Bohring-Opitz Syndrome

National Cancer Institute

Source

National Cancer Institute. Bohring-Opitz Syndrome. NCI Thesaurus. Code C131533.

An autosomal dominant condition caused by mutation(s) in the ASXL1 gene, encoding putative polycomb group protein ASXL1. It is characterized by severe intrauterine growth retardation, profound mental retardation, craniofacial dysmorphisms, and flexion deformities of the upper limbs.