Ohdo Syndrome, Maat-Kievit-Brunner Type

Subjects: Genetics
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Definition

The Maat-Kievit-Brunner type of Ohdo syndrome is a rare condition characterized by intellectual disability and distinctive facial features. It has only been reported in males.

1. Introduction

The intellectual disability associated with this condition varies from mild to severe, and the development of motor skills (such as sitting, standing, and walking) is delayed. Some affected individuals also have behavioral problems.

Distinctive facial features often seen in this condition include a narrowing of the eye opening (blepharophimosis), droopy eyelids (ptosis), prominent cheeks, a broad nasal bridge, a nose with a rounded tip, a large space between the nose and upper lip (a long philtrum), and a narrow mouth. Some affected individuals also have widely set eyes (hypertelorism), an unusually small chin (micrognathia), and small and low-set ears. As people with the condition get older, these facial characteristics become more pronounced and the face becomes more triangular.

Other possible signs of this condition include dental problems, weak muscle tone (hypotonia), and hearing loss.

2. Frequency

The Maat-Kievit-Brunner type of Ohdo syndrome is a very rare condition, with only a few affected individuals reported in the medical literature.

3. Causes

The Maat-Kievit-Brunner type of Ohdo syndrome results from mutations in the *MED12* gene. This gene provides instructions for making a protein that helps regulate gene activity; it is thought to play an essential role in development both before and after birth. The *MED12* gene mutations that cause this condition alter the structure of the MED12 protein, impairing its ability to control gene activity. It is unclear how these changes lead to the particular cognitive and physical features of the Maat-Kievit-Brunner type of Ohdo syndrome.

3.1. The Gene Associated with Ohdo Syndrome, Maat-Kievit-Brunner Type

- MED12

4. Inheritance

This condition is inherited in an X-linked recessive pattern. The *MED12* gene is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder.

Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. Females with only one altered copy of the gene in each cell are called carriers. They do not usually experience health problems related to the condition, but they can pass the mutation to their children. Sons who inherit the altered gene will have the condition, while daughters who inherit the altered gene will be carriers.

A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

5. Other Names for This Condition

- blepharophimosis-mental retardation syndrome, Maat-Kievit-Brunner type
- BMRS, MKB type
- Ohdo syndrome, MKB type
- X-linked Ohdo syndrome

References

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Keywords

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