Open Peer Review on Qeios

Dyggve-Melchior-Clausen Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Dyggve-Melchior-Clausen Syndrome</u>. NCI Thesaurus. Code C124844.

A rare, autosomal recessive inherited syndrome caused by mutations in the DYM gene. It is characterized by abnormal skeletal development, microcephaly, and intellectual disability.