УДК:616-089:617.741:575-053.2

CLINICS AND SURGICAL TREATMENT OF OCULAR MANIFESTATIONS OF MARFAN SYNDROME AMONG JUNIORS

Toshpulatova A.Z., Zakirhodjaeva D,A., Kariev A.V., Tadjieva D.Z.
Tashkent Pediatric Medical Institute

Resume

The most common symptoms of the disease in Marfan Syndrome are the luxation of lens (46.16%) and cataract (26.92%), which are complicated by the progression of secondary glaucoma, lenticular myopia and strabismus. The above-mentioned information indicates the need to improve the traditional methods of surgery on juniors with ocular manifestations of Marfan syndrome in order to reduce the risk of intra - and postoperative complications, and the widespread introduction of different models of the intraocular lens in recent years, greatly expanded the possibilities of surgical rehabilitation of patients with dislocation of the lens.

Key words: Marfan syndrome, ocular clinical signs, lens luxation, intraocular lens.

БОЛАЛАРДА МАРФАН СИНДРОМИДА КЎЗДАГИ КЛИНИК БЕЛГИЛАР ВА ЖАРРОҲЛИК ДАВОЛАШ УСУЛИ

Тошпулатова А.З., Закирходжаева Д.А., Қориев А.В., Таджиева Д.З.
Тошкент педиатрия тиббиёт институти

Резюме

Марфан касаллигининг кўзда энг кўп учрайдиган белгилари (46,16%) гавҳар люксацияси ва (26,9%) катаракта бўлиб, касаллик авж олганда иккиламчи глаукома, гавҳар миопияси ва гилайлик каби асоратларга олиб келади. Келтирилган маълумотлар болаларда Марфан синдромида кўздаги ўзгаришларни операция вақтида ва операциядан кейинги асоратларни келиб чиқиш хавфини камайтириш мақсадида хирургик даволашнинг анъанавий усулларини такомиллаштириш зарурлигини таъкидлайди. Охирги йилларда интраокуляр линзаларнинг ҳар хил моделларини кенг қўллаш натижасида гавҳар дислокацияси бўлган беморларнинг хирургик реабилитацияси имкониятлари сезиларли кенгайди.

Калит сўзлар: Марфан синдроми, кўздаги ўзгаришлар, гавҳар люксацияси, интраокуляр линза.

КЛИНИКА И ХИРУРГИЧЕСКОЕ ЛЕЧЕНИЕ ГЛАЗНЫХ ПРОЯВЛЕНИЙ СИНДРОМА МАРФАНА У ДЕТЕЙ

Тошпулатова А.З., Закирходжаева Д.А., Қориев А.В., Таджиева Д.З.
Ташкентский педиатрический медицинский институт

Резюме

Наиболее часто встречающимися симптомами болезни Марфана является люксация хрусталика (46,16%) и катаракта (26,92%), которые при прогрессировании осложняются вторичной глаукомой, хрусталиковой миопией и косоглазием. Приведенная информация свидетельствует о необходимости усовершенствования традиционных методов хирургических вмешательств у детей с глазными проявлениями синдромом Марфана с целью уменьшения риска возникновения интра - и послеоперационных осложнений.

Широкое внедрение различных моделей интраокулярной линзы в последние годы, значительно расширили возможности хирургической реабилитации больных с дислокацией хрусталика.

Ключевые слова: синдром Марфана, глазные проявления, люксация хрусталика, интраокулярная линза.

Relevance

Among all hereditary connective tissue diseases of greatest interest to pediatricians and general practitioners is Marfan syndrome, as life expectancy of these patients is limited to 30-40 years [3,4], and one patient may experience so many problems with health despite many specialists in the clinic. Since the disease is known to be a serious prognosis for life and disability of patients, diagnosis imposes a special responsibility on the doctor at the first meeting with the patient.

Complexity of diagnosis of Marfan syndrome (MS) is an actual problem of clinical medicine. Every doctor in the practice who had to deal with patients with CM, faces difficulties in establishing the correct clinical diagnosis, which often leads to the appointment of inadequate treatment [2,5].

The disease with autosomal dominant inheritance, which refers to a group of hereditary fibrilinopaty, an inherited disease of the connective tissue with changes in the skeleton, eyes and cardiovascular system [9].

