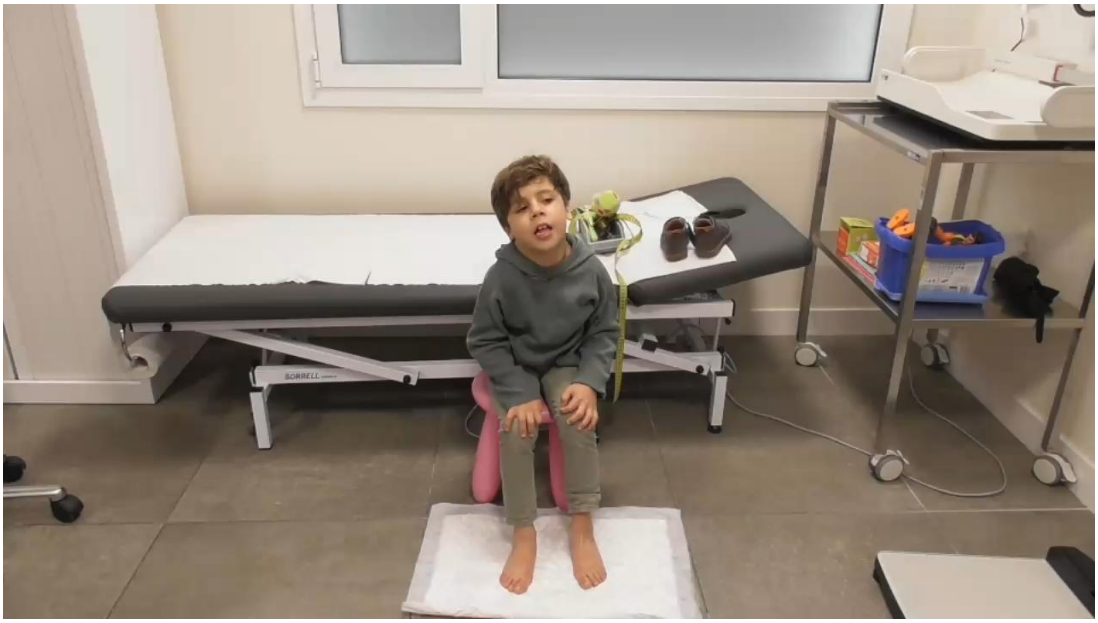
Lasa-Aranzasti A, et al. Clinical and Molecular Profiling in GNAO1 Permits Phenotype-Genotype Correlation. Mov Disord 2024;39:1578-1591.

dystonia → hyperkinetic crisis

股屈曲過活動・股伸展荷重制限
共収縮制御障害＋共収縮を伴う突発性運動



1

chorea・dystpnia → choreic storm (covid)

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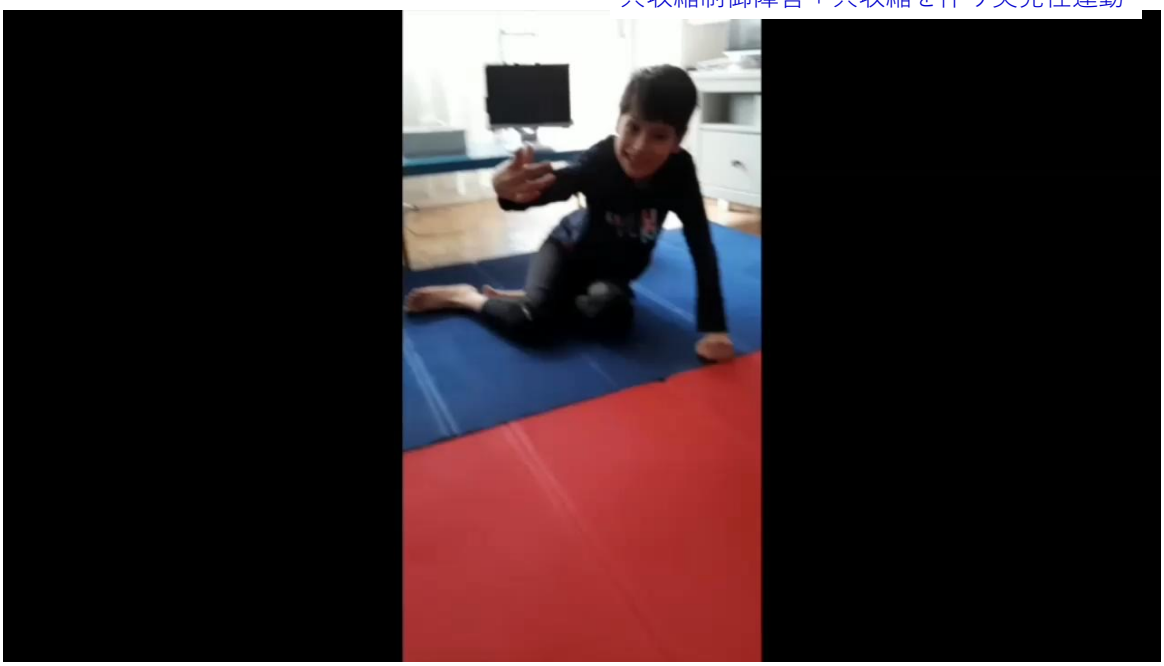

