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EC OPHTHALMOLOGY
Mini Case Study

Visual Loss in Spinocerebellar Ataxia Type 7. A Case Report

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Received: December 29, 2020; **Published:** March 31, 2021

Abstract

The Spinocerebellar ataxias (SCA) are a subset of hereditary cerebellar ataxias with autosomal dominant inheritance, they are a heterogeneous group of neurodegenerative ataxic disorders characterized by degeneration of the cerebellum and its connections with progressive ataxia accompanied by dysarthria and oculomotor deficits [1,2].

Keywords: *Polyglutamine (polyQ); Spinocerebellar Ataxia Type 7 (SCA7); Spinocerebellar Ataxias (SCA)*

The symptoms of SCAs include gait ataxia and incoordination, nystagmus and dysarthria. Patients can present pyramidal, extrapyramidal signs, ophthalmoplegia and cognitive impairment in some specific SCAs [2].

To date, 47 SCA subtypes have been described, and 35 causal genes have been identified. Their most frequent forms are polyglutamine (polyQ) expansion diseases. These diseases manifest above a threshold number of CAG repeats, which is different for each gene [3].

Spinocerebellar ataxia type 7 (SCA7) is a genetic disorder characterized by degeneration of the cerebellum, brainstem, and retina that is caused by abnormal expansion of a CAG repeat located in the *ATXN7* gene encoding sequence on chromosome 3p21.1 [4].

The prevalence of SCA7 is less than 1 per 100,000 individuals. Adult-onset SCA7 is characterized by progressive cerebellar ataxia, slowed ocular saccades, dysarthria, dysphagia, and pyramidal symptoms, such as hyperreflexia and spasticity. Retinal degeneration is a distinctive feature of SCA7 with progressive cone-rod dystrophy leading to eventual blindness [1].

Ataxia-associated symptoms were evaluated using the Scale and Rating of Ataxia (SARA), while extracerebellar features were assessed by the Inventory of Non-Ataxia Symptoms (INAS) [4].

Emerging therapies for neurogenetic diseases. provide physicians and patients of SCA hopes of effective treatments in the near future [2].

Case Report

A 39-year-old female patient with no family history of ataxia and a decreased visual acuity 16 years ago (2004), two years later with difficulty in walking. The best corrected visual acuity was 0.1 in both eyes. Electroretinogram without response. Visual evoked potentials

(VEP) detected no bioelectric responses and fundus with various circumscribed retinal dystrophy, epiretinal membrane in macula, pigments in the periphery, retinal vascular narrowing and optic disc with good coloration (Figure 1-4). Initially, she was diagnosed with macular dystrophy and later, in 2013, the molecular diagnosis has the presence of CAG pathological repeats within one ATXN7 gene. In 2017 SARA score was in 26 and INAS in 5, her vision was hands movements and could walk with great difficult. At present the optic discs show pallidness, physiological excavation and posterior pole of retina with accumulations of pigments in the form of bone spicules. No perception of light.

