Teaching Video NeuroImages: Paroxysmal hyperkinesia with diurnal fluctuations due to sepiapterin-reductase deficiency

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A 42-year-old man, born of consanguineous parents, presented with long-standing severe, nonepileptic jerky movements of the upper body, pronounced during the second half of the day and improving after sleep (video, A and B). There was a history of neurodevelopmental disorder with axial hypotonia, delayed milestones, intellectual disability, and poor speech production. The combination of a neurodevelopmental syndrome and a movement disorder with diurnal fluctuations[1] led to targeted exome sequencing for monoamine metabolism disorders. A homozygous nonsense variant in the *SPR* gene was identified (figure), confirmed by Sanger sequencing (figure). Treatment with levodopa led to marked improvement of abnormal movements (video, C).

MORE ONLINE

Video

Teaching slides
links.lww.com/WNL/B131

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Disclosure
T. Mainka, J. Hoffmann, A.A. Kühn, S. Biskup, and C. Ganos report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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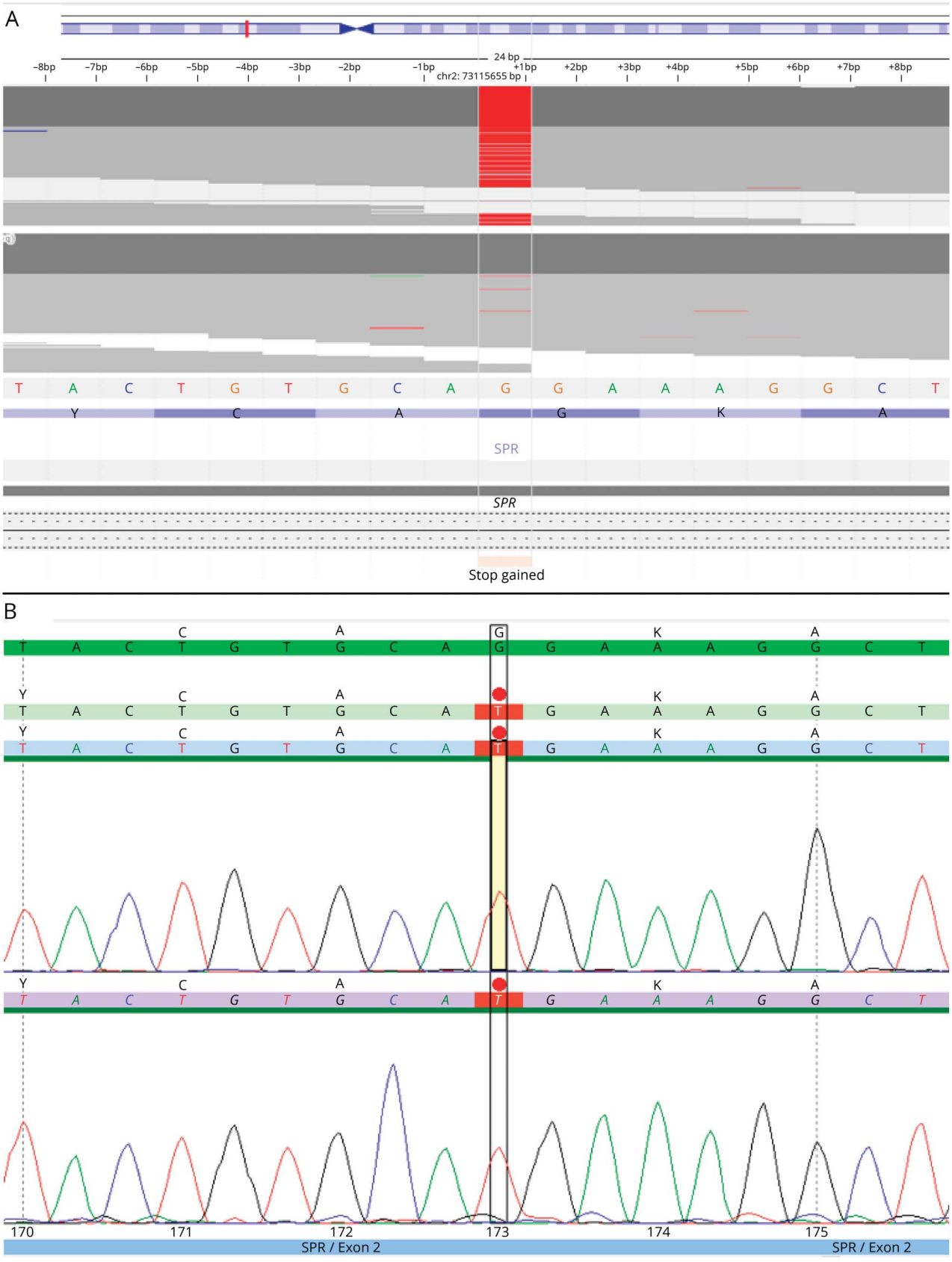
Figure Genetic analysis



(A) Identification of the homozygous nonsense variant *p*. Gly173* in the *SPR* gene using whole exome sequencing. (B) Confirmation of the identified nonsense variant in *SPR* by Sanger sequencing.

Appendix Authors

Name	Location	Contribution
Tina Mainka, MD	Department of Neurology, Charité University Medicine Berlin; Berlin Institute of Health, Germany	Clinical assessment of the patient, collection and editing of video material, interpretation of genetic analysis, drafting the manuscript
Jessica Hoffmann	Center for Genomics and Transcriptomics, Tübingen, Germany	Genetic analysis, interpretation of genetic analysis, drafting the manuscript
Andrea A. Kühn, MD	Department of Neurology, Charité University Medicine Berlin, Germany	Interpretation of genetic analysis, drafting the manuscript
Saskia Biskup, MD	Center for Genomics and Transcriptomics, Tübingen, Germany	Genetic analysis, interpretation of genetic analysis, drafting the manuscript for intellectual content
Christos Ganos, MD	Department of Neurology, Charité University Medicine Berlin, Germany	Clinical assessment of the patient, collection and editing of video material, interpretation of genetic analysis, drafting the manuscript

Reference

1. Friedman J, Roze E, Abdenur JE, et al. Sepiapterin reductase deficiency: a treatable mimic of cerebral palsy. Ann Neurol 2012;71:520–530.