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LETTER TO THE EDITOR

From early psychiatric symptoms to an adolescent diagnosis: Clues from childhood in spinocerebellar ataxia Type-42

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Dear Editor,

Spinocerebellar ataxias (SCAs) comprise a group of genetically inherited, heterogeneous neurodegenerative disorders that primarily affect the cerebellum, brainstem, spinal cord, and cranial nerve nuclei. These disorders are characterized by progressive gait and limb ataxia and may be accompanied by varying degrees of nystagmus, dysarthria, intention tremor, and ophthalmoparesis (1).

Spinocerebellar ataxia type 42 (SCA42) is a rare subtype that typically presents with slowly progressive cerebellar ataxia. It has been associated with a c.5144G>A (p.Arg1715His) mutation in the CACNA1G gene (2, 3). In cases reported by Coutelier, the age of onset ranged from 9 to 78 years, whereas in the series reported by Morino et al. (4), the age of onset ranged from 20 to 70 years (5). A recent study demonstrated that de novo mutations in the CACNA1G gene can result in a clinical picture characterized by prominent neurodevelopmental impairment and cerebellar ataxia beginning in early childhood (6). Increasing reports from different countries, along with the identification of novel mutations, have highlighted the clinical and genetic heterogeneity of SCA42 (7).

Here, we present the case of a 15-year-old male who exhibited developmental and psychiatric symptoms nearly a decade before the onset of motor manifestations. The patient has been followed in a child and adolescent psychiatry outpatient clinic for speech fluency disorder, social anxiety disorder, specific phobias, and mild intellectual disability.

No significant complications were reported during the prenatal period, and both second- and third-trimester screening tests were within normal limits.

Following birth, the patient required 11 days of incubator care due to infection, although respiratory support was not needed. During infancy, he was monitored by pediatric neurology for macrocephaly.

He began walking independently at approximately 24 months, and his first meaningful words emerged between 2.5 and 3 years of age. Toilet training was delayed, and nocturnal enuresis persisted until 9–10 years of age.

At age 4, due to global developmental delay, the patient began receiving special education. Speech difficulties also became apparent around this time and have shown only minimal improvement despite numerous speech and language therapy interventions.

At age 6, he began to exhibit symptoms of social anxiety and specific phobias, which were later diagnosed as social anxiety disorder and specific phobia according to the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5) criteria (8).

At age 8, a Wechsler Intelligence Scale for Children–Fourth Edition (WISC-IV) assessment yielded a full-

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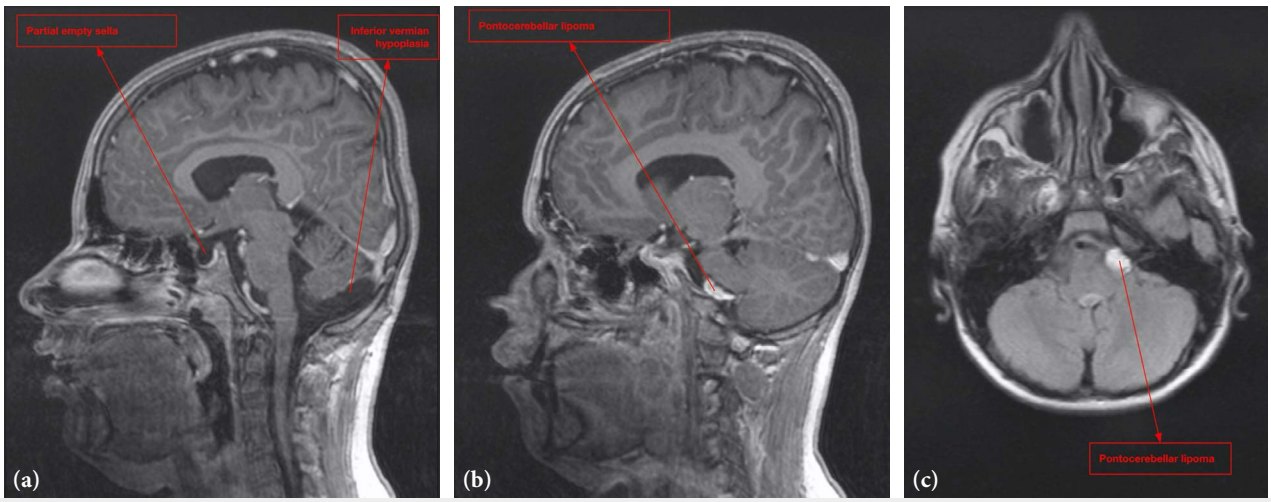

