

Researchers uncover genetic variants linked to blood pressure in African-Americans

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A team led by researchers from the National Institutes of Health today reported the discovery of five genetic variants related to blood pressure in African-Americans, findings that may provide new clues to treating and preventing hypertension. The effort marks the first time that a relatively new research approach, called a genome-wide association study, has focused on blood pressure and hypertension in an African-American population.

Hypertension, or chronic high [blood pressure](#), underlies an array of life-threatening conditions, including heart disease, stroke and [kidney disease](#). Diet, physical activity and obesity all contribute to risk of hypertension, but researchers also think genetics plays an important role.

About one-third of U.S. adults suffer from hypertension. The burden is considerably greater in the African-American community, in which the condition affects 39 percent of men and 43 percent of women.

"This work underscores the value of using genomic tools to untangle the complex [genetic factors](#) that influence the risk for hypertension and other common diseases," said Eric Green, M.D., Ph.D., scientific director for the National Human Genome Research Institute (NHGRI), part of NIH. "We hope these findings eventually will translate into better ways of helping the millions of African-Americans at risk for hypertension, as well as improved treatment options for other populations."

In addition to NHGRI researchers, scientists from the Coriell Institute for Medical Research in Camden, N.J.; Boston University; and Howard University, in Washington, D.C., collaborated on the study, which was published in the July 17 online issue of [PLoS Genetics](#).

To produce their findings, researchers analyzed DNA samples from 1,017 participants in the Howard University Family Study, a multigenerational study of families from the Washington, D.C., metropolitan area who identified themselves as African-American. Half of the volunteers had hypertension and half did not. To see if there were any genetic differences between the two groups, researchers scanned the volunteers' DNA, or genomes, analyzing more than 800,000 genetic markers called single-nucleotide polymorphisms (SNPs).

The researchers found five genetic variants significantly more often in people with hypertension than in those without the condition. The variants were associated with high systolic blood pressure, but not with diastolic blood pressure or combined systolic/diastolic blood pressure.

Blood pressure is measured in millimeters of mercury (mm Hg), and expressed with two numbers; for example, 120/80 mm Hg. The first number (systolic pressure) is the pressure when the heart beats while pumping blood. The second number (diastolic pressure) is the pressure in large arteries when the heart is at rest between beats.

"This is the first genome-wide association study for hypertension and blood pressure solely focused on a population with majority African ancestry," said the study's senior author, Charles Rotimi, Ph.D., NHGRI senior investigator and director of the trans-NIH Center for Research on Genomics and Global Health (CRGGH). "Although the effect of each individual genetic variant was modest, our findings extend the scope of what is known generally about the genetics of human hypertension."

In a genome-wide association study, researchers identify strategically selected markers of [genetic variation](#). If disease status differs for individuals with certain genetic variants, this indicates that something in that chromosomal neighborhood likely influences the disease. Variants detected using this approach can accurately point to the region of the genome involved, but may not themselves directly influence the trait.

In May, two major international studies used the genome-wide association approach to identify 13 genetic variants associated with blood pressure and hypertension in people with primarily European and South Asian ancestry. While each variant was associated with only a slight increase in blood pressure, that work found that the more variants an individual had, the greater his or her risk of hypertension. Two genes identified by one of those studies were also associated with blood pressure in the new study.

In their pioneering study of African-Americans, Dr. Rotimi and his colleagues found that all of the five genetic variants associated with blood pressure were located in or near genes that code for proteins thought to be biologically important in hypertension and blood pressure. Previous research had implicated two of those genes in blood pressure regulation, and additional analyses by Dr. Rotimi's group revealed that all of the variants are likely involved in biological pathways and networks related to blood pressure and hypertension.

An existing class of anti-hypertension drugs, called calcium channel blockers, already targets one of the genes, CACNA1H. However, the additional genes may point to new avenues for treatment and prevention.

To follow up and expand upon their findings in African-Americans, the researchers scanned DNA from 980 West Africans with and without hypertension. The work confirmed that some of the genetic variants detected in African-Americans were also associated with blood pressure

in West Africans. "The Western African population is of particular significance since it is the ancestral population of many African-Americans," said lead author Adebowale Adeyemo, M.D., CRGGH staff scientist.

For more information about [hypertension](#), visit www.nhlbi.nih.gov/health/dci/D.../Hbp/HBP_WhatIs.html.

To learn more about the genome-wide association approach, visit www.genome.gov/20019523.

Source: NIH/National Human Genome Research Institute

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