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Unilateral retinitis pigmentosa. Medical case

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ABSTRACT

Retinitis pigmentosa almost in all cases is revealed in bilateral form. Unilateral retinitis pigmentosa is considered to be an extremely rear pathology and is poorly studied. Unilateral retinitis pigmentosa is proposed to be the result of a somatic mutation, having happened during embryogenesis. It is presented medical case of unilateral retinitis pigmentosa in our practice. Female have been examined since 2006. On the right eye was diagnosed retinitis pigmentosa. On contralateral eye, we did not examine any pathologic changes.

Diagnosis of unilateral retinitis pigmentosa is not taken into consideration that can lead to improper treatment of a patient. Determination of unilateral retinitis pigmentosa requires exclusion of infectious nature of the disease, as well as absence of “interest” of a contralateral eye. Electroretinogram testing is a crucial method in diagnostics of the form of disease.

Using classic common treatment of retinitis pigmentosa during the following 3 years, we kept functions of the right eye at the previous level.

1. Introduction

Retinitis pigmentosa is a group of genetic degenerative inherited disorders of the retina, characterized by progressive photoreceptors damage. Progressive retinal degeneration almost in all cases is revealed in bilateral form and very seldom diseases one eye.

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Defining unilateral retinitis pigmentosa, one should consider the following criteria: exclusion of infectious etiology of the disease (syphilis, rubella, virus retinitis, onchocerciasis), presence of classic symptoms on the diseased eye, and absence of them on the other one (Krill and Iser 1959). For correct diagnosis of retinitis pigmentosa one should pay much attention to electroretinogram and electrooculogram testing (Spadea et al. 1998). Underrepresentation of the disease forces researchers to consider if the pathology is a separate disease or it is just an aborted form of progressive retinal degeneration (Joseph 1951). In literature, there are description of the case histories of the disease, but frequency of such cases is very low (Carr and Siegel 1973; Kolb and Galloway 1964). According to data of Joseph (1951) from 1865 to 1949 there were described only 45 cases, Kolb and Galloway (1964) in 1965 mentioned 3 cases, Spadea et al. (1998) in 1998 marked 4 cases.

Autosomal recessive, autosomal dominant and x-linked inheritance patterns of retinitis pigmentosa is deeply studied. Unilateral retinitis pigmentosa is proposed to be the result of a somatic mutation, having happened during embryogenesis. As a result, some percentage of cells in a patient's body carry gene mutation, with the potential to cause retinitis pigmentosa on one eye. If such mutation is present in a group of cells, which are later developed into cells of retina, retinitis pigmentosa can happen on one eye, whereas the condition of the other eye is normal. In case when mutation causing retinitis pigmentosa is located in the part of an embryo, which is determined to become, for example, skeletal muscles or bone tissue, reveal of the mutation is "silent". It is quite possible that the type of clinical "silent" mutation occurs rather often. In literature on dermatology, one can read about many cases of mutation, being revealed on separate segments of skin, whereas other part of skin is absolutely normal and does not demonstrate any mutation. In case of retinitis pigmentosa, somatic mutation can cause atypical or unilateral forms of the diseases only in case when the disease-producing mutations happen in the part of an embryo, determined to become an eye (Jordan et al. 2008).

2. Results

We would like to present medical case of unilateral retinitis pigmentosa in our practice.

Female have been examined since 2006. For the first time she came to the Institute with complaints of central visual loss of the right eye. She was diagnosed to suffer from weak myopia of both eyes. Also neuroretinitis, secondary wet macular degeneration and partial atrophy of optic nerve of the right eye were diagnosed.

At the moment of admission to the hospital, central visual acuity (CVA) of the right eye was 20/40 and could not be corrected, of the left eye it was 20/20. Visual field (VF) of the right eye was narrowed from temporal side by 10 degrees, from nasal side by 40 degrees, VF of the left eye was normal. According to the data of optical coherent tomography (OCT), we observed macular swelling up to 347 μm in the central sector. Biomicroscopy demonstrated that the anterior segment of both eyes were without pathology. Ophthalmoscopy of the right eye revealed that optic disc was pale, boundaries were obscure, swelling, arteries of normal caliber, veins were considerably broader, foveal reflex was absent, there were swelling

in macular zone. One saw dystrophic transformations in periphery. In case of ophthalmoscopy of the left eye, eye fundus was normal.

