Sickle Cell Disease

Sickle cell disease affects 1 in every 1941 newborns regardless of race and 1 in every 400 African-Americans born in the United States. It is a disease that causes misshaped and stiff red blood cells causing anemia and damage to many organs throughout the body. Even with the best current medical therapy, the lifespan for patients with sickle cell disease remains shortened.
Bone marrow transplant for sickle cell disease
St. Jude Children’s Research Hospital was the first hospital to find the cure for sickle cell disease through bone marrow transplant in 1983. Your child might have a bone marrow (stem cell) transplant to treat their sickle cell disease. But many patients don’t have a well matched donor available.

Testing to find a bone marrow donor
You will meet with our team to discuss whether a bone marrow transplant can help your child. Once you decide, several people within your family will have tests to learn if their cells could match your child’s. If a family member is a good match, this person could be the donor who gives healthy cells to your child. The medical term for this testing is “HLA typing.”

Who is tested for a match?
Your St. Jude team will test your child with sickle cell disease, both of the child’s parents, and all of the child’s full brothers and sisters. A full brother or sister has the same mother and father. A relative who carries sickle cell trait may still be considered as a donor.

Once the testing is performed, it takes about 2 weeks to get the results. Then, the team will know if there is a good match for your child.

The chance of a patient with SCD having an HLA-matched brother or sister is less than 15%. If a full sibling is not a match, it is possible the team will discuss alternative donors for transplant. Sometimes parents, or brothers and sisters who are not fully matched can be a donor for transplant. These donors are called haplo-identical donors (Haplo).

Testing for the bone marrow donor
The bone marrow donor will also need tests before donating stem cells.

Collecting stem cells for transplant
There are two ways to collect stem cells for transplant.

Harvest
If the cells for transplant are obtained from the bone marrow, the collection procedure is called a “bone marrow harvest.” The donor is most often given anesthesia which allows the person to sleep during the procedure. Needles are inserted through the skin over the pelvic (hip) bone and into the bone marrow to draw the blood forming cells out of the bone. Harvesting the marrow takes about an hour.

Apheresis
If the blood-forming cells needed for transplant are obtained from the vein, the process is called apheresis. For several days before apheresis, the donor will be given a medication called granulocyte colony stimulating factor—G-CSF. In apheresis, blood is removed through a large vein in the arm or from a central venous catheter (a flexible tube that is placed in a large vein in the neck, chest, or groin area). The blood goes through a machine that removes the stem cells. The remaining blood is then returned to the donor. Apheresis typically takes 4 to 6 hours.
Clinical Trials for Sickle Cell at St. Jude
Bone marrow transplantation can successfully cure sickle cell disease. However, in many cases, it is hard to find a well matched donor for the patient. As an alternative, St. Jude is currently developing transplant options using half-matched donors (such as parents, also called haploidentical donors) and gene therapy that could change the way the disease is treated in the future. In addition, St. Jude researchers are also trying to reduce the intensity and toxicity related to transplantation to make it safer and more accessible to all.

Gene therapy can be used to change your own cells so they do not sickle. You do not need a donor to undergo gene therapy. This treatment is very new and rapidly changing so talk to your provider to see if this is a good option for you.
Pre-evaluation testing before the transplant

Your child will need testing before the transplant. We can start making plans for this testing after we identify a donor or you decide to undergo gene therapy.

Testing before the transplant takes about 2 weeks. You can expect to spend several hours at St. Jude each day for these tests. They may include:
- MRI, CT (CAT) scans, and X-rays
- Heart, lung, and kidney tests
- Blood tests
- Consultations

You will also spend time talking with the transplant team. A doctor will place a central line in your child that will be used for transplant or blood testing so an IV will not need to be placed frequently. If your child has a different line already, we may replace it with a different central line.

When the tests are complete, you will meet with a doctor to review the transplant information and make sure you understand it. You are welcome to ask any questions that you have about the transplant. Then, you will sign a consent form that allows your child to have the transplant. You also review plans for your child's hospital stay with a nurse or coordinator. Your child will stay at St. Jude inpatient for 5 or 6 weeks, so planning that stay is important.

Preparative Regimen

Your child will start chemotherapy and/or conditioning once they are inpatient. You might hear this being called the “prep” or “preparative regimen”. It prepares your child’s body to get a new transplant and start making healthy cells. We will give you information on each chemotherapy drug, so you and your child know what to expect.

Transplant

Your child will get the cells from the donor after chemotherapy is complete. This is “Day 0”. A team member will give the cells through the central line. The medical term for giving the new cells is “the infusion”. On the day of the infusion, several nurses and a lab technician will stay in your child’s room. They will watch your child closely for signs of a reaction to the transplant cells. You and one (1) other caregiver may stay with your child.

Engraftment

The next step is waiting for the donor cells to grow and multiply in your child’s body. The medical term for this is “engrafting”. It takes several weeks, and allows your child’s body to make healthy cells.

Outpatient Follow-up

Your child will stay in Memphis in local or long term housing for at least 100 days post transplant. This could be longer if complications occur. During this time, your child will have appointments in the outpatient transplant clinic so we can check their progress.

Long Term Follow-up

After your child returns home, he or she will return at regular intervals to St. Jude for checkups. These checkups will continue to space out as long as your child continues to do well.
Side Effects
Most patients who receive chemotherapy experience some degree of hair loss, nausea, vomiting, diarrhea and loss of appetite. We will give you medicines to help control nausea, vomiting and pain during transplant. Hair begins to grow after a few weeks but could have a slight change in appearance.

Graft Rejection
Graft rejection, although uncommon, occurs when the immune system of the patient recognizes the donor cells as being different and destroys them. Since chemotherapy used during transplant destroys the patient’s blood-forming cells, they may regenerate on their own. Patients who experience graft rejection can become quite ill. To prevent graft rejection, the patient receives chemotherapy with or without radiation to destroy the immune system before the transplant occurs. If graft rejection occurs, another transplant or treatment may be an option.

Graft-versus-host Disease (GVHD)
Graft versus host disease is when donor cells attack your body. It can occur early or late. 

Symptoms can include:
- Rash
- Diarrhea
- Yellow skin and eyes
- Scaly skin
- Darkening of skin
- Hardening of skin texture
- Skin scarring/restriction of joints
- Dryness and sores in the mouth and esophagus
- Dry eyes and redness in the eyes
- Dryness of the vagina and other surfaces
- Drying and scarring of lungs
- Liver injury or liver failure

Veno-occlusive disease (VOD)
Patients who have increased iron in their liver due to frequent blood transfusions can develop VOD. VOD is characterized by the elevated concentration of bilirubin (which results in the yellow appearance of the skin and eyes), an enlarged liver and fluid retention or weight gain. VOD is frequently treated with fluid restriction and a medication called defibrotide. Preventive measures may include giving the patient ursodiol or heparin and daily monitoring of weights and fluid balance while the patient is hospitalized.

Infertility
Some people who receive transplant may not be able to have children of their own. If your child is old enough, we may be able to collect their sperm/eggs prior to transplant and freeze them to use at a later date if they choose.

* Transplant only cures you, future generations would still have the same risk of developing sickle cell disease.

Benefits of Transplant/Gene Therapy for Sickle Cell Disease
- No further Blood Transfusion needs
- No further organ damage
- Cure of Sickle cell disease for patient
- Decreased hospital visits
- Decreased pain