Teaching Video NeuroImage: Oculogyric Crises in a 12-Year-Old Girl With Rapid-Onset Dystonia Parkinsonism

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Equal Author Contribution:

Junyu Lin and Chunyu Li have contributed equally to the study.

Contributions:

Junyu Lin: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Study concept or design
Chunyu Li: Drafting/revision of the manuscript for content, including medical writing for content
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A 12-year-old girl presented to the emergency department for forced upward eyes deviation for two hours with craniocervical dystonia and intact consciousness. During the half-year, she experienced several emotionally triggered attacks (Video). Sudden-onset dystonic gait occurred 1 month before the first attack and progressively deteriorated with subsequent attacks. All limbs showed bradykinesia and rigidity without ataxia. Normal electroencephalogram and consciousness suggested oculogyric crises rather than epilepsy. A de novo pathogenic variant in ATP1A3 (c.2767G>A) confirmed rapid-onset-dystonia-parkinsonism (Figure), which is not generally dopa-responsive\(^1\). However, since her oculogyric crises might correlate with a hypodopaminergic state\(^2\), levodopa was beneficial. Adding flunarizine improved her condition.
## Appendix 1. Authors

<table>
<thead>
<tr>
<th>Name</th>
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<tbody>
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interpretation of data
References


Video legend

Video title: A patient with rapid-onset-dystonia-parkinsonism

Segment 1. An episode showed forced upward eyes deviation accompanied by retrocollis and involuntary orofacial movement.

Segment 2. An episode showed forced upward gaze accompanied by retrocollis, sialorrhea, and dystonic opening of the mouth.

Segment 3. Another episode showed forced upward eyes deviation and increased eye blinking.

Segment 4. The patient showed a dystonic gait and dystonic posture of the upper limbs. Segment 5. The patient's gait improved by the 3-month follow-up.
Figure legend

Figure title: Electropherogram of the patient and her parents

Electropherogram showed a heterozygous c. 2767G>A (p. Asp923Asn) mutation in \textit{ATP1A3} of the proband. Her parents did not carry this mutation in \textit{ATP1A3}. 
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