

# What is Alpha Thalassemia?

Alpha thalassemia major (ATM) is a genetic blood disorder. ATM is a serious kind of anemia that prevents the body from making working hemoglobin, a protein that helps red blood cells carry oxygen. Without treatment during pregnancy, a fetus with ATM can become very sick and may not survive.

ATM is most common in families with origins in southern China, the Philippines, Vietnam, Thailand, Laos, Cambodia, India, and in Hmong communities, as well as countries in the West Indies and Middle East. A pregnancy is only at risk if both parents are carriers of the disease. Often parents do not know they are carriers because they have no symptoms.

Your doctor may screen for ATM during or before pregnancy using a simple blood test called a complete blood count (CBC). Anemia or a low red blood cell count can be seen in individuals that carry a change in their genes causing ATM. Some parents may choose a genetic blood test to confirm if they are carriers.

***There are now treatments available for a fetus with ATM. The earlier the fetus can be diagnosed, the better.***

## How do I get more information?

If your fetus has been diagnosed with alpha thalassemia, or you suspect that you or your partner may be a carrier, please contact us at:

**1-800-RX-FETUS (1-800-793-3887)** or  
[fetaltreatmentcenter@ucsf.edu](mailto:fetaltreatmentcenter@ucsf.edu)

You can also watch a video here:  
<https://vimeo.com/251336395>



**UCSF Benioff Children's Hospitals**  
Oakland | San Francisco

*For more information or to refer a patient please contact us at:*



**The Fetal Treatment Center**  
THE BIRTHPLACE OF FETAL SURGERY

**UCSF Fetal Treatment Center**  
1855 4th Street  
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**1-800-RX-FETUS**  
**(1-800-793-3887)**

[fetus@ucsf.edu](mailto:fetus@ucsf.edu)  
[fetus.ucsf.edu](http://fetus.ucsf.edu)



**The Fetal Treatment Center**  
THE BIRTHPLACE OF FETAL SURGERY

## Fetal Intervention for Alpha Thalassemia Major



*If you are pregnant and your fetus has alpha thalassemia, new treatments can help you have a healthier baby.*



**UCSF Benioff Children's Hospitals**

# My fetus has been diagnosed with ATM. What are our options?

## No Interventions

Because ATM is a serious disease, without treatment most fetuses will die before birth. Fetuses with ATM do not have enough oxygen in their growing bodies and brains.

Without healthy red blood cells to deliver oxygen, the heart works harder and harder to pump blood and can develop heart failure. One sign of heart failure is a condition called “hydrops,” which means there is extra fluid and swelling in the body of the fetus. Sometimes, the mother can also develop swelling and become very sick. This is called “mirror syndrome.” When this happens, the only treatment is to deliver the fetus.

Some women choose to terminate a pregnancy after a diagnosis of ATM.

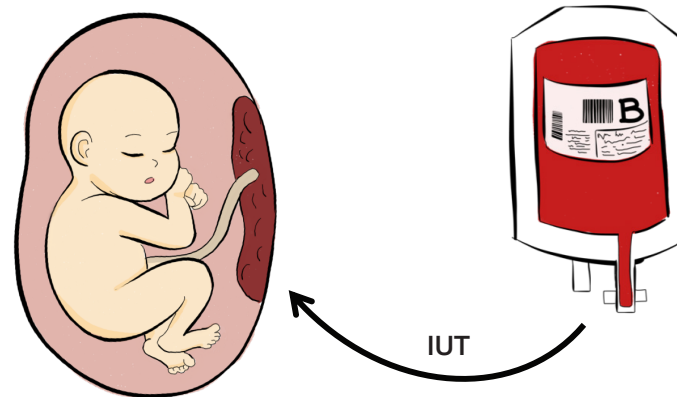
Many fetuses with ATM are never diagnosed, and those pregnancies often end with unexplained miscarriage.

Scan the following QR code with your phone for more information:



## In utero transfusion

If the fetus has ATM, the doctors may offer to give a blood transfusion before birth. A blood transfusion gives the fetus healthy red blood cells that circulate enough oxygen in its body to survive until birth. This treatment can reverse hydrops or swelling in the fetus and allow delivery at full term. This treatment is repeated several times in the pregnancy to keep the fetus healthy with working red blood cells until it is born.



In utero transfusion has a small miscarriage risk (about 5%, or 1 in 20). This treatment is often used to treat other conditions, such as Rh disease (a problem that happens when the blood type of the mother and fetus don't match). Even with proper treatment during pregnancy, babies born with ATM may have learning difficulties, but most of the time these are mild.

After birth, the baby will need blood transfusions for the rest of his or her life. Stem cell transplantation may be done, and this can cure the child of ATM. However, stem cell transplantation could cause serious complications, such as infection and even death.

## In utero transfusion + stem cell transplant

UCSF is testing a new strategy to give stem cells before birth, which may ultimately treat the ATM with fewer risks. In this experimental treatment, the fetus receives a transplantation of the mother's stem cells at the same time as a fetal blood transfusion.

Because it takes time for the mother's stem cells to start working, the fetus still needs several blood transfusions during the pregnancy. After birth, the doctors will do tests to see how well the mother's stem cells are surviving in the baby. Even if the treatment is only partly successful, the baby may still be able to get a “booster” transplant of the mother's stem cells to be cured of ATM. This booster transplant may be safer than if the baby had never been given stem cells before birth.

This treatment is experimental while we do research to see if it is safe for both the mother and fetus. We are also determining whether it will be successful in allowing the fetus to make its own healthy red blood cells.