2

exacerbation of hyperkinetic movements→choreo-dystonic movements

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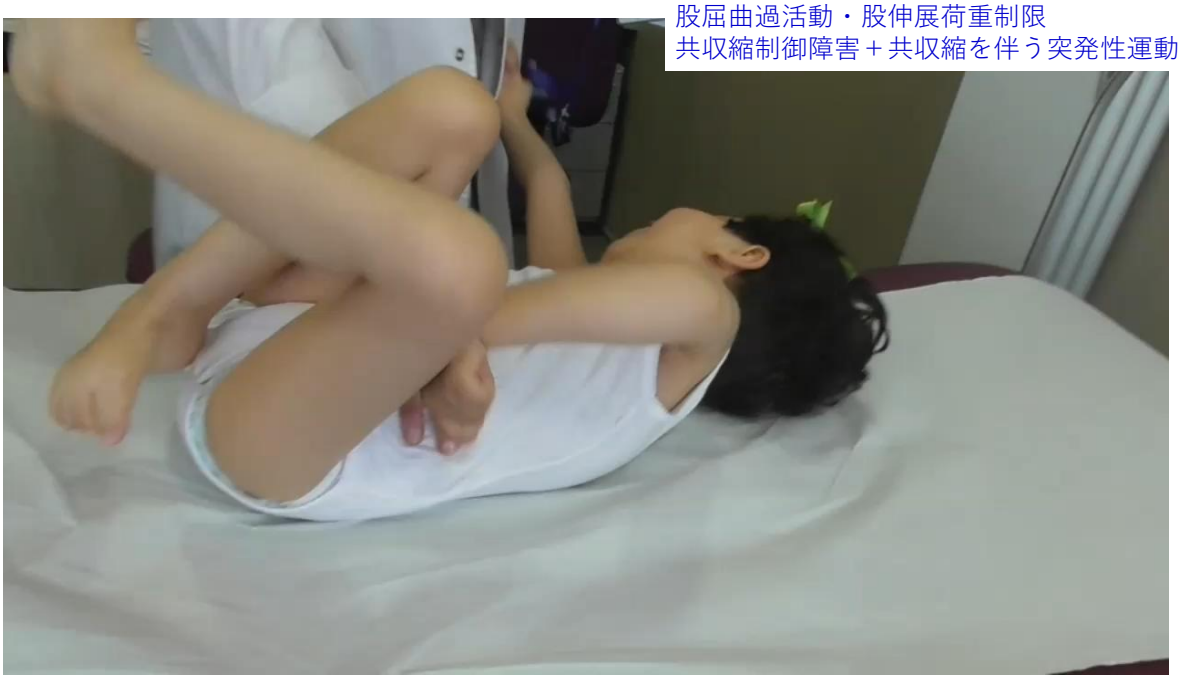

3

hyperkinetic movements (choreic, myoclonic, and dystonic)

