

HAVE YOU CONSIDERED AN UNDERLYING CONDITION FOR PATIENTS WITH SIGNS OF CEREBRAL PALSY?¹⁻³



Your patients with cerebral palsy of unknown etiology may have a rare genetic disorder

Your patients may exhibit signs of cerebral palsy such as dystonia, rigidity/hypertonia, and motor delay, which are also common signs and symptoms of Aromatic L-amino Acid Decarboxylase (AADC) deficiency.¹⁻⁶

AADC deficiency is a genetic disease associated with defects in neurotransmitter synthesis, which can result in life-limiting motor and autonomic dysfunction, developmental delay, and premature death.^{4,5,7-9}

AADC Deficiency and Cerebral Palsy

SIMILAR SIGNS AND SYMPTOMS BETWEEN CEREBRAL PALSY AND AADC DEFICIENCY INCLUDE^{1-3,5,6,10}:

- › Hypotonia
- › Developmental delay
- › Dystonia
- › Rigidity/hypertonia

SYMPTOMS ATYPICAL OF CEREBRAL PALSY THAT MAY INDICATE AN UNDERLYING GENETIC DISORDER, SUCH AS AADC DEFICIENCY:

Oculogyric crises ^{4,11,12}	EEG and neuroimaging inconsistent with symptoms ^{1,5,12,14}	Autonomic symptoms ¹²	Diurnal variation ^{3,5,15}
Episodes of sustained upward or lateral deviation of the eyes, rhythmic orofacial movements, backward and lateral flexions of the neck, tongue protrusion, and jaw spasms that can sometimes be confused with seizures ^{13,14}	One study showed that only a small proportion of patients with AADC deficiency had an abnormal EEG, MRI, or CT ⁴	Multiple signs of autonomic dysfunction, such as hyperhidrosis, hypersalivation, ptosis, or nasal congestion ^{4,14}	Symptoms become exacerbated or more prominent late in the day and improve with sleep ^{3,15}

If you suspect your patient may have these signs and symptoms, consider testing for AADC deficiency. Current consensus guidelines recommend CSF neurotransmitter metabolite panel and/or plasma AADC enzyme activity assay, in combination with genetic testing to confirm a diagnosis of AADC deficiency.⁵



Visit [PTCPinpoint.com](https://www.ptcinc.com) to learn about no-charge genetic testing for your patients.

CSF=cerebrospinal fluid; CT=computed tomography; EEG=electroencephalogram; MRI=magnetic resonance imaging.

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