At the heart of the MS is the accumulation of soluble fraction (immature) collagen with its decay to metabolites containing hydroxyproline, as well as a mutation in the gene for fibrillin-1 (fibrillin-1 - FBN1). Fibrillin - is glycoprotein, the basis of elastic fibers of the connective tissue. It is located in the extracellular matrix, cartilage, vascular walls, lens, etc. Found many different mutations FBN1, which explains the significant clinical polymorphism of the disease. More than 15% of the time - a consequence of new mutations. A typical form of MS can be caused by mutations in other genes, such as in a gene that encodes a receptor for type II transforming growth factor-β [4, 12].

ISSN 2181-1954. EISSN 2181-1962

For the MS characteristic variability of phenotypic manifestations are - cardiovascular, ocular, musculoskeletal and central nervous system [11].

One of the characteristics of Marfan's disease pathology is part of the vision. According to the literature of the vision loss, this results in 50-80% of patients, often being one of the earliest signs of the disease [6]. The early signs of Marfan syndrome include embryotoxon, iris hypoplasia, especially the pigment border [4, 9].

In the future, there is destruction of the vitreous body, the destruction of fibers ciliary zone and therefore ectopic lens. Ophthalmologic manifestations diagnosed in almost all patients Marfan syndrome, regardless of age [3, 10]. However, there is still no consensus on the timing of the appearance of ophthalmologists particular change on the part of the organ of vision in patients with Marfan syndrome.

Functional disparity of many systems development incompatible with life events is the basis for the study of this disease by physicians of various specialties. The disease manifests itself in childhood, most patients - young people. Disease often leads to early disability and death [3, 6, 10, 11].

So, given all of the above object of the present study was to investigate the characteristics of clinical manifestations and evaluation luxation lens surgery in children with Marfan syndrome.

Materials and Methods

A retrospective analysis of the material held in the period from 2008 to 2018 (10 years) in the Ophthalmology Clinic TashPMI. There were analyzed and viewed 2120 records of patients with diseases of the eye (myopia, lens shift).

All children were held a standard eye examination.

To etiologic diagnosis were used genetic methods of the Republican screening center. The diagnosis was verified by clinical and instrumental (radiography of the musculoskeletal system, an ultrasound of the heart) techniques involving an otolaryngologist, a neurologist, cardiologist and trauma.

Research results: According to a retrospective analysis of the data outpatients and histories we have found that 2.5% (26 patients) was diagnosed Marfan syndrome. Age was 6,37 ± 2,14 years.

Among children with Marfan boys dominated, accounting for 62.8%, while the girls made up 37.2%.

When anthropometric examination, all had significant anthropometric changes: correlation brush / growth> 11%, the distance from the pubis to the floor more than half of the increase in the average of 4,9 ± 0,1 cm, limited extension of the elbow joints, arachnodactyly, high-arched palate, a positive test thumb, a positive test wrist.

In 19 patients with arm span was more growth by 5 cm, the ratio of stop / growth> 15%, the length of the index finger of> 10 cm, chest deformity.

There were also changes in the cardiovascular system: expanding the boundaries of the relative cardiac dullness, systolic murmur at the apex, and a 5 point, muted tones of the heart, the tendency to hypotension, ECG changes.

In this case, the majority of patients had the presence of false angina, shortness of breath, the appearance of fatigue during exercise. Cardiac assessment revealed aortic valve replacement in 10 patients, mitral valve prolapse - a 6, the expansion of the aorta - a 5-minute, symptoms of regurgitation - in 3 patients.

With age, these changes tend to progress.

The defeat of the vision recorded in all patients as lyuksatsii lens (46.2%), and complicated cataract (23.1%), which were complicated by the progression of secondary glaucoma (30.8%), myopia and strabismus (26.9%). Refractive error was significantly increased in high school.

Dry eye is caused by disorders of the water and the mucin layer of the tear film was observed in 25 patients out of 26.

Biomicroscopy showed that bilateral lyuksatsiya transparent lens available in 9 (34.62%) patients, and in 7 (26.92%) of which the lens was pulled up, in 2 (7.69%) cases aside.

Lyuksatsiya lens accompanied by a decrease in visual acuity (0.1 - 0.4). In this case, the majority of patients had failure of the lower part of Zinn ligament, which was stretched, had a fringed edge with numerous gaps.

