Inherited Metabolic Disease Service

Patient Information

Fabry disease

What do I need to do next?

Do ask your doctor or nurse for more information.

Do attend your regular appointments with the lysosomal disorders team so that we can ensure you receive the most appropriate care for your current difficulties and can keep you informed of any new developments.

You may find it helpful to contact your patient support group.

MPS society
Tel: 0345 3899901
Email: mps@mpssociety.org.uk
https://www.mpssociety.org.uk/

Contact us

LDU Box 135, Addenbrooke’s Hospital,
Hills Road, Cambridge. CB2 0QQ
Tel: 01223 274634
Email: lducambridge@nhs.net
www.cuh.nhs.uk/addenbrookes-
hospital/services/lysosomal-disorders

Please note the department is open Monday to Friday 08:30-17:00.

If you are unwell outside of these hours you should seek medical advice from your local healthcare team.

If your emergency doctor needs specific advice they should call the hospital switchboard on 01223 245151 and ask for the metabolic consultant on call.

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What is Fabry disease?

Fabry disease is a lysosomal storage disorder. Your body is made up of trillions of individual cells and each cell contains a small part called the lysosome. This is primarily responsible for breaking down and recycling worn out cell components. Inside the lysosome are enzymes which perform this very important housekeeping role. In Fabry disease the necessary enzyme is not working effectively allowing these products to build up and become stored in very specific areas of your body. There are many different enzymes in your lysosomes, and deficiencies of each enzyme will cause a different disease. All of these different diseases together are called lysosomal storage disorders.

How will this affect me?

In Fabry disease the cells most commonly affected can be found in many places throughout your body including your kidneys, heart, nerves, brain, gut and blood vessels.

This means you may experience or be at risk of the following:

- Kidney failure
- Stroke
- Pain in feet/hands
- Gut disturbance
- Abnormal heart rhythms/heart failure
- Hearing loss/tinnitus
- Low mood
- Abnormal sweating
- Fatigue

The progress of the disease in adults is extremely variable with some people not really noticing any symptoms whilst others are more severely affected.

If you have any questions about the effects and progression of the disease please do ask your doctor or specialist nurse

Is there any treatment?

Treatment is currently available for Fabry disease in the form of enzyme replacement therapy (ERT) or chaperone therapy. All medication is supplied to you directly at home using a medical homecare delivery company.

Enzyme replacement therapy

This is an intravenous infusion usually administered every one to two weeks. It provides you with extra enzyme that will supplement your own and help to reduce the amount of stored material in your cells. This is initially administered in hospital but after that you receive this treatment at home with a homecare nurse who will teach you how to manage your infusions for yourself when you are ready.

Chaperone therapy

This is a tablet treatment that you take regularly on specific days of the week. It helps by stabilising your own enzymes and helping them to perform their usual function.

This treatment is only helpful for certain types of disease and your doctor/nurse will tell you more about this if this treatment is suitable for you.

You can only receive this treatment for your Fabry disease if you attend one of the UK national referral centres at least once per year for regular assessment and review.

How did I get Fabry disease?

Your disease is a genetic condition which means that you inherited it from your parents. There are instructions in your genes for making lysosomal enzymes and a small change in the DNA can be enough to stop the enzyme from working properly. You have two copies of every gene, having received one from each parent.

There are two specific chromosomes that determine whether you are male or female – males are XY and females are XX. In Fabry disease the DNA change is found on the X chromosome.

If you are male and have an X chromosome that carries the DNA change you will have the disorder because you do not have another X chromosome that could take over.

If you are female and have an X chromosome that carries the DNA change you may have some symptoms of the disorder although generally not as severely because your other X chromosome can make effective enzyme.