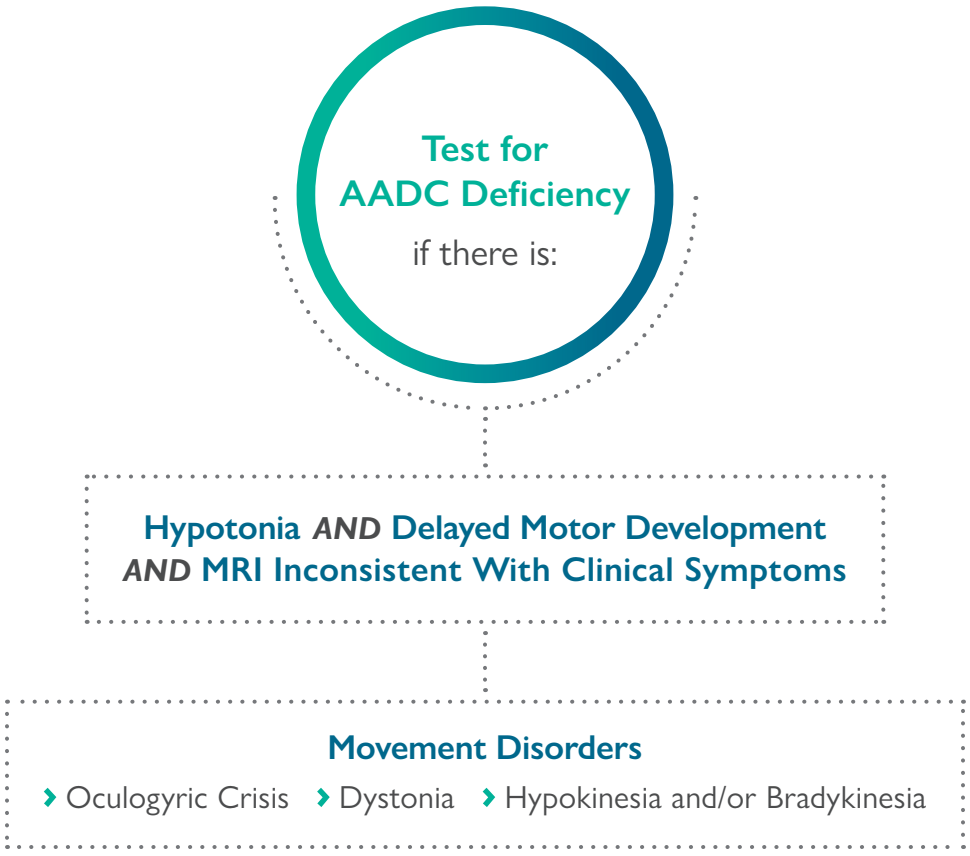
Earlier identification can help improve the care and management of patients with AADC Deficiency[1-3]

Aromatic L-amino Acid Decarboxylase (AADC) Deficiency

is a genetic disease associated with defects in neurotransmitter synthesis, resulting in life-limiting motor and autonomic dysfunction, developmental delay, and premature death.[1-3]

The diagnostic pathway for suspected AADC Deficiency[2,4]



Autonomic symptoms are often present

• Ptosis • Temperature Instability • Nasal Congestion

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.[2,3,5-7]

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Diagnostic tests that identify AADC Deficiency[2]

Following this diagnostic process can help diagnose AADC Deficiency[2]

1 Perform[2]

- CSF neurotransmitter metabolite panel
- Plasma enzyme activity assay

Other diagnostic tests that may be helpful[2]

- Blood level measurement of 3-OMD
- Urinary organic acid analysis

2 Interpret[2]

- **Reduced** HVA, 5-HIAA, and MHPG; **elevated** 3-OMD, L-dopa, and 5-HTP; and normal pterins in CSF

AND/OR

- Low plasma AADC enzyme activity
- Increased urinary VLA

3 Confirm with genetic testing[2]

- Mutation(s) in the *DDC* gene

Blood plasma	CSF	Genetic testing
Decreased AADC enzyme activity	***HIGH LEVELS OF***	Variants in *DDC* gene
	3-OMD L-dopa 5-HTP	
	LOW LEVELS OF	
	5-HIAA HVA MHPG	
	Normal pterins	

3-OMD=3-O-methyldopa; 5-HIAA=5-hydroxyindoleacetic acid; 5-HTP=5-hydroxytryptophan; CSF=cerebrospinal fluid; DDC=dopa decarboxylase; HVA=homovanillic acid; L-dopa=L-3,4-dihydroxyphenylalanine; MHPG=3-methoxy-4-hydroxyphenylglycol; VLA=vanillactic acid.

Consensus guidelines recommend confirming an AADC Deficiency diagnosis with a genetic test[2]

Gene therapy for AADC Deficiency is currently under development in the research setting, offering the possibility of a promising therapy in the future[2]

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MAT-AADC-0254 03/19

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