In 3 cases (11.54%) observed a unilateral lyuksatsiyu lens. In 5 (19.23%) eyes had lyuksirovan lens in the anterior chamber, in 3 eyes (11.54%) - in the back. One patient (3.85%), the lens was placed in the horizontal position of the pupil and always floated between the cameras eye.

Sublyuksatsiya lens was diagnosed in 5 patients (19.23%) of the patients and had a one-sided.

Cataract of various degrees against lyuksatsii lens was detected at 7 (26.92%) eyes and opacities characterized by cortical and subcortical rear lens layers. In this case, visual acuity was significantly reduced, making up 0.04 - 0.07.

2 patients - 3 eyes (11.54%) - have been subjected to surgical treatment, in connection with what they saw aphakia. According to parents, lack of posterior capsule intraocular correction was not made.

One patient (3.85%) was observed expressed gross destruction of the vitreous. Process was one-sided and accompanied by a significant decrease in visual acuity (0.09).

Refractometry showed the presence of high myopia in 3 (11.54%) eyes with lyuksatsiey lens in the posterior chamber and compound myopic astigmatism in one (3.85%). It is characteristic for myopia fundus changes were found, indicating the nature of the lenticular process.

However, despite careful correction, visual acuity does not exceed 0.3. Along with this, one patient (3.85%) were diagnosed with a high degree of hyperopia with the presence of a complex hyperopic astigmatism. Ophthalmoscopy with showed the presence of characteristic changes in the hyperopic eye bottom.

When tanometrii Maklokovu to have 8 patients (30.8%) had a diagnosis of secondary glaucoma, 16 eyes examined. End-stage secondary gloukomy observed in 12.5% of cases. Scleral staphyloma was observed in 12.5% of children. Complicated cataract was found in 3 patients (37.5%). Ultrasound study of children with secondary gloukomoy was found next pathology: floating opacities in the vitreous body (12.5%), destruction of the vitreous body (87.5%) and in 25% of the rear detachment of the hyaloid membrane of the vitreous, 37, 5% of children without ehopatologii.

In the analysis of anamnesis and physical examination we have found that the reasons for the development of secondary gloukomy in children with Marfan syndrome is primarily dependent on the anatomical changes that are characterized by stretching ligaments Qing lens, as well as changes in the vitreous body in the form of destruction and posterior hyaloid detachment membrane.

In addition, the 7 patients (26.92%) were diagnosed with

ISSN 2181-1954. EISSN 2181-1962

exotropia, and given the availability of anisometropia, was secondary.

These ophthalmoscopy showed a hypoplasia of the optic nerve (optic disk) and macula in 3 patients (11.54%), and partial atrophy of the optic disc in 2 (7.69%). This group of patients prevailed changes in the cardiovascular system.

However, our studies have shown that in 7 patients (26.92%) change of the vision were the only sign of Marfan's disease, except for genetic research.

Of the 26 children with Marfan syndrome in 13 patients were performed surgeries are lensoektomiya in 13, basal iridectomy - in 11 and partial vitriektomiya in 9 patients.

The downside transtsiliarnoy lensektomii sublyuksirovannyh lenses is that at the approach to the lens through the flat part of the ciliary body revealed anterior limiting membrane of the vitreous and capsular bag from the back of the capsule and in the removal of the lens of its mass through a defect posterior capsule and the anterior border of the membrane into the cavity vitreous body, resulting in the need to increase - in 10-40% of cases, the amount of produced vitrectomy [1, 7].

Hit lenticular masses in the vitreous body during surgery, marked by several authors [1, 7], 1,4-11,3% of the cases is the starting point of fibroplasticheskih processes in the vitreous body, which then becomes the cause of traction retinal detachment. Surgeon attempts to remove trapped in the vitreous lenticular masses also lead to an unjustified increase in the volume and duration of vitrectomy surgery [7], which led to the development of edema - in 5% of cases and EED cornea - by 4.1% due to endothelial damage in the front lyuksirovannym the camera lens.

Total or subtotal vitrectomy significantly increases the risk of retinal detachment, which is especially dangerous in the eyes of patients with Marfan syndrome, initially predisposed to the development of rhegmatogenous retinal detachment. [8] In general, according to the literature [7], the development of retinal detachment after lens removal sublyuksirovannyh noted in 1,8-33,3% of cases, in our own observations, this percentage was 6.5%.

