Anderson-Fabry disease

Also known as angiokeratoma corporis diffusum, alpha-galactosidase A deficiency, Fabry disease.

Fabry disease is a rare genetic condition, belonging to a group of diseases called lysosomal storage diseases.

What causes Fabry disease?

Fabry disease is an x-linked genetic condition. This means it can be passed on from the mother only. Men are more severely affected; women may have minor symptoms or no symptoms at all. The disease is caused by a genetic mutation or mistake in the encoding of an enzyme called alpha-galactosidase A (AGA). This results in reduced production of the enzyme which this in turn leads to accumulation of a fat molecule (globotriaosylceramide) in various tissues throughout the body.

What does Fabry disease look like?

Multiple angiokeratomas are present on the skin in most affected individuals. These are small dark red or purple lesions scattered on the trunk and limbs, predominantly in a "bathing-suit" distribution. The inside of the mouth is sometimes affected. Angiokeratomas first appear at puberty and can increase in size and number with age. They are harmless and painless. Men with Fabry disease may also have dry skin with decreased sweating and poor exercise tolerance.

What other problems can occur with Fabry disease?

Fabry disease can affect many organs of the body and manifestations vary between individuals. The most commonly affected organs include the kidneys and heart. Other affected organs include the gastrointestinal tract, brain and peripheral nervous system and eyes.

How is Fabry disease diagnosed?

The skin manifestations can offer a clue to the diagnosis of Fabry disease. However, the diagnosis is generally made from a blood test which measures blood AGA enzyme levels. A urine sample may also be required.

Skin biopsies show distinctive findings but are not usually required. Once the diagnosis is confirmed, multiple other tests may be required to determine the type and extent of internal organ involvement.

Genetic testing can be performed on the affected person and other female relatives. Prenatal diagnosis is available for subsequent pregnancies. This tests the foetus to see if it has the same genetic mutation.

How is Fabry disease treated?

Disease monitoring and treatment are performed at specialist centres. Treatment options depend on the type and severity of internal organ involvement. Enzyme replacement therapy is available for some affected individuals through the Life Saving Drugs Program.

Angiokeratomas do not require treatment except for cosmetic purposes. They respond well to treatments with vascular laser or electrocautery.

What is the likely outcome of Fabry disease?

The prognosis depends on the severity of internal organ involvement.