Movement Disorders
CLINICAL PRACTICE

CASE REPORT

Jaw-Opening Oromandibular Dystonia Associated With Spinocerebellar Ataxia Type 2

Antonella Antenora, MD,[1,] Silvio Peluso, MD,[1] Francesco Saccà, MD,[1] Giuseppe De Michele, MD,[1] Alessandro Filla, MD[1]*

SCA2 is an autosomal-dominant neurodegenerative disease caused by expansion of CAG repeats in the *ATX2* gene on chromosome 12.[1] As with other polyglutamine expansion diseases, SCA2 is characterized by anticipation, with earlier onset in more recent generation, associated mostly with paternal transmission and larger expansion. Besides progressive cerebellar ataxia, SCA2 is characterized by slow saccadic movements and decreased/absent tendon reflexes. Cognitive decline, extrapyramidal or pyramidal signs, and myoclonus may occur.[2]

We describe an Italian patient with genetically confirmed SCA2, showing jaw-opening oromandibular dystonia. The patient is a 26-year-old man, followed-up in our clinic since age 18 for progressive limb and gait ataxia, dysarthria, mild dysphagia, and slow saccadic eye movements. The patient's father and paternal grandmother were also affected by ataxia and received genetic diagnosis of SCA2. CAG triplet repeats were 38 and 22 in the father and 39 and 22 in the grandmother. The father had onset at 38 years and the grandmother at 47. The progression was slow. Genetic testing of the *ATX2* gene confirmed the diagnosis of SCA2 in the proband. CAG triplet repeats were 50 and 22, showing a larger expansion, compared to the father. The disease progression was faster, with loss of independent walking after 5 years of disease duration. He also developed a rapidly progressive cognitive decline since age 24 and anarthria at 25. At 26, he developed recurrent episodic dystonic movements restricted to the oromandibular region, characterized by extreme and sustained jaw-opening spasm, tongue dyskinesias within the mouth, and elevation of soft palate, with duration of approximately 30 to 60 seconds (Video). The most severe episodes were associated with dyspnea and cyanosis. No particular trigger events or sensory tricks were associated. He had never been exposed to neuroleptics, and his history was negative for oromandibular-facial trauma or dental procedures. Neurological examination showed severe ataxia with inability to walk, cognitive impairment, anarthria, saccade slowing, brisk knee jerks, and Babinski signs. Brain MRI showed severe cerebellar and brainstem atrophy with hot cross bun sign (cruciform hyperintensity of the pons on T2-weighted images) as well as mild supratentorial atrophy. In the first 5 months after onset of dystonia, the episodes were rather sporadic, with good response to clonazepam. After, frequency and duration of the episodes of jaw-opening dystonia increased and a severe dysphagia to both solids and liquids developed, requiring a percutaneous endoscopic gastrostomy. After the procedure, the patient was transferred to the intensive care unit because of a respiratory failure. At last evaluation, the patient state was that of a decreased level of consciousness with a permanent tracheostomy and gastrostomy. The parents refused botulin toxin or other treatments.

Among features accompanying ataxia, dystonia is present in approximately 14% of SCA2 patients and is usually associated with larger expanded repeats.[3] Only few studies have reported on the localization of dystonia in SCA2, with several cases of cervical dystonia[4] and few cases of writer's cramp and foot dystonia.[5] Oromandibular dystonia, which has been previously described in other SCAs as SCA1,[6] SCA8,[7] and SCA36,[8] may be a serious complication, leading to, or worsening, chewing and swallowing difficulties. On the other hand, dysphagia may be present in approximately 41% of SCA2 patients, especially in the late stage of the disease,[9] or in cases with infantile onset.[10]

Author Roles

(1) Research Project: A. Conception, B. Organization, C. Execution; (2) Statistical Analysis: A. Design, B. Execution, C. Review and Critique; (3) Manuscript: A. Writing of the First Draft, B. Review and Critique.

A.A.: 1A, 1B, 1C, 3A
S.P.: 1A, 1B, 1C, 3A
F.S.: 3B
G.D.M.: 1A, 3B
A.F.: 1A, 1B, 1C, 3B

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[1]Department of Neurosciences, Reproductive and Odontostomatological Sciences, Federico II University, Naples, Italy

[*]**Correspondence to:** Dr. Antonella Antenora, Department of Neurosciences, Reproductive and Odontostomatological Sciences, Federico II University, Via Pansini 5, Naples 80131, Italy; E-mail: antonella.antenora@virgilio.it
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References

1. Imbert G, Saudou F, Yvert G, et al. Cloning of the gene for spinocerebellar ataxia 2 reveals a locus with high sensitivity to expanded CAG/glutamine repeats. *Nat Genet* 1996;14:285–291.
2. Schmitz-Hübsch T, Coudert M, Bauer P, et al. Spinocerebellar ataxia types 1, 2, 3, and 6: disease severity and nonataxia symptoms. *Neurology* 2008;71:982–989.
3. van Gaalen J, Giunti P, van de Warrenburg BP. Movement disorders in spinocerebellar ataxias. *Mov Disord* 2011;26:792–800.
4. Boesch SM, Müller J, Wenning GK, Poewe W. Cervical dystonia in spinocerebellar ataxia type 2: clinical and polymyographic findings. *J Neurol Neurosurg Psychiatry* 2007;78:520–522.
5. Furtado S, Payami H, Lockhart PJ, et al. Profile of families with parkinsonism-predominant spinocerebellar ataxia type 2 (SCA2). *Mov Disord* 2004;19:622–629.
6. Wu YR, Lee-Chen GJ, Lang AE, Chen CM, Lin HY, Chen ST. Dystonia as a presenting sign of spinocerebellar ataxia type 1. *Mov Disord* 2004;19:586–587.
7. Ushe M, Perlmutter JS. Oromandibular and lingual dystonia associated with spinocerebellar ataxia type 8. *Mov Disord* 2012;27:1741–1742.
8. Miyashiro A, Sugihara K, Kawarai T, et al. Oromandibular dystonia associated with SCA36. *Mov Disord* 2013;28:558–559.
9. Filla A, De Michele G, Santoro L, et al. Spinocerebellar ataxia type 2 in southern Italy: a clinical and molecular study of 30 families. *J Neurol* 1999;246:467–471.
10. Abdel-Aleem A, Zaki M. Spinocerebellar ataxia type 2 (SCA2) in an Egyptian family presenting with poliphagia and marked CAG expansion in infancy. *J Neurol* 2008;255:413–419.

Supporting Information

A video accompanying this article is available in the supporting information here.

Video. Oromandibular dystonia in the SCA2 patient, characterized by extreme jaw opening with elevation of soft palate and tongue dyskinesias. The episode concluded with a quick adduction of palatopharyngeal archs and repetitive chewing-like movements.

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