

KBG Syndrome

➤ **What is KBG Syndrome?**

- KBG Syndrome is a rare condition that is mainly characterized by skeletal abnormalities, distinctive facial features, and intellectual disability. Specific signs and symptoms may include delayed bone age, abnormalities of the bones of the spine, ribs, and/or hands, large teeth (macrodonia), short stature, developmental delay, and behavioral or emotional issues. Less common features may include hearing loss, seizures, and congenital heart defects. Currently more than 150 cases have been reported in the medical literature. This disorder can go undiagnosed or misdiagnosed.

➤ **What are the signs/symptoms?**

- The number and severity of symptoms can vary but may include: Craniofacial dysmorphism (physical abnormalities of the head and face), Brachycephaly (flat back of the head), Hypertelorism (widely spaced apart eyes), Strabismus (crossed eyes), Wide bushy eyebrows, Thin, bow-shaped lips, Triangularly shaped face, Macrodonia (abnormally large teeth), Jagged, crowded, misaligned teeth, Short, flattened supporting bones or sockets of the jaw that house the teeth, Microcephaly (small head circumference), Short stature, Speech and hearing impairments, Mild to moderate levels of intellectual disability, Otitis media (recurrent ear infections), Behavioral anomalies (impulsivity, poor concentration, ADHD, temper tantrums, shyness, compulsiveness, aggression, ASD), Epilepsy, Delayed bone age or spinal abnormalities, Syndactyly (webbing of the fingers and toes)

➤ **What is KBG syndrome a result of?**

- Mutations in the ANKRD11 gene and it is inherited in an autosomal dominant manner. Some affected individuals inherit the condition from a parent, while in other people it occurs sporadically.

➤ **What are the complications of KBG Syndrome?**

- Most individuals with this condition have behavioral or emotional difficulties, such as hyperactivity, anxiety, or Autism spectrum disorder. Less common features of KBG syndrome include hearing loss, seizures, and heart defects.

References:

<https://medlineplus.gov/genetics/condition/kgb-syndrome/#frequency>

<https://rarediseases.org/rare-diseases/kgb-syndrome/>

<https://rarediseases.info.nih.gov/diseases/82/kgb-syndrome>

