КЛІНІЧНІ ВИПАДКИ

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PERSISTENT HYPERPLASTIC PRIMARY VITREOUS: DIAGNOSIS AND MANAGEMENT

Persistent Hyperplastic Primary Vitreous (PHPV) also known as Persistent Foetal Vasculature (PFV), is a rare congenital eye disease characterized by failure of involution of the hyaloid vasculature. We have used a multimodal imaging approach with retinal camera, slit lamp biomicrography, ultrasonography and anterior and posterior segment optical coherence tomography (OCT) to demonstrate the pathophysiology of this condition. These investigations have also]played a part in the improvement of diagnosis and management of this rare disease.

Key words: *persistent hyperplastic primary vitreous (PHPV), optical coherence tomography (OCT), cycloplegic refraction, leukocoria, microphthalmos.*

A 5-year-old girl attended the Orthoptic Department at the Prince Charles Eye Unit after her parents noticed an increasing squint, light sensitivity and cloudiness to the left pupil over the previous six months. There was no family history of congenital cataracts and the patient was developing normally. On examination, visual acuities were 6/6 and PL in her right and left eyes respectively. Cycloplegic refraction revealed +3,25/–2,00D right and –12,00D left. Ishihara colour plates were 13/13 in the right eye. Slit lamp biomicroscopy was normal for the right eye. The left eye had a dense central homogenous posterior lens opacity obscuring fundal view (Fig. 1). Intraocular pressures were 15mm Hg bilaterally.

A multimodal imaging approach was undertaken. B-scan ultrasonography of the left eye revealed a persistent foetal vessel extending from the optic nerve head

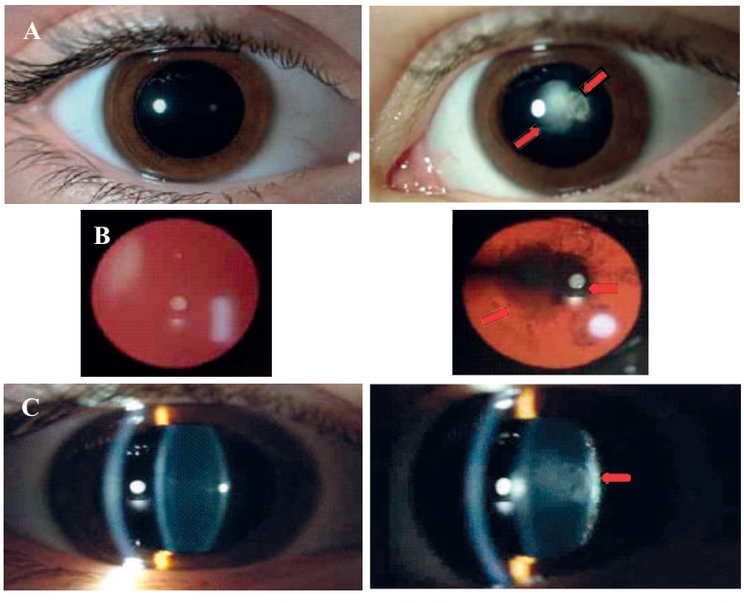

Fig. 1. Colour images illustrating anterior segments of both eyes. Diffuse illumination (A), retroillumination (B) and slit lamp biomicrography (C) showing a dense homogenous lesion covering the central part of the left posterior capsule

