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Teaching Video Neuroimage: "Stop-Motion" Chorea in PURA Syndrome

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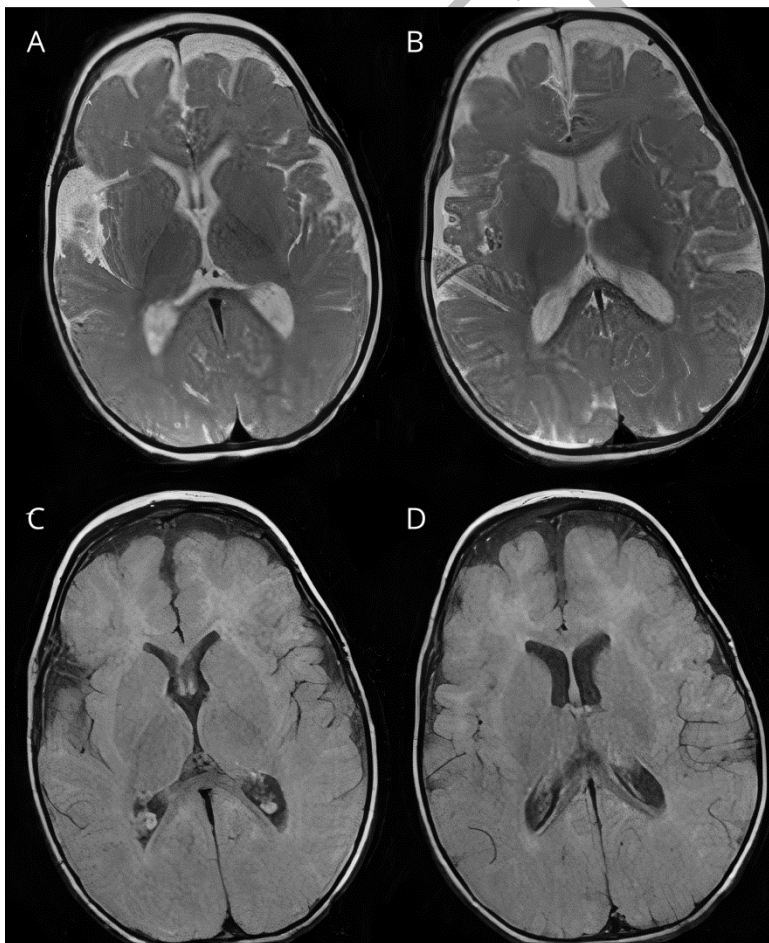
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A 5-year-old female, with abnormal facial features, strabismus, horizontal nystagmus, hypotonia, and a history of hypersomnolence, seizures and developmental delay, began to present a generalized complex movement disorder. Clinically, there was a mixed 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 photographed frame by frame. Brain MRI showed mild frontal cortical atrophy (Figure). Genetic investigation was carried out, and CGH-array was performed, finding a pathogenic variant $\text{arr}[\text{GRCh37}]5\text{q}31.2\text{q}31.3(139033279_140058893)\times 1$ 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. Affected patients may be misdiagnosed with dyskinetic cerebral palsy, if genetic studies are not pursued [2].

Video 1. Patient's Movement Disorder: The patient presents axial hypotonia, and a mixed hyperkinetic movement disorder, consisting of chorea, dystonia, myoclonus, and hand stereotypies, giving the impression of a "stop-motion" chorea.

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



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REFERENCES

1. Johannesen KM, Gardella E, Gjerulfsen CE, et al. *PURA*-Related Developmental and Epileptic Encephalopathy: Phenotypic and Genotypic Spectrum. *Neurol Genet*. 2021 Nov 15;7(6):e613. doi: 10.1212/NXG.0000000000000613.
2. Reijnders MRF, Janowski R, Alvi M, et al. *PURA* syndrome: Clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. *J Med Genet* 2018; 55:104–113.

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