One of the Largest and Most Diverse Studies on Sickle Cell Trait and Blood Clots Reveals Findings That Impact All Populations

September 12, 2024

23andMe, National Human Genome Research Institute, and Johns Hopkins University conducted collaborative research that studied sickle cell trait in a diverse population

SUNNYVALE, Calif., Sept. 12, 2024 (GLOBE NEWSWIRE) -- 23andMe Holding Co. (Nasdaq: ME) (23andMe), a leading genetic health and biopharmaceutical company, in collaboration with lead researchers at National Human Genome Research Institute, part of the National Institutes of Health (NIH), and Johns Hopkins University, conducted one of the largest and most diverse studies on sickle cell trait (SCT). Many prior SCT research studies have only focused on Black/African American populations. This collaborative research leveraged 23andMe's diverse cohort of research-consented participants. The researchers studied the association of sickle cell trait with venous thromboembolism (VTE) (blood clots) and compared this degree of risk to factor V Leiden (FVL). The results from the study were published today in the journal *Blood Advances*.

Sickle cell trait and sickle cell disease

Sickle cell disease is a global public health issue.

- Individuals with sickle cell disease (SCD) have two genes that cause the production of sickle hemoglobin, and these patients can have severe pain and other organ complications
- Individuals with sickle cell trait (SCT) carry only one gene that causes the production of sickle hemoglobin, and these individuals are generally healthy. However, SCT can be a risk factor for select health outcomes such as blood clots.

One hundred million people worldwide have sickle cell trait. In the United States, three million people have sickle cell trait, and it disproportionately affects the Black community. However, sickle cell trait and sickle cell disease aren't just a Black issue. Individuals in all communities can have sickle cell trait.

This study found that sickle cell trait is a modest risk factor for blood clots across populations. "This study uniquely demonstrates that sickle cell trait's association with blood clots extends across diverse genetic backgrounds," said Keng-Han Lin, Ph.D., a Senior Scientist at 23andMe and a co-author of the paper. "It highlights how comprehensive genetic research can reveal health risks relevant to all communities."

Collaborative and diverse sickle cell trait study

Most prior sickle cell trait studies have been limited to the Black community because many assume SCT occurs only in a specific race or population. Combining those assumptions with systemic racism can lead to bias.

"The power of the 23andMe platform is that it enables research that's inclusive of communities historically underrepresented in genetics research at an unprecedented scale. This diversity can help uncover novel biology and can also dispel misconceptions," said Anjali Shastri, Ph.D., a Principal Program Manager at 23andMe who leads research equity programs and advocacy partnerships for research and is a co-author of the study.

With this collaborative study, researchers at National Human Genome Research Institute (NHGRI), part of the National Institutes of Health (NIH), Johns Hopkins University (JHU), and scientists at 23andMe sought to better understand the connection between sickle cell trait and blood clots. The study was one of the largest sickle cell trait studies to date, leveraging 4.2 million 23andMe research-consented participants, which included 19,000 with SCT. The researchers studied the associations with venous thromboembolism (VTE). To contextualize SCT and VTE, the researchers also studied the association of factor V Leiden and VTE.

The study also looked at different types of blood clots: deep vein thrombosis (DVT), which forms in a deep vein like in the legs, and pulmonary emboli (PE), which are caused by blood clots that travel to the lungs.

Sickle cell trait study results

Study results validated the association of sickle cell trait with blood clots. However, the study found that individuals with sickle cell trait are at a lower risk for VTE than carriers of factor V Leiden. The study also validated the association between having SCT and with developing pulmonary emboli (PE), and it similarly validated that sickle cell trait was not associated with developing deep vein thrombosis (DVT). This pattern of blood clots was the opposite of that found in FVL. In individuals with FVL, the risk of DVT was greater than the risk of PE.

"This study suggests a unique mechanism of blood clotting in people with sickle cell trait," said Rakhi Naik, M.D., M.H.S., Clinical Director for the Division of Hematology at Johns Hopkins University, who co-led the study. "Knowing the risks of blood clots in people with sickle cell trait is important for situations such as surgeries or hospitalizations, which add to the risk of developing serious blood clots."

In addition to suggesting novel biology for individuals with sickle cell trait in the development of PE, this study may inform clinical care. "This study provides valuable information to clinicians counseling individuals with sickle cell trait," said Julie Granka, Ph.D., a Principal Scientist at 23andMe and a co-author of the paper. "Our study results are applicable across ancestries, and they show valuable comparisons to Factor V Leiden, where there is more established clinical guidance. Notably, the risk of VTE for individuals with SCT is lower than for those with Factor V Leiden."

About 23andMe

23andMe is a genetics-led consumer healthcare and biopharmaceutical company empowering a healthier future. For more information, please visit www.23andMe.com.

Forward-Looking Statements

This press release contains forward-looking statements within the meaning of Section 27A of the Securities Act of 1933, as amended, and Section 21E of the Securities Exchange Act of 1934, as amended, including. All statements, other than statements of historical fact, included or incorporated in this press release are forward-looking statements. The words "believes," "anticipates," "estimates," "plans," "expects," "intends," "may," "could," "should," "potential," "likely," "projects," “predicts,” "continue," "will," “schedule,” and "would" or, in each case, their negative or other variations or comparable terminology, are intended to identify forward-looking statements, although not all forward-looking statements contain these identifying words. These forward-looking statements are predictions based on 23andMe’s current expectations and projections about future events and various assumptions. 23andMe cannot guarantee that it will actually achieve the plans, intentions, or expectations disclosed in its forward-looking statements and you should not place undue reliance on 23andMe’s forward-looking statements. These forward-looking statements involve a number of risks, uncertainties (many of which are beyond the control of 23andMe), or other assumptions that may cause actual results or performance to differ materially from those expressed or implied by these forward-looking statements. The forward-looking statements contained herein are also subject generally to other risks and uncertainties that are described from time to time in the Company’s filings with the Securities and Exchange Commission, including under Item 1A, “Risk Factors” in the Company’s most recent Annual Report on Form 10-K, as filed with the Securities and Exchange Commission, and as revised and updated by our Quarterly Reports on Form 10-Q and Current Reports on Form 8-K. The statements made herein are made as of the date of this press release and, except as may be required by law, 23andMe undertakes no obligation to update them, whether as a result of new information, developments, or otherwise.

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