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 (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 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 healthcare 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 healthcare 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 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 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 on vaccines at https://mothertobaby.org/fact-sheets/vaccines-pregnancy/pdf/.

**How does pregnancy affect Gaucher disease?**

Most women with Gaucher disease will have healthy children. For some, new symptoms of Gaucher can start during a pregnancy. Women with Gaucher disease are at an increased risk to have bleeding, postpartum infection and bone disease. Some studies have found an increased chance for miscarriage in women with Gaucher disease. The risk...
for other pregnancy complications, including birth defects, 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?

Ideally, you should discuss all of your medications with your healthcare providers before trying to get pregnant. If you become pregnant before talking with your healthcare provider, given them a call as soon as you find out. 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, Miglustat, and Bisphosphonates.

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 healthcare provider about your breastfeeding questions.

Please click here for references.