

FINDING A TREATING PHYSICIAN

Treatment for Pompe disease by Enzyme Replacement Therapy (ERT) 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 the benefits from Enzyme Replacement (ERT) treatment. Before that can happen, it is important to know if Enzyme Replacement in your country will be reimbursed. Then you need to find a qualified physician who is willing and able to treat Pompe. That may take a bit of time and effort. Nevertheless, there are resources that can simplify your search. After you have located a physician, an assessment will be made to ensure that you are a candidate to receive the treatment. This handout describes some places where a physician can be located who can provide you with access to Enzyme Replacement, and the list of medical tests that might be necessary.



Q: How can I find a physician to evaluate me and treat me with Enzyme Replacement?

A: Before receiving treatment with Enzyme Replacement, it is important to find a physician who is willing to treat Pompe disease. 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. <https://www.worldpompe.org/>

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

A: Once a physician is found who is willing to treat you with Enzyme Replacement it is 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?

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- 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, infusion center 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?
- 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?
- Are you willing to monitor my condition on a regular basis?

Q: What must I do before I can receive Enzyme Replacement?

A: Before beginning Enzyme Replacement, it is necessary to have a confirmed diagnosis of Pompe disease. This means that a medical report of your enzyme activity and/or a DNA mutation analysis. Your physician may also want to run additional 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 to determine the level of progression.

Since Enzyme Replacement 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 the chest area under the skin surface using local or general anesthetics. The advantage of this device is that it provides easy access to veins. The disadvantage is that there is a higher risk of infections compared to an intravenous procedure. Your physician should discuss this option with you before any action is taken.

Q: Who else might look after me besides my treating physician?

A: Once having a diagnosis of Pompe disease, it is important to receive multidisciplinary care – that is, medical care from a team of specialists: such as; 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 treatment. For more information regarding multidisciplinary care see Pompe Connections '*Getting the right care for Pompe disease.*'

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