Fabry disease, sometimes referred to as Anderson-Fabry disease, is a rare inherited lysosomal disease. In Fabry disease, an enzyme (a-galactosidase) responsible for the breakdown of waste products in the cells is deficient or absent. This leads to an accumulation of waste biological molecules (called globotriaosylceramide or Gb3) in various types of cell in many organs of the body. This gives rise to the symptoms of Fabry disease which may include pain, skin rashes, bowel, kidney and heart problems.

Fabry disease is an X-linked genetic disorder, therefore women with the disease will have one unaffected X chromosome and one affected X chromosome and can pass either of them onto their children. In men with the disease their only X chromosome is affected. In this way they will always pass the affected chromosome to a daughter but will never pass it to a son. If one member of the family is affected by Fabry disease, it is possible that other relatives may also have the condition. Relatives of an affected individual should be encouraged to seek medical advice.

A more detailed explanation of the pattern of inheritance can be found on the Society for Mucopolysaccharide Disease website:

Inheritance of Fabry Disease

Fabry disease is associated with a wide range of symptoms that usually worsen and change with age. In children, pain is often the most noticeable symptom together with spots on the skin (called angiookeratoma) and harmless changes to the eye (often referred to as corneal verticillata). In teenage years, spots may become more widespread, kidney impairment may be noted and fever associated with an inability to sweat and control body temperature may develop. Stomach and bowel problems may also develop (including diarrhoea, constipation and stomach cramps). In adult life, ringing in the ears (tinnitus), heart problems and stroke may occur. Kidney disease often progresses further. It is not surprising that individuals with Fabry disease often feel depressed. It is important to stress that there are many variations in the severity and symptoms between individuals with Fabry disease.

Treatment for Fabry disease is available in the form of Enzyme Replacement Therapy (ERT). Treatment is only available through a national centre. Enzyme replacement therapy replaces the missing a-galactosidase enzyme and is given by intravenous injection (infusion) every two weeks. Initially this is given in a hospital but most patients then progress to treatment at
home. Some patients learn to administer the treatment themselves and enjoy the greater freedom this gives them or have a friend or relative who can assist. Others receive home treatment administered by a nurse specialised in giving intravenous infusions. A private homecare company provides this service. Treatment is lifelong since the enzyme remains deficient throughout life. The enzyme replacement will reduce the amount of waste material that has built up in the various cell types and it is expected that patients will benefit from reduced pain, improved stomach and bowel symptoms and by preventing or stabilising the effects of the disease on the heart and kidneys.