Original Research Paper

Volume-7 | Issue-9 | September-2017 | ISSN - 2249-555X | IF : 4.894 | IC Value : 79.96

Ophtalmology

MANAGEMENT OF COMBINED HAMARTHOMA OF THE RETINA AND RETINAL PIGMENT EPITHELIUM: CLINICAL, SURGICAL WITH COMBINED INTRAVITREAL/RETROBULBAR INJECTIONS AND REHABILITATION RESULTS

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ABSTRACT This case report describes the case of a 7 years old patient with exotropia and monolateral combined amarthoma of the retina and retinal pigment epithelium(CHRRPE) treated with a combined surgical and rehabilitative method.

The patient underwent a vitrectomy plus membranes peeling and three off label Bevacizumab intravitreal injections plus retrobulbar Triamcinolone injections. After the first Bevacizumab-Triamcinolone injection the Retimax vision trainer rehabilitation method started.

Macular edema (central retinal thickness from 472 to 307µm) ,lesion height (from 805 to 490µm) and lesion vascularization decreased together with a slight improvement in visual function both in terms of visual acuity (BCVA from 6/60 to 6/24) and retinal sensibility at microperimetry (from 12 dB to 18.2 dB).

OCT and Angio-OCT are useful tools to assess the features of the lesion so to plan the best treatment and a prompt amblyopia treatment is mandatory to achieve the best functional outcome in each case.

KEYWORDS : combined retina and retinal pigment epithelium hamarthoma, vision trainer rehabilitation,intravitreal injections

CHRRPE is a rare and benign tumor more typically characterized by pigmentation, elevation, vascular tortuosity and epiretinal membranes formation, as reported by the Macula Society study of CHRRPE. Diagnosis of CHRRPE can be hard to perform mainly due to its rarity, difficult differential diagnosis and clinical presentations. Moreover, despite its benign nature, sight-threatening complications can develop.

In May 2014 a 7-year-old Caucasian child was referred to our center complaining decreased vision in his right eye(RE) since about one month. The patient had a general medical history positive for a pseudotumor cerebri and bilateral papilledema in October 2012, without significant sight-threatening sequelae. The patient presented with a best corrected visual acuity (BCVA) of 6/60,exotropia and a superior rectus muscle deficiency in the affected eye and 6/6 in the fellow eye whose ophthalmic examination was unremarkable. At fundus examination, a pigmented and elevated lesion involving the macular region with diffuse wrinkling was observed at the posterior pole of the RE. The child was diagnosed with CHRRPE according with the clinical appearance, the Optical Coherence Tomography (OCT) and the fluorescein angiography. The patient received a 25G vitrectomy with epiretinal membranes peeling and gas (C3F8) as endotamponade without complications. In the following months a progressive increase in visual acuity was observed, together with an anatomical improvement. At six months BCVA was 6/18.

The patient was regularly monitored in the following months and remained stable. A relapse occurred in October 2015: the child presented again with a decreased visual acuity and metamorphopsia in the affected eye.

BCVA was 6/36 and anterior segment examination was unremarkable. Goldmann manual visual field (VF) test showed a generalized depression and an enlargement of the blindspot. RE fundoscopy revealed a paramacular CHRRPE, few epiretinal membranes, vascular engorgement and possible secondary macular edema. Visual evoked potentials (VEPs) and electroretinograms (ERGs) showed an increased latency and a reduce amplitude in the RE.

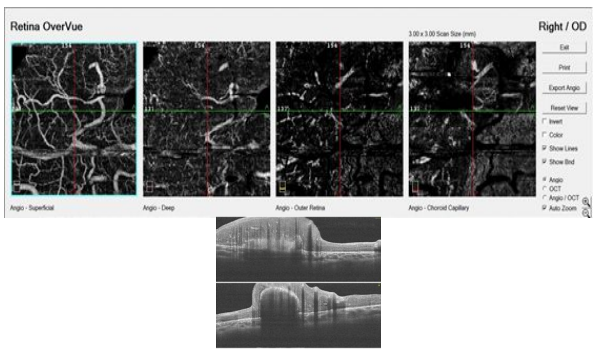
At OCT the central macular thickness was 472 µm in RE and 237 µm in the fellow eye. Cross scans of RE revealed a lesion 805 µm high with high reflectivity of the inner retina and hyporeflective shadowing of the underlying tissue. The presence of intralesion fluid was also detected. OCT Angiography showed abnormal superficial and deep inner retinal vascular plexuses: telangectatic vessels, vasculature engorgement and tortuosity. A marked reduction in central retinal sensitivity (about 12 db) and an extrafoveal fixation in RE resulted from microperimetry examination.

The diagnosis was for a intralesional choroidal neovascularization.

