Lysosomal Disorders

Lysosomal storage diseases are a group of rare inherited metabolic disorders that are characterized by an abnormal build-up of various toxic materials in the body's cells as a result of enzyme deficiencies. There are nearly 50 of these altogether and new lysosomal storage disorders continue to be identified.

They can be classified by the nature of the primary stored material involved, and can be broadly broken into the following groups:

- Glycosphingolipidoses - Including Gaucher, Fabry, Sandhoff, Tay Sachs, Metachromatic Leukodystrophy & Niemann Pick A & B disease
- Mucopolysaccharidoses - MPS I (Hurler), MPS II (Hunter), MPS III (San Filippo), MPS IV (Morquio), MPS VI (Maroteaux Lamy) MPS VII (Sly)
- Glycoproteinoses - eg Schindler disease
- Other - eg Pompe, Danon, Niemann-Pick C, I Cell, Battens disease

Different diseases may affect various parts of the body, including the skeleton, brain, skin, heart, liver, kidney and central nervous system. Some of the lysosomal disorders now have licensed treatments available, and others are currently the subject of clinical and academic research. Symptom support and management is offered for those disorders currently without an approved treatment.