

# The VOICE of FTD

SUMMER 2021

## Featured Study:

### *Genetics of Frontotemporal Degeneration (FTD) in Diverse Populations*

While recent advancements have been made about frontotemporal degeneration (FTD) through genetic research, existing data is often limited to patient groups of European ancestry. This creates a gap in knowledge on genetic factors specific to diverse populations. Identifying genetic factors contributing to disease helps us better understand the biology behind the disease and ultimately improves diagnosis and treatment.

Genetics of Frontotemporal Degeneration in Diverse Populations, a study at the University of Miami (UM), seeks to better understand the genetic factors contributing to FTD primarily in Hispanic populations.

“There is more data out there now by fellow colleagues looking at genetics in FTD patients of European ancestry,” said Dr. Karen Nuytemans, Research Assistant Professor, Human Genetics with the John P. Hussman Institute for Human Genomics and Dr. John T. Macdonald Foundation Department of Human Genetics at UM. “My part will be to contribute that same information but for the Hispanic or Caribbean-Hispanic population.”

Initially, Dr. Nuytemans oversaw a two-year study, funded by a grant from the [National Institutes of Health](#) (NIH), to look at how much genetic factors of FTD identified in patients of European ancestry are

contributing to disease in Hispanic patients. As the genetic background in the Hispanic population has contributions from European but also African and Amerindian ancestry, some overlapping but perhaps also separate factors are to be expected. The goal was to enroll 100 volunteers. However, study recruitment was significantly affected by the coronavirus pandemic.

“We made it to 40 (people) because a year and a half of the study was during corona,” the scientist said. “We were not allowed to go out to recruit, and patients did not want to see us either, which I very much understand.”

Unfortunately, with only 40 percent of the intended enrollment, she was not able to draw many conclusions. Initial analyses in this small group of patients of diverse populations (60% Hispanic, 40% African ancestry) identified a few variants previously identified in individuals of European ancestry in genes known to be implicated in neurodegenerative

Please note that [this glossary](#) is available to help you understand the scientific terms used in this article. Glossary terms are shown in bold the first time they appear.

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disorders. “The data just confirm more research on genetic factors in FTD overall, but specifically in diverse populations, is needed to identify those additional factors contributing to disease on all ancestral backgrounds,” said Dr. Nuytemans.

Starting July 1, 2021, Dr. Nuytemans will begin work on a four-year study that is an extension of the first one, but will focus on Hispanics. The new study, which goes by the same name as the initial one, is funded through a grant from the [Department of Defense](#) (DOD). It seeks to understand the genetic factors underlying disease (both known and new) in this population and thus help improve diagnosis. Understanding disease also ultimately leads to the development of better treatments.

“As different diverse populations have different genetic ancestries, it is important to include all ancestries in genetic research to fully understand the biology of disease in all patients, so that the above outcomes of improved diagnosis and treatments are applicable to all patients in all population groups.”

“Some (minorities) have been hard to reach or there were not a lot of research dollars,” Dr. Nuytemans said. “It’s finally happening that a lot more research effort and dollars are being put to it.”

## Study Details

This study hopes to enroll 120 FTD-diagnosed people who are Hispanic and as many as possible from other diverse populations.

To be eligible, participants must be 18 years or older; have a diagnosis of FTD, PPA, or semantic dementia; and identify as Hispanic.

Participation includes interviews for family and medical history as well as clinical assessments,

## GENETICS of FTD in DIVERSE POPULATIONS



which can be done remotely in one or two sessions (total time required is about 3 hours). There is also a one-time blood draw, which can be completed at the participant's home.

“Since we are just a genetic study, the time commitment from the patients is somewhat less as compared to, say, the ALLFTD study that typically is more time involved because they want patients to come to their facilities,” she noted. “We can do a lot over Zoom, and we can have someone from Quest (Diagnostics) go to the patient’s house to do a quick blood draw.”

The blood samples are sent to the university and processed for genotyping and sequencing. They are screened for the known FTD genes first to see if any of the typical genes and variants originally identified in patients of European ancestry occur in the Hispanic patients. Many times this happens because all Hispanics have some European ancestry, she said. However, there also are those who do not have variants of European ancestry.

“Both are interesting, but for new discovery, the ones without European variants might be more interesting,” she said. “We then do more in-depth sequencing and try to identify new variants and new genes that might tell us more about what is going on with the biology. This will help with diagnosis in diverse populations in

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the future if we can screen for these new ‘bad’ genes, not just the European ones.”

Dr. Nuytemans sees herself as an advocate in both rare diseases and minority populations and wants to make a difference by adding to the scientific knowledge.

“There are so many disorders and populations that have been overlooked,” including FTD, the scientist said. “A lot of research dollars and efforts have gone into (Alzheimer’s and Parkinson’s) diseases. A lot less common disorders like frontotemporal dementia and dementia with Lewy bodies have been overlooked. We don’t see enough patients with these diseases.”

Genetics provides additional knowledge about biology, diagnosis, and future therapies for these diseases.

“It’s interesting to me. I’m an immigrant (from Belgium to the United States), but I’m still non-Hispanic white and fit in the biggest population group in the U.S.,” she said. “Though both places are of European ancestry, we still see differences in genetic factors contributing to disease between them. This shows that we need to understand all diseases in all populations because we’re missing a good portion of information by not including all groups.”

Through DNA tests, people are looked at in terms of how they are genetically assembled. For example, Dr. Nuytemans said that Caribbean Hispanics tend to have more African ancestral background, whereas Hispanics in Peru and Mexico have more Amerindian ancestral background from Native Americans who lived in North or South America before Europeans arrived.

Caribbean Hispanics who have a higher African background are analyzed together, and those who have a higher Native American background are analyzed together.

“It depends on where their ancestors originally came from. Once we do the genetic tests, we can actually determine what kind of admixture they have,” she said. “and then we would analyze similar ancestries together to identify variants on those ancestries. Even for those of European ancestry, , we’re one big group, but we are a combination of many smaller groups together.”

For more information about the Genetics of FTD in Diverse Populations study, email the study coordinator, Anisley Martinez at [FTD-HIHG@miami.edu](mailto:FTD-HIHG@miami.edu), call her at 877-686-6444, or visit the [University of Miami Frontotemporal Dementia Research webpage](#).

[Read More About Dr. Nuytemans](#)

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