

What doctors want patients to know about sickle cell disease

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Sickle cell disease—the most common type is known as sickle cell anemia—is a genetic blood disorder that affects millions worldwide and continues to pose health challenges for those who live with it.

With its roots deeply embedded in the molecular structure of hemoglobin—a protein in red blood cells that carries oxygen—sickle cell disease causes the cells to assume a rigid, sickle-like shape, leading to a cascade of complications. From debilitating pain crises to organ damage and heightened susceptibility to infections, sickle cell anemia takes a toll on patients' health and well-being.

The AMA's What Doctors Want Patients to Know™ series provides physicians with a platform to share what they want patients to understand about today's health care headlines.

In this installment, two physicians took time to discuss what patients need to know about [sickle cell disease](#). These AMA members are:

- Chancellor Donald, MD, a hematologist and medical oncologist at Tulane University in New Orleans who cares for patients with sickle cell anemia and serves as a delegate for the American Society of Hematology in the AMA House of Delegates.
- Sam Milanovich, MD, a pediatric hematologist and oncologist at Sanford Health in Fargo, North Dakota. Sanford Health is a member of the [AMA Health System Program](#), which provides enterprise solutions to equip leadership, physicians and care teams with resources to help drive the future of medicine.

Blood vessels are blocked

“To talk about sickle cell anemia itself, the red blood cell is affected. The red blood cell carries oxygen to deliver it to the different tissues,” Dr. Donald said. “So, if we think about the heart pumping blood, it pumps it through the huge aorta and then the aorta branches off and feeds different arteries throughout the body.

“These arteries get progressively smaller and become capillaries where they can deliver oxygen to the tissues, and then those capillaries start to get bigger again and become veins and return to the heart. So, that’s the complete cycle of blood flow,” he added. “What happens with sickle cell disease is that instead of those red blood cells being able to traverse all of those arteries and then down the capillaries and then back to the heart—because they are misshapen, they can stick to the walls of the vessels and cause a blocking of the vessels.”

“This leads to an impaired delivery of oxygen and then that leads to what we call tissue ischemia. That gives the complication of pain and gives the complication of organ damage as well,” Dr. Donald said.

Sickle cell anemia is inherited

“It’s a genetic condition, so it’s something you’re born with,” said Dr. Milanovich, noting “it’s what’s called an autosomal recessive condition. So that means both of your parents have to be carriers. And if both parents are carriers, there is a 25% chance that a child will have sickle cell disease.”

“It is classically in persons of African descent,” Dr. Donald said, noting “it affects millions of people worldwide. It is the most inherited persistent hemoglobinopathy. With that being said, there are about 100,000 persons in the United States living with sickle cell disease. ... And there are 300,000 people born with sickle cell disease worldwide per year.”

In addition to Black patients, sickle cell disease can affect people of Mediterranean and Latin American heritage, Dr. Donald noted.

“Out of every 365 births to African American parents, one of those children would be affected by sickle cell disease,” Dr. Donald said, adding that there are “millions of persons living with this disease worldwide.”

All newborns are screened

“We screen all babies born in the United States for sickle cell disease,” Dr. Milanovich said. “We’re identifying babies right when they’re born and we’re getting them connected with their pediatrician and with a pediatric hematologist who has experience caring for sickle cell disease so that they can start the appropriate preventative measures right away.”

Pain can occur all over the body

“People with sickle cell disease are at risk for a variety of complications because of the sickling of the red blood cells. The most common symptom is severe episodes of pain,” Dr. Milanovich said. “This can happen anywhere in their body and can be debilitating.

“Sometimes these pain episodes may last for days or weeks, sometimes requiring people to spend many days in the hospital for pain medications,” he added.

“When someone has a painful episode, it’s often localized to different areas of the body, the knees, the back, the shoulders, the chest, rather than just a diffuse aching of the entire body, which can also occur in some people,” Dr. Donald said.

It can cause problems with any organ

“Besides the pain episodes, sickle cell disease can cause problems with just about any other organ,” Dr. Milanovich said, noting “if you have sickle cell disease, you’re at increased risk of getting a stroke even in young children.”

Additionally, “you can have sickling of the blood cause damage to the lungs,” he said. “It can also cause damage to the kidneys and the eyes over time. And that also damages the spleen, which is important for your body to fight infection.”

Young adults must continue getting care

“Painful episodes can occur at a very early age, but what happens is that we have the effects of those painful episodes,” Dr. Donald said. “The infarction or the blocking of oxygen delivery to different tissues and organ dysfunction ... creates chronic problems on top of having acute pain issues. So, it’s something that continuously builds.”

And those painful episodes continue into adulthood, which is why it is important for young adults to continue receiving care for sickle cell disease after transitioning from their pediatrician.

Luckily “there are programs universally established within communities that allow for a smooth transition from that pediatric side to the adult side,” he said.

Keep warm and stay hydrated

“When the winter gets really cold a child may be playing at recess and that exposure to the cold can accelerate sickling in the blood and lead to a pain crisis,” Dr. Milanovich said. “I’ve seen kids go swimming in the summertime and when you get cold from coming out of the pool— that may trigger a sickle cell crisis.”

Also, “as soon as possible, try and stay hydrated. If your blood is dehydrated, it may make the sickling worse,” he said. “If it’s mild, we can tell people to drink plenty of fluids and take some oral pain medicines—things like Tylenol or ibuprofen—but for more severe episodes, patients will need to come to the emergency department for intravenous fluids and pain medications.”

“What we want to do is to prevent the complications from sickle cell disease or the sickling of these red blood cells,” Dr. Donald said. “Staying hydrated is key,” which means [drinking eight to 10 glasses of water per day](#).

