A 5-year-old girl with abnormal facial features, strabismus, horizontal nystagmus, hypotonia, and a history of hypersomnolence, seizures and developmental delay began to experience a generalized complex movement disorder. Clinically, there was a mixed hyperkinetic movement disorder, consisting of chorea, dystonia, myoclonus, and hand stereotypies. The presence of generalized jerks, interposed with those complex movements, resembled a stop-motion animation (Video 1), similar to the animation technique in which objects are physically manipulated in small increments and

(A) and (B): Axial T2-weighted brain MRI; (C) and (D): Axial T1-weighted brain MRI, all showing volumetric reduction bilaterally, notably in frontal lobe.

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Teaching slides
links.lww.com/WNL/C556
photographed frame by frame. Brain MRI showed mild frontal cortical atrophy (Figure). Genetic investigation was performed, and CGH array was performed, finding a pathogenic variant arr[GRCh37]5q31.2q31.3(13903279_140058893)x1 in PURA gene, compatible with PURA syndrome.\(^1\) The presence of complex hyperkinetic movement disorders in infants with global developmental delay may be an important clue to diagnose PURA syndrome.\(^1\) Affected patients may be misdiagnosed with dyskinetic cerebral palsy if genetic studies are not pursued.\(^2\)

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### Appendix

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