Aromatic L-amino Acid Decarboxylase (AADC) Deficiency is an autosomal recessive disease associated with defects in neurotransmitter synthesis, resulting in profound motor and autonomic dysfunction and developmental delay. 

AADC Deficiency can manifest with a broad spectrum of symptoms. The most common are:
- Hypotonia
- Developmental delay
- Movement disorders, especially oculogyric crisis

Despite symptom onset during infancy, diagnosis is typically delayed.

Mean age of diagnosis
3.5 years

Age range of diagnosis
2 months to 23 years

How AADC Deficiency is diagnosed

- Earlier identification can help improve the care and management of patients with AADC Deficiency.
- Currently, these core tests can help diagnose AADC Deficiency:
  - CSF neurotransmitter metabolite panel
  - Plasma enzyme activity assay
  - Genetic testing

The challenge of a correct diagnosis

- Many of the most common symptoms of AADC Deficiency can also be attributed to a number of other conditions such as:
  - Epilepsy
  - Cerebral palsy
  - Neuromuscular weakness

Other tests that may be helpful

- Blood level measurement of 3-OMD (3-O-methyldopa)
- Urinary organic acid analysis

Consensus guidelines recommend confirming an AADC Deficiency diagnosis with a genetic test.

To learn more about AADC Deficiency and no-cost diagnostic testing supported by PTC, visit AADCinsights.com

References: