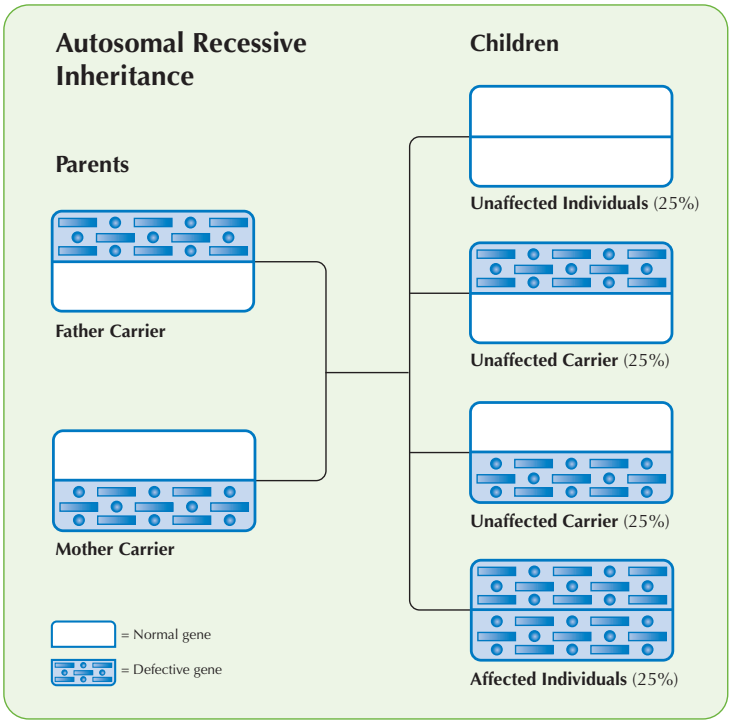
POMPE Connections

Having children when you have Pompe disease

Pompe disease is an inherited disorder, which means it can be passed on to children when both parents have the genetic defect that causes the disease. For this reason, men and women with a family history of Pompe disease may be concerned about having children. Partners of people with Pompe disease may want to know if they are carriers of the defective gene. Women who have Pompe disease may also worry about the health risks of becoming pregnant. If you are thinking about having children, it is important to be aware of both the chances of passing on the disease, and the problems that could arise before, during, and after pregnancy. If you already have children, you may want to know their risk for having the disease or passing it on. This handout talks about some of the issues you will want to think about. It also describes the tests that can help predict whether your baby will be affected by Pompe disease.



Q How does someone get Pompe disease?

A Pompe disease is one of many rare disorders that are passed down through families because of a mutation, or change, in the genes. Genes are made of DNA, the chemical material that determines the unique features of each human being. Each gene contains codes for making a protein that helps the human body function. But when a mutation in the gene occurs, it disrupts this process. Pompe disease is caused by a genetic mutation that blocks the production of an enzyme (a type of protein) called *acid alpha-glucosidase*. This can lead to muscle damage throughout the body. You can only get Pompe disease when you inherit, or receive, 2 copies of the defective gene – 1 from each parent, as shown in the diagram above. This is called *autosomal recessive inheritance*. (Autosomal means the defective gene affects both sexes equally. Recessive means 2 defective copies of the gene are needed to produce the disease.) If you inherit a copy of the defective gene from only 1 parent, you do not get Pompe disease, but you will be a carrier. Although carriers will not develop symptoms of Pompe disease, they can pass on the disease to their children if their partner is also a carrier of the defective gene. While all of us carry mutations in genes, it is rare to find a partner who carries a mutation in the same gene.

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Other names for Pompe disease

Acid alpha-glucosidase deficiency, acid maltase deficiency (AMD), glycogen storage disorder (GSD) type II, glycogenosis II, and lysosomal alpha-glucosidase deficiency. In different parts of the world, Pompe may be pronounced *"pom-PAY," "POM-puh,"* or *"pom-PEE."*

Refer to the diagram on the previous page. If you and your partner are both carriers, with *each* pregnancy you will have:

- A 25% chance of having an unaffected child who inherits 2 normal copies of the gene
- A 50% chance of giving birth to an unaffected child who is a carrier (who inherits the defective gene from only 1 parent)
- A 25% chance of having a child with Pompe disease

It is possible that someone with Pompe disease could have a partner who is a carrier. In truth, the chance of this happening is low. But if it were the case, there is a 50% chance of having either a child with the disease or an unaffected child who is a carrier.

And if 1 parent has the disease and the other is *not* a carrier, all of their children will be carriers but none will have Pompe disease.

Q **Are there tests to determine if my partner and I are Pompe carriers?**

A The only way to know for sure if someone is a carrier of the genetic mutation that causes Pompe disease is by doing DNA testing, or direct mutation analysis. This involves taking a sample of blood, separating the DNA from the cells, and then looking for the specific mutations that are known to cause Pompe disease. Molecular testing of DNA mutations is possible because more than 150 mutations of the GAA gene have been identified in people with Pompe disease. Some of these mutations are limited to particular ethnic groups.

Since Pompe disease is so rare and the risk for being a carrier is so small, carrier testing is not done unless there is a family member with the disease whose mutations are known. DNA testing can tell you if you or your partner are carriers. It can also help you know your risk for having a baby with Pompe disease. If you already have children, you can learn about *their* chances for having the disease or being carriers. To find out more about carrier testing for Pompe disease, see *Where to learn more* on page 4.

