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 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/pdf/.

ERT gives the enzyme that is missing or not working properly with a lab-made form of the enzyme. ERTs are usually given IV (intravenously) every two weeks. Some of the enzyme replacement therapies for Gaucher disease are imiglucerase (Cerezyme®), velaglucerase (VPRIV®), alglucerase (Ceredase®), eliglustat (Cerdelga®) and taliglucerase alfa (Elelyso®). 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.

A woman with Gaucher disease who is planning a pregnancy may consider enzyme replacement therapy before trying to get pregnant so that she can be as healthy as possible in a pregnancy. If you have Gaucher disease but have not been experiencing symptoms, it may not be necessary to start enzyme replacement therapy in pregnancy. However, if symptoms do start in pregnancy, enzyme replacement therapy can be considered.

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

Discuss the appropriate treatment for you with your healthcare team. While ERT hasn’t been well studied for use during a pregnancy, there are reports of women 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 have been on ERT and just found out that I am pregnant. Should I stop?

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 woman to be in her 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.

Can taking ERT during pregnancy cause a birth defect in my baby?

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. 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.

Can I continue ERT while I am breastfeeding?

There are no studies looking at enzyme replacement therapies and breastfeeding. There are some case reports of infants who had no health problem when nursing while their mothers took ERT. Based on a couple case reports,
there was only a small amount of the enzyme detected in the first milk produced following administration of the enzyme to the mother. 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 (GI) tract. All of this information suggests that breastfeeding would be a very low risk to the nursing infant. 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 have are unlikely to increase risks to a pregnancy. For more information, please see the MotherToBaby fact sheet ‘Paternal Exposures and Pregnancy’ at https://mothertobaby.org/fact-sheets/paternal-exposures-pregnancy/pdf/.

Selected References: