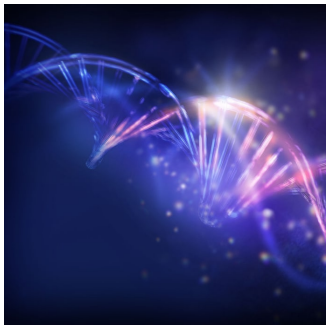
HealthManagement
Promoting Management and Leadership

Genetic Variant Among Blacks Contributes to Cardiovascular Disease Burden



According to a new study by researchers at Brigham and Women's Hospital and Duke University School of Medicine, a genetic variant carried by 3-4% of self-identified Black Americans increases the risk of heart failure and death, leading to a significant decrease in population longevity.

While the association between the V142I variant and heart failure has been previously described, precise estimates of how the variant increases risk were unclear until now. Considering approximately 48 million Americans self-identify as Black, an estimated 1.5 million across the lifespan carry this variant. However, since the variant's effects typically manifest after age 50, the researchers focused on the risk among Black Americans in mid-to-late life.

The study reveals that individuals with the V142I transthyretin variant face a significantly increased risk of heart failure beginning in their 60s and a heightened risk of death starting in their 70s. On average, carriers of the variant die 2 to 2.5 years earlier than expected. With nearly half a million Black American carriers over age 50, researchers estimate that approximately one million years of life will be lost due to this variant among currently living Black individuals in mid-to-late life. The findings are published in JAMA.

Study researchers examined data from 23,338 self-reported Black individuals, 754 (3.23%) of whom carried the V142I genetic variant.

The study found that V142I increased the risk of heart failure hospitalisation by age 63 and the risk of death by age 72. The variant's contribution to heart failure risk increased substantially with age and was not influenced by other known risk factors like diabetes and hypertension. The research also showed that female and male carriers of the variant were equally at risk, contrary to some previous studies that suggested men were more affected, indicating that women are likely underdiagnosed. The researchers estimated that individual carriers with the V142I variant live 2-2.5 years less than expected.

The researchers believe these data will inform clinicians and patients regarding risk when these genetic findings are known through family screening, medical, or genetic testing.

The V142I variant causes the transthyretin protein in the blood to misfold, leading to deposits of abnormal amyloid protein in the heart and other body parts. In the heart, these deposits cause the muscle to become thick and stiff, a condition known as cardiac amyloidosis, which can ultimately lead to heart failure. Several therapies have been developed to treat cardiac amyloidosis, including treatments that prevent the protein from misfolding, reduce the protein amount, remove the protein, and a gene-editing therapy currently undergoing clinical trials. Understanding the epidemiology of V142I and cardiac amyloidosis could help physicians connect patients with appropriate treatments at the right age.

Future studies should investigate why some carriers of the V142I variant develop cardiac amyloidosis while others do not.

Source: Brigham and Women's Hospital

Image Credit: iStock

Published on : Tue, 14 May 2024