Preimplantation Genetic Testing (PGT)
for Sickle Cell Disease

PGT is a procedure to decrease the chance of having a baby with sickle cell disease.

This handbook was created to educate families about PGT. You may have questions after reading the handbook; please share your questions with your health care team.

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Sickle Cell Genetics

Sickle cell disease is a genetic disease that is passed on to a child from his or her parents. A child gets two copies of a gene, one copy from each parent.

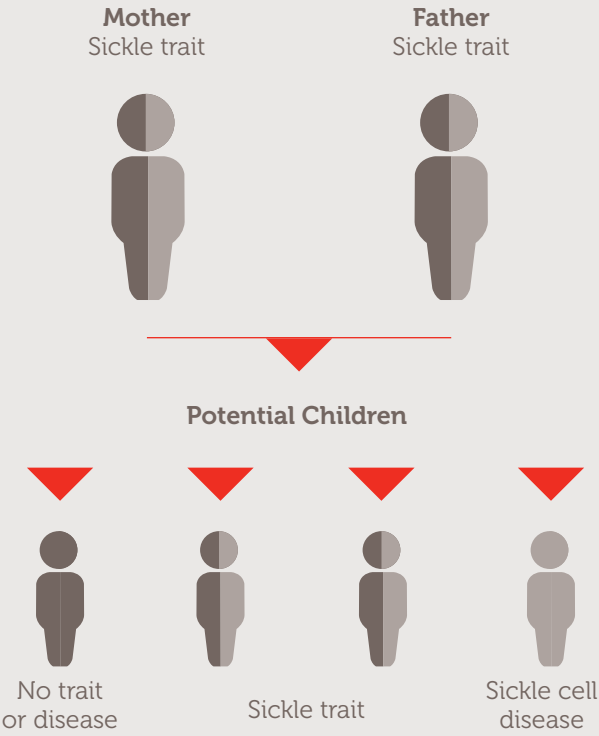
People with sickle cell trait have one copy of the sickle cell gene "S" and one copy of the normal hemoglobin gene "A". People with sickle cell trait (also called "AS") do not have the serious health problems of sickle cell disease.

If both parents have sickle cell trait, there is a

1 in 4

(25%) chance of having a child with sickle cell disease.

The number of previous children with or without the disease does not change this chance.



There are different types of sickle cell disease: hemoglobin SS, SC, S beta zero thalassemia, S beta plus thalassemia, and others. **Talk to your health care team to learn more about your child's sickle cell disease and how it was inherited.**

What is PGT?

PGT stands for Preimplantation Genetic Testing. PGT was also called **PGD** (Preimplantation Genetic Diagnosis).

PGT is done by a fertility specialist and is used with in-vitro fertilization (IVF).

In-vitro fertilization (IVF) means that the mother's egg and father's sperm are placed together to make an embryo in the laboratory. An **embryo** is the early group of cells that can grow into a baby when placed back into a woman's uterus (womb).

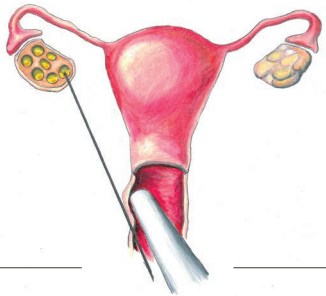
A microscopic picture of an embryo

With PGT, embryos in the laboratory are tested for certain genes. This can include the gene for sickle cell disease. This can also include the HLA genes to see if a sibling is a "match" for blood and marrow transplant (BMT). Based on the test results, embryos without disease and (if desired) the best HLA match are used for transfer. One embryo is placed into the woman's uterus to grow into a baby.

IVF and PGT Steps

1. **HORMONE INJECTIONS:** To start an IVF cycle, the woman will get hormone injections to stimulate her ovaries to make multiple eggs.

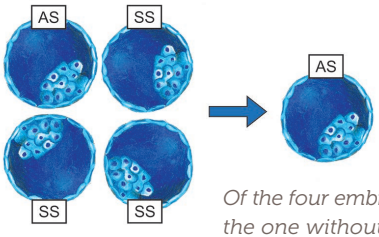
2. **EGG REMOVAL:** The woman will next have a procedure to remove her eggs. This is a minor outpatient surgery. The eggs are collected with an ultrasound-guided needle through the vagina.



3. **FERTILIZATION:** Sperm is obtained from the man and then injected into each collected egg in the laboratory. This mixing of the egg and sperm makes an embryo. Most IVF cycles will make multiple embryos.

Intracytoplasmic sperm injection: sperm being injected into an egg

4. **EMBRYO BIOPSY:** After a few days, a biopsy is done on the embryos in the laboratory. A biopsy involves removing a few cells for testing.



Of the four embryos tested, the one without sickle cell disease (AS) is chosen

5. **GENETIC TESTING:** Testing is done to find out if an embryo has sickle cell disease. Testing can also be done to see if the embryo is a match for transplant. Healthy embryos can be chosen for transfer.

6. **EMBRYO TRANSFER:** An embryo is put into the woman's uterus through the vagina. If there are many healthy embryos, embryos can be safely frozen.

PGT Risks & Benefits

IS IT SAFE?

Yes, IVF is safe for most women but there are some risks. It is important to talk to your doctor about your risks.

SUCCESS IS NOT GUARANTEED

PGT is less likely to work in an older woman. An embryo put into a woman after IVF and PGT may not grow into a baby. Some couples must go through more than one IVF cycle to have a healthy baby. There is also a small chance that the genetic testing could be wrong.

COST

Many insurance companies will not cover all of the costs of PGT.

CHILD WITHOUT SICKLE CELL DISEASE

PGT can help families have a baby who will not have sickle cell disease.

CURE ANOTHER CHILD WITH SICKLE CELL DISEASE

Blood and marrow transplant (BMT) is a proven cure for sickle cell disease that works best with a sibling donor who is an HLA "match." The chance that a full sibling (child with the same mother and father) is an HLA match is less than 1 in 4 (25%). A couple can use PGT to have another child who will not have sickle cell disease and will match a sibling for BMT.

Is PGT for my family?

The decision to have PGT is usually not easy.

Deciding to have PGT is a very personal choice.

PGT is a good option for some but not all families.

THERE ARE ALTERNATIVES TO PGT INCLUDING:

- Having a baby naturally and taking the chance that they will have sickle cell disease.
- Having prenatal testing (chorionic villus sampling "CVS" or amniocentesis). Some families may decide to terminate a pregnancy based on the test results.
- Using an egg or sperm donor to have a baby.
- Adopting a child.

Talk to your health care team to learn more about these options.

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