

α -Gal A enzyme/alpha-galactosidase A enzyme

An enzyme involved in breaking down complex molecules to simpler molecules within the body, notably so they can be eliminated. α -Gal A helps prevent a fatty substance called Gb3 (globotriaosylceramide) from building up in the tissues, which causes disease in vital organs.

Classical Fabry disease

People with classical Fabry disease do not produce any α -Gal A enzyme or very little enzyme. The disease can significantly impact the normal functioning of many vital organs in the body, mainly the heart, kidneys and skin.

Clinical study

Medical research studies involving people. Researchers test possible new drugs in the laboratory to begin with and then, if they look promising, they are carefully tested in people in clinical studies.

Discontinuation/study discontinuation/termination

If you decide to leave the clinical study, or if you are withdrawn from it.

Efficacy

The ability of the medicine to achieve the intended result.

Enzyme

A substance that acts as a catalyst (stimulus) in living organisms, regulating the rate at which chemical reactions happen.

Enzyme replacement therapy (ERT)

A medical treatment which replaces an enzyme that is deficient or absent in the body. It is a treatment option for people with Fabry disease.

Fabry disease

A debilitating rare genetic disorder that can significantly impact the normal functioning of many vital organs in the body, mainly the heart and kidneys.

Gb3 (globotriaosylceramide)

A fatty substance that, in Fabry disease, can build up in the tissues of the body when the α -Gal A enzyme is in low quantities in the blood, causing disease in vital organs.

Gene therapy

The treatment of disease by delivery of a new gene into a patient's cells to replace an incorrect or damaged gene. Most often, gene therapy works by introducing a corrected copy of a defective gene into the patient's cells, without removing or modifying DNA. The goal of gene therapy is to treat, or potentially cure, a genetic disease by adding back a normal copy of the gene involved in the disease.

Genetic testing

Fabry International Network

Registered address Alice Nahonlaan 7, 9120 Melsele - BE

Visiting address: Floralaan 35A, 9120 Beveren - BE

Charity Registration Number 04080030 The Chamber of Commerce Drenthe The Netherlands

info@fabrynetwork.org

www.fabrynetwork.org

The study of a person's DNA in order to identify genetic differences or susceptibility to particular diseases or abnormalities.

GLA gene

The GLA gene is responsible for providing the body with instructions for cells to produce an enzyme called alpha-galactosidase A (α -Gal A).

Infusion

A medicine administered directly into the vein with a needle or catheter.

Non-classical Fabry disease

People with non-classical Fabry disease, also referred to as late-onset or atypical Fabry disease, have low levels of α -Gal A enzyme, and may have a more variable disease course than those with classical Fabry disease. They are generally less severely affected and disease may be limited to a single organ.

Phase I/II clinical study

A study that tests the safety, side effects and best dose of a new treatment. Phase I/II clinical trials can also test how well a certain type of disease responds to a new treatment.

Prednisone

A steroid that can be used to decrease the body's immune response and decrease inflammation.

Progressive disease

Sometimes also called a chronic disease. The symptoms may gradually worsen over time as the disease progresses. For example, in Fabry disease the symptoms may get worse over time as more and more of the body's cells and organs are affected by the build-up of the fatty substance Gb3. The symptoms of many chronic diseases can be improved with various treatments.

Quality of life

The standard of health, comfort and happiness felt

Safety (clinical study)

How safe an investigational agent is, and what side effects it may cause.

Screening

The process to identify whether a person is suitable to take part in a clinical study.

Side effect [treatment]

An undesirable effect caused by a treatment.

Tolerability

The degree to which side effects of a treatment can be tolerated (accepted) by the participant in a clinical study.

Fabry International Network

Registered address Alice Nahonlaan 7, 9120 Melsele - BE

Visiting address: Floralaan 35A, 9120 Beveren - BE

Charity Registration Number 04080030 The Chamber of Commerce Drenthe The Netherlands

info@fabrynetwork.org

www.fabrynetwork.org