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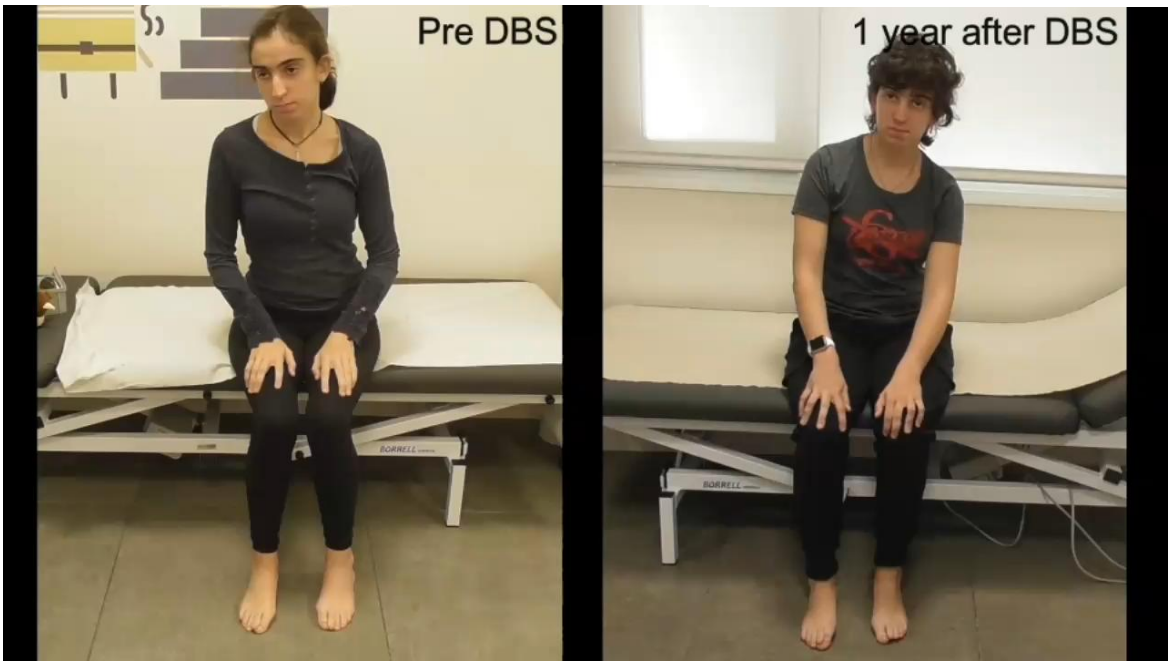


4

Video S1 Combined Hyperkinetic Movement Disorders. Patient 50 (GNAO1). Two videos of the same patient are presented: in the first one, he is sitting in his baby stroller and in the second, he is lying in bed. In both videos, prominent and **generalized choreo-dystonic movements** are observed. **Stereotypies** of the hands are present, as well as **intermittent involuntary mouth opening**. The movements are continuous and in the second video there is an impression of discomfort.

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共収縮制御障害＋共収縮を伴う突発性運動

Pérez-Dueñas B et al. The Genetic Landscape of Complex Childhood-Onset Hyperkinetic Movement Disorders. Mov Disord 2022;37:2197-2209.

5

Video S2 Dystonia. Patient 127 (GNAO1). The patient is walking. He has **generalized dystonic movements** that affect gait stability.

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6

**Video S1: Patient 1, with *GNAO1* mutation**

**Patient 1 (currently 8 years)** had a history of infantile-onset neurodevelopmental delay associated with severe axial hypotonia, generalised chorea and dystonic posturing of hands. From 2 years of age, he showed prolonged episodes of paroxysmal hyperkinesia, with partial response to Tetrabenazine. From 3 years of age he suffered well-controlled focal seizures with secondary generalisation. The c.139A>G [p.(Ser47Gly)] *GNAO1* mutation was detected by whole exome sequencing. Video S1 (age 7 years) illustrates an episode of **exacerbated hyperkinetic movements with generalised ballismus and chorea**, as well as **dystonic posturing** of the hands and intermittent orolingual dyskinesia.

Apostolos Papandreou A, et al. The expanding spectrum of movement disorders in genetic epilepsies. Dev Med Child Neurol 2020;62:178-191.

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*共収縮制御障害*+共収縮を伴う突発性運動

7


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8