A Case of Blepharospasm with Retinitis Pigmentosa

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ABSTRACT
Retinitis pigmentosa is a retinal dystrophy characterized with a progressive degeneration of photoreceptors that result with visual impairment. Blepharospasm is a primary focal dystonia presented with involuntary and persistent contraction of the orbicularis oculi muscle causing spasmodic closure of the eyelids. Most of the cases of blepharospasm have no other identifiable associated disease. We report here a patient with retinitis pigmentosa who developed severe blepharospasm.

Keywords: retinitis pigmentosa, blepharospasm, retina, focal dystonia

ÖZET
Retinitis pigmentoza fotoreseptörlerin ilerleyici dejenerasyonu ile gelişen ve görme bozukluğu ile seyreden bir retinal distrofidir. Blefarospazm orbikularis okuli kasında istemsiz kasılmalarla seyreden primer fokal distoni durumudur. Vak’aların çoğununda blefarospazm izole olarak ortaya çıkmakta, başka bir hastalığa eşlik etmemektedir. Bu yazida retinitis pigmentozaya ciddi blefarospazmın eşlik ettiği bir vak’a sunulmuştur.

Anahtar Kelimeler: retinitis pigmentoza, blefarospazm, retina, fokal distoni

INTRODUCTION
Retinitis pigmentosa is a retinal dystrophy that is characterized with a progressive degeneration of photoreceptors, and it is one of the major causes of untreatable blindness in young adults. Clinical characteristics include night blindness and visual field loss accompanied by bone spicule-like pigmentary deposition of the retina (Xia et al 2004, Oh et al 2003).

Blepharospasm is an involuntary and persistent contraction of the orbicularis oculi muscle that causes spasmodic closure of the eyelids, usually begins in mid-age (Misbahuddin et al 2002, Grandas et al 1988). Most cases of blepharospasm have no other identifiable associated disease, but it occasionally has been reported in patients with retinitis pigmentosa (Coppeto and Lessell 1990). We report here a patient with retinitis pigmentosa who developed severe blepharospasm.

CASE
A 54-years-old woman was admitted to our outpatient neurology clinic with intermittent bilateral eyelid closure as a result of involuntary contractions of the orbicularis oculi muscles. Also she suffered visual loss and she was diagnosed retinitis pigmentosa at 13-years-old. On ophthalmic examination visual acuity was recorded positive light perception and projection. Slit-lamp examination revealed bilateral posterior subcapsular cataract. Intraocular pressure was 16 mmHg at the right side and 18 mmHg at the left side. Binocular and indirect ophthalmoscopy of eyes showed narrowed retinal vessels, depigmentation of the retinal pigment epithelium, intra-retinal bone spicule pigmentation and waxy pallor of the optic discs which were characteristically for retinitis pigmentosa. Her neurological examination was normal except for the visual loss and blepharospasm.

Surface electromyography showed tonic discharges in the orbicularis oculi muscles when she attempted to open her eyes. The blink reflex was studied and the supra-orbital nerve was electrically stimulated with supra-maximal intensity with 0.2 msec duration and normal latency values of R1, R2 and R2 consensual obtained bilaterally. Her liver, renal and thyroid function tests and hematological findings were normal.

There was no achantocytosis. Serum lactate, pyruvate, ceruloplasmine and betalipoprotein levels were within normal values. Magnetic resonance imaging of the brain was normal.

Patient was treated with botulinum toxin A for blepharospasm, and she obtained good relief.
DISCUSSION

In this report we present a patient with retinitis pigmentosa who developed severe blepharospasm. Most cases of blepharospasm have no other identifiable associated disease. In the literature there is only one report describing retinitis pigmentosa associated with blepharospasm. Coppeto and Lessell (1990) reported 2 siblings with a syndrome of pigmented retinopathy, blepharospasm, dystonia. Although these findings suggested Hallervorden-Spatz disease they described that as a distinct disorder.

Retinitis pigmentosa is a progressive retinal dystrophy resulting in severe visual impairment (Xia et al. 2004). Many neurodegenerative diseases are associated with abnormal retinal pigmentation. These are familial disorders, and sporadic or mitochondrial syndromes. Our case had no family history, also no other clinical signs and additional laboratory findings, so familial disorders such as Hallervorden-Spatz disease, abetalipoproteinemia, ceruloplasmin deficiency were excluded (Coppeto and Lessell 1990, Miyajima et al 1987). Diffuse cerebral sclerosis with pigmented retinopathy, Rud’s syndrome and Tuck-McLeod syndrome are sporadic disorders and these were excluded because our patient’s had no clinical and laboratory findings resembling these syndromes (Kayden et al 1973, Marxmiller et al 1985, Tuck and McLeod 1983). Pigmentary retinopathy sometimes occurs in mitochondrial cytopathies such as Kearns-Sayre syndrome, MERRF (myoclonus epilepsy with ragged-red fibers) or MELAS (mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes). But our patient had no ophthalmoplegia, deafness, myoclonic epilepsy, encephalopathy, stroke-like episodes, and lactic acidosis (Mullie et al 1985, DiMauro et al 1987).

Blepharospasm is a disorder characterized by bilateral episodic contractions of the orbicularis oculi muscle that cause spasmodic closure of the eyelids (Grandas et al 1988, Kerrison et al 2003). Most cases of blepharospasm have no other identifiable associated disease. Blepharospasm can occasionally associate with degenerative extrapyramidal disorders such as Idiopathic Parkinson’s disease (PD), progressive supranuclear palsy (PSP), multi-system atrophy (MSA) and cortico-basal degeneration (CBD) (Janati et al 1989, Rivest et al 1990, Gibb et al 1989, Boesch et al 2002). But it has been reported that blepharospasm develop in the advanced stage of the extrapyramidal disorder (Janati et al 1989). Our patient had no clinical signs and radiological findings of PD, PSP, MSA and CBD. So we think that blepharospasm is a distinct finding other than neurodegenerative disorder.

Coppeto and Lessell (1990) reported 2 siblings with a syndrome of pigmented retinopathy, blepharospasm, and dystonia resembling to Hallervorden-Spatz disease. But unlike those with Hallervorden-Spatz disease, their patients had not a fulminant course, and one of the patients’ illness began in late teens rather than in childhood. Also Newell et al (1979) reported that cases of Hallervorden-Spatz disease with pigmented retinopathy have an earlier onset and more fulminant course than cases without retinopathy.

In this report we presented a patient with retinitis pigmentosa who developed severe blepharospasm. In our case retinitis pigmentosa developed at age 13, blepharospasm developed at age 45. We think that retinitis pigmentosa and blepharospasm represents an independent entity. But this entity may be distinct disorder without identifiable neuroimaging and biochemical abnormalities.

REFERENCES


