LEARN ABOUT AADC DEFICIENCY

A rare genetic disorder of the central nervous system

- 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[1-3]
- AADC Deficiency can manifest with a broad spectrum of symptoms. The most common are[1-3]:
  - Hypotonia
  - Developmental delay
  - Movement disorders, especially oculogyric crisis

Despite symptom onset during infancy, diagnosis is typically delayed[2,3]

Mean age of diagnosis
3.5 years[2]

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

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[2,4-7]:
  - Epilepsy
  - Cerebral palsy[4,5]
  - Neuromuscular weakness

How AADC Deficiency is diagnosed

- Earlier identification can help improve the care and management of patients with AADC Deficiency[2,3]
- Currently, these core tests can help diagnose AADC Deficiency[2]
  - CSF neurotransmitter metabolite panel
  - Plasma enzyme activity assay
  - Genetic testing

Other tests that may be helpful[2]

- 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[2]

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

References: 1. Manegold C, Hoffmann GF, Degen I, et al. Aromatic L-amino acid decarboxylase deficiency: clinical features, drug therapy and follow-up. *J Inherit Metab Dis.* 2009;32(3):371-380. **2.** Wassenberg T, Molero-Luis M, Jeltsch K, et al. Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. *Orphanet J Rare Dis.* 2017;12(1):12. doi: 10.1186/s13023-016-0522-z. **3.** Brun L, Ngu LH, Keng WT, et al. Clinical and biochemical features of aromatic L-amino acid decarboxylase deficiency. *Neurology.* 2010;75(1):64-7. **4.** Krigger KW. Cerebral palsy: an overview. *Am Fam Physician.* 2006;73(1):91-100. **5.** Ng J, Papandreou A, Heales SJ, et al. Monoamine neurotransmitter disorders—clinical advances and future perspectives. *Nat Rev Neurol.* 2015;11(10):567-584. **6.** Kurian MA, Dale RC. Movement disorders presenting in childhood. *Continuum (Minneap Minn).* 2016;22(4 Movement Disorders):1159-1185. **7.** Lee WT. Disorders of monoamine metabolism: inherited disorders frequently misdiagnosed as epilepsy. *Epilepsy & Seizure.* 2010;3(1):147-153. https://www.jstage.jst.go.jp/article/eands/3/1/3_1_147/_article/-char/en. Accessed February 6, 2019.

MAT-AADC-0248 03/19

PTC THERAPEUTICS