SYNDROMES OF FUNCTIONAL VISUAL DISTURBANCES - A CASE REPORT AND OVERVIEW OF THE LITERATURE

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Abstract: *The large spectrum of syndromes related to functional visual disturbances is an important topic for both ophthalmologists and neurologists. Because of the diversity of these medical conditions, the clinician faced with such cases should thoroughly investigate the patient in order to exclude a structural disorder of the eye, visual pathways or cerebrum. The patient's medical condition is best approached when a multidisciplinary team of experts in ophthalmology and neurology is involved. In this paper, we present the case of a patient who suffers from visual snow syndrome, a very rare type of functional visual disturbance and who does not have currently an optimal treatment. In our continuous struggle to diagnose and treat diseases, we must always consider the advice of our fellow colleagues, and remember that we have a duty to the patient.*

INTRODUCTION

There are numerous clinical entities in which visual disturbances appear and the physicians cannot find structural anomalies, even with the help of ancillary investigations. By "functional disturbance", the medical community generally refers to a disorder in which no abnormality can be detected by the use of any of the available investigation methods, despite the presence of symptoms.(1,2,3)

The causes are various and are believed to be disorders of subcellular mechanisms in different functional organ systems. In the past, the preferred term was "cryptogenic mechanisms", as if the cause were hidden. With the continuous advances in medical research, more and more of the formerly called cryptogenic causes are being explained and understood. Still, there is a long way ahead until we can identify and treat the ever-growing number of peculiar syndromes.(1,2,3)

It is believed, that a large part of the human cerebrum is dedicated to processing the information from the visual apparatus. The visual processing begins in the 10 layered retina, a special part of the cerebrum. Next, the visual information is transferred through complex pathways to the central processing neurons, the primary visual cortex in the occipital lobe. Visual association areas of the cortex also pay an important role in visual processing and image perception. On an anatomical basis, we can assume that a functional disorder can have its disordered basis in any of the visual pathway component: from the eye to the cortex.(1-7)

CASE REPORT

This paper has been devoted to a case of a 29- year old patient, who presented in our clinic after being investigated and treated by several clinicians. No physician could find an explanation or a proper cure. The patient suffered a mild cranial trauma during his childhood, after which frequent headache attacks appeared later in 2008. Presently, our patient suffers from continuous scintillating dots in both visual fields, which appear as "snowflakes" and persist even on eye closure.

The patient had been formerly examined by numerous ophthalmologists and neurologists and was treated periodically with many neurotrophic agents, antimigraine drugs, anticonvulsants, benzodiazepines, anti-inflammatory drugs and many others. He reported that only Clonazepam 1-2 mg bid had a slight favourable effect on his symptoms.

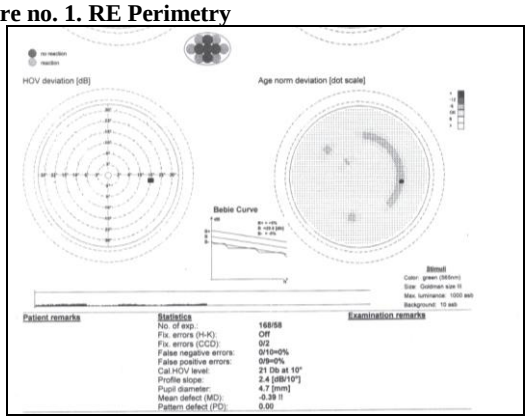
No abnormal signs could be detected during the neurologic exam. During the ophthalmologic exam, we found the following: visual acuity (VA) of the right eye (RE) = 1 wc, VA of the left eye (LE)= 1 wc, intraocular pressure (IOP) of the RE = 16 mmHg, IOP LE = 14 mmHg, anterior segment BE – normal, normal ocular motility. BE Ophthalmoscopy reveals vital papilla, well delimited, normal retinal vessels and macula with normal reflex.

The optical coherence tomography (OCT) reveals a central macular thickness of 280 µm in the RE, 278 µm in the LE, and a normal optic nerve bilaterally, without fiber loss.

The angiofluorography of the retina shows normal retinal vessels in both arterial and venous phases, bilaterally.

A computed perimetry reveals a lowering of the retinal sensitivity threshold.

Figure no. 1. RE Perimetry



All the blood tests were within normal parameters. Borrelia burgdorferi antibodies were negative for both IgM and

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IgG. Antineuronal antibodies were negative for: amphiphysin, CV2, PNMA2, Ri Yo, Hu, recoverina, SOX1 and titin. Serum B12 levels were 463 pmol/L (normal range: 156 – 672 pmol/L). Also, the thyroid panel tests were normal: TSH 4,7 mIU/L, anti-tireoperoxidase < 10 U/ml (normal range < 20 U/ml), anti TSH receptor < 0,3 IU/L (normal range < 1,75 IU/L).

