

Finding a treating physician

Treatment for Pompe disease is now available to many patients worldwide which is a tremendous accomplishment. However, reimbursement remains an issue in some countries. As a Pompe patient, you are probably anxious to find out more about this opportunity and how you can benefit from Myozyme treatment. Before that can happen, you need to know if Myozyme will be reimbursed. Then you need to find a qualified physician who is willing and able to treat you. That may take a bit of time and effort. Nevertheless, there are resources that can simplify your search. After you've located a physician who will take on your case, you need to be assessed to ensure that you are a candidate to receive the treatment. This handout describes some places where you may locate a physician who can provide you with access to Myozyme, and what sort of medical tests you may need to take.



Q How can I find a physician to evaluate me and treat me with Myozyme?

A Before you can receive treatment with Myozyme, you need to find a physician who is willing to treat you. This varies according to the country in which you live. In some countries, there are expert centers that supervise the treatment of Pompe patients. In other countries, patients must locate a willing physician at a local hospital.

To find an expert in Pompe disease in your country, a good starting point is to contact the International Pompe Association

(IPA) contact person or organization in your country. Go to the website www.worldpompe.org/interpom.html. If you are not able to find your country's contact person, then get in touch with the IPA through the website www.worldpompe.org/secretariat.html.

In some areas, your local Genzyme representative may also be able to assist you in finding an expert.

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."

Q After I locate a physician who agrees to treat me with Myozyme, what are some questions that I may want to ask?

A When you have found a physician who is willing to treat you with Myozyme it's important to have a conversation prior to starting treatment. Be well prepared for this conversation. Make a list of all of your questions, including things like pre-examination, reimbursement, allergic reactions, medical tests and rehabilitation.

Here are some examples of the type of questions you may want to ask. Depending on your country of residence, some questions may be more relevant to you than others:

- Are you also treating other Pompe patients?
- Are you in contact with other specialists who are experienced in Pompe disease and ERT?
- What is your experience with infusion treatments?
- Are you willing to support me with insurance related formalities?
- Do I need to have any medical examinations or tests before the infusion treatments begin?
- On what days, and at what times would it be possible to receive treatment?
- How long will the infusion take?
- Where will the infusion take place – at the nearest hospital or at home?
- What should I expect on an infusion day?
- Will I need a port-a-cath or will I get a new infusion needle every time?
- What will happen if I have an allergic reaction?

- Would it be possible for you to administer the enzyme at my home?
- What happens when you are on vacation? Do you have an acting representative?
- After I start treatment, I may need referrals to other physicians such as rehabilitation specialists, physiotherapists or speech therapists. Can you assist me in these referrals?
- Have you used disease registries in the past? Would you be willing to monitor my disease status using the Pompe Registry?
- Are you willing to register my condition on a regular basis?

Q Who else might look after me besides my physician?

A Once you have been diagnosed with Pompe disease, it is important that you receive multidisciplinary care – that is, medical care from a team of specialists. Typically, a geneticist, a pediatrician or a neuromuscular specialist monitors your progression and provides referrals to various medical specialists who manage specific symptoms such as a physical therapist, speech therapist, dietician, and pulmonologist. Clinicians and allied health care professionals may also provide supportive care. Members of this multidisciplinary team communicate with each other on an ongoing basis throughout your treatment. For more information regarding multidisciplinary care see Pompe Connections at www.worldpompe.org/pompeconnections.html 'Getting the right care for Pompe disease.'

Q **What must I do before I can receive Myozyme?**

A Before you can begin receiving Myozyme, you must have a confirmed diagnosis of Pompe disease. This means that you need a medical report of your enzyme activity and/or a DNA mutation analysis. Your physician may also want you to have several tests before you begin your treatment. These tests may include a heart examination (to measure your heart's rate and rhythm and visualize electrical activity of heart muscle tissue), neurological tests (to measure the strength and function of muscle groups), lung function tests (to record the amount and rate of air that you breathe in and out over specific time periods), blood tests (to measure the level of electrolytes, fats, proteins and enzymes in your blood), a pregnancy test or a hearing test. The results of these tests provide your physician with a baseline or starting point. As your treatment progresses, these tests may be repeated so that your level of progress can be assessed.

Since Myozyme treatment is delivered intravenously (through your bloodstream) your physician will likely examine you to determine how easy it is to access your veins. If your veins are difficult to access, or if you are fearful about receiving needles, your physician may suggest that you have a port-a-cath (or a similar device) implanted. A port-a-cath is a device that is placed in your chest area under the skin surface using local or general anesthetics. The advantage of this device is that it provides easy access to your

veins. The disadvantage is that there is a higher risk of infections compared to an intravenous procedure. You and your physician should discuss this option before any action is taken.

This publication is designed to provide general information in regard to the subject matter covered. It is distributed as a public service by the International Pompe Association, with the understanding that the International Pompe Association is not engaged in rendering medical or other professional services. Medicine is a constantly changing science. Human error and changes in practice make it impossible to certify the precise accuracy of such complex materials. Confirmation of this information from other sources, especially one's physician, is required. Please keep in mind that the effectiveness of Myozyme varies from person to person.