Despite symptom onset during infancy, diagnosis of AADC deficiency is typically delayed[1]

Aromatic L-amino Acid Decarboxylase (AADC) deficiency is a genetic disorder associated with defects in neurotransmitter synthesis that can result in life-limiting motor and autonomic dysfunction, developmental delay, and premature death.[1-6]

Mean age of diagnosis
3.5 years[1]

Age range of diagnosis
2 months to 23 years[1]

The challenge of diagnosing AADC deficiency

Symptoms of neurotransmitter disorders can overlap with those of other neurological disorders, which can make diagnosis challenging. Many of the most common symptoms of AADC deficiency can also be attributed to a number of other conditions such as cerebral palsy and epilepsy, resulting in potential misdiagnosis.[1,2,7-11]

POSSIBLE AADC DEFICIENCY MISDIAGNOSES

AADC deficiency symptoms[1,12]	May be diagnosed as[7-9,13]
Oculogyric Crisis	Epilepsy
Dystonia • Rigidity • Motor Delay	Cerebral Palsy
Hypotonia • Akinesia • Ptosis	Neuromuscular Disorders

Look for key red-flag diagnostic clues that should prompt investigation for a neurotransmitter disorder, like AADC deficiency:

Oculogyric crises[2,12,14]

Autonomic symptoms[12]

Normal EEG and neuroimaging[1,9,11,12]

Diurnal variation[1,8,15]

CSF=cerebrospinal fluid; CT=computed tomography; EEG=electroencephalogram; MRI=magnetic resonance imaging.

Signs and symptoms associated with AADC deficiency

In a clinical study of 78 patients who were diagnosed with AADC deficiency, the following symptoms were documented[2]:

Hypotonia
was present in

95%

of patients (n=74)[2]

- Most commonly reported symptom

Developmental delay
exhibited in

63%

of patients (n=49)[2]

- Impairments in head control, sitting, crawling, or standing[1,16]
- Speech delays[1,16]

Oculogyric crises
occurred in

86%

of patients (n=67)[2]

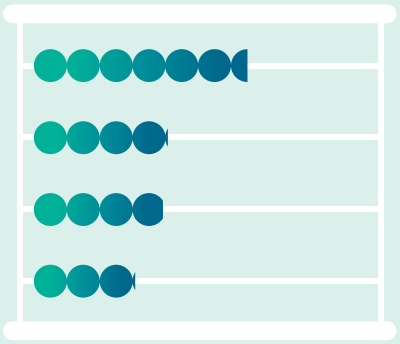
- Episodes of sustained upward or lateral deviation of the eyes, rhythmic orofacial movements, backward and lateral flexions of the neck, tongue protrusion, and jaw spasms[4]
- Can last a few seconds or persist for several hours, and occur several times a day or week[17]
- May not be present in all cases[2]
- Often misdiagnosed as a seizure, epilepsy, or mitochondrial disease[13,17]

Patients may present with other movement disorders or symptoms, including[2]:

- Dystonia (53%) n=41
- Hypertonia (44%) n=35
- Hypokinesia (32%) n=25

Autonomic symptoms
observed in patients included[2]:

- Hyperhidrosis (65%) n=51
- Hypersalivation (41%) n=32
- Ptosis (39%) n=30
- Nasal congestion (31%) n=24



Visit **AADCInsights.com** to learn more about the signs, symptoms, and no-cost testing to aid in diagnosing AADC deficiency ›

A diagnostic pathway for suspected AADC deficiency[1,12]

Test for AADC deficiency if there is:

Hypotonia, hypertonia
and
Delayed motor development
and
MRI inconsistent with clinical symptoms

+

Movement disorders

- Oculogyric crisis
- Dystonia
- Hypokinesia and/or bradykinesia

Autonomic symptoms are often present

- Ptosis
- Temperature instability
- Nasal congestion
- Excessive sweating

Current consensus guidelines recommend genetic testing in combination with CSF neurotransmitter metabolite panel and/or plasma AADC enzyme activity assay to confirm a diagnosis of AADC deficiency.[1]

Access no-cost genetic testing with PTC Pinpoint™

PTC Therapeutics and Invitae have partnered to offer no-cost genetic testing programs—including testing, genetic counseling, and family screening—for individuals of any age with a suspected neurotransmitter disorder or symptoms suggestive of cerebral palsy in the absence of risk factors for an acquired brain injury. While specimen collection can be done in office, PTC Pinpoint also offers the option to ship saliva or buccal specimen collection kits directly to patients or their caregivers.

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