Scheie Syndrome

National Cancer Institute

Source

National Cancer Institute. Scheie Syndrome. NCI Thesaurus. Code C61265.

An autosomal recessive disorder representing the milder form of mucopolysaccharidosis type I. It is characterized by deficiency of the enzyme alpha-L-iduronidase. Signs and symptoms include broad mouth with full lips, cloudy cornea which may lead to blindness, stiff joints, and hirsutism.