Say-Barber-Biesecker Syndrome

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Ohdo syndrome, Say-Barber-Biesecker-Young-Simpson variant

Description

The Say-Barber-Biesecker-Young-Simpson (SBBYS) variant of Ohdo syndrome is a rare condition characterized by genital abnormalities in males, missing or underdeveloped kneecaps (patellae), intellectual disability, distinctive facial features, and abnormalities affecting other parts of the body.Males with the SBBYS variant of Ohdo syndrome typically have undescended testes (cryptorchidism). Females with this condition have normal genitalia.Missing or underdeveloped patellae is the most common skeletal abnormality associated with the SBBYS variant of Ohdo syndrome. Affected individuals also have joint stiffness involving the hips, knees, and ankles that can impair movement. Although joints in the lower body are stiff, joints in the arms and upper body may be unusually loose (lax). Many people with this condition have long thumbs and first (big) toes.The SBBYS variant of Ohdo syndrome is also associated with delayed development and intellectual disability, which are often severe. Many affected infants have weak muscle tone

(hypotonia) that leads to breathing and feeding difficulties.The SBBYS variant of Ohdo syndrome is characterized by a mask-like, non-expressive face. Additionally, affected individuals may have distinctive facial features such as prominent cheeks, a broad nasal bridge or a nose with a rounded tip, a narrowing of the eye opening (blepharophimosis), droopy eyelids (ptosis), and abnormalities of the tear (lacrimal) glands. About one-third of affected individuals are born with an opening in the roof of the mouth called a cleft palate. The SBBYS variant of Ohdo syndrome can also be associated with heart defects and dental problems.

Frequency

The SBBYS variant of Ohdo syndrome is estimated to occur in fewer than 1 per million people. At least 19 cases have been reported in the medical literature.

Causes

The SBBYS variant of Ohdo syndrome is caused by mutations in the KAT6B gene. This gene provides instructions for making a type of enzyme called a histone acetyltransferase. These enzymes modify histones, which are structural proteins that attach (bind) to DNA and give chromosomes their shape. By adding a small molecule called an acetyl group to histones, histone acetyltransferases control the activity of certain genes. Little is known about the function of the histone acetyltransferase produced from the KAT6B gene. It appears to regulate genes that are important for early development, including development of the skeleton and nervous system.The mutations that cause the SBBYS variant of Ohdo syndrome likely prevent the production of functional histone acetyltransferase from one copy of the KAT6B gene in each cell. Studies suggest that the resulting shortage of this enzyme impairs the regulation of various genes during early

development. However, it is unclear how these changes lead to the specific features of the condition.

Learn more about the gene associated with Ohdo syndrome, Say-Barber-Biesecker-Young-Simpson variant

KAT6B

Inheritance

This condition has an autosomal dominant inheritance pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Almost all reported cases have resulted from new mutations in the gene and have occurred in people with no history of the disorder in their family.

Other Names for This Condition

Blepharophimosis and mental retardation syndrome, Say-Barber/Biesecker/Young-Simpson type
Blepharophimosis-intellectual deficit syndrome, Say-Barber/Biesecker/Young-Simpson type
BMRS
SBBYS
Ohdo syndrome, Say-Barber-Biesecker variant
Ohdo syndrome, SBBYS variant

Say-Barber-Biesecker-Young-Simpson syndrome Say-Barber-Biesecker-Young-Simpson variant of Ohdo syndrome SBBYS variant of Ohdo syndrome SBBYSS Young-Simpson syndrome

Additional Information & Resources

Genetic Testing Information

Genetic Testing Registry: Blepharophimosis - intellectual disability syndrome, SBBYS type

Genetic and Rare Diseases Information Center

Blepharophimosis-intellectual disability syndrome

Patient Support and Advocacy Resources

National Organization for Rare Disorders (NORD)

Catalog of Genes and Diseases from OMIM

OHDO SYNDROME, SBBYS VARIANT; SBBYSS

Scientific Articles on PubMed

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