Gaucher Disease

This sheet is about having Gaucher disease during pregnancy and while breastfeeding. This information should not take the place of medical care and advice from your healthcare provider.

What is Gaucher disease?

Gaucher disease is a genetic condition that results in people having low levels of the enzyme glucocerebrosidase (gluco·ce·re·bro·si·dase). This enzyme helps break down fatty substances in the body. Without enough of this enzyme, fatty material builds up in many tissues including 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), enlarged spleen (splenomegaly), lack of healthy red blood cells (anemia), fatigue, low blood platelet levels (thrombocytopenia), and easy bruising.

There are three major types of Gaucher disease, referred to as Type 1, Type 2 or Type 3, and 2 subtypes. Symptoms can range from mild to severe and depend on the type of Gaucher disease.

I have Gaucher disease and would like to get pregnant. What should I talk about with my healthcare team?

It is important to talk to your healthcare team (including obstetrician, anesthesiologist, hematologist, and Gaucher disease specialist) about treatment plans before and during pregnancy, during delivery, and after delivery. Talk with them before getting pregnant, if possible. If your pregnancy is unplanned, contact your healthcare providers as soon as you learn you are pregnant.

Things to talk about with your healthcare team include:

- Ways to monitor your pregnancy and your Gaucher disease symptoms.
- Any 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.
- Checking your bones. Pregnancy can increase the chance for severe bone pain (bone crisis).
- Your vaccines (immunizations) and getting any necessary vaccines either before pregnancy or as soon as you find out you are pregnant, especially if you have had your spleen removed. Many vaccines can be given in pregnancy. For more information, please see the MotherToBaby fact sheet on vaccines at https://mothertobaby.org/fact-sheets/vaccines-pregnancy/.
- Any other questions or concerns.

Does having Gaucher disease increase the chance for miscarriage?

Miscarriage is common and can occur in any pregnancy for many different reasons. Based on the studies reviewed, Gaucher disease may increase the chance for miscarriage.

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

Based on the studies reviewed, Gaucher disease is not expected to make it harder to get pregnant.

Does having Gaucher disease increase the chance of birth defects?

Every pregnancy starts out with a 3-5% chance of having a birth defect. This is called the background risk. Based on the studies reviewed, Gaucher disease is not expected to increase the chance for birth defects above the background risk.

Does having Gaucher disease increase the chance of other pregnancy-related problems?

A small number of studies have not found a higher chance for pregnancy complications such as high blood pressure,
preterm delivery (birth before week 37), low birth weight (weighing less than 5 pounds, 8 ounces [2500 grams] at
birth), or gestational diabetes among those with Gaucher disease.

For some with Gaucher disease, new symptoms can start during pregnancy. People with Gaucher disease can have a
higher chance for bleeding, infection after delivery, and bone disease.

Some people with Gaucher disease can develop anemia and thrombocytopenia in pregnancy. Those with severe
thrombocytopenia and/or clotting disorders could be at risk for bleeding around the time of delivery. Having very low
numbers of red blood cells and blood platelets increases the chance for complications with anesthetic medications,
such as the use of an epidural anesthesia during delivery. It is important to talk with your healthcare team about your
plans for delivery.

**Does having Gaucher disease affect future behavior or learning for the child?**

Studies have not been done to see if Gaucher disease can cause behavior or learning issues for the child.

**Breastfeeding while I have Gaucher disease:**

Gaucher disease does not appear to affect the ability to breastfeed. In general, a person will lose 3-7% of their bone
density while breastfeeding. The lost bone is usually regained after breastfeeding stops. However, this loss could be
significant for people with Gaucher disease who already have low bone density. For some, breastfeeding more than 6
months may not be recommended. Be sure to talk to your healthcare provider about all of your breastfeeding
questions.

**If a male has Gaucher disease, could it affect fertility (ability to get partner pregnant) or increase the
change of birth defects?**

Studies have not been done to see if Gaucher disease could affect fertility or increase the chance of birth defects
above the background risk. In general, exposures that fathers or sperm donors have are unlikely to increase the risk to
a pregnancy. For more information, please see the MotherToBaby fact sheet Paternal Exposures at
https://mothertobaby.org/fact-sheets/paternal-exposures-pregnancy/.

Please click [here](https://mothertobaby.org/fact-sheets/paternal-exposures-pregnancy/) for references.

---

**Questions? Call 866.626.6847 | Text 855.999.3525 | Email or Chat at MotherToBaby.org.**

Disclaimer: MotherToBaby Fact Sheets are meant for general information purposes and should not replace the advice of your health care provider. MotherToBaby is a service of the non-profit Organization of Teratology Information Specialists (OTIS). OTIS/MotherToBaby encourages inclusive and person-centered language. While our name still contains a reference to mothers, we are updating our resources with more inclusive terms. Use of the term mother or maternal refers to a person who is pregnant. Use of the term father or paternal refers to a person who contributes sperm. Copyright by OTIS, February 1, 2022.