Pseudoproptosis in a case of familial hereditary cerebellar ataxia

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Key words: Cerebellar disorders, pseudoproptosis, spinocerebellar ataxia

Familial hereditary cerebellar ataxia is a rare congenital disorder wherein there is difficulty with gait and balance. As the disease progresses, there can be poor coordination of hand movements, eye movements, and speech.[1] These disorders are due to degeneration of the cerebellum. This disorder is very rare with an incidence rate of one to five per 100,000 population.[2] Almost 40 variants have been identified, and ophthalmic manifestations vary depending on the subtype involved. We report a case of spinocerebellar ataxia (SCA) with classical family history and progressive proptosis (pseudoproptosis).

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A 48-year-old female presented to us with complaints of inability to wear glasses as her right eye was bulging gradually for the past 2 years. She had no vision in her right eye since birth due to the opacity of the cornea. She was bound to wheelchair as she could not walk independently. She gave a history of gait disorder and was diagnosed to have cerebellar ataxia. She also gave a similar history in father, brother, and paternal aunt. She was given vitamin E supplements for the ailment. On examination, she had a proptosed right eye with no light perception. She had a total retinal detachment in the right eye. The magnetic resonance imaging (MRI) scan taken a few years back at the time of diagnosis of SCA showed almost the same axial length in both eyes. That did not show significant pseudoproptosis. Our patient had a progressive thinning and pseudoproptosis only for the past 2 years. Unfortunately, we did not do any MRI; we only did ultrasound B scan in the right eye. The ocular motility was restricted in all the gazes in the right eye. Her left eye had best-corrected vision of 6/9, n6. The axial length of the left eye was 25 mm. Her anterior and posterior segment findings of the left eye were normal. MRI showed an atrophic cerebellum characteristic of spinocerebellar ataxia [Fig. 1]. We gave an option of evisceration with a ball implant in the right eye to give her better comfort.

Discussion

SCA can be hereditary or nonhereditary. Nonhereditary causes of ataxia include intoxication (alcohol), cerebellar masses/infarcts, vitamin deficiencies (vitamin E), multiple sclerosis, and vascular disease, and paraneoplastic diseases should be ruled out. It is important to rule out that as they can be cured with treatment. There is no definitive treatment for hereditary

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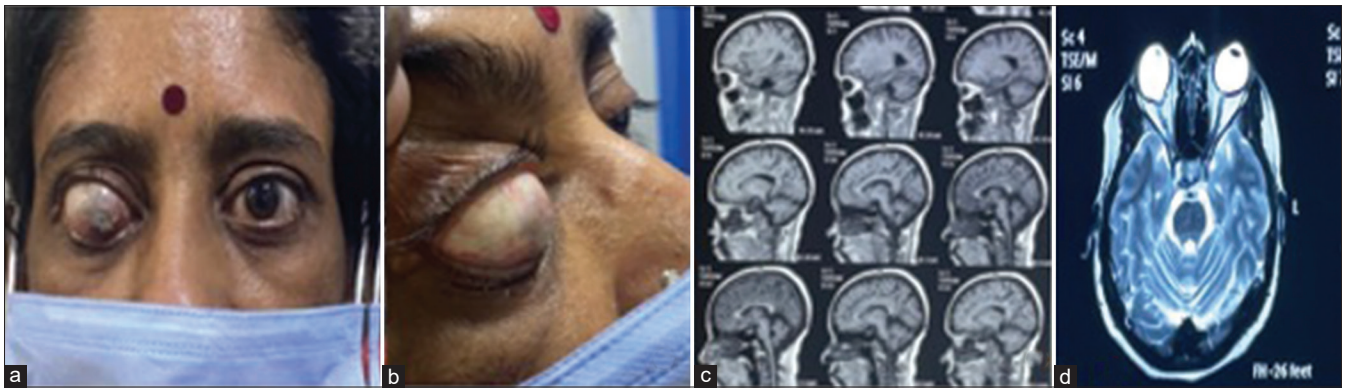

Figure 1: (a) External photograph of the patient with right pseudoproptosis. (b) Enlarged side view showing the prominence. (c) MRI brain showing atrophic cerebellum characteristic of spinocerebellar ataxia. (d) MRI orbit–axial scans of orbit at the time of diagnosis of spinocerebellar ataxia (no pseudoproptosis earlier)

SCA; however, a partial agonist at α4β2 nicotinic acetylcholine receptors (varenicline) approved as a smoking cessation drug was reported to improve symptoms of cerebellar dysfunction in patients with SCA in several case studies.[3]

Various ophthalmic findings reported are ophthalmoparesis, gaze-evoked nystagmus, bulging eyes, downbeat nystagmus, retinal degeneration, optic atrophy, and ptosis. Differentiating the subtypes based on clinical signs might be difficult, but there are few pathognomonic ophthalmic signs to give clues to the subtype.[4-6] Bulging eyes secondary to eyelid retraction with ophthalmoparesis is correlated with spinocerebellar ataxia type 3 (SCA3) but is not pathognomonic,[7] but retinal detachment could be incidental.

Our patient had no vision since birth with an opaque cornea and a physical eye, but gradually she developed progressive bulging of eyes. The reason for pseudoproptosis could be due to retracted lids due to overacting retractors and proliferation of intraorbital fat pushing the globe anteriorly.[8]

Our patient did not have a genetic karyotyping but most likely could fall in this category. The other affected family members had no significant ophthalmic findings.

The prognosis depends on the subtype of the SCA, but most of them are bound to wheel chair in 15 to 20 years. We present this case to report this rare ophthalmic finding in an uncommon systemic disease.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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