Figure no. 2. LE Perimetry

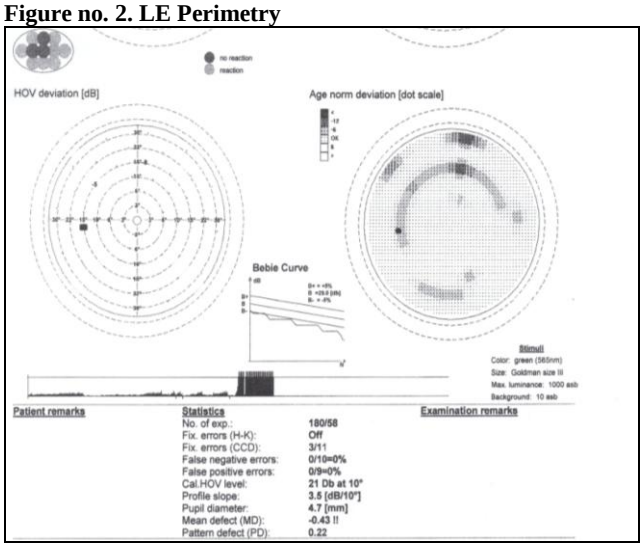
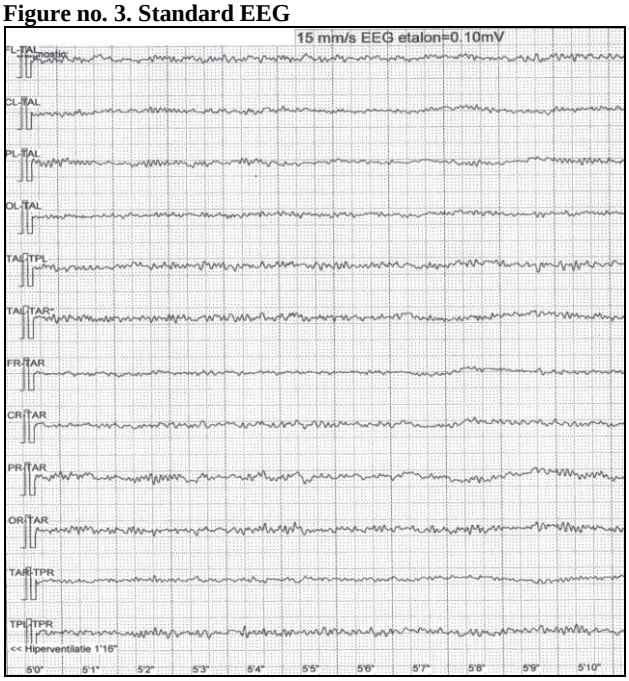


Figure no. 3. Standard EEG



Multiple cerebral MRI scans revealed no inflammation or other structural anomalies of the orbit, optic nerve or cerebrum.

A standard, long-run electroencephalography (EEG) showed a normal background rhythm, with normal anterior-posterior gradient, normal posterior dominant rhythm and no pathological discharges during or after the activating procedures.

Visual evoked potentials (VEP) showed normal P 100 latencies with normal amplitudes bilaterally and simetrically.

Psychological and psychiatric examinations were normal. At the one-year follow-up, the symptoms and repeated investigations were similar.

Figure no. 4. Visual evoked potentials (VEP)

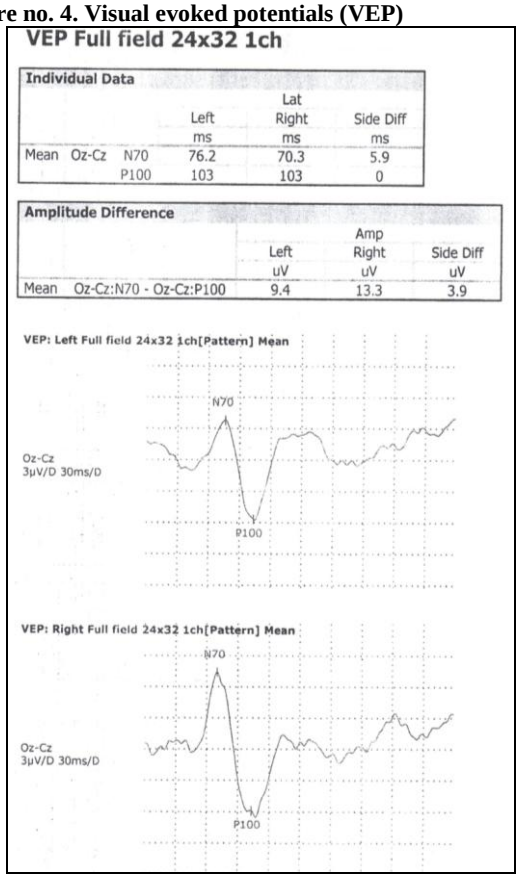
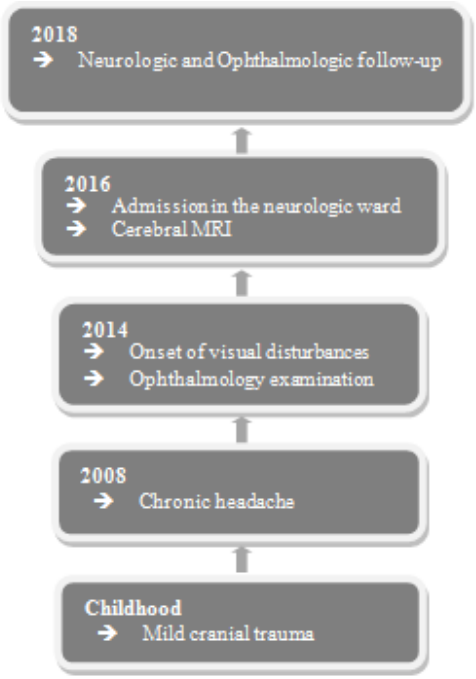


