Steinert myotonic dystrophy – a multisystemic disorder with occular implication

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Abstract

The Steinert Myotonic Dystrophy is the most common systemic disease in adults with dominant autosomal transmission. We present two patients, who were hospitalized in the 2nd Clinic of Ophthalmology, at the "Prof. Nicolae Oblu" Emergency Hospital of Iasi, with the diagnosis of Pathological Cataract and Steinert Myotonic Dystrophy.

Key words: Steinert myotonic dystrophy, pathologic cataract, palpebral ptosis.

Introduction

Myotonic Dystrophy is the most common dystrophy in adults, it is an autosomal dominant disease with two distinct genetic types: type 1 – the classical form described by Steinert and type 2 identified by Ricker (1). The Steinert disease or type 1 myotonic dystrophy is a multisystem genetic disease that affects muscles (delayed muscle relaxation), nervous system, eyes and heart (2). The main ocular manifestations of the disease are: pathological cataract, myogenic palpebral ptosis (3) and reticular retinopathy. Cataract associated with Steinert disease have a pathognomonic aspect: the crystalline lens cortex shows polychromatic iridescent crystals formed from lens fiber plasmolemma (3).

We present two clinical cases: A.A., aged 51 years, A.E., 58, siblings diagnosed with Pathological Cataract Bilateral, Myogenic Ptosis grade 2-3 and Steinert Myotonic Dystrophy.

Case 1

Patient A.A., female, 51 years, from Iasi, comes for the symptoms: L.E. gradual decrease of visual acuity and palpebral ptosis. Medical history: R.E. operated pathological cataract (10 years ago), O.U. Myogenic Bilateral Ptosis, Steinert Myotonic Dystrophy. Local ocular examination: O.U. grade 2-3 ptosis (Figure 1). Biomicroscopy examination highlights: R.E. pseudophakia, L.E. anterior and posterior cortical and nuclear subcapsular opacities; nucleus sclerosis +3; crystalline with a pathognomonic aspect, iridescent crystals in the cortex and nucleus.

V.A.R.E. 1/500, V.A.L.E. 3/500, Ophthalmoscopic examination: R.E. - pale papilla, narrowed vessels with central emergence, macula with pigment dispersion; L.E. cannot be examined, due to lens opacification. P.R.E.=18 mmHg; P.L.E.=17 mmHg. M.R.I. Exam shows cerebral atrophy (Figure 2).

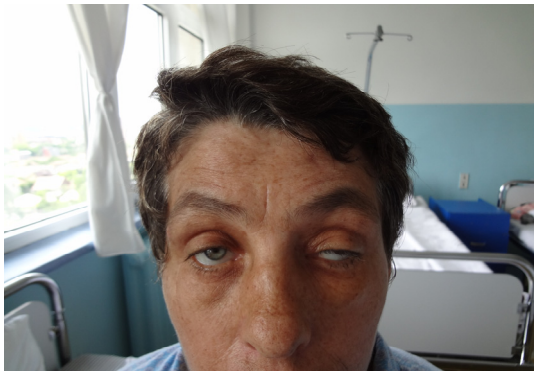


Figure 1
Patient A.A., 51 years, with Steinert Myotonic Dystrophy

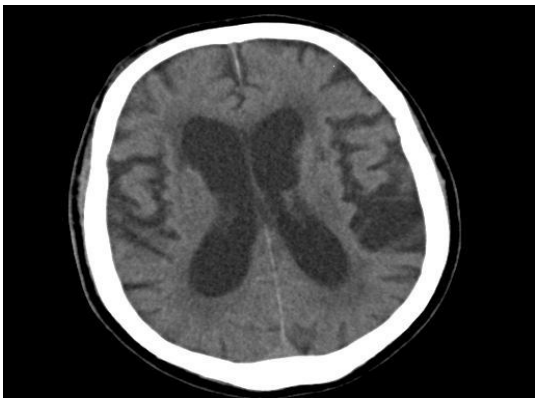
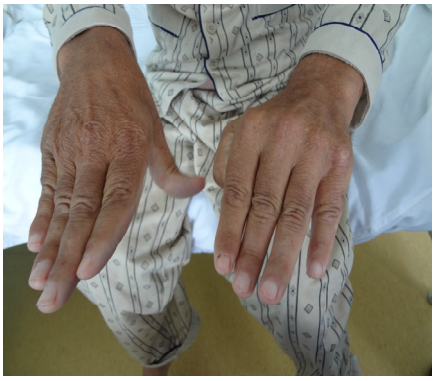


Figure 2
Patient A.A., 51 years, M.R.I. imagines shows cerebral atrophy

A



B

Figure 3 A, B
Patient A.E., 58 years, with Steinert Myotonic Dystrophy

Neurological examination shows motor disorders with central and peripheral causes. The biomicroscopy of the cataract indicates a favourable diagnosis (presence of iridescent crystals in the lens nucleus and cortex) and pathological neurological and neurosurgical associations.

Differential Diagnosis: is made in relation to other pathological cataracts (diabetic, chronic eye inflammation, cortisone), senile cataract, traumatic cataract, persistent primary vitreous and Coats disease.

Treatment

Surgery was performed at LE: extracapsular lens extraction and implantation of the intraocular lens in the anterior chamber.

Postoperative evolution with appropriate medical treatment was favourable.

The evolution was favourable after surgery: the corneal edema resolved, the inflammatory syndrome healed, V.A.L.E. 1/8 without correction.

