



Results of Surgical Management of Epiretinal Membrane Due to Combined Hamartoma of the Retina and Retinal Pigment Epithelium in Children

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Abstract

Purpose: To study of the effectiveness of microinvasive vitrectomy with membrane peeling for epiretinal membrane (ERM) due to combined hamartoma of the retina and retinal pigment epithelium (CHRRPE) in children.

Material and Methods: The object of the study was 14 children aged 1-8 years with a CHRRPE. 8 children (9 eyes) underwent 23G microinvasive vitrectomy with membrane peeling. In addition to the standard ophthalmological examination, all children underwent optical coherence tomography (OCT) of the macula before and during the follow-up period ranged from 2 months to 4 years.

Results: Improvement of the vitreoretinal contour was noted in all cases. The maximum retinal thickness in the hamartoma area before surgery ranged from 476 to 841 μm (average, 691 μm), after surgery - from 414 to 607 μm (average, 535 μm). The best corrected visual acuity before surgery ranged from 0.03 to 0.3 (on average, 0.15), after surgery it increased by 0.01 to 0.4 in 5 eyes, remained stable in 2 eyes, and decreased by 0.02 and 0.05 in 2 eyes.

Conclusion: Microinvasive vitrectomy with membrane peeling is an effective intervention that stabilizes or improves visual functions in patients with macular ERM due to CHRRPE.

Keywords: Combined Hamartoma of the Retina and Retinal Pigment Epithelium; Children; Epiretinal Membrane; Microinvasive Vitrectomy; Optical Coherence Tomography

Abbreviations

BCVA: Best Corrected Visual Acuity; CHRRPE: Combined Hamartoma of the Retina and Retinal Pigment Epithelium; ERM: Epiretinal Membrane; OCT: Optical Coherence Tomography; RPE: Retinal Pigment Epithelium

Introduction

Combined hamartoma of the retina and retinal pigment epithelium (CHRRPE) is a rare congenital developmental anomaly. It was first described by J.D. Gass in 1973, however, there are a rather limited number of publications in the literature devoted to this disease, most of which contain descriptions of isolated clinical cases [1-8]. The pathology is usually unilateral, sporadic and isolated.

Histologically, CHRRPE consists of different proportions of melanocytic, vascular and glial cells. The glial cells dominate in its clinical manifestations [2,5]. Variations in the composition of cell types are responsible for heterogeneous clinical manifestations.

The main complaints in CHRRPE are low visual acuity, metamorphopsia and strabismus [5]. The main diagnostic methods are ophthalmoscopy and optical coherence tomography (OCT). Sources of additional diagnostic information include fluorescence angiography, OCT angiography, autofluorescence, and microperimetry [6]. Ophthalmoscopically, a CHRRPE is defined as a protruding lesion of a grayish color with unclear contours, 1 to 10 mm or more in size [10], with an abnormal course of retinal vessels above the formation and perifocally. Localization of a CHRRPE in the peripapillary zone (most often), in the macula, and in the middle periphery has been described [9]. A characteristic diagnostic criterion of CHRRPE in OCT images is the epiretinal membrane (ERM), which is determined in all cases of hamartoma of macular localization; OCT signs reflecting the presence and degree of traction syndrome have also been described - the so-called mini-peaks, maxi-peaks, "omega sign", "shark teeth" [10-13]. The main complications of CHRRPE, which lead to an even greater decrease in visual functions, include an increase in the traction effect of ERM on the retina and the formation of a choroidal neovascular membrane (CNM). The tactics of treating these complications are ambiguous: in the case of CNM, the method of choice is intravitreal injection of an antiangiogenic drug, but the question of the advisability of removing ERM in CHRRPE is debatable. There is a point of view according to which ERM is "woven" into the dysplastic retina itself and its removal is accompanied by significant trauma to the retinal tissue and is not functionally justified [6]. At the same time, contraction of ERM in these patients causes secondary formation of radial retinal folds, local deformation of retinal vessels, traction retinal detachment, macular retinoschisis and chronic macular edema, which leads to a decrease in visual function [5]. In the literature there is a description of only one case of spontaneous separation of ERM due to CHRRPE [14].

