Sickle cell's threat to adults

For those carrying the chronic blood disease, vigilance is key

THE DAILY CHECKUP

BY KATIE CHARLES

The specialist: Dr. Patricia Shi on adult sickle cell disease

As director of the Adult Sickle Cell Program at Mount Sinai Hospital, Patricia Shi devotes 60% of her time to treating adult patients with sickle cell disease. Shi is board-certified in transfusion medicine and hematology.

Who's at risk

Most adults with sickle cell disease have been receiving care since childhood, but some may discover the disease only as adults or during pregnancy.

Even without symptoms, the disease can progress silently, so it's important to be screened if you're in a high-risk group. "People of African descent are the group at highest risk, with 1 in 12 African-Americans carrying the trait and about 1 in 400 with the disease," says Shi. "Sickle cell disease also affects people whose ancestors came from India and the Caribbean, the Middle East, South and Central America, and the Mediterranean."

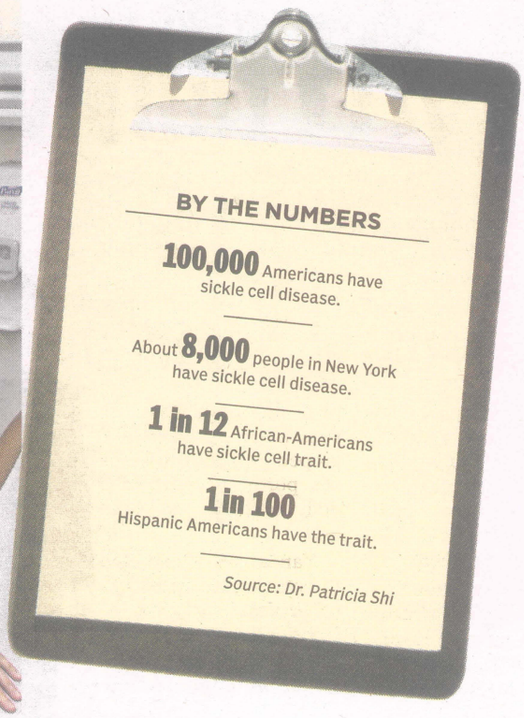
There are three major subtypes of sickle cell disease. "Fifty percent of patients have inherited the sickle cell mutation from both parents," says Shi, "and 30% have inherited the sickle cell gene mutation from one parent and a hemoglobin C mutation from the other parent."

The remainder have inherited one sickle cell mutation along with a beta-thalassemia hemoglobin mutation.

"People who have inherited one copy of the sickle cell mutation and one normal hemoglobin gene don't have sickle disease," says Shi. "But they are carriers, so they can pass the gene on to their children." (For more info, check out last week's column on sickle cell anemia in children.)

MARIELA LOMBARD

Dr. Patricia Shi says some patients don't learn they have sickle cell until adulthood.



WHAT YOU CAN DO

Get screened.
Screening can be done with a simple blood test.

Get informed.
Dr. Patricia Shi recommends the patient advocacy groups Sickle Cell Thalassemia Patients Network (sctpn.org) and Queens Sickle Cell Advocacy Network (qscan.org), along with the national organization sicklecelldisease.org.

Find an experienced provider.
"There are many additional complications that come with this disease, so you need to seek a provider who is experienced with these patients," says Shi.

Take steps to reduce complications.
Adequate hydration is essential for sickle cell patients because dehydration causes higher viscosity in the blood, exacerbating blockages. Avoiding overexertion and extreme temperatures is also important.

Sickle cell disease changes the structure of hemoglobin, the protein in red blood cells that carries oxygen. "The red cells get rigid and sickle-shaped under low oxygen conditions," says Shi. "This causes blood vessel blockage."

One of the biggest challenges of treating sickle cell disease is helping patients transition from pediatric to adult care. "A lot of patients fall off the map," says Shi. "They go off their parents' insurance and have to get their own." Because treating pediatric sickle cell is often an intensive process, these patients tend to depend a lot on their parents, and it can be hard for parents to let them take over their own care. "This transition may happen as late as age 22, rather than 18," says Shi.

Signs and symptoms

Sickle cell disease shows up in adult patients in several ways. "Adults experience severe pain episodes that can occur anywhere in the body," says Shi. "They usually affect the leg and arm bones, the chest, abdomen and back." Acute pain episodes can last from several days to weeks and often require hospital stays.

Another serious complication with sickle cell disease is called acute chest syndrome, in which the vessels of the lungs become blocked. "This causes problems in breathing, so it's a life-threatening complication," says Shi. The treatment is red cell transfusion, which, if given often, can cause iron overload. That requires treatment in its own right.

Rigid sickle cells break apart easily, which causes anemia and a host of related symptoms. "Patients often have as low as one half to two thirds of a normal red blood cell count," says Shi. "This anemia can cause chronic fatigue." When the cells break, they release billirubin, which may cause yellowing of the eyes and stones in the gallbladder. The damage builds up in various organs over time.

"Adults may increasingly experience chronic pain, the destruction of hip and shoulder joints, eye damage, lung damage, increased risk of blood clots and worsening anemia," says Shi. "That's why it's so important to seek care from an adult provider familiar with this disease." Adult patients can have three times the risk of stroke as other patients.

Traditional treatment

Adults with sickle cell disease must work closely with their doctors to develop and adhere to a treatment plan. "It's a chronic disease and treatments are ongoing," says Shi. "Unless you have a bone-marrow transplant, there's no cure for sickle cell disease." One vital therapy all patients need is additional folic acid, which helps create new red cells.

Like pediatric patients, some adults are candidates for hydroxurea, a drug that increases fetal hemoglobin in the red blood cell. "Hydroxurea decreases the incidence of pain, acute chest syndrome and probably even the risk of death, perhaps by decreasing the progression to end organ damage," says Shi. "However, new treatments would definitely be helpful."

About 25% of patients may not get relief from hydroxurea, a therapy most doctors now advise only for patients who have at least three severe episodes of pain in a year or acute chest syndrome.

Research breakthroughs

Researchers are looking for better ways to treat sickle cell disease, and the experimental therapies range from intravenous gammaglobulin to improved bone-marrow transplant techniques.

"Sinai has just started a multicenter clinical trial on IV gammaglobulin, which in mouse models drastically relieved blood vessel blockage," says Shi. "There's also a recent recognition that decreased levels of nitric oxide in the blood contributes to the disease by causing blood vessels to constrict."

New transplant regimens don't require the elimination of the patients' own bone marrow, which allows patients to get lower doses of chemo.

Questions for your doctor

If you're in a high-risk population and don't know if you have sickle cell disease, ask, **"What's my sickle gene status?"** Some patients don't show any symptoms even though the disease is at work.

"Chronic damage can be done even if the patient isn't showing symptoms," says Shi. "Since some states didn't start screening until 2006, there may be a lot of people walking around who don't know that they have sickle cell disease."

If you have been transfused, ask, **"Do I have red cell antibodies?"** Some patients have a difficult blood match for transfusions, and Shi recommends that all sickle cell patients carry that info on a card.