Simpson Golabi Behmel Syndrome Type 1

National Cancer Institute

Source

National Cancer Institute. Simpson Golabi Behmel Syndrome Type 1. NCI Thesaurus. Code C118787.

A rare, X-linked inherited syndrome caused by mutations in the GPC3 and GPC4 genes. It is characterized by pre-and postnatal overgrowth, coarse facial features, macrocephaly, macroglossia, congenital heart defects, and intellectual disability.