Aromatic l-amino acid decarboxylase deficiency

Description

Aromatic l-amino acid decarboxylase (AADC) deficiency is an inherited disorder that affects the way nerve cells (neurons) transmit information to other cells.

Signs and symptoms of AADC deficiency typically appear in the first six months of life. Affected infants may have feeding problems, weak muscle tone (hypotonia), and sleep disturbances. Most children with AADC deficiency experience episodes called oculogyric crises that involve involuntary upward-rolling movements of the eyes. Additional signs and symptoms typically include developmental delays and intellectual disabilities.

Movement disorders are common in people with AADC deficiency. Involuntary movements that can occur in people with AADC deficiency include muscle contractions that may cause unusual body positions (dystonia) and writhing movements of the limbs (athetosis). Dystonia tends to become worse when the individual is tired, but it usually improves after sleep. Affected individuals may also experience slow or diminished movements (hypokinesia).

AADC deficiency may also affect the autonomic nervous system, which controls involuntary body processes such as the regulation of blood pressure and body temperature. This can lead to signs and symptoms such as droopy eyelids (ptosis), constriction of the pupils of the eyes (miosis), inappropriate or impaired sweating, nasal congestion, drooling, poor control of body temperature, low blood pressure (hypotension) , low blood glucose (hypoglycemia), and abnormal heart rhythms.

People with AADC deficiency have an increased risk of infection, which can lead to life-threatening complications.

Frequency

AADC deficiency is a rare disorder. Approximately 350 people with this condition have been reported in the literature. The condition is more prevalent in certain Asian populations, particularly among people from Taiwan, Japan, and China.

Causes

Variants (also called mutations) in the *DDC* gene cause AADC deficiency. The *DDC*

gene provides instructions for making the AADC enzyme, which plays an important role in the nervous system. Within neurons, the AADC enzyme helps produce dopamine and serotonin, which are two types of neurotransmitters. Neurotransmitters are chemical messengers that transmit important signals from neurons to other cells in the body. Dopamine can be used to create additional neurotransmitters called epinephrine and norepinephrine.

Variants in the *DDC* gene can reduce the activity of the AADC enzyme. This causes neurons to produce less dopamine and serotonin than normal, which in turn reduces the production of epinephrine and norepinephrine. Changes in the levels of these four neurotransmitters contribute to the developmental delays, intellectual disabilities, abnormal movements, and autonomic nervous system dysfunction seen in people with AADC deficiency.

Learn more about the gene associated with Aromatic l-amino acid decarboxylase deficiency

- DDC

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- AADC deficiency
- AADCD
- DDC deficiency
- Dopa decarboxylase deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Deficiency of aromatic-L-amino-acid decarboxylase (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1291564/)

Genetic and Rare Diseases Information Center

- Aromatic L-amino acid decarboxylase deficiency (https://rarediseases.info.nih.gov/diseases/770/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Aromatic l-amino acid decarboxylase deficiency%22)

Catalog of Genes and Diseases from OMIM

- AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY; AADCD (https://omim.org/entry/608643)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28aromatic+l-amino+acid+decarboxylase+deficiency%5BTIAB%5D%29+OR+%28aadc+deficiency%5BTIAB%5D%29+OR+%28ddc+deficiency%5BTIAB%5D%29+OR+%28dopa+decarboxylase+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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