# ISOLATED GENERALIZED CHOREA IN A PATIENT WITH SMALL-EXPANDED ALLELE SPINOCEREBELLAR ATAXIA 17

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**BACKGROUND:** Spinocerebellar ataxia type 17 (SCA17) is an autosomal dominant cerebellar ataxia caused by a polyglutamine-encoding CAG/CAA repeat expansion within the TATA box-binding protein (TBP) gene. It is characterized by a markedly heterogeneous phenomenology and complex genotype–phenotype relationships. This includes decreased penetrance forms where the clinical presentation ranges from pure cerebellar ataxia to a Parkinson's disease-like phenotype.

**OBJECTIVE:** We report the case of a patient carrying 41 CAG/CAA repeats in the TBP gene, presenting with isolated chorea and depressive symptoms, in order to contribute to the understanding of the phenotype heterogeneity associated with small-expanded alleles of SCA17. To date, in fact, only a few SCA17 patients with 41 CAG/CCG repeats have been reported.

**METHODS:** We describe the clinical, neuropsychological, and neuroimaging findings of a 73-year- old patient who presented with depressive syndrome and a 10-year history of generalized hyperkinetic movements. The patient's family history was unremarkable. Neurological examination revealed choreic movements affecting the upper and lower limbs, the face and the trunk with no additional neurological signs **(*VIDEO 1*)**. Blood sample analysis, brain imaging (***FIG. 1***), and neuropsychological evaluation revealed normal results. Genetic analysis identified, in the TBP gene, the 41-CAG pathological allele with reduced penetrance.

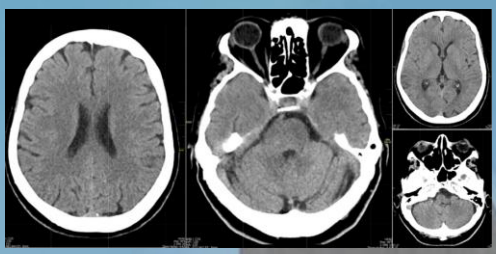

***FIG. 1****. Axial views of the patient's brain computed tomography (CT) scan. The images reveal hypodensity in the periventricular white matter, semioval centers, and corona radiata bilaterally, indicative of mild chronic hypoxic-ischemic encephalopathy. Additionally, there is minimal enlargement of the cerebrospinal fluid spaces at the base and vault, suggestive of slight cortical atrophy. The rest of the exam appears normal.*

***VIDEO 1.*** *The patient exhibits irregular, non-stereotyped involuntary movements consistent with choreoathetosis, predominantly involving the head and upper limbs, with occasional involvement of the lower limbs and orobuccal region. These movements are present both at rest and during postural maintenance. Extraocular movements are normal, except for questionable saccadic intrusions. Motor impersistence is observed, particularly during tongue protrusion and hand grip (manifesting as the "milkmaid's sign"). Muscle strength is preserved. There are no overt signs of dysmetria, adiadochokinesia, or bradykinesia, although choreic movements interfere with repetitive motor tasks. Mild left-sided dysdiadochokinesia, right-sided dysrhythmia during finger tapping, and slight bilateral dysmetria on heel-to-knee testing are noted. Gait is normal, though choreic movements of the head and upper limbs become more apparent during ambulation. Tandem gait is possible with only slight unsteadiness, and postural reflexes are preserved. The remainder of the neurological examination is unremarkable.*

**RESULTS:** Our case of depressive symptoms and generalized chorea, without additional neurological features, in a patient carrying 41 CAG/CAA repeats, supports the recently proposed new clusters of repeat expansion sizes for SCA17.

**CONCLUSIONS:** The present case report provides further insight into the small-expanded allele SCA17 associated phenotype.

***MAJOR REFERENCES:***

- [1] Rossi M, Hamed M, Rodríguez-Antigüedad J, et al (2023) Genotype-Phenotype Correlations for ATX-TBP (SCA17): MDSGene Systematic Review. Mov Disord 38:368–377. https://doi.org/10.1002/mds.29278
- [2] Paolini Paoletti F, Prontera P, Nigro P, et al (2021) Small-expanded allele spinocerebellar ataxia 17: imaging and phenotypic variability. Neurol Sci 42:4309–4315. https://doi.org/10.1007/s10072-021-05313-z 3.

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