Genetic ancestry test beats self-reports in predicting bleeding stroke risk

24 January 2018

A genetic ancestry test more accurately identified patients at risk for bleeding stroke than traditional self-reports of race, cultural identity or ethnicity, according to preliminary research presented at the American Stroke Association's International Stroke Conference 2018, a world premier meeting dedicated to the science and treatment of cerebrovascular disease for researchers and clinicians.

Stroke risk can be lessened with lifestyle changes such as controlling high blood pressure and cholesterol; not smoking; being physically active; eating a healthy diet; and treating other heart and blood vessel diseases. Like many diseases, stroke risk also may be affected by inherited as well as environmental factors. An example of an inherited risk is a family history of stroke. Also, studies have shown that blacks are at greater risk of stroke than whites, which may result from higher rates of high blood pressure, obesity and diabetes.

"Treatment and prevention of risk factors represent the most crucial components of efforts to limit the burden of stroke," said study lead author Sandro Marini, M.D., post-doctoral research fellow in the Center for Genomic Medicine at Massachusetts General Hospital in Boston. "Accurate identification of patients at higher risk is the first step towards tailored prevention strategies."

Investigators reviewed data on 4,935 patients in the Ethnic/Racial Variations of Intracerebral Hemorrhage (ERICH) study, a large, multi-center U.S. project examining stroke due to blood vessel rupture and examined risk factors for a type of stroke called an intracerebral hemorrhage, which occurs when a ruptured blood vessel causes bleeding within the brain. Thirty-five percent of participants were black, 35 percent were white, and 30 percent were Hispanic. More than half were male, and they ranged in age from 49-79 years.

Compared to study participants' self-reports of race and ethnicity, a genetic ancestry test was more likely to identify patients with four known risk factors for stroke, including diabetes, high blood cholesterol levels, plaque buildup in the heart's arteries and irregular heartbeat. This was particularly true for black and Hispanics.

Thus, genetic ancestry markers may provide a more detailed assessment of risk levels compared to traditional risk factors alone.

"Genetic ancestry represents an accurate way to control for both genetic and environmental exposures that vary across races and ethnicities, in association with risk factors for intracerebral hemorrhage," Marini said. "Limiting our definitions of race and ethnicity to standard self-reports leaves out valuable information that could be used to better predict risk of at least some complex diseases."

Human DNA is nearly identical, with only slight variations among different populations, Marini explained. Researchers tested for these slight variations by looking for 15 changes in the sequence of DNA molecules. They then linked the test results to geographic populations that possess four known risk factors for stroke and used statistical methods to calculate participants' stroke risk.

The study may have been limited by patients reporting incorrect information about their diagnosis of vascular risk factors. In addition, the study focused only on blacks, whites and Hispanics so its findings cannot be applied to other groups. And, for diseases other than stroke, it is possible that self-reported race and ethnicity may be more accurate than genetic testing in predicting risks.

In the United States, stroke is the fifth-leading cause of death and a major cause of long-term disability. It occurs when a damaged blood vessel to the brain becomes blocked or ruptures,

preventing blood and oxygen from reaching the brain. Sudden warning signs that require immediate medical treatment include numbness in the face, arms, or legs; confusion; vision impairment; dizziness or trouble walking; and severe headache without apparent cause.

Provided by American Heart Association