

WHEN SHOULD YOU SCREEN FOR AROMATIC L-AMINO ACID DECARBOXYLASE (AADC) DEFICIENCY?

Here are the key red flags:

CENTRAL NERVOUS SYSTEM RED FLAGS

Motor dysfunction¹

- Delayed motor development^{2,3}



- Hypotonia (mainly truncal)
- Hypertonia (mainly limbs)
- Movement disorders
- Oculogyric crises (bilateral one-sided eye deviation)
- Delayed cognitive development
- Delayed speech development
- Hypokinesia
- Dystonia
- Autism-like symptoms
- Epileptic seizures

AUTONOMIC NERVOUS SYSTEM & OTHER RED FLAGS

Autonomic¹

- Ptosis
- Nasal congestion
- Excessive sweating
- Temperature instability
- Excessive crying/irritability⁴
- Hypotension (orthostatic)
- Heart rhythm abnormalities
- Diarrhea
- Sleep disturbance

Other signs and symptoms¹

- Feeding/swallowing problems
- Gastrointestinal reflux
- Failure to thrive
- Hypoglycemia



OBSERVE⁵

The red flags listed above are important, especially **delayed motor development** and **hypotonia/hypertonia** and **structurally normal MRI/EEG**. Consider differential diagnosis from CP, epilepsy, autism and mitochondrial diseases.



TEST^{1,6,7}

Patients with AADC deficiency often show **elevated plasma 3-OMD levels**. **Free 3-OMD dry blood spot testing** is offered by PTC Therapeutics to help your diagnosis of patients with suspected AADC deficiency. Questions on testing for AADC deficiency can be directed to: AADCTesting@ptcbio.com.



REFER

Promptly refer to a pediatric neurologist or geneticist.

CP, cerebral palsy; EEG, electroencephalogram; MRI, magnetic resonance imaging; 3-OMD, 3-O-methylidopa.

1. Wassenberg T, et al. *Orphanet J Rare Dis*. 2017;12(1):12; 2. Denver II Developmental Milestone Chart. Available at: https://www.ccmedical.org/forms/1428352937_171971.pdf (Accessed: March 2021); 3. Hwu WL, et al. *JIMD Rep*. 2018;40:1-6; 4. Lee HF, et al. *Eur J Paediatr Neurol*. 2009;13(2):135-140; 5. Himmelreich N, et al. *Mol Genet Metab*. 2019;127(1):12-22; 6. Chen PW, et al. *Clin Chim Acta*. 2014;431:19-22; 7. Brennenstuhl H, et al. *J Inherit Metab Dis*. 2020;43:602-610.