



MotherToBaby

Medications & More During Pregnancy & Breastfeeding
Ask The Experts

Fact Sheet

by the **Organization of Teratology Information Specialists (OTIS)**

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Gaucher Disease

In every pregnancy, a woman starts out with a 3-5% chance of having a baby with a birth defect. This is called her background risk. This sheet talks about whether exposure to Gaucher disease may increase the risk for birth defects over that background risk. This information should not take the place of medical care and advice from your health care provider.

What is Gaucher disease?

Gaucher disease is a genetic disorder. People with Gaucher disease have low levels of the enzyme called glucocerebrosidase. This enzyme helps break down fatty substances in the body. Without enough of this enzyme, fatty material build up in the liver, spleen, lungs, bone marrow and, less commonly, in the brain. The build-up of this fatty material causes many of the body's organs to stretch and not work well. Common symptoms include thinning of the bone (osteopenia), bone pain/fractures, enlarged liver (hepatomegaly) and/or spleen (splenomegaly), decreased red blood cells (anemia), fatigue, decreased number of blood platelets (thrombocytopenia), and easy bruising.

There are three major types of Gaucher disease, referred to as Type I, Type II or Type III. Symptoms can range from mild to severe and depend on the type of Gaucher disease. Gaucher disease occurs in approximately 1 in 60,000 individuals. Type I is the most common form of the disorder. It occurs more often in individuals of Ashkenazi (Eastern European) Jewish ethnicity than in those from other backgrounds. It affects about 1 in 900 individuals of Ashkenazi Jewish descent.

Can Gaucher disease make it harder for me to get pregnant?

Most women with Gaucher disease do not have problems with fertility related to their disease.

I have Gaucher disease and would like to become pregnant. What should I do?

It is important to make a treatment plan before getting pregnant, if possible. A discussion with your health care team (including obstetrician, anesthesiologist, hematologist and Gaucher disease specialist) about treatment strategies before and during pregnancy, during delivery and postpartum is recommended.

If your pregnancy is unplanned, you should contact your health care provider as soon as you learn you are pregnant. Assessing your bone disease before pregnancy or as soon as you find out you're pregnant is important because pregnancy increases the risk for severe bone pain (bone crisis).

Your treatment plan will include ways to monitor your pregnancy and Gaucher disease symptoms. Talk to your health care provider about what medications or vitamins you should take during pregnancy. People with Gaucher disease might not have enough of certain vitamins and nutrients such as Vitamin D or calcium. Vitamin B12 and folic acid supplementation should be started prior to pregnancy. It is recommended that you review your immunization records and obtain any of the necessary immunizations either before pregnancy or as soon as you find out you're pregnant, especially if you have had your spleen removed. Many vaccines are compatible with pregnancy. For more information, please see the MotherToBaby fact sheet Vaccines and Pregnancy at <https://mothertobaby.org/fact-sheets/vaccines-pregnancy/pdf/>.

How does pregnancy affect Gaucher disease?

Most women with Gaucher disease will have healthy children. If you have Gaucher disease and become pregnant, you are at risk for an increase in symptoms of the disease. You may also have new symptoms start during pregnancy. Women with Gaucher disease are at an increased risk to have bleeding, postpartum infection and bone disease. Some studies have found an increased risk for miscarriage in untreated women with Gaucher disease. The risk

for other pregnancy complications, including high blood pressure, preterm delivery, and gestational diabetes is not thought to be increased for women with Gaucher disease.

Some women with Gaucher disease can develop anemia (low levels of red blood cells, needed to carry oxygen) and thrombocytopenia (low blood platelet count, which can affect clotting) in pregnancy. Women with severe thrombocytopenia and/or clotting abnormalities could be at risk for bleeding around the time of delivery. Since having very low numbers of red blood cells and blood platelets increases the risk for complications with anesthetic medications, the use of an epidural anesthesia during delivery may not be recommended for women with Gaucher disease.

Do I continue my treatment for Gaucher disease during pregnancy?

You should discuss all of your medications with your health care provider before conception, if possible, or as soon as you find out you're pregnant. Enzyme replacement therapy has been used successfully in pregnancy. For more information on Gaucher disease treatments in pregnancy, please see the MotherToBaby fact sheets on Enzyme Replacement Therapy (<https://mothertobaby.org/fact-sheets/enzyme-replacement-therapy-treatment-gaucher-disease-pregnancy/pdf/>), Miglustat (<https://mothertobaby.org/fact-sheets/miglustat-zavesca-pregnancy/pdf/>), and Bisphosphonates (<https://mothertobaby.org/fact-sheets/bisphosphonate-treatment-pregnancy/pdf/>).

Since I have Gaucher disease, will my baby have it too?

A baby can only have Gaucher disease if both the mother and the father carry a specific genetic change for Gaucher disease. Since you have Gaucher disease, you have two non-working genes for Gaucher disease, one from your mother and one from your father. You will pass on one of these non-working genes for Gaucher disease to your children. A person who has only one non-working gene for Gaucher disease is called a carrier for Gaucher disease. Carriers of Gaucher disease are healthy.

If the father of the baby does not have Gaucher disease and is not a carrier, your children will not inherit Gaucher disease, but they will all be carriers. However, if you have children with someone who is a carrier of Gaucher disease, then there is a 50% chance for each child to have Gaucher disease. Finally, if you have children with someone who also has Gaucher disease, all of your children will have Gaucher disease. Testing to find out if a partner is a carrier of Gaucher disease is possible, and if a specific genetic change is found, prenatal testing may also be available. A genetic counselor or other health care provider can provide more information.

Can I breastfeed if I have Gaucher disease?

Gaucher disease does not appear to affect a woman's ability to breastfeed. It is important to consider possible bone complications. In general, a woman who is breastfeeding will lose 3-7% of her bone density during lactation, which is normally regained after she stops breastfeeding. This loss could be significant for a woman with Gaucher disease who already has low bone density. Breastfeeding beyond 6 months may not be recommended. It is important to balance the benefits of breastfeeding with the maternal health risks. Be sure to talk to your health care provider about all your breastfeeding questions.

References Available By Request

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