

Rare conditions – lysosomal storage disorders

Most services provided by the NHS are funded by primary care trusts (PCTs) to meet the general requirements of their local population via contracts negotiated with GP practices, hospitals and other healthcare providers. However, the NHS also provides treatments for a number of rare conditions which affect only a small number of patients: these services are commissioned on a national level by the National Commissioning Group (NCG) on behalf of all the PCTs in the country.

The Trust is one of only seven designated centres in the country providing a service for lysosomal storage disorders (LSD) – a group of rare genetic conditions characterised by specific lysosomal enzyme deficiencies.

The Lysosomal Disorders Clinic at the Trust provides services for investigation and management of the diseases which the condition gives rise to in children and adults. The clinic which sees patients from as far afield as Aberdeen and Penzance and is also involved in clinical and academic research.

What is a lysosome?

The human body contains many different cells. A basic cell has many contents including a small compartment called the lysosome – this contains all the enzymes the cell needs to manage its recycling of waste products.

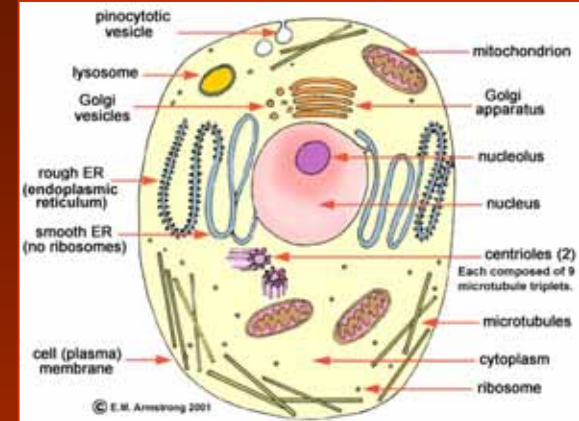
A deficiency of any one of the enzymes within the lysosome causes a build-up of the product it is intended to break down. This product is then stored in different specific sites within the body producing the symptoms experienced by the patient.

What is a lysosomal storage disorder (LSD)?

About one in 7,000 births are affected. LSDs are a group of rare inherited metabolic diseases characterised by an abnormal build up of various toxic materials in the body's cells as a result of enzyme deficiencies. There are nearly 50 diseases and new disorders continue to be identified. The most common of these seen at Addenbrooke's are Gaucher, Fabry and Pompe disease.

Different diseases may affect various parts of the body including the skeleton, brain, skin, heart, liver, kidney and central nervous system.

Patients are cared for by a highly specialised multi-disciplinary team including radiology, pathology, genetic analysis, and other specialties including orthopaedics, respiratory medicine, cardiology, renal medicine, neurology, ophthalmology, physiotherapy, occupational therapy, speech therapy and dietetics.



Treatment

ERT – enzyme replacement therapy is the direct replacement of the missing enzyme

SRT – substrate reduction therapy aims to reduce the amount of waste (substrate) that the cells make so the burden of clearance is reduced

Other – recently licensed therapies and some which are still in research focus mainly on producing an oral treatment. Licensed treatments range from £100,000 to £200,000 per patient per year



A patient receives ERT