The purpose to study of the effectiveness of microinvasive vitrectomy with membrane peeling for ERM due to CHRRPE in children.

Material and Methods

The object of the study was 14 children (10 boys and 4 girls, ages 1-8 years) with CHRRPE who were examined and treated in the department of eye pathology of children «Helmholtz National Medical Research Center of Eye Diseases» from 2016 to 2024. A retrospective analysis of the data from the anamnesis, standard ophthalmological examination and optical coherence tomography (OCT) (RS-3000 Advance 2, Nidek, OCT Spectralis, Heidelberg) in the pre- and postoperative period was performed.

Ethics approval

The study was conducted in accordance with the World Medical Association's Declaration of Helsinki (2013 edition). Parental consent was obtained in accordance with the principles approved by the ethics committee of the Helmholtz National Medical Research Center for Eye Diseases, Moscow, Russia.

Results

In 13 children, the lesion was isolated, in 1 child the pathology was associated with neurofibromatosis type I. In 2 children, the hamartoma was binocular (including in a child with neurofibromatosis type I), in 12 – was monocular. The main complaints were strabismus and low vision.

In 13 eyes, the CHRRPE was localized in the macular zone, in 1 eye - paramacular, in 1 eye - peripapillary, in 1 eye - on the middle periphery. Clinically, a protruding lesion of a grayish color with unclear contours, ERM and an abnormal course of retinal vessels above the lesion and perifocally were determined on the fundus (Figure 1).



Figure 1: Ophthalmoscopic picture of CHRRPE of macular localization - a protruding lesion of a grayish color with unclear contours, ERM and abnormal course of retinal vessels.

According to OCT, ERM with pronounced retinal traction, leading to the formation of tangential tension folds across the entire retinal thickness, was diagnosed in 10 children (in 11 out of 13 (84.6%) eyes with CHRRPE of macular localization) and was an indication for vitreoretinal surgery (Figure 2).

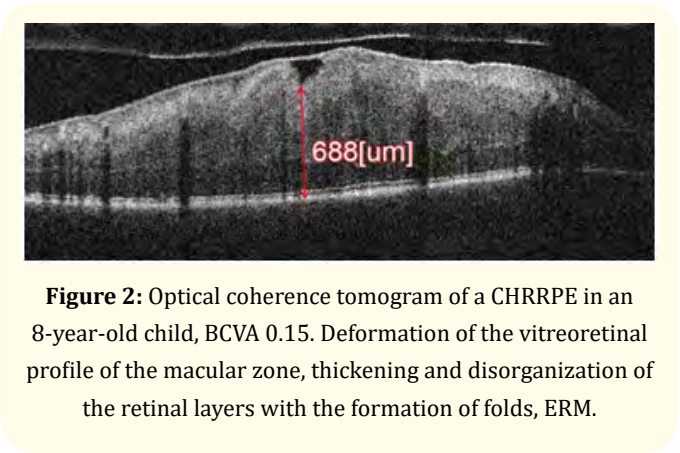


Figure 2: Optical coherence tomogram of a CHRRPE in an 8-year-old child, BCVA 0.15. Deformation of the vitreoretinal profile of the macular zone, thickening and disorganization of the retinal layers with the formation of folds, ERM.

The best corrected visual acuity (BCVA) at the time of hamartoma diagnosis with macular localization was from 0.02 to 0.3 (in 2 children under 2 years of age it was defined as “object vision”), with paramacular and peripheral localization - 1.0.

8 out of 10 children with severe traction syndrome underwent standard 3-port microinvasive vitrectomy 23G with removal of ERM and internal limiting membrane. 2 parents refused from surgical intervention.

3 children from surgical group were dynamically observed in Helmholtz National Medical Research Center of Eye Diseases, the periods of increasing ERM traction in them were 2, 5 and 8 years. In 5 children, surgery was performed after the first examination.

A brief description of all children included in the study is presented in Table 1.

All examined: 14 children (16 eyes)	
Surgical intervention was performed on 8 children (9 eyes)	6 children (7 eyes) remained under dynamic observation

Table 1

ERM was removed in all cases of surgical intervention (Figure 3).

