

# Early-Onset Dystonia, Exacerbation With Fever, and Striatal Signal Changes

## Emerging Phenotype of DYT-PRKRA

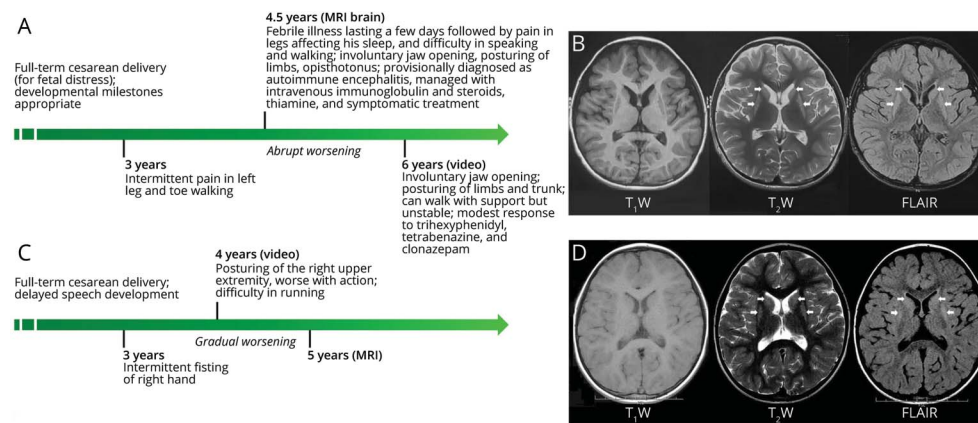
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### Figure 1 Clinical Features and MRI Abnormalities



Timeline showing clinical features of the boy (A) and his sister (C); MRI of the boy (B) and his sister (D) showing striatal T2W and FLAIR hyperintensities (arrows) and mild cerebral atrophy (B).

A 6-year-old boy from Maharashtra, India, presented with subacute-onset generalized dystonia after a febrile illness; his sister manifested insidious-onset multifocal dystonia (Video 1). T2-weighted MRI showed striatal hyperintensities (Figure 1). Clinical exome sequencing detected a novel homozygous variant (c.127 G > T; G43C) in the *PRKRA* gene which encodes PACT, a stress-response protein. The variant was confirmed by Sanger sequencing (eFigure 1, links.lww.com/WNL/C111). DYT-PRKRA is a childhood-onset progressive dystonia.<sup>1</sup> Some patients have exacerbation with fever and evidence of striatal degeneration in neuroimaging.<sup>2</sup> PACT-dependent activation of protein kinase R (PKR) leads to apoptosis.<sup>2</sup> Perhaps, the G43C PACT triggered abnormal activation of PKR and intensified neuronal apoptosis during febrile illness.

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**▶ Video**

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### Disclosure

The authors report no disclosures relevant to the manuscript. Go to [Neurology.org/N](http://Neurology.org/N) for full disclosures.

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## Appendix Authors

Name	Location	Contribution
<b>Suvorit Subhas Bhowmick, MD, DM</b>	Movement Disorders Clinic, Vadodara Institute of Neurological Sciences, India	Drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; study concept or design; analysis or interpretation of data
<b>Sarbani Raha, MD</b>	Child Neurology and Epilepsy Clinic, Vadodara, India	Drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; analysis or interpretation of data

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## Appendix *(continued)*

Name	Location	Contribution
<b>Amita Bohora, MD</b>	Baroda Imaging Center, Vadodara, India	Drafting/revision of the manuscript for content, including medical writing for content; analysis or interpretation of data

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## References

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2. Masnada S, Martinelli D, Correa-Vela M, et al. PRKRA-related Disorders: bilateral striatal degeneration in addition to DYT16 spectrum. *Mov Disord*. 2021;36(4):1038-1040.

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