Open Peer Review on Qeios

Mucopolysaccharidoses

National Institute of Neurological Disorders and Stroke (NINDS)

Source

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

The mucopolysaccharidoses are a group of inherited metabolic diseases in which a defective or missing enzyme causes large amounts of complex sugar molecules to accumulate in harmful amounts in the body's cells and tissues. This accumulation causes permanent, progressive cellular damage that affects appearance, physical abilities, organ and system functioning, and, in most cases, mental development. Depending on the type of mucopolysaccharidosis, affected individuals may have normal intellect or may be profoundly impaired, may experience developmental delay, or have severe behavioral problems. Physical symptoms generally include coarse or rough facial features, thick lips, an enlarged mouth and tongue, short stature with a disproportionately short trunk (dwarfism), abnormal bone size or shape (and other skeletal irregularities), thickened skin, enlarged organs such as the liver or spleen, hernias, and excessive body hair growth.