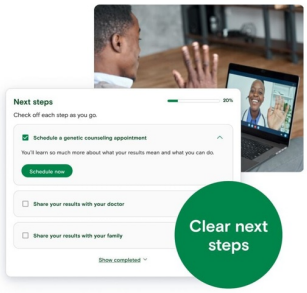

Quest Diagnostics Genetic Insights Test Results Sickle Cell Anemia User Guide

Genetic Insights: quick referenceguide for healthcare providers

This guide is intended to facilitate a discussion between a provider and their patient.

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Genetic Insights test results: Sickle cell anemia

Key results
The c.20A>T (p.Glu7Val) DNA variant was found in each of the 2 copies of the HBB gene; this result is associated with sickle cell anemia.

Next steps

Clinical recommendations	Resources
Genetic Insights is a screening test and n ot intended to diagnose whether an indiv idual has sickle cell anemia. Consider a referral to a specialist for diagnostic eval uation of sickle cell anemia, if not previo usly completed.	The American Society of Hematology maintains a searchable list of specialists found at: www.hematology.org/Patients/FAH.aspx **Have questions?** Call 1.866.GENE.INFO (1.866.436.3463) to speak to a specialized Quest genetic counselor or geneticist available to healthcare provid ers to discuss test selection and results.
Refer your patient to a genetic counselor.	Your patient can schedule a 1-on-1 remote genetic counseling sessi on through their online Genetic dashboard at no additional cost.
Genetic counselors can provide counseli ng on the implications of this test result and next steps for your patient.	To find a genetic counselor practicing in your patient's area for an in -person session, please visit FindAGeneticCounselor.NSGC.org

Patient conversation starters:
Patient conversation starters summarize the preceding information in plain language to support meaningful conversations between you and your patient.
FindAGeneticCounselor.NSGC.org

Your Genetic Insights test is a genetic screening test. That means it is not meant to tell you for sure if you have a certain health condition.
If you haven't already, it's important to talk to a specialist about additional testing for sickle cell anemia. It's also important that you talk with a genetic counselor.
Genetic counselors are experts in genetics and can help you understand this result and potential next steps.
You can access a genetic counselor through your online Genetic Insights dashboard at no additional cost to you, and we can discuss a referral to a local genetic counselor.

Additional resources
The following advocacy groups have additional information and resources about sickle cell anemia:

ckle Cell Disease Coalition: SCDCoalition.org
Sickle Cell Disease Association of America: SickleCellDisease.org

What is sickle cell anemia?

Sickle cell anemia (SCA) is the most common and severe form of sickle cell disease (SCD).[1] SCD is a group of inherited blood disorders that affect the function of the red blood cells, leading to severe anemia and pain crises.

Signs of SCA such as jaundice, fatigue, and dactylitis typically appear in the first year of life. Additional complications can include acute pain crises, chronic pain, severe anemia, infections, and stroke.[2]
Diagnostic evaluation of SCA typically includes analysis of hemoglobin in addition to genetic testing. Treatment and management for individuals confirmed to have SCA is complex and best coordinated by a specialty care team. Management is individualized and may include medications, blood transfusions, and in some cases stem cell transplant. Expert clinical care guidelines are available.[1]
SCA is the result of a DNA variant in each of the 2 copies of the HBB gene.[1] People with a variant linked to SCA in only 1 copy of HBB are considered carriers (also called sickle cell trait) and do not have the condition.

What this result means for family members

SCA is an inherited condition. Biological parents of people with SCA are most likely carriers. Full siblings have a 1 in 4 chance to also have SCA, a 1 in 2 chance to be carriers, and a 1 in 4 chance to neither be a carrier nor have the condition. Children of people with SCA are carriers and would only be at risk of having SCA if the other biological parent (or sperm/egg donor) is also a carrier.
People with SCA should talk with their reproductive partners and at-risk family members. Comprehensive evalution and carrier testing may be indicated, especially for family planning.
Note that other β-globin disorders (such as β-thalassemia) can interact with an HBB SCA variant to cause clinically significant disease. To determine the risk to children, it is recommended that partners of people with SCA be tested with a thalassemia panel that includes hemoglobin electrophoresis, CBC and reticulocyte count, and a measure of iron status to screen for carrier status for both sickle cell trait and other β-globin disorders. This test only looked for the HBB DNA variant associated with SCA.
In people with sickle cell anemia, testing for other family members may help inform their risks. Therefore, it is strongly recommended that individuals share these results with their biological relatives and reproductive partners.

Patient conversation starters:
Sickle cell anemia is the most common and severe form of sickle cell disease.
Signs of sickle cell anemia generally appear in the first year of life, but not always.
People with sickle cell anemia are cared for by a team of doctors and other healthcare providers who are experts in the condition.

Sickle cell anemia runs in families. That means the DNA variants linked to it can be passed down from parents to their children.
For people who have sickle cell anemia, their biological parents are most likely carriers. Their siblings may have sickle cell anemia, may be a carrier, or may not have any DNA variants linked to sickle cell anemia.
Talking to your family members and reproductive partners about your sickle cell anemia status is important so they can decide if they want to have genetic testing or other testing for sickle cell anemia or related blood disorders.

References

1. Bender, MA. Sickle Cell Disease. September 15, 2003. Updated November 22, 2022. In: Adam MP, Ardinger HH, Pagon RA, et al, editors. GeneReviews® . Seattle (WA): University of Washington; 19932023. https://www.ncbi.nlm.nih.gov/books/NBK1377
2. National Heart, Lung, and Blood Institute. Sickle Cell Disease. Accessed December 15, 2022. www.nhlbi.nih.gov/health-topics/sickle-cell-diseases

Documents / Resources

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