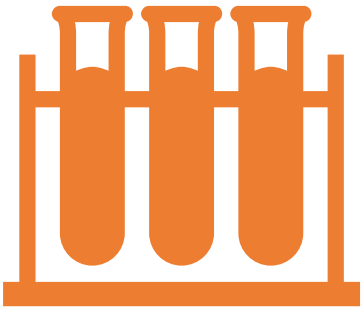
POMPE Connections

ABOUT ENZYME REPLACEMENT THERAPY

As a person diagnosed with Pompe disease, you want to understand as much as possible about the treatment that you may receive. Enzyme replacement therapy (ERT) represents the result of years of scientific research and development. Many healthcare professionals, science experts and patients (through participation in clinical trials) around the world have contributed to the development of these medicines. This brochure offers insight into what an enzyme replacement therapy is and how it works in the human body.



Q: What is enzyme replacement therapy?
A: Enzyme replacement therapy is a medical treatment that replaces a deficient or absent enzyme.

In the case of Pompe disease, one of the lysosomal enzymes called acid alpha glucosidase or GAA is deficient or missing. As a result, glycogen builds up within the lysosome in the cell. This is usually seen in muscle tissue in the body, such as cardiac, respiratory, skeletal and smooth muscles (muscles found in blood vessels, the bladder or the gastrointestinal tract).

With ERT, a patient with Pompe disease receives regular amounts of the deficient GAA enzyme using a genetically engineered form of the enzyme. The enzyme therapy is delivered intravenously (through the bloodstream). The enzyme travels to the muscles and breaks down the glycogen that causes damage when it builds up in the cells.

Enzyme replacement therapy is a lifelong treatment that is given at regular intervals (for example, twice a month). The total dose is based on the patient's weight.

ERT's are also available for some other lysosomal storage disorders such as Gaucher disease, Fabry disease, and MPS diseases (I, II & VI).

Q: How does Enzyme Replacement Therapy work?
A: Enzyme Replacement Therapy provides you with the enzyme that you are lacking. Upon being infused into a patient, it travels through the blood stream, across the blood vessel barrier and into an area of the body called interstitial space.

The final destination for ERT is the microscopic region (or sub-organelle) of a muscle cell called a lysosome. The lysosome is where the glycogen accumulates. Once the ERT arrives in the lysosome, it takes over the job of the absent natural enzyme alpha-glucosidase. That is, it breaks down the buildup of glycogen into glucose.

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Q: What is Enzyme replacement used for?
A: If you have Pompe disease, then your body has a deficiency of an enzyme called alpha-glucosidase. This enzyme normally breaks down glycogen (a carbohydrate) into glucose. If the enzyme is not present, glycogen builds up in certain tissues, particularly your heart (more common in infants) and muscle tissue (including the diaphragm, the main breathing muscle under the lungs, as well as skeletal muscles). The progressive build-up of glycogen causes a wide range of signs and symptoms, including an enlarged heart, breathing difficulties and muscle weakness. This may cause severe disability and even premature death. Enzyme replacement therapy replaces the missing or deficient enzyme.

Q: How is Enzyme Replacement Therapy given to Pompe patients?
A: Enzyme replacement therapy is administered through the blood stream, either through an IV (intravenous line), or through a special access called a port-a-cath. The prescription and administration of ERT is supervised by a physician with experience in the management of patients with Pompe disease, or other inherited diseases of the same type. This is usually a Geneticist, a pediatrician or Neuromuscular specialist.

Q: What is the process for developing new treatments?
A: The process for developing new treatments for diseases starts in the laboratory where researchers begin to test their theories. This is followed by animal studies and, if these go well, by research studies in humans called clinical trials. The purpose of these studies is to gather information about the safety of the therapy and how well it works. It takes many years of strict and careful testing before an experimental treatment is approved for use in humans. To learn more, read the Pompe Connections brochure entitled 'Medical Progress in Pompe Disease.'