

Genetic mutation may have allowed early humans to migrate throughout Africa

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A genetic mutation that occurred thousands of years ago might be the answer to how early humans were able to move from central Africa and across the continent in what has been called "the great expansion," according to new research from Wake Forest Baptist Medical Center.

By analyzing <u>genetic sequence</u> variation patterns in different populations around the world, three teams of scientists from Wake Forest Baptist, Johns Hopkins University School of Medicine and the University of Washington School of Medicine, Seattle, demonstrated that a critical genetic variant arose in a key <u>gene cluster</u> on <u>chromosome 11</u>, known as the fatty acid desaturase cluster or FADS, more than 85,000 years ago. This variation would have allowed early humans to convert plant-based polyunsaturated fatty acids (PUFAs) to brain PUFAs necessary for increased brain size, complexity and function. The FADS cluster plays a critical role in determining how effectively medium-chain PUFAs found in plants are converted to the long-chain PUFAs found in the brain.

This research is published online today in **PLOS One**.

Archeological and <u>genetic studies</u> suggest that *homo sapiens* appeared approximately 180,000 years ago, but stayed in one location around bodies of water in central Africa for almost 100,000 years. Senior author Floyd H. "Ski" Chilton, Ph.D., professor of physiology and pharmacology and director of the Center for Botanical Lipids and Inflammatory Disease Prevention at Wake Forest Baptist, and others have hypothesized that this location was critical, in part, because early



humans needed large amounts of the long-chain PUFA docosahexaenoic acid (DHA), which is found in shellfish and fish, to support complex <u>brain function</u>.

"This may have kept early humans tethered to the water in central Africa where there was a constant food source of DHA," Chilton said. "There has been considerable debate on how early humans were able to obtain sufficient DHA necessary to maintain <u>brain size</u> and complexity. It's amazing to think we may have uncovered the region of genetic variation that arose about the time that early humans moved out of this central region in what has been called the 'great expansion.'"

Once this trait arose, the study shows that it was under intense selective pressure and thus rapidly spread throughout the population of the entire African continent. "The power of genetics continually impresses me, and I find it remarkable that we can make inferences about things that happened tens of thousands of years ago by studying patterns of genetic variation that exist in contemporary populations," said Joshua M. Akey, Ph.D., lead scientist at the University of Washington.

This conversion meant that <u>early humans</u> didn't have to rely on just one food source, fish, for brain growth and development. This may have been particularly important because the genetic variant arose before organized hunting and fishing could have provided more reliable sources of long-chain PUFAs, Akey said.

To investigate the evolutionary forces shaping patterns of variation in the FADS gene cluster in geographically diverse populations, the researchers analyzed 1,092 individuals representing 15 different human populations that were sequenced as part of the 1000 Genome Project and 1,043 individuals from 52 populations from the Human Genome Diversity Panel database. They focused on the FADS cluster because they knew those genes code for the enzymatic steps in long-chain PUFA synthesis



that are the least efficient.

Chilton said the findings were possible because of the collaboration of internationally recognized scientists from three distinct and diverse disciplines – fatty acid biochemistry (Wake Forest Baptist), statistical genetics (Johns Hopkins) and population genetics (University of Washington). This new information builds on Chilton's 2011 research findings published in BMC Genetics that showed how people of African descent have a much higher frequency of the gene variants that convert plant-based medium-chain omega-6 PUFAs found in cooking oils and processed foods to long-chain PUFAs that cause inflammation. Compared to Caucasians, African Americans in the United States have much higher rates of hypertension, type 2 diabetes, stroke, coronary heart disease and certain types of cancer. "The current observation provides another important clue as to why diverse racial and ethnic populations likely respond differently to the modern western diet," Chilton said.

Provided by Wake Forest University Baptist Medical Center

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