In the absence of surgical treatment, the prognosis is reserved, the disease progressing to peripheral blindness.

Case 2

Patient A.E. 58 years, male, from Iasi, presents the same clinical picture as in the previous case. The medical history shows: R.E. operated pathological cataract (10 years ago), O.U. Myogenic Bilateral Ptosis, Steinert Myotonic Dystrophy. Local ocular examination: O.U. Palpebral Ptosis grade 2-3. The biomicroscopy highlights: R.E. Pseudophakia, L.E. full nucleus and cortex opacification, +3 nucleus sclerosis; polychromatic crystals are found in the lens nucleus and cortex (Figure 3 A, B).

V.A.R.E. 1/500, V.A.L.E. less than 1/500, P.R.E.=17 mmHg; P.L.E.=16 mmHg. Ophthalmoscopic examination R.E.: net shape of optic nerve papilla, temporally pale, central emergence of narrow vessels macula with pigment dispersion;L.E. cannot be examined due to lens opacification.

The internal medicine examination establishes the following diagnoses: Chronic Alcoholism, Chronic Ischemic Cardiomyopathy, Thrombocytopenia. M.R.I. Exam shows cerebral atrophy (Figure 4).

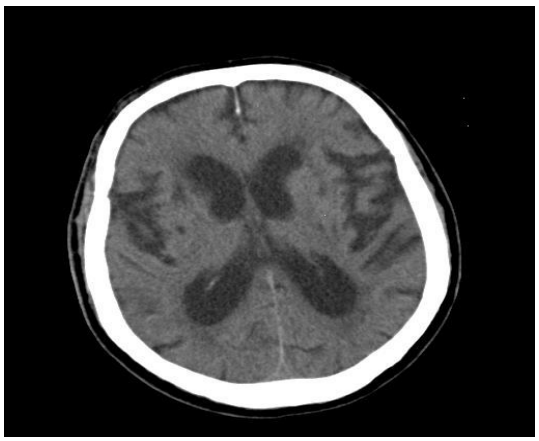


Figure 4
Patient A.E., 58 years M.R.I. imagines shows cerebral atrophy

The neurological examination shows motor disorders with central and peripheral causes.The favourable diagnosis that emerges is: L.E. Pathological Cataract, R.E. Pseudophakia, O.U. Bilateral Myogenic Ptosis grade 2-3, Steinert Myotonic Dystrophy, Chronic Alcoholism, Chronic Ischemic Cardiomyopathy, Thrombocytopenia. The diagnosis of Pathological Cataract is supported by the muscle dystrophy and the particular aspect of the crystalline (presence of iridescent crystals in the lens nucleus and cortex).

Differential Diagnosis: is made in relation to other pathological cataracts (diabetic, chronic eye inflammation, cortisone), senile cataract, traumatic cataract, persistent primary vitreous and Coats disease.

Treatment

Surgery was performed on L.E.: the phacoemulsification of the lens and implantation the intraocular lens in the posterior chamber with no intraoperative complications.The postoperative evolution under medical treatment was favorable.

Evolution after surgery was favorable: inflammatory syndrome was completely resolved and V.A. at L.E. 1/3 without correction.The prognosis under surgical treatment is favorable.

In the absence of surgical treatment, the prognosis is reserved, the disease progressing to peripheral blindness.

Case particularity

In the first case of pathological cataract due to the anterior and posterior capsule adhesion to the lens nucleus and cortex, a 6-mm wide lens sclerocorneal incision was performed, with extracapsular lens extraction and pseudophak implantation in the anterior chamber.

The second case was solved by the method of phacoemulsification with clear corneal incision of 2.7 mm and pseudofak implantation in the posterior chamber. In both cases the cortex and nucleus were very dense and could cause posterior capsule rupture during surgery, resulting in the release of the vitreous and developing secondary glaucoma. In both cases the posterior capsule was very fragile presenting numerous cracks, and the intraocular lens was implanted in the anterior chamber.

Discussions

Eyes disorders in Steinert disease are common and are characterized mainly by pathological cataract. Other possible ocular manifestations are: myogenic ptosis, ocular hypotony due to ciliary body detachment (4) and reticular dystrophy of the retinal pigment epithelium, responsible for variable decrease in visual acuity (5). Gjertsen et al. report three cases diagnosed with myotonic dystrophy which developed secondary recurrent opacification of the posterior capsule after cataract surgery. These patients undergo an increased risk of posterior capsule opacification and intraocular fibrosis after cataract surgery (6). In the literature, for myogenic ptosis the treatment recommended is blepharoplasty or eyelid levator resection or frontalis muscle suspension.

The eye disorder in our cases was pathological cataract and myogenic palpebral ptosis grade 2-3. Patients showed no significant ocular hypotony or retinal changes. We only solved surgically the pathological cataract and later remained to operate ptosis by upper eyelid levator resection. After operation there was no posterior capsule opacification.

Conclusions

Patients diagnosed with pathological cataract and Steinert Myotonic Dystrophy are advised to undergo cataract surgery, when there is no severe impairment of respiratory and cardiovascular systems and the surgical technique must be adapted to each case to minimize intraoperative and postoperative complications. Myogenic palpebral ptosis is recommended to be solved at a later stage. The long-term vision quality will depend on the progression of neurological damage, which adds to the retinal changes and brain damage (cerebral atrophy).

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