CASE REPORT

Cerebellar cognitive affective syndrome due to cerebellar atrophy: case report

Síndrome cerebelar cognitivo-afetiva secundária à atrofia cerebelar: relato de caso

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Abstract

Cerebellar atrophy is a rare and challenging disease with few descriptions in the medical literature. Motor impairment is mild, but behavioral and linguistic alterations stand out, in what is known as the cerebellar cognitive affective syndrome secondary to cerebellar atrophy. We report the case of an older woman with early-onset (age 45) signs and symptoms of this syndrome, including impairment of executive functions and visuospatial cognition, personality changes, and language deficits, who was followed at a geriatric medical center for 14 years. Neuropsychological, imaging, and behavioral aspects during this period are discussed in light of scientific evidence. This case report contributes to the scientific literature by describing the progression of the signs and symptoms of cerebellar atrophy over the years, which can help guide medical management and support advice for patients and their families.

Keywords: cerebellar ataxia; cerebellar diseases; case reports.

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Resumo

A atrofia cerebelar é uma doença rara, desafiadora e com poucas descrições na literatura médica. O prejuízo motor é discreto, mas as alterações comportamentais e de linguagem se destacam, caracterizando a síndrome cognitivo-afetiva cerebelar secundária à atrofia cerebelar. Apresentamos o relato de caso de uma paciente idosa, que apresentou sinais e sintomas dessa síndrome precoce (aos 45 anos de idade) — tais como déficits na função executiva, prejuízo visuoespacial, alterações de personalidade e déficits de linguagem — e foi acompanhada em um centro médico geriátrico por um período de 14 anos. Aspectos neuropsicológicos, de imagem e comportamentais durante esse período são comentados à luz das evidências científicas. O caso relatado contribui com a literatura científica ao descrever a evolução dos sinais e sintomas da atrofia cerebelar ao longo dos anos, balizando as condutas médicas e amparando as orientações ao paciente e seus familiares.

Palavras-chave: ataxia cerebelar; doenças cerebelares; relatos de casos.

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INTRODUCTION
The differential diagnosis of dementia is a challenge in medical practice, especially in those patients with early-onset dementia (which occurs before age 65 years) and those in whom amnestic symptoms are not prominent.1

Few studies address the role of the cerebellum in dementias. The cerebellum has long been considered critical for motor functioning, but a growing body of evidence demonstrates that it also plays a significant role in cognitive and affective processing.2,3 There appears to be a functional dichotomy of the cerebellum, so that the anterior portion of the organ is responsible for controlling the sensorimotor system, while its posterior portion would control cognitive-affective function.4

We report herein a case of cerebellar cognitive-affective syndrome (CCAS) secondary to cerebellar atrophy, followed over 14 years at a geriatric specialist center.

The authors of the case report obtained written consent. The signed Informed Consent Term has been submitted alongside the manuscript.

CASE DESCRIPTION
A Brazilian widow and housewife with 8 years of formal education presented in 2021 at age 65. According to her daughter, at 45 years of age the patient exhibited behavioral changes such as isolation, sadness, irritability, flat affect, childish behavior, and slowed speech, with dysprosody and a nasal-sounding voice, reported as “stuttering”. These symptoms coincided with financial difficulties after her husband’s death, which led to a diagnosis of depression and the prescription of fluoxetine 20 mg daily. The patient remained under psychiatric care for the following six years, with several changes in medication but no significant improvement in her clinical picture.

Progressively, the patient developed abnormal (“drunken”) gait, postural instability, and experienced multiple falls. At age 51 years, she was referred for evaluation at a specialist dementia center due to a complaint of “forgetfulness”. At the time, her Mini-Mental State Examination (MMSE) score was borderline at 24. The neuropsychological assessment demonstrated impairment of language, executive function, and praxis; below-average performance in activities related to perceptual organization, working memory, and processing speed (Figure 1); good orientation in space and time; and satisfactory scores on verbal recall tests, with an ascending learning curve. She exhibited dysdiadochokinesia, dysmetria, and a slightly ataxic gait on physical examination. Laboratory tests were normal. A computed tomography scan of the head showed cerebellar atrophy with no vascular involvement (images not available). The patient was independent for basic and semi-dependent for instrumental activities of daily living.

She continued to attend regular follow-up visits in subsequent years and take citalopram 20 mg. At 60 years of age, 15 years after her first symptoms, she experienced a slight function deterioration. According to her daughters, the patient’s language skills worsened (naming and fluency), her reasoning was flawed, and she experienced episodes of mental confusion. There were also behavioral changes, such as a preference for sweet foods and overall apathy. The patient’s daughters reported that she could not have her whims denied or contradicted and is easily excited. Repeat neuropsychological

FIGURE 1. Clock-drawing test (A) and pentagon test (B) components of the Mini-Mental State Examination done during a 2012 neuropsychological assessment, demonstrating visuospatial disorganization.
assessment in 2018 (compared to 2012) was consistent with a slight decline in tests related to executive function, abstraction capacity, judgment, and critical thinking, a deterioration that was also clinically slight. Conversely, there was no change in tests of episodic memory (immediate and delayed recall) (Table 1).

