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CASE REPORTS

Changing Visual Defects in a Patient with Gilles de la Tourette Syndrome

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ABSTRACT

Gilles de la Tourette syndrome (GTS) is a complex disorder characterized by the presence of motor and vocal tics, as well as neuropsychiatric pathological features. Visual field defects have also been described in GTS patients by Enoch et al. in the 1980s. In the current paper, the authors discuss Enoch et al. studies showing visual field defects in patients with GTS, presenting a similar case evaluated in the context of newer structural and functional examination modalities.

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Gilles de la Tourette; visual field defects; hemifield visual evoked potentials

Introduction

Gilles de la Tourette syndrome (GTS) is a heritable movement disorder characterized by the simultaneous presence of multiple motor and at least one vocal tics for more than 12 months, with typical onset before age of 18. GTS is often associated with obsessive-compulsive and attention deficit hyperactivity disorders.[1]

Visual fields abnormalities associated with GTS were first described by J. Enoch et al. in *Neuro-Ophthalmology* in the 1980s.[2–4] They described step-like field defects not only in patients with GTS, but also in blood relatives of these patients. However, Whitefield et al.[5] reported no significant visual field changes between patients with GTS and controls, concluding that the visual field examinations were not useful as biological marker of this pathology.

We describe a case report of a patient with GTS and visual field defects, and show, for the first time, modern functional (Humphrey Visual Field (HVF), Hemifield Visual Evoked Potentials (VEP)), and structural (Optical Coherence Tomography (OCT)) examinations in such a patient.

Case report

We here report the case of a 24-year old female who was referred to our clinic by a local optometrist for a bilateral nasal hemianopia. This visual field test was part of a routine screening and the defect was not present the previous year. She was totally unaware of this problem.

The patient had been diagnosed with GTS and medically treated for 10 years. She was currently taking daily Atomoxetine 10 mg, Bupropion 150 mg, Fluvoxamine 100 mg, Guanfacine 1 mg, Haloperidol 1 mg, Modafinil 100 mg, Quetiapine 100 mg.

Our examination showed best corrected visual acuities of 20/40 right eye and 20/50 left eye. She read only 7/14 Ishihara plates with her right eye and 8/14 with her left eye. She had no afferent pupillary defect and her fundus examination was unremarkable.

A 30-2 HVF examination was performed (Figure 1a), and showed an almost complete bilateral nasal hemianopia. Mean deviation (MD) was -16.07 in the right eye and -10.57 in the left eye. Retinal nerve fibre layer (RNFL) and ganglion cell layer (GCL) were examined with Cirrus OCT machine (Carl-Zeiss, Meditech, Dublin, CA, USA) without showing any defects. Autofluorescence of the optic discs was negative for optic disc drusen and for abnormalities of the surrounding retina, including the macular region. A magnetic resonance imaging (MRI) of the brain was normal.

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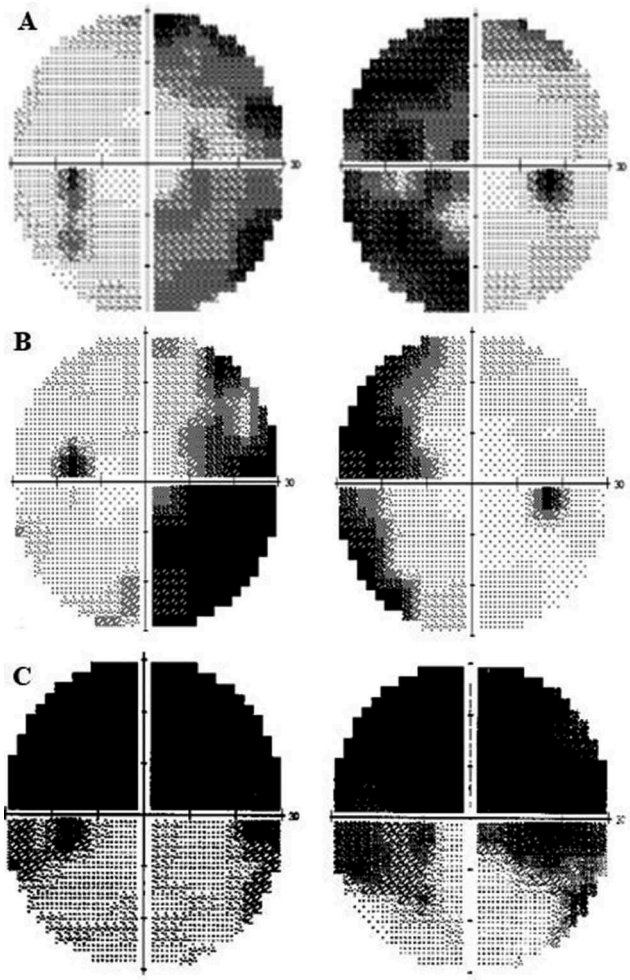

Figure 1. Gray-scale representation of HVF at first (A), second (B) and last examination (C).

