

Paroxysmal jerks with diurnal fluctuations in an adult

Teaching NeuroImages

Neurology

Resident & Fellow Section



Vignette

- 42-year-old male
- Consanguineous parents
- History of neurodevelopmental disorder with axial hypotonia, delayed milestones, intellectual disability and poor speech production
- Presentation due to long-standing severe, non-epileptic jerky movements of the upper body
- Jerks appeared during the second half of the day and improved after sleep



Video



Paroxysmal jerks with diurnal fluctuations in an adult due to sepiapterin-reductase deficiency

- Characteristic combination of a neurodevelopmental syndrome and a movement disorder with diurnal fluctuations¹
- Targeted exome sequencing for monoamine metabolism disorders revealed homozygous nonsense variant (p.Gly173*) in the *SPR* gene, confirmed by Sanger sequencing
- Treatment with levodopa led to marked improvement of abnormal movement