“Also, if someone knows that becoming cold is a trigger, we ask them to stay warm and bundle up,” he added.

Stay on top of recommended vaccines

“Because certain infections can also not only occur at an increased rate in people with sickle cell disease, once that infection happens, it can be more severe,” Dr. Donald said. “So, part of the prevention is immunizations,” which means it is important to not only stay up to date on [family immunizations](#), but [yearly flu shots](#) and COVID-19 vaccines too.

“One thing we do then for babies is we get them started on preventive antibiotics when they’re young, before their first birthday,” Dr. Milanovich said. “We also recommend some additional immunizations for infants and young children with sickle cell disease to help boost their immune system to fight the kind of infections they may be at increased risk.”

“There are a couple bacteria that are higher risk for people with sickle cell disease. One that people have heard of is called meningococcus, which can cause meningitis or a severe bloodstream infection,” he said. “Babies and young children with sickle cell disease should have extra immunizations against meningococcal disease.”

“The other is pneumococcal infection. All babies are recommended to get pneumococcal vaccines, but we do recommend some additional pneumococcal vaccinations as babies get older. And we also start babies on penicillin to prevent pneumococcal infection,” said Dr. Milanovich. “The last infection is called *haemophilus influenzae*, or HIB. It’s recommended for all babies to get immunizations against HIB, but especially babies with sickle cell disease—they’re at higher risk of a complicated infection if they were to contract *haemophilus*.”

Life expectancy has improved

“Unfortunately, sickle cell anemia confers a shortened lifespan. Traditionally, that lifespan has been limited to the mid-40s,” said Dr. Donald. But now “we are seeing increases in the lifespan as we have better supportive, comprehensive care, and we are having a development of newer therapies that have come online as of late.

“But unfortunately, as of now, it’s a shortened lifespan for individuals who have sickle cell anemia,” he added.

Medications help prevent pain

“There are medicines that have been approved in recent years and we’ve been using to help prevent pain episodes,” Dr. Milanovich said. “The one we’ve been using the longest is called hydroxyurea. We usually like to start that early in childhood. It decreases the amount of sickle hemoglobin in the blood and can decrease pain episodes.”

“In recent years there’s been a few new treatments that have come out like Oxbryta [voxelotor] and Endari [L-glutamine] that also can decrease pain episodes when used regularly,” he said. “These medications do not cure the disease. They only can decrease the symptoms, particularly the pain symptoms, but not necessarily the long-term damage that sickle cell disease can cause on multiple organs such as the kidneys, brain and lungs.”

New therapies have been approved

In December 2023, the Food and Drug Administration approved [“two new kinds of gene therapy”](#), which offers a brand-new potential cure for people with sickle cell disease,” Dr. Milanovich said. “Gene therapy works by taking a patient’s own bone marrow stem cells and then using the gene therapy to change their hemoglobin so that there’s less sickle cell hemoglobin.”

“That requires genetically modifying their bone-marrow stem cells outside of the body and then infusing them back into the patient so their bone marrow can start making red blood cells that do not sickle or do not have hemoglobin,” he said. “It’s approved for people who are 12 or older with sickle cell disease.”

The use of gene therapy causes a big decline in pain episodes, but more needs to be understood about the long-term impacts and side effects. This is not a one-and-done treatment, and the FDA recommends 15 years of patient follow up.

“Deciding who’s the best candidate for these therapies really comes down to the patient and their family having those discussions with their hematologist and discussions with those centers that offer the gene therapy,” Dr. Milanovich said, noting “it’s quite expensive.

The cost is estimated at up to \$2 million for one patient.”

That is a big price, he added, but noted that “sickle cell disease is a very expensive disease in terms of the lifelong medical care that people need, let alone how do you measure the impacts of having the disease that might make you sick unexpectedly or cause you to miss work when you have pain.”

Despite “that sticker shock of a nearly \$2 million price tag, it may be beneficial over the long run from a cost standpoint.”

A bone-marrow transplant is a cure

“The one curative treatment for sickle cell disease is bone-marrow transplantation, and that is where a person with sickle cell anemia would see a bone-marrow transplant specialist,” Dr. Donald said, noting “they would have to match their” human leukocyte antigens “type to ensure that there’s engraftment and not rejection.”

“But unfortunately, only about 14% to 20% of people would have a match that would allow for a transplant,” he said. “And then—just as not everyone is a candidate for transplantation as they have to weigh the risks and the benefit of performing that procedure—they have to weigh the risks and the benefit of performing that procedure at very specialized centers.

“So, that is not widely and easily available throughout different cities in the United States,” Dr. Donald added.

Discriminatory care exists

“This is primarily a Black persons’ disease, so within this country there is an irrefutable history of discriminatory care that has occurred,” Dr. Donald said. “Another challenge that occurs is that about 50% of all patients with sickle cell disease are on Medicaid, which is the state-level provided insurance plan. So that presents its own challenges for patients as well.

“And then because we we’re talking about a larger system of discrimination that has occurred historically in this country regarding Black people, then [it becomes a question of resources](#),” Dr. Donald said. “It becomes a question of housing. It becomes a question of where someone lives if they’re in a food desert, if they have all these other things that could really complicate care.”

“Patients with sickle cell anemia do have recurrent painful episodes and do at times require narcotic medication,” he said, noting “there is a cohort of people who having been on narcotics for several years could possible develop abuse or dependency.”

“But this country is facing an opioid epidemic, so to label this cohort of patients as having a stigma for being on pain medication or needing pain medication is disingenuous when you look at the huge amount of inappropriate prescription level narcotics that are abused by citizens throughout this country,” Dr. Donald said. That is why it is important to not allow “the stigma to be placed on patients with sickle cell anemia.”