Gaucher Disease

What is Gaucher disease?

Gaucher disease is a genetic disorder. People with Gaucher disease have low levels of the enzyme called glucocerebrosidase (glu·co·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 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 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 become pregnant. What should I discuss with my healthcare team?

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

Remember to discuss ways to monitor your pregnancy and your Gaucher disease symptoms. Talk to your healthcare providers 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 before pregnancy. Ask if your bones should be checked since pregnancy can increase the chance for severe bone pain (bone crisis). Review your immunization records and get any of the 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 are compatible with pregnancy. For more information, please see the MotherToBaby fact sheet on vaccines at https://mothertobaby.org/fact-sheets/vaccines-pregnancy/pdf/.

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

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

How does pregnancy affect Gaucher disease?

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

Some people 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. 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 risk for complications with anesthetic medications, such as the use of an epidural anesthesia during delivery. Discuss delivery options with your healthcare team.

Does Gaucher disease increase the chance for miscarriage?

Miscarriage can occur in any pregnancy. Gaucher disease might increase the chance for miscarriage.

Does having Gaucher disease increase the chance of birth defects?

Every pregnancy starts with a 3-5% chance of having a birth defect. This is called the background risk. Among those with Gaucher disease, the chance for birth defects in their pregnancy is not thought to be higher than the background risk.
**Does having Gaucher disease cause other pregnancy complications?**

A small number of studies have not found a higher chance for other pregnancy complications, such as high blood pressure, preterm delivery (birth before 37 weeks of pregnancy), or gestational diabetes among those with Gaucher disease.

**Does having Gaucher disease in pregnancy cause long-term problems for the baby?**

Based on the data available, it is not known if Gaucher can cause behavior or learning issues.

**Can I breastfeed if I have Gaucher disease?**

Gaucher disease does not appear to affect the ability to breastfeed. It is important to consider possible bone complications. In general, a breastfeeding person will lose 3-7% of their bone density during lactation, which is normally regained after breastfeeding stops. This loss could be significant for a people 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 any health risks. Be sure to talk to your healthcare provider about your breastfeeding questions.

**I have Gaucher disease. Can it make it harder for me to get my partner pregnant or increase the chance of birth defects?**

This has not been studied. 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/pdf/.

Please click [here](#) for references.