Smith-Lemli-Opitz Syndrome

National Cancer Institute

Source

National Cancer Institute. Smith-Lemli-Opitz Syndrome. NCI Thesaurus. Code C85071.

A rare, autosomal recessive syndrome caused by mutations in the DHCR7 gene. It is characterized by deficiency of the enzyme 3 beta-hydroxysterol-delta 7-reductase resulting in defects in the cholesterol synthesis. It is manifested with multiple congenital malformations, including facial abnormalities, microcephaly, and syndactyly. Behavioral abnormalities may also be present.