Monoamine Oxidase A Deficiency

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Monoamine oxidase A deficiency is a rare disorder that occurs almost exclusively in males. It is characterized by mild intellectual disability and behavioral problems beginning in early childhood.

Keywords: genetic conditions

1. Introduction

Most boys with monoamine oxidase A deficiency are less able to control their impulses than their peers, causing aggressive or violent outbursts. In addition, affected individuals may have features of other behavioral disorders, including autism spectrum disorder and attention-deficit/hyperactivity disorder (ADHD). These features can include obsessive behaviors, difficulty forming friendships, and problems focusing attention. Sleep problems, such as trouble falling asleep or night terrors, can also occur in monoamine oxidase A deficiency.

Some people with monoamine oxidase A deficiency have episodes of skin flushing, sweating, headaches, or diarrhea. Similar episodes can occur in female family members of males with monoamine oxidase A deficiency, although females do not experience other signs or symptoms of the condition.

In some cases, certain foods, such as cheese, appear to worsen symptoms of monoamine oxidase A deficiency.

2. Frequency

Monoamine oxidase A deficiency is thought to be very rare. Its prevalence is unknown.

3. Causes

Monoamine oxidase A deficiency is caused by mutations in the *MAOA* gene. This gene provides instructions for making an enzyme called monoamine oxidase A. This enzyme breaks down chemicals called monoamines, including serotonin, epinephrine, and norepinephrine. These particular monoamines act as neurotransmitters, which transmit signals between nerve cells in the brain. Monoamine oxidase A helps break down the neurotransmitters when signaling is no longer needed. Signals transmitted by serotonin regulate mood, emotion, sleep, and appetite. Epinephrine and norepinephrine control the body's response to stress. Monoamine oxidase A also helps break down monoamines found in the diet.

Mutations in the *MAOA* gene reduce monoamine oxidase A activity, which causes serotonin and other neurotransmitters to build up in the brain. It is unclear how this buildup leads to the signs and symptoms of monoamine oxidase A deficiency. Researchers suspect that high levels of serotonin may impair an affected individual's ability to control his impulses, leading to aggressive outbursts. In addition, the outbursts may be an overreaction to stress, possibly due to the impaired breakdown of epinephrine and norepinephrine.

A reduction of monoamine oxidase A activity also impairs breakdown of monoamines found in foods. An excess of these molecules can contribute to the behavioral problems, flushing, sweating, and other symptoms associated with monoamine oxidase A deficiency, which may be why foods high in monoamines sometimes worsen the symptoms of the condition.

Monoamine oxidase A plays a role in normal brain development. Some studies suggest that reduced monoamine oxidase A activity alters the development of certain regions of the brain, which may contribute to intellectual disability and behavioral problems in people with monoamine oxidase A deficiency.

Research suggests that environmental factors, such as mistreatment in childhood, may impact the severity of the condition and the behavioral problems that develop.

3.1. The Gene Associated with Monoamine Oxidase A Deficiency

- MAOA

4. Inheritance

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition 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. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

5. Other Names for This Condition

- Brunner syndrome
- deficiency of monoamine oxidase A
- X-linked monoamine oxidase deficiency

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