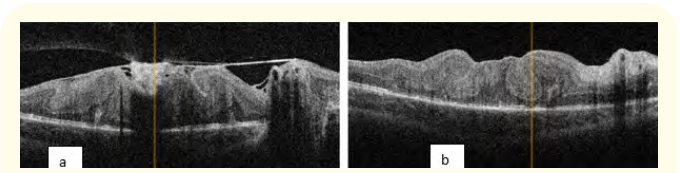


Figure 3: Optical coherence tomogram of CHRRPE in a 5-year-old child. a. Severe deformation of the vitreoretinal contour of the macular zone, thickening and disorganization of the retinal layers with the formation of folds across the entire thickness of the retina, ERM, maximal retinal thickness 619 μm, BCVA 0.03. b. 6 months after surgery. The ERM is not detected, the maximal retinal thickness has decreased (544 μm) with preservation of its folds, BCVA 0.04.

Improvement of morpho-functional parameters was noted: maximum retinal thickness in the hamartoma area before surgery was from 476 to 841 μm (average, 691 μm), after surgery - from 414 to 607 μm (average, 535 μm); BCVA before surgery was from 0.03 to 0.3 (average, 0.15), after surgery - in 5 eyes it increased by 0.01 to 0.4, in 2 eyes it remained stable, in 2 eyes it decreased by 0.02 and 0.05. Response to the reviewer’s last comment: two children under 2 years of age with objective vision had no indications for surgical intervention and remained under dynamic observation, therefore there is no assessment of the dynamics of visual acuity.

In 1 child, hemophthalmos developed on the first postoperative day, which was completely resorbed against the background of conservative treatment. No other post- and intraoperative complications were observed. The observation period after surgery ranged from 2 months to 4 years. No cases of recurrence of ERM were detected.

Discussion and Conclusion

Due to the low incidence of CHRRPE, the number of studies devoted to the analysis of the results of surgical treatment of ERM in this pathology is very limited. Most publications describe individual clinical cases, the largest study includes 15 patients. In total, about 50 outcomes of surgical intervention for ERM due to CHRRPE have been analyzed from the 1980s to the present, 2/3

of which are in children. It has been demonstrated that surgical intervention in most cases leads to an improvement in the retinal architecture and an increase or stabilization of visual acuity. Cases of unsatisfactory functional results of surgical intervention date back to the period of the 1980s and are explained by the low level of technical equipment of the vitreoretinal surgery, as well as the age of patients who had persistent macular edema, which is a consequence of prolonged retinal traction and leads to damage to photoreceptors [2,15].

We believe that the indication for surgical intervention is severe vitreoretinal traction and retinal deformation. In our work, visual acuity improvement was observed in 5 of 9 eyes (55.5%). 2 cases where visual acuity did not change after treatment and 1 case of BCVA decrease by 0.02 can be explained by the short observation period of these patients (up to 2 months), insufficient for functional rehabilitation of the retina. Another case of BCVA decrease in the postoperative period was observed in a child, in whom the observation period from the moment of ERM diagnosis was 8 years. According to OCT performed in the preoperative period, dense adhesion of the ERM and hamartoma, its hyperreflectivity with shielding of the underlying retina, and the formation of retinal folds across the entire thickness were noted. In the postoperative period, destruction of the outer layers of the retina in the fovea zone was diagnosed.

The issues of predicting visual functions after ERM removal, the timing of surgical intervention and improving functional outcomes remain open.

Microperimetry and OCT data are considered as prognostic criteria for visual functions: lower retinal light sensitivity is associated with more pronounced vitreoretinal adhesion and, consequently, the risk of intraoperative complications that reduce the anatomical and functional prognosis of surgical intervention; according to OCT, the probability of surgical success decreases with the spread of hamartoma to the outer layers of the retina and the absence of a visible border between ERM and hamartoma [6]. There is an opinion that the functional results of ERM removal in CHRRPE depend on the age of the intervention - the younger the patient, the less impaired the macular anatomy [16].