Despite this clinical progression, cerebellar size did not change substantially between 2011 (Figure 2A) and 2018 (Figure 2B). 5

**DISCUSSION**

CCAS, or Schmahmann’s syndrome, was first described in 1998, based on a careful neurological examination, detailed neuropsychological tests, and anatomical neuroimaging of a group of 20 patients with focal cerebellar disorders. 6 It is characterized by four groups of symptoms, including:

1. Executive function deficits (planning, cognitive flexibility, abstract reasoning, verbal fluency, and working memory);
2. Visuospatial impairment;
3. Personality changes, with blunted affect or abnormal behavior; and
4. Language deficits (agrammatism, dysprosody and mild anomia). 7

In this case, a thorough workup did not identify any secondary causes for the signs and symptoms observed, and neuroimaging ruled out the possibility of stroke or tumors. The absence of a clinical profile consistent with Alzheimer’s disease or other dementias and the long-term follow-up of this patient led to a diagnosis of CCAS, which can be a challenge in clinical practice, as this syndrome is often confused with countless others. Family distress, when faced with the lack of a definitive diagnosis and the difficulty in establishing therapeutic goals is a particular challenge in the follow-up of these patients.

CCAS can have various etiologies, such as cerebellar injuries (hemorrhage, infarctions, or cerebellar tumors), post-infectious cerebellitis, hereditary and non-hereditary idiopathic cerebellar atrophy, alcoholic cerebellar degeneration, and drug toxicity (e.g., phenytoin). 2,3,6,8 In addition to cerebellar-specific conditions, several neurodegenerative syndromes can affect the cerebellum, including Alzheimer’s disease, multiple system atrophy, amyotrophic lateral sclerosis, frontotemporal dementia, and progressive supranuclear palsy. 2

The neuropsychological picture is particularly interesting in CCAS. Episodic and semantic remote memories are preserved; however, other cortical features are absent, leading to aphasia, apraxia, and agnosia. 6 Affective changes are manifold and include personality changes, lack of inhibitory regulation, inappropriate behavior, altered mood regulation, blunted affect, obsessive-compulsive tendencies, and psychotic thinking, which, to some authors, is secondary to an impairment in so-called social cognition. 8

**TABLE 1.** Progression of selected clinical, neuropsychological, and imaging findings over a 14-year follow-up period.

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<tr>
<td>Clinical</td>
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<tr>
<td>Postural instability</td>
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<td>+++</td>
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<tr>
<td>Ataxic dysarthrophonia</td>
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<td>Urinary incontinence</td>
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<td>Tremor</td>
<td>+</td>
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<tr>
<td>Dysphagia</td>
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<tr>
<td>Dependent for instrumental activities of daily living</td>
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<td>++</td>
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<tr>
<td>Dependent for basic activities of daily living</td>
<td>-</td>
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<td>+</td>
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<tr>
<td>Dysautonomia (syncope and change in bowel habits)</td>
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<tr>
<td>Cerebellar atrophy</td>
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<tr>
<td>Neuropsychological</td>
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<td>Episodic memory, immediate and delayed recall</td>
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<td>Executive functions</td>
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<td>Orientation in space and time</td>
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<td>Semantic memory and abstraction ability</td>
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<td>Judgment and critical reasoning</td>
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</tbody>
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- : absent; +: mild; ++: moderate; +++: marked; Ø: not assessed.

Neuropsychological assessments carried out in 2012 and 2018. Brain MRI performed in 2011 and 2018; head CT performed in 2007. Clinical examination was performed at all time points.

Source: Own work, based on patient records.
As in the case of the patient reported herein, the early onset of symptoms is the rule in most published descriptions, with manifestations almost always appearing before 60 years of age. For instance, in the original study of Schmahmann and Sherman, age ranged from 24 to 56 years. Life expectancy has not been reported frequently in studies, but Ota et al. described neuropathological findings in four patients and reported one case of survival for 26 years after diagnosis (54 to 80 years). Therapeutic options are limited; as in many other neurodegenerative diseases, there is no specific treatment, only management of mood symptoms and, to a lesser extent, preservation of language through speech therapy interventions. Gait and balance training can also help reduce falls, although there is no scientific evidence of effectiveness. Meetings with family members and caregivers helped elucidate any issues and provided guidance on dealing with the patient’s mood changes and language difficulties.

This report has some limitations inherent to any clinical case, such as the retrospective design, the limited generalizability of findings, and the impossibility of establishing a causal relationship between cerebellar atrophy and the patient’s clinical abnormalities. However, considering the few cases of this condition described in the literature, the present case report can contribute by generating hypotheses and helping clinicians in everyday practice. No genetic studies were carried out, which is considered a limitation.

Finally, CCAS should be included in the differential diagnosis of dementia syndromes, especially in those cases which feature deficits in executive function, visual-spatial impairment, personality changes, and language deficits. In addition, the early onset of symptoms (before age 60) and evidence of cerebellar atrophy on neuroimaging are commonly seen in this syndrome.

**Conflict of Interest**
The authors report no conflicts of interest.

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Authors’ Contributions
EFC: Conceptualization, investigation, methodology, visualization, writing – original draft. YM: Conceptualization, investigation, methodology, visualization, writing – original draft. LLL: Conceptualization, investigation, methodology, visualization, writing – original draft. JLQ: Conceptualization, investigation, methodology, visualization, writing – original draft.

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