in immunity, as stated in the AAIC’s press release.

“This research demonstrates that genetics may contribute to differences in risk and progression of Alzheimer’s disease between men and women,” said Dr. Kunkle. “More research is needed to understand how much these genes contribute to Alzheimer’s risk, and whether they can be used to specifically identify men and women at risk for this disease.”

The study, which Dr. Kunkle presented on July 17 before a
group of industry and national journalists, received widespread national media coverage, including stories by the BBC, The Guardian, NPR, NBC News and The Associated Press, among other outlets.

“What’s most exciting to me — and did get much play in the news — is that some of the genes have opposite effects in men and women. A particular variant, for example, might increase a man’s risk, but decrease a woman’s risk. If you didn’t analyze them separately, you might think the gene had no effect at all on disease risk,” said Eden R. Martin, Ph.D., professor of human genetics and director of the Center for Genetic Epidemiology and Statistical Genetics (CGESG) at the HIHG, who is senior author and contact principal investigator on the research project, funded by the National Institutes of Health’s National Institute on Aging. “We’ve just begun to scratch the surface when it comes to genetic studies of sex differences in Alzheimer’s disease.”

Other important original research presentations from HIHG researchers include:

- “Genome-Wide Linkage Analysis of Caribbean Hispanic Puerto Rican Families Supports Evidence of Linkage to C9orf72 Region of Chromosome 9” by Farid Rajabli, Ph.D.
- “Genome-Wide Linkage Analysis of African-American Alzheimer’s Disease Families” by James Jaworski, Ph.D., and Farid Rajabli, Ph.D.
- “Genetic Variants in Alzheimer’s Disease-Associated Regions Have Different Effects on RNA Editing Rates in African-American and Non-Hispanic White Populations” by Miller School of Medicine M.D/Ph.D. student Olivia
The HIHG is a world-renowned human genomics research institute that has made several breakthrough discoveries in Alzheimer’s disease and has been instrumental in training the next generation of Alzheimer’s disease scientists.

Dr. Barry S. Baumel.

The Miller School’s Department of Neurology is also a leading Alzheimer’s and memory disorders care and research center, with a wide range of ongoing clinical trials, including stem cell research. It’s also home to the Brain Endowment Bank, a leading Alzheimer’s basic science research center and one of
six designated brain and tissue biorepositories in the U.S.

At the AAIC, several Miller School neurologists investigating Alzheimer’s and memory disorders provided expert commentary on innovative areas of research and new studies.

Barry S. Baumel, M.D., assistant professor of neurology, interim chief of the Cognitive Disorder Division and director of the Memory Disorders Division, spoke to Neurology Live on stem cell research for Alzheimer’s.

“The meeting allowed us to exchange ideas with investigators about bold new ideas to fight Alzheimer’s,” said Dr. Baumel.

Christian J. Camargo, M.D., a UHealth and Miller School cognitive neurologist and assistant professor of clinical neurology, provided secondary commentary for several newly released studies. In his commentary for a MedPage Today story on the use of PET imaging to track the Alzheimer’s protein amyloid and disease progression, he noted that clinical staging can provide predictive data on disease course and a patient’s capabilities.

For a Healio Psychiatry story on a new study in the Journal of the American Medical Association (JAMA) on how lifestyle may impact the genetic risk for dementia, Dr. Camargo said, “As more genetic risk factors are identified, many older adults may find themselves at higher genetic risk of dementia. It is thus reasonable for these individuals to ask, ‘Is there anything I can do to lower my risk?’”