Teaching Video NeuroImage: Stop-Motion Chorea in PURA Syndrome

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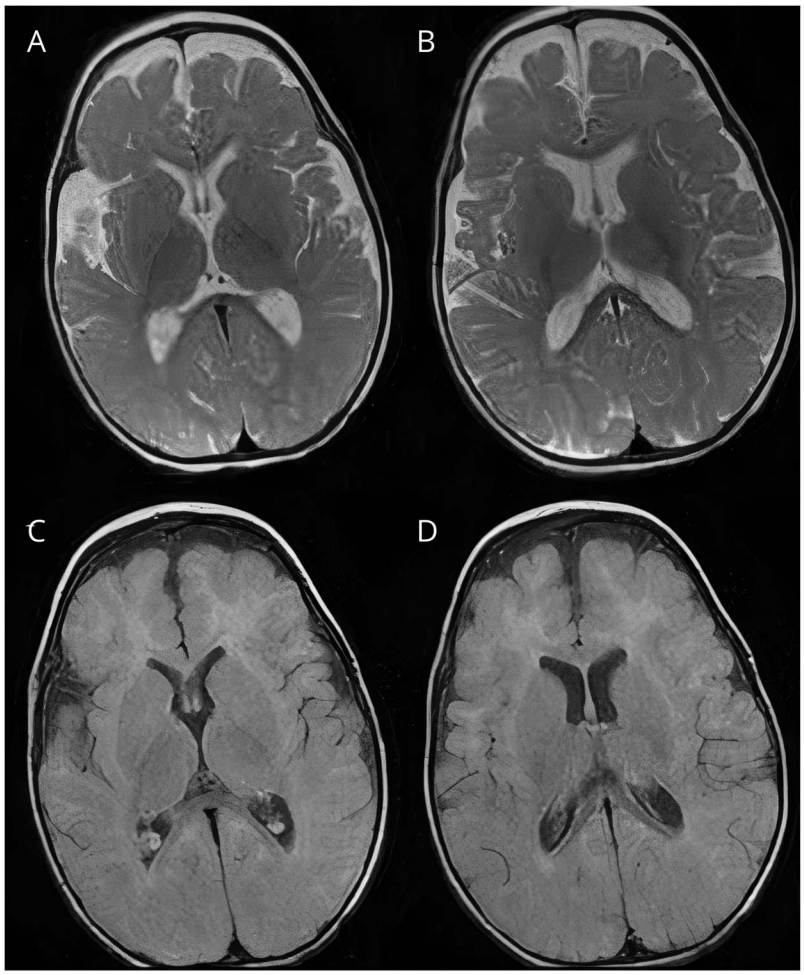
Figure Brain MRI



(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.

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

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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(139033279_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. Affected patients may be misdiagnosed with dyskinetic cerebral palsy if genetic studies are not pursued.[2]

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