**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.**

  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.

**Key clinical symptoms include:**

- **DECREASED MUSCLE TONE**
  - 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 used.

- **DEVELOPMENTAL DELAY**

- **MOVEMENT DISORDERS**
  - Most children will never be able to:
    - Stand
    - Sit by themselves
    - Feed themselves
  - Children with AADC deficiency typically require round-the-clock support with:
    - Eating
    - Dressing
    - Bathing

- **DECREASED MUSCLE TONE**

- **CARDIOVASCULAR**
  - Most children with AADC deficiency do not live through childhood.

- **ENDOCRINE**

- **NEUROLOGICAL SYSTEM**

- **RESPIRATORY PROBLEMS**

### Patient burden

- **Most children will never be able to:**
  - Stand
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- **Caregiver burden**

- **Healthcare burden**

### Diagnosis and treatment

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.

### Patient burden

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

- 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.

### Caregiver burden

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

- **Typically, caregivers need:**
  - Spend an average of 15 hours per day caring for their child, and performing administrative tasks
  - Sleep less than 4 hours during 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.

### Healthcare burden

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