One month later, she came back without reporting any changes in her condition. 30–2 HVF showed a reduction of the scotomas in the right eye with a change in the MD to -8.57 in the right eye and -16.38 in the left eye (Figure 1b). An Esterman binocular visual field was performed, showing no significant defects. Full-field and hemifield VEP with 0.8 and 0.24 checks degree size (Dyagnosys LLC, Lowell, MA, USA) were normal and showed no significant difference between nasal and temporal stimulation.

HVF was repeated 1 month later. The MD was -22.51 in the right eye and -20.01 in the left eye with scotomas denser in the superior quadrants, almost respecting the horizontal meridian (Figure 1c). All the data regarding visual acuity and visual field examination for each visit are summarized in Table 1.

Table 1. Overview of the best corrected visual acuity and visual field data (mean deviation, fixation losses, false positive, and negative rate) for each visit. N.A., not available.

	First visit		Second visit		Third visit	
	Right	Left	Right	Left	Right	Left
Visual acuity	20/40	20/50	N.A.	N.A.	20/50	20/40
Mean deviation	−16.07	−10.57	−8.57	−16.38	−22.51	−20.01
Fixation losses	2/15	5/14	2/15	3/16	2/14	0/14
False positive rate (%)	13	5	0	3	0	0
False negative rate (%)	29	17	11	0	0	22

Discussion

J. Enoch et al.[2–4] first described shifting and step-like visual field defects not only in patients with GTS, but also in their blood relatives. The abnormalities they noted fluctuated day by day, showing equal distribution between nasal and temporal steps.[6]

The pattern of visual field defect of our patient was similar and peculiar, respecting at first the vertical meridian with bilateral nasal scotomas (mildly improved at the second HVF), and showing a completely different pattern in the last examination, with bilateral superior altitudinal defects.

To better understand the pathophysiology of this singular behaviour, we combined, for the first time, modern structural and functional examinations. Importantly, we did not find any OCT changes in RNFL and GCL, nor any electrophysiological defects on VEP stimulating the abnormal binasal hemifield depicted in the HVF. Furthermore, high-quality MRI did not show any abnormalities. This suggests that the visual pathways were not compromised in this case of GTS. The pathophysiology of this problem remains uncertain in this complex syndrome. Insofar as the VEP did not corroborate the visual field defects, we cannot support a functional and transient impairment of the visual pathways as an explanation for the variable visual field defects described in this disease.

Enoch suggested that the ephemeral visual field defects may be caused by daily alterations in dopaminergic retinal neurotransmission.[6] This hypothesis is consistent with current understandings

about the role of dopamine (DA) in GTS and in retinal function.

The pathogenesis of GTS probably relates to multiple neurotransmitter systems dysfunction in the ganglia-thalamo-cortical network. The improvements of tics after administration of DA antagonist drugs suggests the main role of the DA pathway.[7] The most accepted hypothesis is that striatal DA receptor supersensitivity is the mechanism for tics.[8] DA has multiple roles in retinal function, such as light adaptation, ocular growth, and development. DA is released under the effect of light by amacrine and/or interplexiform cells.[9] The activity of DA-dependent retinal signalling can be assessed using photopic electroretinography (ERG). The light-adapted ERG responses of retina-specific deficient dopamine mice are reduced in contrast with the dark-adapted ERG responses, which appear normal.[10] Moreover, the light-adapted ERG signal of a normal retina is positively correlated with the spinal fluid dopamine concentration.[11] We did not perform an ERG because the first visual field defect was more suggestive of an optic pathway damage, which is more detectable by a VEP. In literature there is actually no available data regarding the study of GTS patients with ERG. It would be interesting to study a wide population of GTS patients with visual field defects by using multifocal ERG to try to correlate the site of the functional defect with the ERG response.

From a clinical perspective, the peculiar pattern of visual field changes in our patient is more indicative of an attention deficit rather than a structured retinal defect.

The current study would suggest, then, that DA receptor supersensitivity in the forebrain modulates visual attention rather than directly affecting the retinal-geniculate-cortical visual pathways.

This case report also serves as a reminder of the uncommon behaviour of GTS with respect to their visual field defects.

Declaration of interest

The authors declare no conflicts of interest.

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