

# AADC DEFICIENCY GLOSSARY

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This glossary defines key terms that you may encounter while discussing rare diseases, in particular Aromatic L-amino Acid Decarboxylase (AADC) deficiency. Refer to this glossary to understand medical terminology when talking with your healthcare provider or doing research.

## A

**AADC deficiency:** Aromatic L-amino Acid Decarboxylase (AADC) deficiency is a rare genetic disorder that affects the brain, causes weak muscle tone, and affects how a child develops.

**AADC enzyme:** an enzyme required to form dopamine and serotonin.

**Amino acid:** a type of compound that is the building block of proteins.

**Approved treatments:** in most countries, a central agency or organization studies medicines before they become available to the public. In the United States, this is the FDA (US Food and Drug Administration). The FDA studies the safety, dosing, and effectiveness of every medicine.

**Autosomal recessive disorder:** with autosomal recessive disorders, a child must inherit two abnormal copies of a gene (one from each parent) to develop the disorder.

## B

**Byproduct:** a substance that is created in the process of making something else.

## C

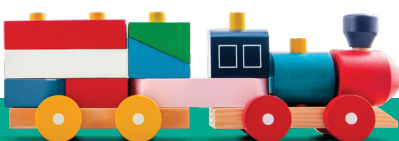
**Caregiver:** someone who regularly looks after and provides care or assistance to someone, such as a child, an elderly person, or someone who is sick or disabled.

**Cerebral palsy:** a disability resulting from abnormal development of, or damage to, the brain before, during, or shortly after birth that causes issues with body movement, muscle tone, speech disturbances, and/or other medical issues.

**Cerebrospinal fluid (CSF):** the fluid surrounding the brain and spinal cord.

**Clinical geneticist:** a physician who is trained to help identify, study, and manage genetic disorders.

**Computed tomography (CT):** a scan that uses a series of X-ray images taken from different angles around the body and uses computer processing to create a cross-sectional image of the inside of the body.



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## D

**Deficiency:** a lack or shortage of something.

**DDC (dopa decarboxylase) gene:** a gene that provides instructions for making the enzyme aromatic L-amino acid decarboxylase (AADC), which is important in the brain and nervous system.

**Developmental pediatrician:** a pediatrician with additional training in developmental and behavioral problems who evaluates children who aren't developing, learning, or behaving the way they should for their age.

**Dopamine:** a neurotransmitter made in the brain that is involved in many brain activities, including movement and emotion. Patients with AADC deficiency lack or have low levels of dopamine.

**Dystonic movement:** movement problems, including involuntary muscle contractions, sometimes called dystonia.

## E

**Electroencephalography (EEG):** a test that looks at abnormalities in the brain by attaching small metal electrodes to the scalp in order to detect tiny electrical charges from brain activity.

**Enzyme:** a substance that helps to bring about, speed up, or regulate chemical reactions.

**Epilepsy:** a chronic disorder that causes sudden episodes in which the patient may lose consciousness, have his/her senses disrupted, shake uncontrollably, and/or show other symptoms (for example, seizures or fits); often caused by abnormal electrical activity in the brain.

## F

**Flailing:** waving or swinging wildly.



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## G

**Gastroenterologist:** a medical professional who specializes in identifying and treating disorders of the stomach and intestines.

**General practitioner:** a medical professional who is trained to provide health care to patients of any gender or age.

**Gene mutation:** see *Mutation*.

**Gene therapy:** a way to treat or help prevent health conditions using genes. With gene therapy, a new, working copy of a gene is introduced into the body to make up for a gene that is not working properly or is missing. By introducing a working copy of the gene, the function of the protein can be restored.

**Genetic:** relating to or determined by genes; something that is passed down through families and considered inherited.

**Genetic testing:** a study of DNA to find differences, abnormalities, or changes that could cause disorders or to determine if a person has or will develop a certain disease.

## H

**Hypotonia:** low muscle tone, often involving reduced muscle strength.

## I

**Involuntary:** done without conscious control.

## J

**Juvenile parkinsonism:** a rare type of Parkinson's, a neurological disease. It is considered a movement disorder because it affects body movements, including walking and talking, and it may cause tremors.

## L

**Lumbar puncture:** a medical procedure in which fluid from the spine is removed with a needle; often done for diagnostic testing. Also called a spinal tap.



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## M

**Magnetic resonance imaging (MRI):** a test that uses a magnetic field and radio waves to create detailed images of the body's organs and structures.

**Metabolite:** any substance that is made during the changing of one chemical compound into another.

**Mitochondrial disease:** a disorder that causes a malfunction in the structure of a cell that produces energy, which can affect the nerves and the muscle cells.

**Movement disorder specialist:** a healthcare professional who has special training in disorders that affect someone's ability to move, such as Parkinson's disease, or that involve low muscle tone (hypotonia) or tremors.

**Muscle spasm:** an uncontrolled tightening of a muscle.

**Muscle tone:** the amount of tension or resistance in a muscle at rest or in response to stretching.

**Mutation:** a change within a gene. In AADC deficiency, there is a mutation in the *DDC* gene.

## N

**Nervous system:** an organ system of the body made up of the brain, spinal cord, and nerves.

**Neurological:** relating to the makeup, functions, and disorders of the nervous system.

**Neuromuscular:** relating to nerves and muscles.

**Neuromuscular disorders:** disorders that affect the neuromuscular system that can cause problems with the nerves that control muscles and communication between nerves and muscles.

**Neurotransmitter:** a chemical substance that works as a messenger to send signals from one nerve cell to another.

**Neurotransmitter disorders:** a group of genetic conditions that are characterized by problems processing neurotransmitters.



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## O

**Occupational therapist:** a healthcare professional who works to help patients with mental, physical, and developmental disabilities improve their ability to participate in activities that use the upper extremities, such as feeding or bathing.

**Oculogyric crises:** periods of involuntary eye movements during which the eyes suddenly roll upward involuntarily; can last anywhere from a few seconds to hours, and can happen several times a day or several times a week.

## P

**Pediatrician:** a medical professional who specializes in the development, care, and treatment of children and their diseases.

**Pediatric neurologist:** a medical professional who focuses on studying and treating diseases and conditions that affect the nervous system of children.

**Physical medicine and rehabilitation (PM&R) physician:** a medical doctor, also known as a physiatrist, who treats a variety of medical conditions that affect the brain, spinal cord, bones, joints, ligaments, muscles, and tendons with the goal of improving movement ability and quality of life.

**Physical therapist:** a trained healthcare professional who helps patients reduce pain and strengthen muscles to improve mobility.

**Plasma:** the colorless fluid part of blood.

**Precursor:** a substance from which another substance is created.

## R

**Reflux:** to flow backward in the body, such as from the stomach.



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## S

**Seizure:** a sudden attack, spasm, or convulsion, as in epilepsy or another disorder.

**Speech therapist:** a healthcare professional who specializes in the treatment of speech and communication disorders.

**Symptom:** an indication of a disease or a change in a condition that the patient notices.

For additional information and resources about  
AADC deficiency, visit [AboutAADC.com](https://www.aboutaadc.com)



# AADC DEFICIENCY GLOSSARY



Use the space below to expand your glossary by adding additional useful terms and definitions as you come across them.

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