Specialists' examination excluded syphilis, rubella, tuberculosis. Consultation of otolaryngologist, dentist, neuropathist did not detected any pathology. X-ray of lungs was without pathology.

The patient was treated with a course of intensive anti-inflammatory, dissolving therapy, as well as urinative medicines. After the course of treatment, CVA of the right eye increased to 20/25. Swelling of optic disc and macular area decreased (according to the data of OCT to 280 μm in the central sector). Conditions of visual functions and eye fundus of the left eye stayed without pathological changes.

One year later the patient was admitted for the second time, complaining on decrease of CVA of her right eye (20/32). At the moment of admission to the hospital, signs of inflammation were not observed, no swelling of optic disc. Nevertheless, there were signs of progressive atrophy of optic nerve (narrowing of visual field, growth of threshold of electric sensitivity to phosphine, decoloring of optic disc), as well as dystrophic transformations in macular area and cystoid macular edema. The patient was treated with a course of trophic, nootropic and antioxidant as well as vascular therapy, resulting in the increase of CVA to 20/20 of the right eye.

Eight months later examination determined that CVA decreased to 20/25. Signs of inflammation were absent, but cystoid macular edema did not disappear. It was decided to use panmacular laser coagulation on a base of trophic, anti-inflammatory and dissolving therapy. As a result of the treatment, CVA increased to 20/20, and cystoid macular edema decreased.

While examining 4 months later, signs of inflammation were absent, atrophy of optic nerve and macular swelling prevailed. Trophic therapy was carried, but in spite of improvement of visual functions, cystoid macular edema left at the same level.

Refractory cystoid macular edema, high CVA and rapid narrowed field of VF were particular for the next two years. At the same time, pathological changes of the left eye were not observed.

In 2010, next visit of the patient and ophthalmoscopy of peripheral retina of the right eye detected deposit of a pigment by a bone spicule-like changes. They guessed that the patient suffered from unilateral retinitis pigmentosa. It is known, that a similar ophthalmologic picture, imitating retinitis pigmentosa, can be observed in some infectious chorioretinitis. Nevertheless, infectious cause of the mentioned pathological changes was excluded because of the following factors. Staying in hospital, the patient submitted analyses on syphilis (RW was always negative, PCR -negative). The patient had not suffered from rubella, proved by laboratory analyses. She was also under observation from the moment of the first episode of uveitis and clinical event was not particular for syphilis neuroretinitis and onchocercosis. Dystrophic processes were on the foreground in the clinical picture.

To prove retinitis pigmentosa we made electro-physiological examinations and darkness adaptation as well as early made investigation (VF, phosphine diagnostics, OCT). Data of electroretinogram demonstrated lowering of waves range. Darkness adaptation decreased. VF was concentrically narrowed to: 5 degrees from a nasal side, 30 degrees from a temporal side, an upper limit – to 10, a lower limit – to 20 degrees. CVA was 20/32. According to data of OCT, a cystoid macular edema made 438 μm. One should mention, that retinitis pigmentosa is classically complicated by refractory cystoid macular edema (Ikeda et al. 2012;

Rajarshi et al. 2011; Scorolli et al. 2007). At the same time, the condition of the right eye was absolutely normal, and physiological examination did not found out any pathological changes.

Thus, the patient got a final diagnosis of unilateral retinitis pigmentosa.

Using classic common treatment of retinitis pigmentosa during the following 3 years, we kept functions of the right eye at the previous level. However, one noted persistence of cystoid macular edema, refracting to all kinds of treatment. On contralateral eye, we did not examine any pathologic changes.

3. Conclusions

Unilateral retinitis pigmentosa is considered to be an extremely rear pathology and is poorly studied. In most cases, signs of unilateral reveal of retinitis pigmentosa force diagnosis, directed to search infectious nature of the disease. Diagnosis of unilateral retinitis pigmentosa is not taken into consideration that can lead to improper treatment of a patient. Determination of unilateral retinitis pigmentosa requires exclusion of infectious nature of the disease (syphilis, rubella onchocercosis), as well as absence of “interest” of a contralateral eye. Electroretinogram testing is a crucial method in diagnostics of the form of disease.

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