Treating Fabry Disease

Fabry disease is a rare, genetic condition which is estimated to affect around **11N 100,000** people. In Fabry, an enzyme called α -galactosidase A (α -Gal A) is missing or there is a reduced amount. This means that the body cannot break down a certain type of fat called globotriaosylceramide (Gb3). Gb3 continues to build-up in body cells causing damage to tissues and organs. Gradually, this leads to a range of physical symptoms and complications, which vary from one person to another.¹

Treatment Approaches

There is no cure for Fabry disease. Current treatments such as Enzyme Replacement Therapy (ERT) and chaperone therapy provide the enzyme needed to break down the accumulation of Gb3 on an ongoing basis. Other treatments are being studied including gene therapy, a one-time treatment to provide continuous production of the α -Gal A enzyme.





Enzyme Replacement Therapy



Enzyme Replacement Therapy (ERT) is the current standard of care for Fabry disease.

The goal is to provide the missing α -Gal A enzyme in the blood through regular IV infusions, usually once every two weeks.

Gene Therapy

Gene therapy is a way of altering the genetic instructions inside the body's cells to treat or stop disease.

- 1. To introduce a corrected copy of a defective gene into a patient's cells, a new healthy gene is created in a laboratory.
- 2. The healthy gene is placed in a modified (harmless) version of a virus called a "vector" to carry the working gene into targeted cells.
- **3.** Depending on the gene therapy, vectors target specific cells in the body, such as cells in the liver or heart.
- 4. The new working gene instructs cells to make the α -Gal A enzyme and release it via the bloodstream for delivery to other organs.





Ex Vivo and In Vivo - In vivo and ex vivo approaches can be used to deliver the working gene into the cells with new instructions. In vivo means that the treatment is delivered directly into the body. Ex vivo means the person's own cells are modified outside the body, and then returned.²

Multiple gene therapy approaches are being studied as one-time treatments that provide stable, continuous production of α -Gal A to slow or stop the progression of Fabry disease.

New approaches to the treatment of Fabry disease are ongoing. Visit this resource to learn more about clinical trials or support for people living with Fabry disease:

Fabry International Network | <u>fabrynetwork.org</u>

References

- 1. Fabry International Network. What is Fabry? Retrieved from https://www.fabrynetwork.org/what-is-fabry/
- 2. American Society of Gene & Cell Therapy. Fabry Disease and Gene Therapy. Retrieved from https://asgct.org/global/documents/ patient-ed-infographics/sept-launch-website-material/fabry-disease-and-gene-therapy.aspx