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KBG SYNDROME FOUNDATION
#everylinkmatters

The KBG Foundation is a 501(c)(3) nonprofit organization, dedicated to providing support, assisting in research programs and advocating to raise awareness about KBG syndrome.

To stay up to date and join the private community follow us on Facebook & Twitter.

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INTERNATIONAL KBG SYNDROME AWARENESS DAY JUNE 11TH

What is KBG Syndrome?

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KBG syndrome is caused by a variant in the ANKRD11 gene at location 16q24.3 (maybe more) which creates a shortening of a scaffolding protein in that region.

This short protein may be completely ineffectual or may be somewhat functioning which could contribute to the variety and severity of the symptoms.

Incidence

New research suggests that less than 1000 individuals worldwide have been diagnosed with KBG syndrome. For no known reason, males seem to be more affected than females.

Appearance

Nearly all patients present with large upper front teeth, bushy eyebrows and triangular faces.

The 'KBG appearance' is quite distinctive and is usually the first noticeable trait along with developmental delay and other bone anomalies

Prognosis

KBG has an excellent prognosis with no known impact on longevity.

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Treatment

There is no singular treatment for KBG syndrome but is determined on a case by case basis as symptoms arise.

Common Traits/Symptoms

Listed in no particular order

Unique Facial Features
- Hairline *(low in front or back of skull)*

Autistic Characteristics *(Behavior, self control, social issues)*

Brachy-clinodactylous 5th finger *(short/curved pinky)*

Macrodontia *(large teeth)*

Abnormal EEG with or without seizures

Cognitive deficits/psychomotor delay

Anteverted nostrils *(upturned nose)*

Hip dysplasia

Cutaneous syndactyly, *(interconnections between the fingers)*

Palatal defects *(including uvula)*

Abnormal spine curvature

Webbed/short neck

Mild synophrys *(unibrow - wide eyebrows)*

Brachycephaly/turricephaly *(head shape)*

Chest abnormalities

Cryptorchidism *(undescended testes)*

Abnormal ribs/vertebrae

Epicanthal folds *(skin covering corner of eye)*

Delayed bone age

Ptosis *(droopy eyelid)*

Prominent/high nasal bridge

Long philtrum *(space between nose and lip)*

Short hand bones

Hearing loss

Thin upper lip

Prominent/anteverted ears

Strabismus

Congenital heart defects