

AROMATIC L-AMINO ACID DECARBOXYLASE (AADC) DEFICIENCY: A RARE DISEASE



AADC deficiency is a rare autosomal recessive disorder of neurotransmitter synthesis^{1,2}

Documented in the literature

Symptoms develop early in life²

2.7 months

is the mean age of symptom onset (n=68)

3.5 years

is the mean age of diagnosis (n=68)

Similar symptomatology may lead to misdiagnosis^{2,4-6}

Key clinical signs and symptoms of AADC deficiency include:^{1,2}

- Hypotonia
- Movement disorders such as oculogyric crisis, dystonia, and hypokinesia with diurnal fluctuation and improvement of symptoms after sleep observed in some patients
- Developmental delay
- Autonomic dysfunction such as ptosis, excessive sweating and nasal congestion



Differential diagnosis^{1,2,4-6}

AADC deficiency may be challenging to diagnose as it can present with signs and symptoms similar to those of more common conditions, including:

Hyperekplexia

Epilepsy

Cerebral palsy

Mitochondrial disorders

Neuromuscular disorders

Early diagnosis improves outcomes^{2,7,8}

To confirm a diagnosis of AADC deficiency, the following tests are recommended:

- Lumbar puncture to analyse cerebrospinal fluid (CSF) neurotransmitter profile
- Single gene or genetic panel testing to identify a mutation of the *DDC* gene
- Measurements of AADC enzyme activity in plasma

If you have a child presenting with signs and symptoms of AADC, consider referring to a paediatric neurologist for further investigation

References

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