

AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY (AADC DEFICIENCY) BURDEN OF DISEASE

AADC deficiency is a fatal, rare genetic disorder that typically causes severe disability and suffering from the first months of life.¹⁻⁴

How AADC deficiency affects people



A defect in the dopa decarboxylase (DDC) gene causes a deficiency in the production of the AADC enzyme.¹

The AADC enzyme plays a vital role in the production of chemical messengers in the brain, like dopamine, that regulate:

- Body control and movement
- Cognition
- The central nervous system



When the enzyme is deficient, the production of these messengers is disrupted.¹

Key clinical symptoms include:^{1,2}

BEHAVIOUR AND SLEEPING PROBLEMS

CARDIOVASCULAR
GASTROINTESTINAL
ENDOCRINE
& RESPIRATORY PROBLEMS



DEVELOPMENTAL DELAY

AUTONOMIC SYMPTOMS (problems with the regulation of involuntary body processes such as heart function and temperature control)

MOVEMENT DISORDERS

DECREASED MUSCLE TONE



Suffering is exacerbated by episodes of **distressing seizure-like oculogyric crises** which can happen daily and last for hours, causing the eyes to involuntarily roll up in the head.^{1,3,6-7}

Patient burden

Most children with AADC deficiency do not live through childhood.⁵

Their shortened life is typically one of **severe disability**, often reliant on **feeding tubes** and/or **breathing support**, frequent **hospitalizations** and periods of **intensive care**.^{1,2}



Most children will never be able to:²

HOLD THEIR HEAD UP



SIT BY THEMSELVES



STAND



SPEAK



Caregiver burden

Caring for a child with AADC deficiency impacts the whole family physically, emotionally and financially.

Children with AADC deficiency typically require round-the-clock support with:⁸⁻⁹



Typically, caregivers need to:⁸⁻⁹

- Spend an average of **15 hours per day** caring for their child, and performing **administrative tasks**
- **Stop work** or **reduce their working hours**
- **Receive additional paid or unpaid support**



Caregivers have little time to themselves, impacting their ability to carry out **household tasks**, go to **work** or **socialize**. Caregivers have described feeling **depressed**, **anxious** and **fearful of the future**, as well as problems with **back pain** and **sleep**.⁹



Healthcare burden



Managing patients with AADC deficiency requires a **multidisciplinary team of specialists** to address the patient's many significant health issues.¹

Diagnosis and treatment



AADC deficiency remains **largely undiagnosed**.¹⁰

The **journey to diagnosis** can be **extremely challenging** for patients and families, exacerbated by the **financial** and **emotional pressures** of a complex diagnostic journey.



Currently there are no licensed treatments for AADC deficiency.

Affected children may be prescribed **treatments** to manage symptoms, but they yield little improvement for the majority of patients.¹

This is often complicated by interactions between the multiple drugs used¹



One natural history study found that **97%** of patients (n=37) receiving current treatment options **failed to achieve any motor milestones**.²

97% OF AADC PATIENTS FAIL TO ACHIEVE MOTOR MILESTONES



Abbreviations

AADC, Aromatic L-amino acid decarboxylase; DDC, Dopa decarboxylase

References

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