Sickle Cell Disease

Diagnosis and Treatment

Diagnosis

The first step in treating your child is providing an accurate and complete diagnosis. Early diagnosis of sickle cell disease is essential in providing proper preventative treatment for some of the devastating potential complications. Sickle cell disease can be identified by the following tests:

- review of newborn screening results
- haemoglobin electrophoresis
- complete family history
- additional blood tests

In addition, using genetic testing, we are able to identify the specific type of sickle cell disease as well as your child's unique genetic variations.

Treatment Options

Most treatments are aimed at relieving symptoms or preventing complications.

Prevention of bacterial infections and sepsis
Most children with sickle cell disease must:

- take preventative dosing penicillin until at least age 5
- complete all routine childhood vaccinations and additional sickle cell specific vaccinations
- maintain vigilance regarding fevers and signs of infection

Hydroxyurea therapy
Hydroxyurea is an oral medication that causes the body to produce fetal haemoglobin (HbF), a type of haemoglobin normally found only in fetuses and very young children. Increasing the healthy fetal haemoglobin can significantly reduce the side effects and complications of sickle cell disease.

Transfusion therapy
Blood transfusions may be given acutely in order to treat severe anaemia, acute chest syndrome, and other life-threatening complications of sickle cell disease. Children with sickle cell disease may receive chronic blood transfusions to prevent complications such as stroke.<

Iron chelators
Sickle cell anaemia patients who require chronic red blood cell transfusions to prevent a stroke gradually become iron overloaded. Without removal of this iron, it can build up and become toxic to several vital organs. Iron can be removed from the body by medications called chelators. Children may take chelators orally or by injection in order to remove excess iron from the body. Your child's physician may also recommend specific treatments based on the symptoms your child experiences.

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For anaemia

Prevention:

- folic acid
- hydroxyurea

Treatment:

- blood transfusion

For pain

Prevention:

- drinking plenty of water daily (8 to 10 glasses)
- avoiding dehydration
- hydroxyurea medication to prevent painful events

Treatment:

- receiving fluids intravenously
- pain medications such as non-steroidal anti-inflamatories (e.g. ketrolac, ibuprofen) and opioids (e.g. morphine, oxycodone)

For acute chest syndrome (ACS)

Prevention:

- Hydroxyurea
- Incentive spirometry (deep breathing)

Treatment:

- broad spectrum antibiotics
- blood transfusions to dilute HbS with normal haemoglobin

For splenic sequestrations

- blood transfusions to dilute HbS with normal haemoglobin and promote 'release' of blood trapped in the spleen
- penicillin for those who have their spleen surgically removed

For stroke

- blood transfusion

Stem cell transplant

Right now, the only cure for sickle cell disease is stem cell transplant (also called bone marrow transplant) – the transplantation of normal blood stem cells from another person (the "donor") to your child. In sickle cell disease, the best transplant outcomes are almost always when the donor is a healthy sibling with compatible stem cells. The stem cells replace the sickle blood stem cells and restore normal blood production in children with sickle cell disease.

The first step is to determine if the patient has a compatible full-sibling donor. This is done with a blood test or a swab of the cheek.

Reference

Sickle Cell Disease | Diagnosis & Treatment | Boston Children's Hospital
https://www.childrenshospital.org/conditions-and-treatments/conditions/s/sickle-cell-disease/diagnosis-and-treatment (accessed 12/06/21)

Live well with Sickle Cell

The following steps will help relieve symptoms and reduce the chances of serious problems, such as pain crises and other complications of sickle cell disease.

Receive routine follow-up care.

- **See your doctor regularly.** Most people who have sickle cell disease should see their doctor every 3 to 12 months, depending on their age.
- **Get regular vaccines,** including an influenza or flu shot every year. Your doctor may also recommend a second pneumococcus (PPSV23), in addition to the pneumococcus one (PCV13) that all children get as part of their regular immunizations. This second vaccine is given after 24 months of age and again 5 years later. Adults who have sickle cell disease and have not received any pneumococcal vaccine should get a dose of the PCV13 vaccine. They should later receive the PPSV23 if they have not already received it or if it has been more than 5 years since they did. Follow these guidelines even if you or your child is still taking penicillin.

