Opitz G/BBB Syndrome

National Cancer Institute

Source

National Cancer Institute. Opitz G/BBB Syndrome. NCI Thesaurus. Code C125487.

An X-linked syndrome caused by mutations in the MID1 gene or autosomal dominant syndrome caused by changes in chromosome 22. It is characterized by ocular hypertelorism, and defects of the larynx, trachea, or esophagus. Most males have hypospadias, cryptorchidism, underdeveloped scrotum, or a scrotum divided into two lobes. Mild intellectual disability and developmental delays occur in approximately half of the affected individuals.