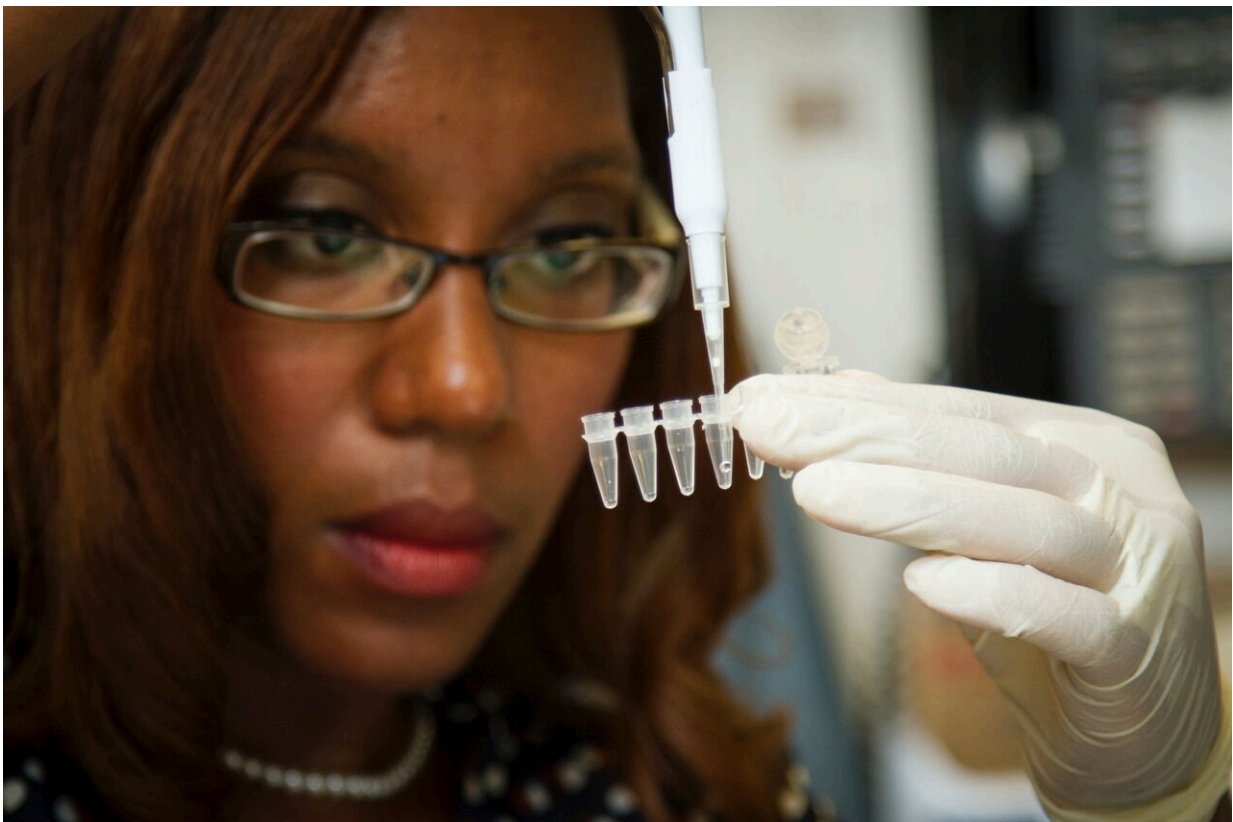


# Study offers first glimpse of how many suffer from previously unknown VEXAS syndrome

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About 13,200 men and another 2,300 women in the United States over age 50 are estimated to have VEXAS syndrome, according to a new

study. Long considered a mystery illness until its genetic basis was identified in 2020, the latest findings, led by researchers at NYU Grossman School of Medicine, offer the first indication of how common the illness is domestically.

Although a rare disorder, the [syndrome](#) carries a [high mortality rate](#), with up to half of people, mostly men, dying within five years of diagnosis. The syndrome most often involves unexplained fevers and low blood oxygen levels in people diagnosed with other diseases, such as rheumatoid arthritis, lupus, and [blood cancer](#). Some of the symptoms have been linked to an overactive immune system, which can cause inflammation and classifies the syndrome as an autoimmune condition.

Researchers say they hope their findings will raise awareness of the disorder among physicians, particularly because high-dose steroids, JANUS kinase inhibitors, and bone marrow transplantation have proven effective in controlling some symptoms.

"Now that we know VEXAS syndrome is more common than many other types of rheumatologic conditions, physicians need to add this condition to their list of potential diagnoses when confronted by patients with persistent and unexplained inflammation and low blood cell counts, or anemia," says geneticist and study lead investigator David Beck, MD, Ph.D.

Beck, an assistant professor in the Department of Medicine and the Department of Biochemistry and Molecular Pharmacology at NYU Langone Health, also led the federal research team that initially identified the shared UBA1 mutation among VEXAS patients.

In the new study, published in the *Journal of the American Medical Association (JAMA)*, researchers analyzed the [electronic health records](#) of 163,096 mostly white men and women in Pennsylvania who agreed to

have their blood DNA screened for signs of genetic disease. Twelve were found to have the UBA1 mutation, with all experiencing VEXAS symptoms.

Statistically, this corresponded to one in 4,269 American men over age 50 and one in 26,238 women over age 50 having or are likely to develop the syndrome. This, researchers say, is a higher prevalence figure than many other inflammatory conditions, including vasculitis and myeloid dysplasia syndrome.

"Our study offers the first glimpse of just how common VEXAS syndrome is in the United States, particularly among men, who also happen to be the most to die from it," says Beck, who is leading several clinical research efforts into VEXAS syndrome at NYU Langone's Center for Human Genetics and Genomics.

Previous research, led by Beck, traced the origins of the syndrome to a mutation, or change in the letter code that makes up DNA, in the gene UBA1 (short for ubiquitin-like modifier activating enzyme 1.) The enzyme usually assists in protein breakdown.

VEXAS stands for many of its biological characteristics: vacuoles in [blood cells](#), the E1 enzyme, X-linked, autoinflammatory, and somatic.

For the study, researchers analyzed the electronic medical records of adult patients who volunteered to participate in the Geisinger MyCode Community Health Initiative. The program has been collecting data for more than 25 years from patients in Geisinger's 10-plus hospitals in Central and Northeastern Pennsylvania. Almost all study participants who agreed to have their blood DNA tested were white; half were over the age of 60.

Beck says the team next plans to analyze patient records in more racially

diverse groups, especially among those with higher rates of rheumatologic and blood disease, to gain a more precise picture of who is most at risk of VEXAS syndrome. They also plan to look for additional genetic causes, test new therapies for the syndrome, and develop a simple blood test for UBA1 to make it easier to diagnose.

**More information:** David B. Beck et al, Estimated prevalence and clinical manifestations of UBA1 variants associated with 2 VEXAS syndrome in a clinical population, *JAMA* (2023). [DOI: 10.1001/jama.2022.24836](https://doi.org/10.1001/jama.2022.24836). [jamanetwork.com/journals/jama/... cle-abstract/2800661](https://jamanetwork.com/journals/jama/abstract/2800661)

Provided by NYU Langone Health

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