



MotherToBaby

Medications & More During Pregnancy & Breastfeeding  
Ask The Experts

Fact Sheet

by the **Organization of Teratology Information Specialists (OTIS)**

For more information about us or to find a service in your area,  
call **(866) 626-6847**. Visit us online at **www.MotherToBaby.org**.

Find us! Facebook.com/MotherToBaby or @MotherToBaby on Twitter

## Enzyme Replacement Therapy for Treatment of 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 enzyme replacement therapy (ERT) 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 enzyme replacement therapy?***

Enzyme replacement therapy is a treatment of 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 <http://mothertobaby.org/fact-sheets/gaucher-disease-pregnancy/pdf/>.

The treatment replaces the enzyme that is missing or not working properly with a lab-made form of the enzyme. This medicine (the enzymes) is usually given IV (intravenously), in high doses every two weeks. Some brand names for enzyme replacement therapy are Cerezyme® (imiglucerase), VPRIV® (velaglucerase) and Ceredase® (alglucerase). 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 enzyme replacement therapy. I would like to become pregnant. What should I do?***

Current guidelines suggest that a woman with Gaucher disease who is planning a pregnancy should consider enzyme replacement therapy before trying to get pregnant so that she can be as healthy as possible in a pregnancy. If you become pregnant on enzyme replacement therapy, discuss continuation of treatment throughout pregnancy with your health care provider.

### ***I have been on enzyme replacement therapy and just found out I am pregnant. Should I stop?***

You should not stop taking any medication without first talking to your health care provider. The current recommendation is to not stop treatment if pregnancy occurs. Studies suggest that continued use reduces 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.

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. 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 pregnancy puts on the body.

### ***Can taking enzyme replacement therapy during pregnancy cause a birth defect in my baby?***

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 the infant, and the enzyme is likely to be digested (broken down) in the infant's gastrointestinal tract. All of this information suggests that breastfeeding would be a very low risk to the nursing infant. Be sure to talk to your health care 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 <http://mothertobaby.org/fact-sheets/paternal-exposures-pregnancy/pdf/>.

**Selected References:**

- Dornelles AD, et al. 2014. Breastfeeding in Gaucher disease: is enzyme replacement therapy safe? Clin Ther.36:990-1.
- Elstein D et al. 2014. Outcome of pregnancies in women receiving velaglucerase alfa for Gaucher disease. J Obstet Gynaecol Res.40(4):968-975.
- Gene Review: Gaucher Disease: ncbi.nlm.nih.gov access on 5/31/2011.
- Giannubilo SR, et al. 2015. Replacement therapy for Gaucher disease during pregnancy: A case report. J Reprod Infertil 16(1): 53-57.
- Granovsky-Grisaru, S et al. 2011. The management of pregnancy in Gaucher disease. Eur J Obstet Gynecol Reprod Biol. 156: (1) 3-8.
- Korkmazer E, et al. 2015. Pregnancy and lactation in a patient with Gaucher disease receiving enzyme replacement therapy: Case report. Turkey J Clin Obstet Gynecol.25:224-6.
- Mamopoulos A M et al. 2009. Gaucher disease and pregnancy. J Obstet Gynaecol 29: 240-242.
- Sekijima Y et al. 2010. Successful pregnancy and lactation outcome in a patient with Gaucher disease receiving enzyme replacement therapy, and the subsequent distribution and excretion of Imiglucerase in human breast milk. Clin Ther 32(12): 2048-2052.
- Zimran, A et al. 2009. The female Gaucher patient: the impact of enzyme replacement therapy around key reproductive events. Blood Cells Mol Dis. 43:264-288.

December, 2016