Q **I have Pompe disease and I'm thinking about having a baby. My healthcare provider has advised me to get genetic counseling before I get pregnant. How would that help me?**

A If you or anyone else in your family has Pompe disease, or if you think you may be a carrier, genetic counseling can help you understand your chances for having a baby with the disease. Meeting with a genetic counselor **before you get pregnant** will help you sort out all the issues that may affect your decision to have children. The healthcare provider can explain the benefits and risks of DNA testing and help you understand what the results may mean.

If you are already pregnant, the genetic counselor can talk with you about prenatal screening for your unborn child. Should you choose to go ahead with genetic testing, the genetic counselor will make appointments for the tests and provide the support you need once you get the results. For example, if you find out your unborn child is affected by Pompe disease, the

genetic counselor can help you explore your options and cope with the difficult choices ahead of you. Since the process of getting tested and waiting for results takes time, it is important to seek genetic counseling as early as possible.

Q **Can I get pregnant if I have Pompe disease?**

A Pompe disease does not appear to affect fertility (the ability to conceive a child). And women with Pompe disease do not seem to have a higher risk for miscarriage (loss of the pregnancy). However, if 1 partner has severe muscle weakness, scoliosis (curvature of the spine), or contractures (muscle tightness), having sexual intercourse may be difficult. Also, if you are concerned about your risk for having a child with Pompe disease (for example, because you and your partner are both carriers), you may want to consider other options, such as adoption or conception with donor eggs or sperm. It is important to explore these choices with your healthcare provider and a genetics counselor so that you can make informed decisions.

Q **I have Pompe disease and I want to start a family. How will pregnancy affect my health?**

A Though having Pompe disease should not affect your ability to get pregnant or carry a pregnancy to term, there are some health concerns to be aware of, especially if you are severely affected.

Weight gain: The biggest concern is the extra weight you will gain when you are pregnant. If you already have a lot of muscle weakness, the weight you gain during your pregnancy can cause lower back pain and make it harder to walk or keep your balance. Scoliosis can make these problems worse. You may need to use a wheelchair as you get further along in the pregnancy.

Breathing: Whether your muscle weakness is mild or severe, you may have more trouble breathing as you gain weight. Be sure to tell your healthcare provider if you notice these symptoms: shortness of breath, morning headaches, fatigue, dizziness, confusion, or sleeping problems. Using a ventilator may help you breathe more easily. Because of these concerns, you will need to be seen by both a healthcare provider who treats high-risk pregnancies and one who treats your Pompe disease. It is important to have these doctors work together to manage your care.

Delivery: If muscle weakness or scoliosis is severe, your baby may need to be delivered through the abdomen instead of the vagina. This is called a *Cesarean section* (C-section). If a C-section must be performed, it will be necessary to plan in advance for the anesthesia you may need (see the handouts *Common health concerns* and *Breathing problems in Pompe disease).*

Recovery: After the baby is born, it may take longer for you than for others to recover and lose the extra weight you gained. It may also be hard to lift, carry, or nurse your newborn. Be prepared to seek advice from other Pompe parents and your healthcare team, and to get help at home if you need it.

Q **Is there a way to find out if my unborn child has Pompe disease?**

A Yes, there are 2 prenatal screening tests that can be done early in pregnancy to see if your unborn child (the fetus) is affected with Pompe disease. Because there is a slight risk for miscarriage with these tests, they are usually done only if the baby is clearly at risk for the disease — when 1 parent has Pompe disease, both parents are carriers, or you already have a child with the disease. Both tests give accurate results, but one can be done earlier than the other.

Chorionic villus sampling, or CVS, is done before the 12th week of pregnancy. This test involves taking a small sample of tissue from the growing placenta (the sac that surrounds the fetus) and checking to see if the cells contain acid alpha-glucosidase. DNA testing may also be done to compare the DNA from the fetus with the DNA of the parents and an affected sibling (a brother or sister of the unborn child).

Another prenatal test, called *amniocentesis,* is done around the 15th week of pregnancy. It checks for enzyme activity and allows for DNA analysis by testing cells taken from fluid in the womb. The results of these prenatal tests can help guide choices about the pregnancy and prepare for the baby's arrival.

Where to learn more

These groups can help you find answers to any other questions you may have about pregnancy or family planning issues related to Pompe disease:

- The **International Pompe Association (IPA)** is a global federation of Pompe disease patient groups. The IPA helps patients, family members, and healthcare providers from around the world share their experiences and knowledge across continents and cultures. To find the contact for your country, visit the IPA Web site at www.worldpompe.org
- The **Muscular Dystrophy Campaign** offers fact sheets on genetic testing, inheritance patterns, and pregnancy for people affected by neuromuscular diseases. Visit www.muscular-dystrophy.org and click on "Information and resources"
- The **National Society of Genetic Counselors (NSGC)** has an online directory of genetics counselors in the United States and around the world. To search the listings, visit www.nsgc.org and click on "Find a Counselor"
- The **Pompe Center at Erasmus Medical Center** in the Netherlands maintains the most up-to-date record of the mutations that have been identified for the GAA gene. To learn about advances in research, treatment, and genetic testing for Pompe disease, visit the Pompe Center Web site at www.pompecenter.nl.