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FABRY DISEASE IN OUR FAMILY

You have been given this leaflet because maybe you, or someone in your family has been diagnosed with Fabry. It is important that you read this leaflet carefully as it contains information that is relevant to you.

What is Fabry Disease?

Fabry Disease, also known as Anderson-Fabry Disease, is one of a number of disorders known as lysosomal storage disorders. It is caused by deficiency of an enzyme called alpha-galactosidase A, which is essential in the breaking down of waste products within the body. These waste products build up in the cells causing progressive damage.

Fabry typically affects males but it is now clear that females may show signs and symptoms of the condition similar to that seen in males. These symptoms tend to occur at a later age, but this is not always the case.

What are the symptoms?

The first signs and symptoms of Fabry can start during early childhood although they can often be mistakenly diagnosed as growing pains and other common complaints. However, not all patients will develop symptoms this early and the symptoms can be variable. It is unlikely that any two patients will be affected in the same way.

The signs and symptoms of Fabry include:

- Burning sensations or pain in your hands and feet
- Small raised dark-red spots (angiokeratomas) on your body
- Fever
- Sweating too little, intolerance to heat
- Abdominal pain, vomiting and diarrhoea
- Impaired hearing
- Alterations in the eye, leading to opacity of the cornea
- As they get older, patients typically develop more serious symptoms that affect the kidneys, heart and brain.

Fabry is not common and the symptoms can often be mistaken for other illnesses. This may make an accurate diagnosis difficult for non-specialist doctors so it can be important to get expert assessment.

Treatment

Although there is no cure for Fabry there is treatment available. Enzyme Replacement Therapy is designed to replace the missing enzyme alpha-galactosidase A and is given via repeated intravenous infusion in order to stabilise and reduce the symptoms and clinical manifestations associated with the condition. Other treatments such as Chaperone Therapy are in development.

The sooner Fabry is diagnosed, the earlier treatment can begin. If you would like more information on treatment options and clinical trials, please contact the MPS Society. If you have been diagnosed with Fabry Disease you may want to consider discussing this with your relatives. Not only will this help your family understand about Fabry, but it also is important because Fabry is a genetically inherited condition.

Inheritance of Fabry

Fabry is not contagious, but it is a genetically inherited condition which means that you can inherit it from your parents. It may be passed from parent to child for several generations, resulting in many family members being affected, for example, nephews, nieces, sisters, brothers etc.

The condition is linked to the x- chromosome (females have two x- chromosomes and males have one x- and one y- chromosome). This means that if a father has the condition he will always pass it to all of his daughters through his affected x- (female) chromosome. None of his sons will be affected since the father's y- chromosome (which is normal) will be passed on to produce a male child. For an affected female there will be a 50/50 chance with every pregnancy of passing on the condition to her children whether the child is male or female.

Are there tests for Fabry?

Yes. A genetic test can be used to confirm whether or not an individual has Fabry. The doctor may also check the individual's levels of the enzyme alpha-galactosidase A. Levels of this enzyme are usually lower than normal in individuals with the condition.

What should I do now?

As you or someone in your family has been diagnosed with Fabry, it is possible other members of your family may also be at risk of having inherited the condition.

If you suspect this may be the case it is very important that you contact your doctor. As Fabry is very rare, it may be helpful if you take this leaflet to your GP.

Alternatively, there are a number of specialist centres (known as Adviser Group for National Specialised Services or AGNSS) where you can go to be tested and see a specialist in Fabry Disease and a genetic counsellor. A list of these AGNSS centres is shown below.

Adviser Group for National Specialised Services (AGNSS)

Paediatric

Great Ormond Street Hospital, London
Royal Manchester Children's Hospital
Birmingham Children's Hospital

Adult

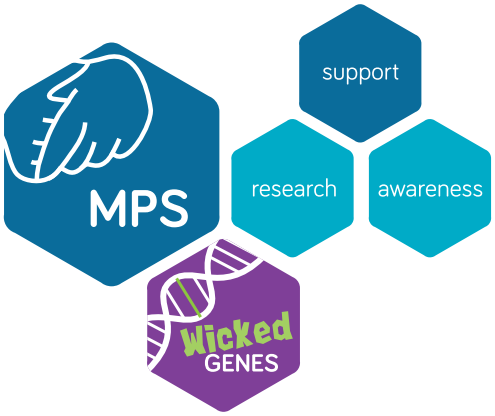
National Hospital, London
Royal Free Hospital, London
Addenbrooke's Hospital, Cambridge
Hope Hospital, Manchester
Queen Elizabeth Hospital, Birmingham

Genetic counsellors can explain the pattern of inheritance and advise about family planning, genetic screening and other issues. The medical specialists can manage your condition and assess for treatment.

There is lots of support available to individuals with Fabry. It is vitally important that once a diagnosis of Fabry Disease is received, that other family members are encouraged to consider undergoing tests. This means that informed decisions can be made over treatment options and reproductive choices.

Further information

For more information, please contact the MPS Advocacy Team by email at **advocacy@mpssociety.org.uk** or by phone on **0845 389 9901**.



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