Recognizing AADC deficiency in your patients

Learn how to detect and confirm this rare genetic disorder

Aromatic L-amino Acid Decarboxylase (AADC) deficiency is an autosomal recessive disorder associated with defects in neurotransmitter synthesis, resulting in profound motor and autonomic dysfunction and developmental delay.[1-3]

Many symptoms of AADC deficiency are similar to the symptoms of other more common conditions, making diagnosis a challenge.[1-4]

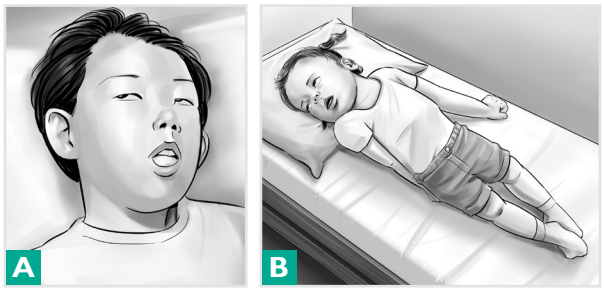
Key differentiating signs and symptoms

One or a combination of red-flag diagnostic clues should prompt investigation for AADC deficiency.

A 2010 study (n=78) recorded the prevalence of these common symptoms, which can help to identify AADC deficiency.[1]

Oculogyric crisis[1]

- Occurred in 86% of patients (n=67/78)[1]
- 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 that can sometimes be confused with seizures[5,6]
- Can occur several times a day or several times a week, and last from a few seconds to hours[6]
- Often confused with a seizure, epilepsy, or cerebral palsy[6,7]



Illustrations depicting (A) an oculogyric crisis and (B) involuntary movements that may accompany an episode

Normal EEG and neuroimaging[1,2,4,8,9,a]

- History of normal EEG, MRI, or CT
- Inconsistent with presentation

Autonomic symptoms[1]

- Hyperhidrosis (65%; n=51/78)
- Hypersalivation (41%; n=32/78)
- Ptosis (39%; n=30/78)
- Nasal congestion (31%; n=24/78)

Diurnal variation[2,10,11]

- Motor symptoms become exacerbated or more prominent late in the day[10,11]
- Symptoms improve with sleep[10,11]

[a]Some patients presented with abnormalities in imaging (24/78) and EEG (10/78).[1]

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

Common signs and symptoms

Hypotonia[1]

- Present in 95% of patients (n=74/78)[1]
- Most commonly reported symptom[1]

Developmental delays[1]

- Present in 63% of patients (n=49/78)[1]
- May include impairments in head control, crawling, or standing, and speech delays[2,12]

Movement disorders[1]

- Dystonia (53%; n=41/78)
- Hypertonia (44%; n=35/78)
- Hypokinesia (32%; n=25/78)

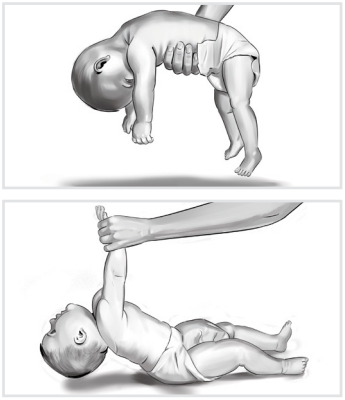


Illustration of hypotonia

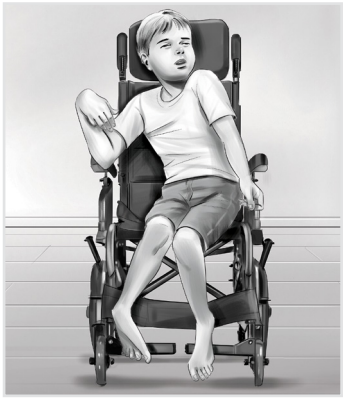


Illustration depicting developmental delay and/or dystonia; lack of head control or dystonic posturing of body

Diagnosing AADC deficiency

Currently, these core tests can help diagnose AADC deficiency[2]:

- CSF neurotransmitter metabolite panel
- Plasma enzyme activity assay
- Genetic testing

Current consensus guidelines recommend CSF neurotransmitter metabolite panel and/or plasma AADC enzyme activity assay in combination with genetic testing to confirm AADC deficiency.[2]

For further information on signs and symptoms, testing, and diagnosing AADC deficiency in your patients, visit AADCInsights.eu.

CSF=cerebrospinal fluid.

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