Hurler-Scheie Syndrome

National Cancer Institute

Source

National Cancer Institute. Hurler-Scheie Syndrome. NCI Thesaurus. Code C122782.

An autosomal recessive disorder representing the intermediate form of mucopolysaccharidosis type I. It is characterized by deficiency of the enzyme alpha-L-iduronidase. Signs and symptoms include short stature, cloudy cornea, umbilical hernia, joint stiffening, hepatosplenomegaly, and mental retardation.