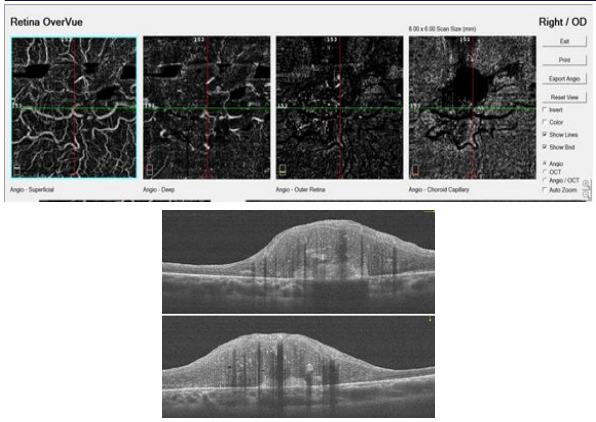
The patient was prescribed a 6 hours of patching of the fellow eye per day and was treated with three combined off-label intravitreal bevacizumab (1.25 mg in 0.05 mL) and retrobulbar Triamcinolone (40 mg/1ml) injections at one-month intervals, from January till March 2016.

The first week after the second injections the Retimax vision trainer(VT) program started with 2 sessions weekly lasting 10 mins for 4 consecutive weeks. VT is a new noninvasive rehabilitation strategy conceived to provide visual improvement in adulthood and childhood disorders. In this method the visual pathway is stimulated by a pattern stimulus; an acoustic feedback allows the patients' fixation and cooperation to be checked. It was shown that the electrophysiology VT rehabilitation acts mainly at the level of the inner retina in amblyopic patients with an age outside the plastic period1. The reasons supposed for the improvement of the retinal ganglion cells activity and BCVA in amblyopic eyes were the reactivation of dysfunctional or quiescent retinal ganglion cells or a magnification of the activity of the healthy ones, the potential role of neurotrophic factor and its retrograde action induced by periodic selective stimulation [1].

A month after each intravitreal injection, biomicroscopy with fundoscopy, OCT, OCT angiography, microperimetry and manual VF tests, electrophysiology assessment were repeated. At fundoscopy a reduction in retinal exudation was detactable at each FU visit. At last FU visit BCVA was 6/24,mean retinal sensitivity within the central 20°improved to 18.2 dB, the amplitude of electrophysiology recordings slightly improved but VF didn't significantly change over the FU. At OCT CRT decreased to 307 µm. B scans and angio-OCT scans showed a significant decrease of lesion height as well as a reduction in vascular engorgement (Figure 1).



A



B

Figure 1. B scans OCT and angio OCT scans of the lesion before (A) and after the three combined intravitreal bevacizumab (1.25 mg in 0.05 mL) and retrobulbar Triamcinolone (40 mg/1ml) injections. (B)

Treatment protocol for CHRRPE involved either a "watch and wait" or an interventional approach, mainly basing on patient's clinical assessment. Surgical treatment in such cases is tricky and functional gain is often slight and not satisfying so the surgical management of CHRRPE is still controversial2. Gass et al suggested that the glial membrane causing the retinal wrinkling is often part of the mass so it's impossible to peel the membrane intrinsic to the dysplastic retina without damaging the retinal fiber layer and Muller cells and this leads to little chance of vision recovery 3. The significant improvement in visual acuity after surgery observed in this case suggests that the surgical treatment is beneficial in some cases. There are not clear criteria to determine if and at what degree the epiretinal components and the underlying hamarthoma are integral; we used the OCT scans for highlighting the position of the membranes, their cleavage plans and their relation with the retinal layers.

In recent years, a new approach has been applied for CHRRPE: anti-VEGF therapy could be considered if a related macular edema (non tractional) or CNV are detectable4. Anti-VEGF treatment for macular edema has the advantage over photodynamic and photocoagulation therapy to be less invasive on retinal and choroidal tissues and to not induce scarring.

In our report good anatomical results were achieved after this combined treatment (Figure 2) without cataract development but the eccentric fixation as well as the macular atrophic-degenerative changes didn't allow a satisfying recovery of visual function: a timely amblyopia and strabismus therapy after surgery are very important in these patients.

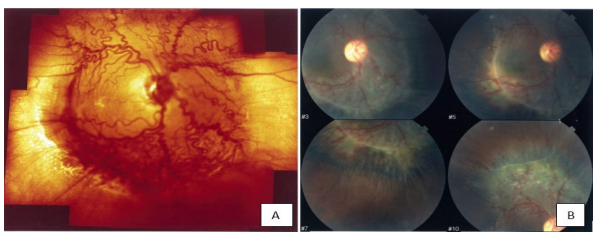


Figure 2. Fundus retinography before (A) and after surgical and medical treatment (B).

Moreover our patient presented with low BCVA and low retinal sensitivity at microperimetry : Bruè et al demonstrated that patients with lower sensitivity preoperatively tend to have a worse functional outcome 2.

In conclusion a combined surgical, medical and rehabilitative approach could be the best strategy in these cases where the challenges of the surgical procedure compound the difficulties related to the paracentral lesion location and the risk of permanent amblyopia if a recover of VA is not achieved before the critical visual developmental period.

Conflicts of interest

The authors declare no conflict of interest.

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