Learn how to manage pain.

- **When an acute crisis is just starting,** drink lots of fluids and take a nonsteroidal anti-inflammatory (NSAID) pain medicine, such as ibuprofen. If you have kidney problems, acetaminophen is often preferred.
- **If you cannot control the pain at home,** go to a sickle cell disease day hospital/outpatient unit or an emergency room to receive additional, stronger medicines and intravenous (IV) fluids. You may be able to return home once your pain is under better control. You may need to be admitted to the hospital to fully control an acute pain crisis.

Learn more about how to manage pain.

Adopt a healthy lifestyle.

- Get regular physical activity. You may tire easily, so be careful to pace yourself and avoid very strenuous activities.
- Choose heart-healthy foods**,** including limiting alcohol. Drink extra water to avoid dehydration.
- Quit smoking. Visit Smoking and Your Heart and the National Heart, Lung, and Blood Institute's Your Guide to a Healthy Heart. Although these resources focus on heart health, they include basic information about how to quit smoking.

Prevent problems over your or your child's lifetime

- **Avoid situations that may set off a crisis.** Extreme heat or cold, as well as abrupt changes in temperature, are often triggers. When going swimming, ease into the water rather than jumping right in.
- **Do not travel** in an aircraft cabin that is unpressurized.
- **If you experience priapism** (prolonged, painful erection), you may be able to relieve your symptoms by doing light exercise, emptying your bladder by urinating, drinking more fluids, and taking medicine recommended by your doctor.
- **If your child attends day-care, preschool, or school,** speak to his or her teacher about the disease. Teachers need to know what to watch for and how to accommodate your child.

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- **Learn how to palpate, or feel, your child's spleen.** Because of the risk of splenic sequestration crisis, caretakers should learn how to palpate a child's spleen. They should try to feel for the spleen daily and more often when the child is ill. If the spleen feels larger than usual, they should call the care provider.
- **Seek help** if you have feelings of depression or anxiety. Supportive counselling and, sometimes, antidepressant medicines may help.

Get more tips for healthy living with sickle cell disease.

Know when to seek emergency medical care.
Sickle cell disease can lead to serious and life-threatening health problems. If you think you or someone else is having any of the following symptoms or complications

Symptoms of severe anaemia, including extreme tiredness (fatigue), shortness of breath, dizziness, or irregular heartbeat. Splenic sequestration crisis or an aplastic crisis can cause severe anaemia symptoms. These conditions can be life-threatening.

- **Fever.** All children and adults who have sickle cell disease and a fever of more than 101.3 degrees Fahrenheit or 38.5 degrees Celsius must be seen by a doctor and treated with antibiotics right away. Some people will need to be hospitalized.
- **Symptoms of acute chest syndrome,** including chest pain, coughing, fever, and shortness of breath. You will need to be admitted to the hospital, where you may receive antibiotics, oxygen therapy, or a blood transfusion.
- **Signs and symptoms of a stroke,** such as sudden weakness, numbness on one side of the body, confusion, or trouble speaking, seeing, or walking.
- **Priapism.** If you experience priapism that lasts for 4 hours or more, go to the hospital to see a haematologist and urologist.

Pregnancy and sickle cell disease
Pregnant women who have sickle cell disease are at greater risk for problems. If you are pregnant or planning for pregnancy, meet with a doctor who specializes in high-risk pregnancies and has experience with patients who have sickle cell disease.
Your doctor may prescribe certain vitamins and will be careful to prescribe pain medicines that are safe for you and your baby. You should *not* use hydroxyurea during pregnancy.
You may need to have one or more blood transfusions during pregnancy to treat problems, such as anaemia symptoms that get worse. You may also experience more pain crises or be at higher risk of having acute chest syndrome. Your doctor will talk to you about how to help prevent these complications.

References
Content reproduced from https://www.nhlbi.nih.gov/health-topics/sickle-cell-disease