Figure 1. (a) Sagittal MRI scan revealing additional radiological findings: Partial empty sella (left arrow) and inferior vermian hypoplasia (right arrow). **(b)** Sagittal T1-weighted MRI scan demonstrating the localization of the pontocerebellar lipoma (indicated by the red arrow). **(c)** Axial T1-weighted MRI scan showing a pontocerebellar lipoma (indicated by the red arrow) in the posterior fossa.

scale IQ score of 50, consistent with a diagnosis of mild intellectual developmental disorder according to DSM-5 criteria (8).

Metabolic evaluations performed as part of the differential diagnosis (including lysosphingolipid panel, oligosaccharide analysis, and mucopolysaccharide screening) were within normal limits and did not support a diagnosis of mucopolysaccharidosis. The patient had no history of epileptic seizures. He was also followed by ophthalmology for strabismus.

At age 14, the patient was referred to neurology because of frequent falls. Neurological examination revealed a broad-based gait, dysmetria, and slowed fine motor performance. Brain magnetic resonance imaging (MRI) demonstrated inferior vermian hypoplasia (Fig. 1a), partial empty sella (Fig. 1a), right pontine atrophy, and a 17×13×11 mm left ponto-cerebellar lipoma (Fig. 1b, c).

Due to anxiety symptoms, the patient has been receiving fluoxetine at a dose of 30 mg/day for the past year and continues to benefit from the treatment.

Genetic analysis identified a pathogenic CACNA1G variant (p.Arg1715His) as well as a heterozygous variant in the SACS gene. These findings confirmed the diagnosis of SCA42. Both parents tested negative for the CACNA1G mutation; the patient's mother was identified as a heterozygous carrier of the SACS variant. Genetic counseling was provided to the family.

Neuropsychiatric symptoms are frequently observed in hereditary ataxias, although they are often underrecognized or misdiagnosed. Depression is the most commonly reported psychiatric comorbidity, while anxiety, apathy, agitation, and psychotic symptoms have also been reported to varying degrees (9). In SCA, anxiety is more prevalent than in the general population and often co-occurs with depression (10, 11). Additionally, impulsive and compulsive behaviors, as well as sleep disturbances, have been described (12).

Beyond its role in motor control, the cerebellum also plays a key role in cognitive and emotional regulation. Cerebellar Cognitive Affective Syndrome (CCAS), as defined by Schmahmann and Sherman, is characterized by impairments in executive functioning, social cognition, affect regulation, and language (e.g., verbal fluency and prosody). This syndrome can be observed in both children and adults with cerebellar dysfunction and underscores the importance of recognizing non-motor features in cerebellar disorders (13–15). In the present case, the presence of speech difficulties, social anxiety, and intellectual disability may be considered within the spectrum of CCAS.

Cases in which psychiatric symptoms begin in childhood and a diagnosis of spinocerebellar ataxia type 42 (SCA42) is established during adolescence are extremely rare in the literature (16). Although psychiatric and cognitive manifestations have been reported in SCAs, these features are most commonly described in adult-onset cases, and reports of childhood-onset presentations remain very rare (14, 15).

Given the broad clinical spectrum of SCA42, a comprehensive assessment integrating neurological, developmental, and psychiatric evaluations is essential for timely and accurate diagnosis. Recognition of early neuropsychiatric and developmental symptoms may facilitate earlier suspicion of SCA42, thereby enabling genetic counseling and anticipatory guidance for families. In the absence of disease-modifying treatments, multidisciplinary management focused on rehabilitation, educational support, and psychiatric care remains crucial.

Future studies should aim to clarify genotype–phenotype correlations and explore potential targeted therapies (17).

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