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RESEARCH FEATURE

Questions and answers about sickle cell trait

September 22, 2010

Questions

What is sickle cell trait?

Sickle cell trait occurs when a person carries a single copy of the sickle globin gene inherited from one parent along with a normal globin gene from the other parent. The globin genes direct red blood cells in the bone marrow to produce hemoglobin, a protein that carries oxygen in red blood cells throughout the body. In most cases, people living with sickle cell trait experience no symptoms and lead normal lives. Because some persons with sickle cell trait have complications from the condition, research is needed to better understand when and how sickle cell trait might affect a person's health. About 2.5 million people in the United States live with sickle cell trait. Sickle cell trait is different from sickle cell disease, also known as sickle cell anemia.

Return to "Questions"

What is the difference between sickle cell trait and sickle cell disease?

People with sickle cell trait carry only one copy of the altered hemoglobin gene and rarely have any clinical symptoms related to the disease. In contrast, people with sickle cell disease carry two copies of the altered hemoglobin gene. With two copies of the altered gene, the red blood cells are destroyed rapidly and patients have chronic, severe anemia, or low hemoglobin levels. Red blood cells become misshapen, many of which take the "C" or sickle shape that gives the disease its name. Without proper treatment, a person with sickle cell disease can develop recurrent episodes of pain and may have life-threatening complications, including damage to organs such as brain, bones, lungs, kidneys, liver and heart. The disease affects between 70,000 and 100,000 Americans and is most common in people of African, Middle Eastern, Mediterranean, Central and South American and Asian Indian origin or descent.

Return to "Questions"

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What does it mean if someone has sickle cell trait?

Most people who have sickle cell trait will never experience any medical complications. However, in rare instances, some people who have sickle cell trait can experience medical complications when performing intense physical activity. Several of the risks to persons with sickle cell trait occur when they engage in high intensity physical activity or are active at higher elevations such as mountains or unpressurized airplanes. Persons with sickle cell trait occasionally experience damage to their kidneys from sickling in sections of the kidney. People with sickle cell trait should be aware of their condition for family planning purposes because they can pass the gene onto their children. If both parents have sickle cell trait, there is a greater chance that one or more of their children will be born with sickle cell disease.

Return to "Questions"

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What research is being done to better understand sickle cell trait?

The National Heart, Lung, and Blood Institute (NHLBI) is currently defining which research questions need to be answered about sickle cell trait. In June 2010, the NHLBI and the Advisory Committee of Blood Safety and Availability in the Office of Public Health and Science, U.S. Department of Health and Human Services, held a two-day scientific workshop to discuss the current evidence and to examine the ethical, legal, social, and public health impacts of the sickle cell trait.

There is growing interest in how sickle cell trait can affect a person's health and well-being. For example, several student-athletes with sickle cell trait have died following intense training sessions over the last decade. The magnitude of risks for such events, and how to best prevent and manage them are not known. In addition to its on-going research efforts in sickle cell disease, the NHLBI is currently determining what types of research is needed to better understand these issues.

Return to "Questions"

Source: National Heart, Lung, and Blood Institute (Sept. 2010)

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