Enzyme Replacement Therapy for Treatment of Gaucher Disease

This sheet talks about exposure to enzyme replacement therapy (ERT) in a pregnancy or while breastfeeding. This information should not take the place of medical care and advice from your healthcare provider.

What is enzyme replacement therapy?

Enzyme replacement therapy (ERT) is a treatment for some lysosomal storage disorders. This sheet focuses on ERTs used to treat Gaucher disease. People with Gaucher disease do not have enough of the enzyme called glucocerebrosidase. This enzyme helps to break down fatty substances in the body. When the enzyme levels are too low, fatty substances build up in parts of the body and cause damage. For more information, please see the MotherToBaby fact sheet Gaucher Disease at https://mothertobaby.org/fact-sheets/gaucher-disease-pregnancy/.

ERT gives the body a lab-made form of the enzyme that is missing or not working properly. Some of the enzyme replacement therapies for Gaucher disease are imiglucerase (Cerezyme®), velaglucerase alpha (VPRIV®), alglucerase (Ceredase®), eliglustat (Cerdelga®), taliglucerase alfa (Elelyso®) and Miglustat (Zavesca®). In many individuals with Gaucher disease, especially Type I, enzyme replacement therapy can reduce the size of the liver and spleen and can help maintain normal blood factors.

I have Gaucher disease and am currently on ERT. I would like to become pregnant. What should I do?

Discuss the appropriate treatment for your pregnancy with your healthcare team. An individual with Gaucher disease who is planning a pregnancy might consider enzyme replacement therapy before trying to get pregnant so that they can be as healthy as possible during pregnancy. If you have Gaucher disease but have not been experiencing symptoms, it may not be necessary to start enzyme replacement therapy.

While ERT hasn’t been well studied for use during a pregnancy, there are reports of individuals who have successfully continued their treatment throughout pregnancy. Some treatments have more information available on use while pregnant than others. If you become pregnant on ERT, discuss continuation of treatment throughout pregnancy with your healthcare providers. You may also contact a MotherToBaby specialist to see what information is available on the specific ERT you are using or are planning to start.

I take ERT. Can it make it harder for me to get pregnant?

The current data does not suggest that ERT affects one’s ability to become pregnant.

I just found out that I am pregnant. Should I stop ERT?

You should not stop taking this medication without first talking to your healthcare providers. Studies have suggested that treatment with enzyme replacement prior to and during pregnancy can help the person who is pregnant to be in their best health to deal with the demands a pregnancy puts on the body. Continued use can reduce the chance of complications during pregnancy and delivery. Treatment during pregnancy might decrease the risk for miscarriage and bleeding that can be related to having Gaucher disease.

Does taking ERT increase the chance for miscarriage?

Miscarriage can occur in every pregnancy. Current data doesn’t indicate that ERT increases the chance for miscarriage. Individuals treated with ERT have had similar live birth rates to those not treated with ERT.

Does taking ERT during pregnancy increase the chance of birth defects?

In every pregnancy, there is a 3-5% chance of having a baby with a birth defect. This is called the background risk. There are no published studies that have addressed the question of birth defects and the use of enzyme replacement therapy. A small number of case reports have not suggested an increased chance for birth defects.

Could taking ERT cause other pregnancy complications?

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The data does not suggest that ERT increases the chance for pregnancy complications. ERT has actually been shown to decrease bleeding during pregnancy, delivery, and postpartum. In this regard it is thought that ERT can decrease pregnancy complications that can arise due to bleeding and infection in people with Gaucher disease.

**Does taking ERT in pregnancy cause long-term problems in behavior or learning for the baby?**

Based on the data available, there is not enough information to know if ERT can cause behavior or learning issues.

**Can I breastfeed while taking ERT?**

ERT has not been well studied for use during breastfeeding but there are some case reports of infants who had no health problem who had exposure to ERT while nursing. The replacement enzyme is similar to the naturally occurring enzyme in breast milk and in the infant. Furthermore, the enzyme is likely to be digested (broken down) in the infant’s gastrointestinal (stomach and intestines) tract. This information suggests that breastfeeding would be of low risk to the nursing infant. Talk with your healthcare providers or a MotherToBaby specialists about the specific product that you use. Be sure to talk to your healthcare provider about all your breastfeeding options.

**Is there a concern if my partner was on ERT when I got pregnant?**

There are currently no studies looking at men on enzyme replacement therapy at the time of conception. In general, exposures that fathers or sperm donors 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.