Figure no. 5. Timeline of events



DISCUSSIONS

The large spectrum of syndromes related to functional visual disturbances is an important topic for both ophthalmologists and neurologists. Because of the diversity of these medical conditions, the clinician faced with such cases should thoroughly investigate the patient in order to exclude

structural disorders of the eye, visual pathways or cerebrum before certifying a functional disorder.

Stargardt retinopathy, Leber's hereditary optic neuropathy (LHON), Vitamin A deficiency, Alcohol-tobacco amblyopia syndrome, Demyelinating diseases, Autoimmune diseases, Chronic intoxications, Visual snow syndrome, Retinal migraine variant, focal occipital seizures, Functional visual loss syndrome and various vision disorders due to other genetic defects, are some of the functional disorders of the visual apparatus.

Stargardt disease is due to mutations in the ABCA4 gene which codes a component protein of the photoreceptors. Its onset is before 20 years with a progressive macular degeneration.(1,2,3,4,5)

The LHON has its onset in the second decade of life, with a chronic progressive visual loss in both eyes. The first symptom is a bilateral central scotoma (similar to Neuromyelitis optica), progressing to amaurosis, with atrophic optic discs.(6,7,8,9,10)

The retinal migraine variant is a rare disorder in which visual impairments such as temporary scotomata, blindness or scintillations appear in one eye, just before the headache.(11,12,13) The diagnostic criteria of the retinal migrain according to the International Headache Society (IHS) are:(11)

A. Attacks fulfilling criteria for migraine with aura
B. Aura characterized by both of the following: 1. fully reversible, monocular, positive and/or negative visual phenomena (scintillations, scotomata or blindness) confirmed during an attack by either or both of the following: – clinical visual field examination – the patient's drawing of a monocular field defect 2. at least two of the following: – spreading gradually over ≥5 minutes – symptoms last 5-60 minutes – accompanied, or followed within 60 minutes, by headache
C. Not better accounted for by another ICHD-3 diagnosis, while other causes of amaurosis fugax have been excluded.

Furthermore, occipital seizures must be excluded by continuous electroencephalographic monitoring. (7)

The Functional visual loss syndrome refers to a pathological entity in which the visual acuity of the patient is affected, but no structural anomalies can be found. This syndrome is also known as the nonorganic visual loss syndrome (NOVL). The incidence is 5-12% and affects more frequently female patients (63%) between ages of 11-20 years. The underlying causes remain unknown, but the onset has been linked to excessive exposure to computer screens or mental fatigue. No objective neurologic or ophthalmologic abnormalities are present. The visual acuity should be measured by testing the small symbols first, in order to exclude the organic disorders. When testing the visual field, some patients complain about the appearance of scotomata in the entire field. The visual field can have the appearance of a tunnel or spiral.(1,2,3)

The visual snow syndrome (the presented case) is the most peculiar functional disorder of all. It has been described very rarely worldwide. The patient suffers from continuous scintillating dots in both visual fields, which appear as snowflakes and persist in any light condition, on eye closure, giving the appearance of badly tuned TV image. Little is known, but PET-CT scans have shown that an increased metabolic activity can take place in the cerebellum and lingual gyrus.(9)

As mentioned above, the most common differential diagnoses to the functional visual disturbances are maculopathies (prematurity, diabetic or age-related types), optic neuropathy due to demyelinating diseases and various structural disorders of the occipital lobes. A multidisciplinary team of experts in both ophthalmology and neurology delivers the maximum efficiency of diagnosis, by which the ophthalmologist objectifies the retinal and anterior optic nerve damage, and then the neurologist searches for visual pathway and cerebral lesions. Thus, a systematic search for lesions in the involved system is the correct approach.(7,11)

CONCLUSIONS

The functional visual disturbances require the full effort of both ophthalmologists and neurologists in order to exclude structural lesions of the involved system.

Functional visual disturbances are diagnosed only when no structural abnormality is detected by complete ancillary examinations.

Functional visual disturbances have at date mostly ineffective symptomatic treatments, and the goal of this paper is to challenge the medical community to focus on studying rare disorders in order to increase the quality of life of these patients.

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