Open Peer Review on Qeios

Borjeson-Forssman-Lehmann Syndrome

National Cancer Institute

Source

National Cancer Institute. Borjeson-Forssman-Lehmann Syndrome. NCI Thesaurus. Code C157122.

An X-linked recessive condition caused by mutation(s) in the PHF6 gene, encoding PHD finger protein 6. It is characterized by severe intellectual disability, epilepsy, hypogonadism, hypometabolism, and obesity.