Thus, microinvasive vitrectomy with ERM removal is an effective intervention that stabilizes or improves visual functions in patients with CHRRPE. Factors that reduce the functional prognosis in the postoperative period, in addition to the severity of structural retinal changes, include the duration of traction on the retina from the ERM, leading to an increase in secondary retinal changes.

Competing Interests

Authors declare to have no competing interest.

Author Contribution

- Katargina L.A. - concept and design of the study, scientific editing, approval of the final version of the article;
- Denisova E.V. - examination of patients, surgical treatment, scientific editing;
- Getadaryan V.R. - examination of patients, surgical treatment, scientific editing;
- Osipova N.A. - examination of patients, writing of text, design of bibliography, responsibility for the integrity of all parts of the article.

Bibliography

1. Gass JD. "An unusual hamartoma of the pigment epithelium and retina simulating choroidal melanoma and retinoblastoma". *Transactions of the American Ophthalmological Society* 71 (1973): 171-183. discussions 184-185.
2. Schachat AP, *et al.* "Combined hamartomas of the retina and retinal pigment epithelium". *Ophthalmology* 91.12 (1984): 1609-1615.
3. Ledesma-Gil G., *et al.* "Presumed Natural History of Combined Hamartoma of the Retina and Retinal Pigment Epithelium". *Ophthalmology Retina* 5 (2021): 1156-1163.
4. Shields CL, *et al.* "Combined hamartoma of the retina and retinal pigment epithelium in 77 consecutive patients visual outcome based on macular versus extramacular tumor location". *Ophthalmology* 115.12 (2008): 2246-2252.e3.
5. Font RL, *et al.* "Combined hamartoma of sensory retina and retinal pigment epithelium". *Retina* 9.4 (1989): 302-311.

6. Zhang X., *et al.* "Description and surgical management of epiretinal membrane due to combined hamartoma of the retina and retinal pigment epithelium". *Advances in Ophthalmology Practice and Research* 3.1 (2022): 9-14.
7. Gupta R., *et al.* "Peripapillary Versus Macular Combined Hamartoma of the Retina and Retinal Pigment Epithelium: Imaging Characteristics". *American Journal of Ophthalmology* 200 (2019): 263-269.
8. Yarovoy AA., *et al.* "Combined hamartomas of the retina and retinal pigment epithelium: clinical cases". *Bashkortostan Medical Journal* 15.4 (2020): 47-51.
9. Shields JA., *et al.* "Tumors and related lesions of the pigment epithelium". *Intraocular Tumors: an Atlas and Textbook* (3rd ed) Lippincott Williams & Wilkins; Philadelphia, PA, USA (2016): 1188-319.
10. Shields CL. "Optical coherence tomographic Findings of combined hamartoma of the retina and retinal pigment epithelium in 11 patients". *Archives of Ophthalmology* 123.12 (2005): 1746.
11. Arepalli S., *et al.* "Combined hamartoma of the retina and retinal pigment epithelium: Findings on enhanced Depth imaging optical coherence tomography in Eight eyes". *Retina* 34.11 (2014): 2202-2207.
12. Kumar V., *et al.* "Omega sign: a distinct optical coherence tomography finding in macular combined hamartoma of retina and retinal pigment epithelium". *Ophthalmic Surgery Lasers Imaging Retina* 48.2 (2017): 122-125.
13. Arrigo A., *et al.* "Optical coherence tomography and optical coherence tomography angiography evaluation of combined hamartoma of the retina and retinal pigment epithelium". *Retina* 39.5 (2019): 1009-1015.
14. Kumar V. "Spontaneous separation of ERM in combined hamartoma of retina and retinal pigment epithelium". *Ophthalmology* 124.9 (2017): 1402.
15. McDonald HR., *et al.* "Clinicopathologic results of vitreous surgery for epiretinal membranes in patients with combined retinal and retinal pigment epithelial hamartomas". *American Journal of Ophthalmology* 100.6 (1985): 806-813.
16. Bonnin S., *et al.* "Long-term outcome of epiretinal membrane surgery in young children". *Retina* 36.3 (2016): 558-564.