Frequently (12%) occurred in the iris damage during surgery. In our opinion, this is due to the fact that when one of his lens sublyuksatsii pole is visible in the pupil, while the opposite, in the area of the greatest displacement, usually upward, covered the front of the iris. Even maximizing pupil at bias lens II-III degree it possible to see the pole in the area of maximum lens shift it. Removal of the lens which is under the iris, is «blind», which is particularly inconvenient because intervention is also usually set at the top. Place the tip of the lower vitreotoma meridians prevent bony wall of the orbit, which also highlights other authors [8].

Iris damage contributed to the development of bleeding during surgery in 15.4% of cases, and the postoperative period - by 7.6%. Must take into account the rigidity of the pupil in Marfan's syndrome caused by congenital structural abnormalities of the iris, which does not allow to reach the maximum mydriasis. Violation of the shape, size and location of the pupil subsequently significantly reduces cosmetic result of the operation and in 3 cases was the cause of reduced vision.

One of the major complications of surgery is incomplete removal and lenticular masses that cause postoperative inflammation membranes of the eye, which in 3 patients led to the development fakogennogo cyclites and later according to some researchers may cause complications such as pupillary block, the secondary hypertension - 3.5% - 12.7%, and the development of exudative vitreous opacities - 24.3%, and macular edema - 5.4%, which is a cause of serious, sometimes irreversible visual loss [8, 10].

Conclusion

The information demonstrates the need for knowledge of the major clinical manifestations of Marfan syndrome in children by physicians of all specialties, not to take the treatment of ex juvantibus (and thus - delay setting correct clinical diagnosis, leaving no chance to save the patient's life), and the need to improve the provision of health With this group of patients. Also need to improve the traditional methods of surgery in children with ocular manifestations of Marfan's syndrome in order to reduce the risk of intra - and postoperative complications, and the widespread introduction of different models of intraocular lens, in recent years, greatly expanded the possibilities of surgical rehabilitation of patients with dislocation of the lens.

REFERENCES:

1. Ahmed Samir and Ahmed Moemen Gad. Sulcus Suture Fixation of a Foldable IOL over Preserved Capsule for Managing Lens Subluxation in Marfan's syndrome // J Clinic Experiment Ophthalmol. – 2011. - №2. – P. 4
2. Vatutin NT, Sklyanov EV, EV Keting Marfan syndrome. // Cardiology. - 2006. - № 1. - S. 92-98.
3. Zerbino DD Pathologia aorta klasifikatsiya, hvorobi i syndrome, Problems etiologiï // Medical Transport of Ukraine. - 2006. - № 2. - S. 6-14.
4. Zerbino DD, Olkhova OV Juraev RK (2010) Marfan syndrome: istorichny view that suchasny Looking at the problem // Ukr. honey. Journal. - 2010. - № 6 (80). - S. 97-100.
5. Kadurina TI, Gorbunova VN (2009) Connective tissue dysplasia. //Guide for physicians. - St. Petersburg: Albee, 2009. - 704 p.
6. Fischenko VY, YV Fischenko Semiotika Marfan Syndrome //Ukr. honey. almanac. - 2007. - № 10 (2). - S. 172-175.
7. Dean J.C. (2007) Marfan syndrome: clinical diagnosis and management. // Eur. J. Hum. Genet. – 2007. - №15(7). – C. 724–733.
8. Frydman M. The Marfan syndrome // Isr. Med. Assoc. J. – 2008. - №10(3). – C. 175–178.
9. He R., Guo D.C., Sun W. Characterization of the inflammatory cells in ascending thoracic aortic aneurysms in patients with Marfan syndrome, familial thoracic aortic aneurysms, and sporadic aneurysms. // J. Thorac. Cardiovasc. Surg.. – 2008. - №136(4). - C. 922–929.
10. Gonnermann J., Torun N., Klamann M. Posterior iris-claw intraocular lens implantation in subluxated lenses due to Marfan syndrome // Eur. J. Ophthalmol. - 2014. - Vol. 24, №3. - P. 352-357.
11. Toshpulatova A.Z. Marfan syndrome and its geniologic characteristics // European scence review. – Vienna, 2015. – P. 164–166 (14.00.00; № 19).
12. Toshpulatova A.Z. Clinic and ocular displays of marfan syndrome in children and their surgical treatment // European applied scences, 2015. – P. 20–22.

Поступила 15.05. 2019

ISSN 2181-1954. EISSN 2181-1962