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Letter to the Editor

Color Vision Pathway Follows the Expansion Repeats in SCA7

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Spinocerebellar ataxia type 7 (SCA7) is an autosomal dominant neurodegenerative disorder caused by gene ataxin7 mutation with a polymorphic CAG repeat tract that falls in first axon. The gene which encodes ataxin7 normally contains a ten CAG repeat tract starting a codon 30, in affected patients the CAG repeat tract expands from 37 to 250 triplets, decreasing the transcription of an antisense non-coding RNA. Clinically SCA7 is characterized by progressive cerebellar ataxia, slow saccadic eye movements and progressive blindness.

SCA7 neurotoxicity can be showed by the cone-rod dystrophy which follows in the generations the CAG repeats expansions in the affected patients. As retinal disease progresses in SCA7, the photoreceptors cells do become involved, and then loss of central visual acuity proceeds to complete blindness. The presence in the long run of the altered cone function, allows SCA7 to be classified as a cone dystrophy type of retinal degeneration [1].

Considering the X-linked inheritance of color vision deficiency, fixed sampling of only males, that compensation's phenomenon in the heterozygous females [2], which can hid a real acquired color vision deficiency. The acquired color vision deficiency is real in the males where the anomaly has not hidden, and due to this reason we suggest sampling only male patients when color vision has studied both in neurological and metabolic diseases [3].

In the present study, the father and son of a familiar SCA7 were subjected to Ishihara test [4] to exclude the inherited red-green colorblindness, Farnsworth D15 test [5], and the City University Test [6] both monocularly, and binocularly, which are reliable to confirm the acquired color vision deficiency, especially regarding blue-yellow cone axis not identifiable by Ishihara test or by the most part of the pseudo-isochromatic clinical tests, and/or the cone dystrophy.

A father, 63 years old, showed a SCA7 ataxia diagnosed in 2017. Reading the three test showed a red-green acquired color vision deficiency coupled a blue-yellow acquired color vision deficiency.

A son, 23 years old, showed a SCA ataxia diagnosed in 2014. He does not read any clinical test showing a complete cone dystrophy.

The genetic advance is a phenomenon regarding the disorders caused by nucleotide repeats expansions. Based on it, the disorders manifest more severe symptoms than their parents, and early in each generation [3].

Color vision resulted to be an useful biological marker as previously [7] to be placed side by side the Nuclear Magnetic Resonance, in order to confirm the progression of SCA7, to two different generations.

The different color vision status in the father and his son follows the clinical greatness of the genetic disorder, too.

At the end, the comparison in our between two generations in familiar SCA7 study confirms the reliability of the simultaneous use of three tests identifying and scoring the acquired color defects in some neurodegenerative diseases, while also highlighting real brain manifestations.

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