Mucolipidoses

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Mucolipidoses Information Page.

The mucolipidoses (ML) are a group of inherited metabolic diseases that affect the body's ability to carry out the normal turnover of various materials within cells. In ML, abnormal amounts of carbohydrates and fatty materials (lipids) accumulate in cells. Because our cells are not able to handle such large amounts of these substances, damage to the cells occurs, causing symptoms that range from mild learning disabilities to severe intellectual impairment and skeletal deformities.

The group includes four diseases:

- Mucolipidosis I (sialidosis)
- Mucolipidosis II (inclusion-cell, or I-cell, disease)
- Mucolipidosis III (pseudo-Hurler polydystrophy)
- Mucolipidosis IV

The MLs are classified as lysosomal storage diseases because they involve increased storage of substances in the lysosomes, which are specialized sac-like components within most cells. Individuals with ML are born with a genetic defect in which their bodies either do not produce enough enzymes or, in some instances, produce ineffective forms of enzymes. Without functioning enzymes, lysosomes cannot break down carbohydrates and lipids and transport them to their normal destination. The molecules then accumulate in the cells of various tissues in the body, leading to swelling and damage of organs.

The mucolipidoses occur only when a child inherits two copies of the defective gene, one from each parent. When both parents carry a defective gene, each of their children faces a one in four chance of developing one of the MLs.