Gaucher Disease

This sheet is about having Gaucher disease in pregnancy or while breastfeeding. This information is based on published research studies. It 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 causes low levels of an enzyme, called glucocerebrosidase, that helps break down certain fatty substances in the body. When this enzyme is low, these fats build up in parts of the body, such as the liver, spleen, lungs, bone marrow, and sometimes the brain. Over time, this buildup can cause these organs to get larger and not work as well.

Common symptoms of Gaucher disease include thinning of the bone (osteopenia), bone pain/fractures, enlarged liver (hepatomegaly), enlarged spleen (splenomegaly), lack of healthy red blood cells (anemia), fatigue (low energy), low blood platelet levels (thrombocytopenia), and easy bruising and bleeding.

Symptoms can range from mild to severe and depend on the type of Gaucher disease a person has. The 3 major types of Gaucher disease are called Type 1, Type 2, and Type 3; and there are 2 subtypes.

I have Gaucher disease. What should I talk about with my healthcare team before I get pregnant?

It is important to talk to your healthcare team (including your obstetrician, anesthesiologist, hematologist, and Gaucher disease specialist) about plans for treating your condition before and during pregnancy, during delivery, and after delivery. If possible, talk with your healthcare team before getting pregnant. If your pregnancy is unplanned, contact your healthcare providers as soon as you find out 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 vitamins and nutrients, such as Vitamin D or calcium.
- Getting your bones checked. Pregnancy can increase the chance for severe bone pain (bone crisis).
- Getting any necessary vaccines before and during pregnancy, 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 you have.

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

Gaucher disease is not expected to make it harder to get pregnant.

Does having Gaucher disease increase the chance for miscarriage?

Miscarriage is common and can happen in any pregnancy. Some studies show that Gaucher disease might increase the chance of miscarriage, especially in women with more severe symptoms or who are not receiving treatment. Other studies suggest that miscarriage rates are similar to women without Gaucher disease.

Does having Gaucher disease increase the chance of birth defects?

Birth defects can happen in any pregnancy for different reasons. Out of all babies born each year, about 3 out of 100 (3%) will have a birth defect. We look at research studies to try to understand if an exposure, like having Gaucher disease, might increase the chance of birth defects in a pregnancy. Gaucher disease is not expected to increase the chance of birth defects.

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

Studies have shown mixed results about pregnancy-related problems in women with Gaucher disease. Some studies have found a higher chance of preterm delivery (birth before week 37) and low birth weight (weighing less than 5 pounds, 8 ounces [2500 grams] at birth). Other pregnancy complications, such as high blood pressure and gestational diabetes, have been reported but are not common.

Some women who have Gaucher disease start to have new symptoms during pregnancy. In addition, symptoms of Gaucher disease, such as anemia, thrombocytopenia, enlarged spleen or liver, and bone problems can worsen during pregnancy. Having severe thrombocytopenia increases the chance of bleeding around the time of delivery.

This bleeding risk can affect decisions about anesthesia for delivery. It is important to talk with your healthcare team about your plans for delivery, including the use of anesthesia.

Does Gaucher disease in pregnancy 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. However, breastfeeding for more than 6 months may not be recommended for some women with Gaucher disease who have low bone density. This is because most women will lose 3% to 7% of their bone density while breastfeeding. Although the lost bone is usually regained after breastfeeding stops, this loss might be too much for someone who already has low bone density. Be sure to talk to your healthcare provider about all your breastfeeding questions.

If a man has Gaucher disease, can it affect his fertility or increase the chance of birth defects?

Studies have not been done to see if Gaucher disease could affect male fertility (ability to make healthy sperm) or increase the chance of birth defects above the background risk. In general, exposures that men have are unlikely to increase risks 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 for references.

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