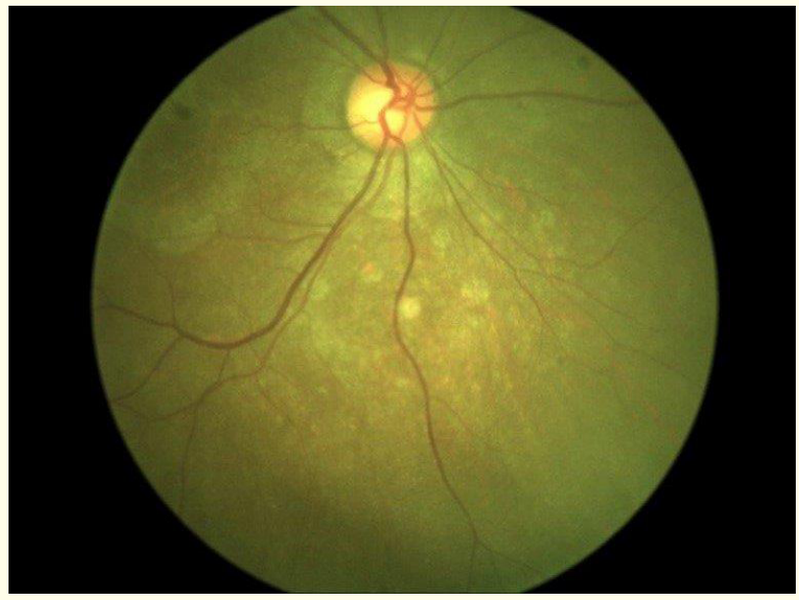

Figure 1: *Fundus Right eye.*

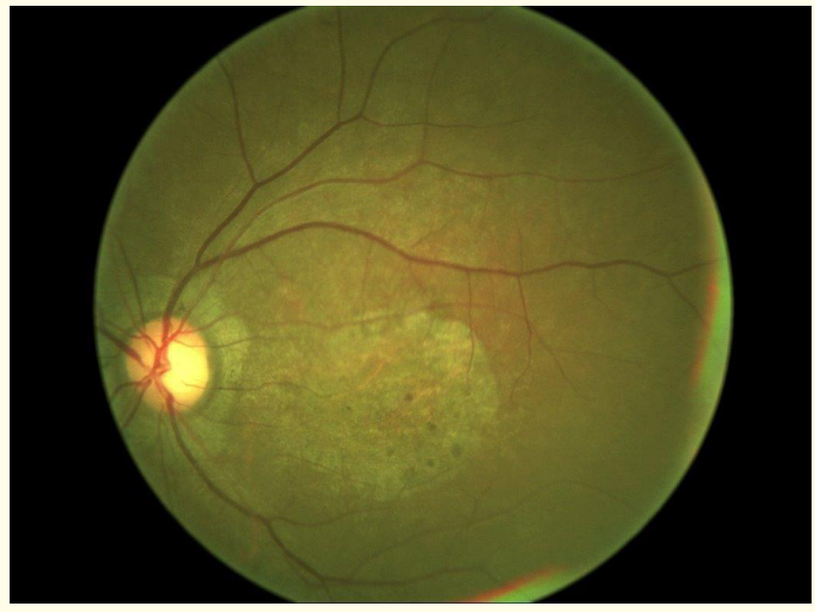

Figure 2: *Fundus Left eye.*

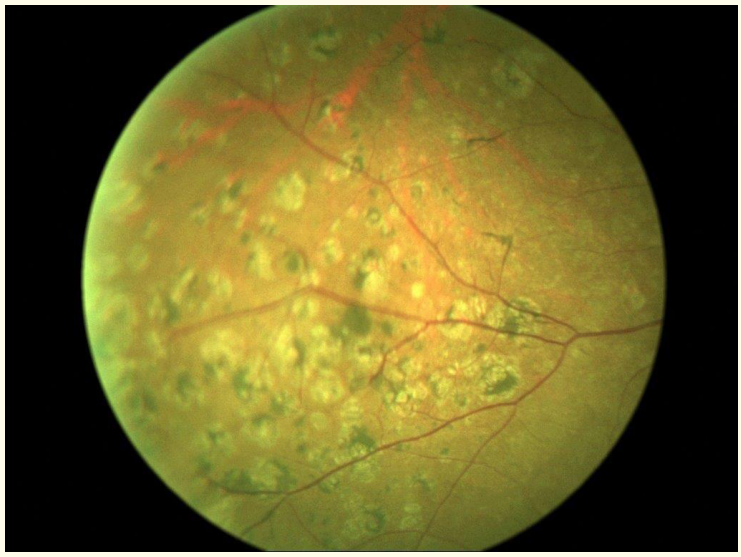

Figure 3: *Periphery right eye.*

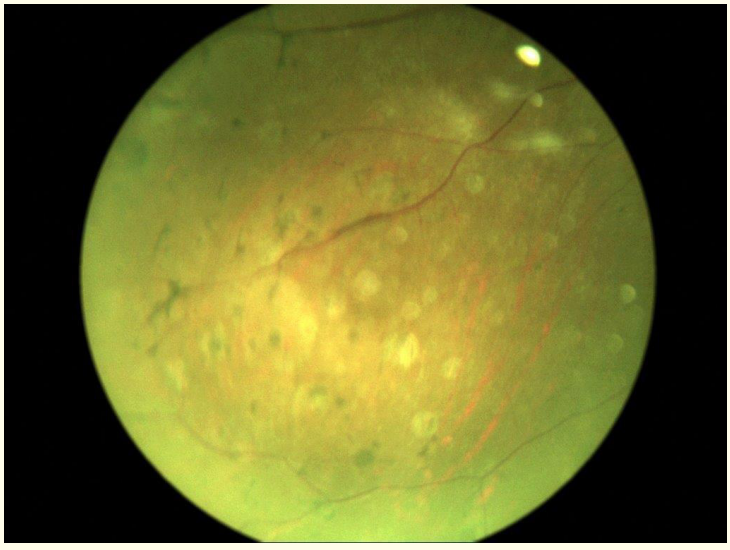

Figure 4: *Periphery right eye.*

Conclusion

This has been the usual evolution in a case of SAC 7 for which there are no possibilities of visual or neurological treatment to prevent visual loss in patients affected by this disease. She is in a terminal stage of her neurological disorder.

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Volume 12 Issue 4 April 2021