Immunoglobulin Superfamily Member 1 Deficiency Syndrome

National Cancer Institute

Source

National Cancer Institute. Immunoglobulin Superfamily Member 1 Deficiency Syndrome. NCI Thesaurus. Code C130989.

An X-linked recessive syndrome caused by loss-of-function mutation(s) in IGSF1, encoding immunoglobulin superfamily member 1. This condition can result in central hypothyroidism, macroorchidism, delayed puberty, and variable prolactin deficiency.