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 during infancy[2–4]

<12 months is the age of symptom onset

3.2 years is the mean age of diagnosis (n=185)

Similar symptomatology may lead to misdiagnosis[2,5–7]

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[1,2,5–7]

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[2,8,9]

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 deficiency, consider referring to a paediatric neurologist for further investigation

References

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