A 7-month-old boy was referred with developmental delay and axial hypotonia (video 1). Screening for inborn errors of metabolism was negative and single nucleotide polymorphism array was normal (46,XY). Myotonic dystrophy (type 1) and spinal muscular atrophy were excluded. Whole exome sequencing yielded biallelic mutations in the tyrosine hydroxylase gene (c.698G>A, p.Arg233His and c.1211C>T, p.Thr404Met). Subsequent CSF analysis revealed a significantly lowered homovanillic acid/5-hydroxyindoleacetic acid ratio, confirming tyrosine hydroxylase deficiency. Treatment with monotherapy levodopa resulted in profoundly improved motor development (video 1). After several weeks of treatment, the patient developed levodopa-induced dyskinesias (video 1), insomnia, and hyperactive behavior. All symptoms ameliorated with levodopa reduction.

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Disclosure
The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

Appendix

<table>
<thead>
<tr>
<th>Name</th>
<th>Location</th>
<th>Contribution</th>
</tr>
</thead>
<tbody>
<tr>
<td>Etienne Janssen, MD</td>
<td>Maastricht University Medical Center, the Netherlands</td>
<td>Designing first draft of manuscript and participation in revision, selection of video data</td>
</tr>
<tr>
<td>Mayke Oosterloo, MD, PhD</td>
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<td>Participation in revising manuscript, treating genetic metabolic physician</td>
</tr>
<tr>
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<tr>
<td>Joost Nicolai, MD, PhD</td>
<td>Maastricht University Medical Center, the Netherlands</td>
<td>Participation in revising manuscript, treating pediatric neurologist</td>
</tr>
</tbody>
</table>

References
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