Genetic Insights: quick reference guide for healthcare providers

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

Genetic Insights test results: **Sickle cell disease - carrier**

Key results

The DNA variant c.20A>T (p.Glu7Val) in 1 copy of the *HBB* gene is associated with being a carrier of sickle cell anemia. Carriers do not have the condition. Their children are at higher risk for the condition if the other parent is also a carrier of sickle cell anemia or a related blood disorder.

Next steps

Clinical recommendations	Resources
Genetic Insights is a screening test and not intended for diagnosis or to replace routine carrier screening for family planning or during pregnancy. *Follow-up genetic testing should be performed in a clinical setting, especially if biological children are planned. Comprehensive hemoglobinopathy and/or thalassemia testing in a clinical setting should be considered for the individual and reproductive partners/ gamete donors.*	**Ready to order?** Check with your institution and/or patient's insurance about the preferred testing laboratory. Blueprint Genetics® offers sickle cell anemia genetic testing. To confirm this result you can order a confirmation test here: Blueprint Genetics **Blueprint Genetics/TVT** **Have questions?** Call **1.866.GENE.INFO** (1.866.436.3463) to speak to a specialized Quest genetic counselor or geneticist available to healthcare providers to discuss test selection and results.
Refer your patient to a genetic counselor.	Your patient can schedule a 1-on-1 remote genetic counseling session through their online Genetic dashboard at no additional cost.
Genetic counselors can provide counseling 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 screening test. The next step is to have your result confirmed with a second genetic test, especially if you are planning to have children.

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:

Sickle Cell Disease Coalition: SCDcoalition.org

Sickle Cell Disease Association of America: SickleCellDisease.org

Visit QuestDiagnostics.com/Genetic-Health-Screening for more information about this test.

What is sickle cell anemia?

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

SCA is the result of having 2 DNA variants associated with SCA, 1 in each copy of the *HBB* gene.[1] The *HBB* gene provides instructions for the protein beta-globin, a subunit of hemoglobin.

People with a DNA variant associated with SCA in 1 copy of *HBB* are considered carriers (also called sickle cell trait) and do not have the condition. Rarely, SCA carriers have some symptoms.[1]

In people with SCA, treatments may include medications, blood transfusions, and in some cases, a bone marrow transplant.[1]

Patient conversation starters:

Sickle cell disease (SCD) affects the blood. The most common and severe form of SCD is called sickle cell anemia.

To have sickle cell anemia, a person has to inherit the DNA variant in both copies of the *HBB* gene, 1 from each parent. A person who has the variant in only 1 copy of the *HBB* gene will not have sickle cell anemia. They have sickle cell trait. This means they are carriers of the variant and can pass it on to their children.

It is rare, but a carrier could have some symptoms of the condition.

What this result means for family members

Sickle cell anemia is an inherited condition. If both biological parents are carriers, each child has a 1 in 4 chance of having sickle cell anemia.

If only 1 parent is a carrier, each child has a 1 in 2 chance of being a carrier (and a 1 in 2 chance that they will not be a carrier). Parents and siblings of sickle cell anemia carriers may also be carriers.

Carriers should discuss their carrier status with their reproductive partner, especially if planning to have children, as well as with other family members who may also be carriers.

Non–sickle cell anemia beta-globin disorders (such as beta-thalassemia) can interact with the sickle cell anemia *HBB* variant to cause clinically significant disease in individuals.[1] Therefore, for the purposes of determining risk to children, it is recommended that partners or gamete donors of *HBB* carriers 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 beta-globin disorders. This test only looked for the *HBB* DNA variant associated with SCA.

Patient conversation starters:

Sickle cell anemia runs in families. That means the DNA variants linked to it can be passed down from parents to their children. One of your biological parents is likely a sickle cell anemia carrier.

Each of your siblings has a 1 in 2 (50%) chance of also being a carrier. Your biological children's chance of being a sickle cell anemia carrier or having sickle cell anemia depends on if their other parent is a carrier.

Sharing these results with your family members is important so family members can decide if they want to have genetic or other testing.

In people with a confirmed DNA variant, testing for family members may help inform their risks. Therefore, it is recommended that individuals share these results with their biological relatives and reproductive partners.

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; 1993-2023. https://www.ncbi.nlm.nih.gov/books/NBK1377
2. The American College of Obstetricians and Gynecologists Committee Opinion. Carrier Screening for Genetic Conditions. Number 691, March 2017 (Reaffirmed 2023). https://www.acog.org/Clinical-Guidance-and-Publications/Committee-Opinions/Committee-on-Genetics/Carrier-Screening-for-Genetic-Conditions