Teaching Video NeurolImage: Improvement in Motor Development After Start of Levodopa in Tyrosine Hydroxylase Deficiency

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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.698 G>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.¹ 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),² insomnia, and hyperactive behavior. All symptoms ameliorated with levodopa reduction.

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

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Name	Location	Contribution
Etienne Janssen, MD	Maastricht University Medical Center, the Netherlands	Designing first draft of manuscript and participation in revision, selection of video data
Mayke Oosterloo, MD, PhD	Maastricht University Medical Center, the Netherlands	Selection of video data and participation in revising manuscript
Estela Rubio-Gozalbo, MD, PhD	Maastricht University Medical Center, the Netherlands	Participation in revising manuscript, treating genetic metabolic physician
Koen van Gassen, PhD	University Medical Center Utrecht, the Netherlands	Genetic analysis and interpretation
Joost Nicolai, MD, PhD	Maastricht University Medical Center, the Netherlands	Participation in revising manuscript, treating pediatric neurologist

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