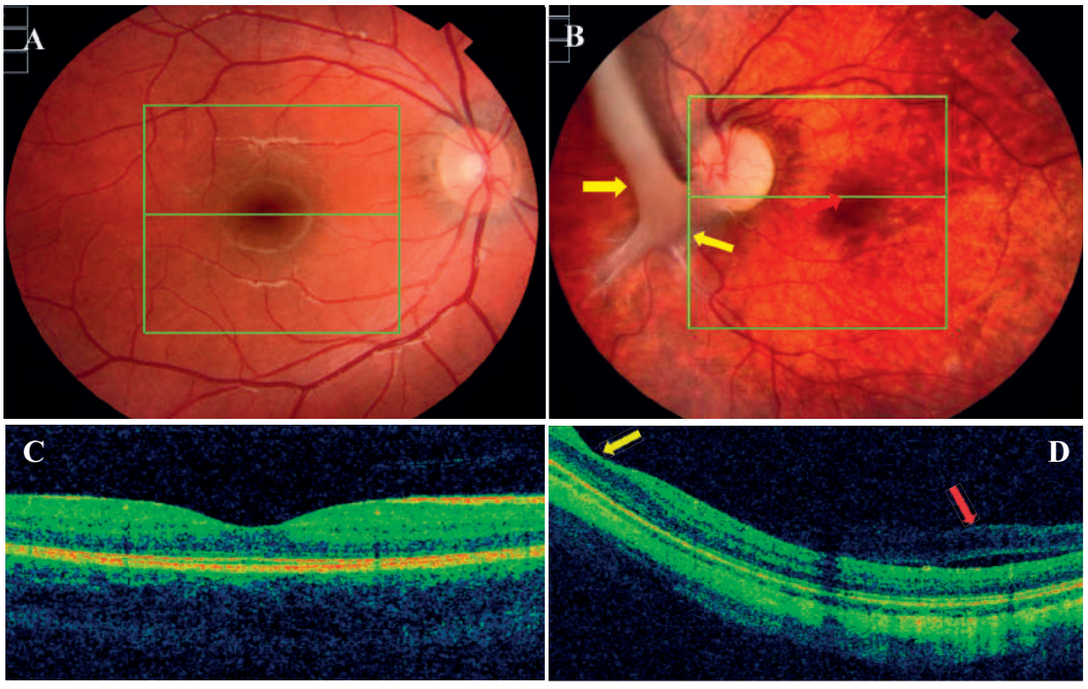

Fig. 2. Colour and OCT images of both eyes. (A and C) The colour and OCT images of the right eye showing no pathological features. (B) Colour image of the left fundus revealing a dense greyish stalk (yellow arrows) emanating from the optic nerve head into the vitreous with tenting of the surrounding retina and dragging of the macula (red arrow). (D) A horizontal scan of the left macula illustrating drugging of the fovea (yellow arrow) and the fibrovascular tissue (red arrow) overlying the macula which corresponds to the fibrovascular plaque

to the back of the lens, where a fibro-vascular plaque gave rise to the previously noted lens opacity. Doppler ultrasonography confirmed the absence of blood flow through the persistent foetal vascular stalk. Biometry showed increased axial lengths of 26.8 mm right and 22.5 mm left.

The clinical examination and imaging demonstrated the severity and progression of PHPV and provided the reason for undertaking surgery. A combined left anterior vitrectomy, lensectomy and intraocular lens insertion was performed. Following surgery, refractive correction and patching for a year resulted in increased left eye visual acuity from perception of light (PL) to 6/60.

Postoperative left fundal examination revealed a white stalk of a fibrovascular tissue extending from the peripheral part of the optic nerve head around 6–8 o'clock (yellow arrows), that caused distortion of the surrounding retinal vessels (Fig. 2B). OCT illustrated opaque tissue (red arrow) on the surface of the macular, corresponding to the fibrovascular plaque. The fovea was dragged to the optic nerve head (yellow arrow) and caused structural changes (Fig. 3).

B-scan ultrasonography of the left eye revealed a bright hyperechoic signal from the optic nerve head projecting into the vitreous towards the lens (Fig. 4).

Differential diagnoses. In order to plan the correct management options, it is important to differentiate PHPV from retinoblastoma, retinopathy of prematurity, Coats' disease, Norrie's disease, toxoplasmosis, congenital cataract and other potentially serious conditions.

Based on location, PHPV is divided into three types: anterior, posterior and combined. Important features of the anterior PHPV are microphthalmos, leukocoria, strabismus and ambliopia. This type is associated with cataract formation, elongated ciliary processes, development

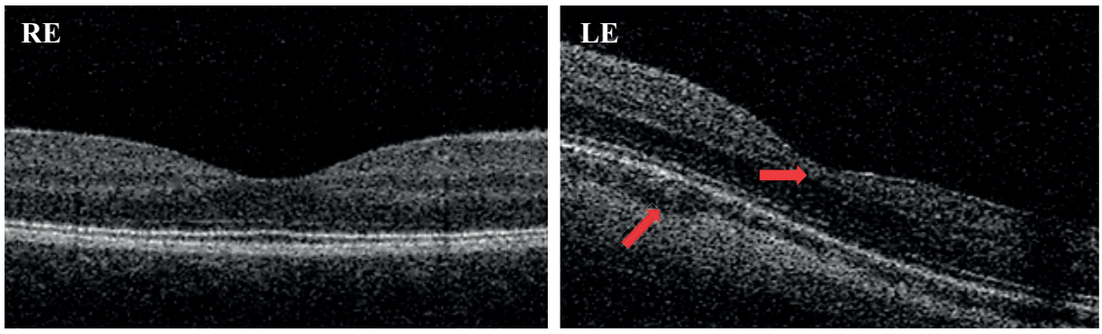

Fig. 3. Horizontal scans of the fovea of both eyes. The tomography of the right fovea is normal. The scan of the left fovea showing changes at the choriodal level and also thickening of the central part of the fovea

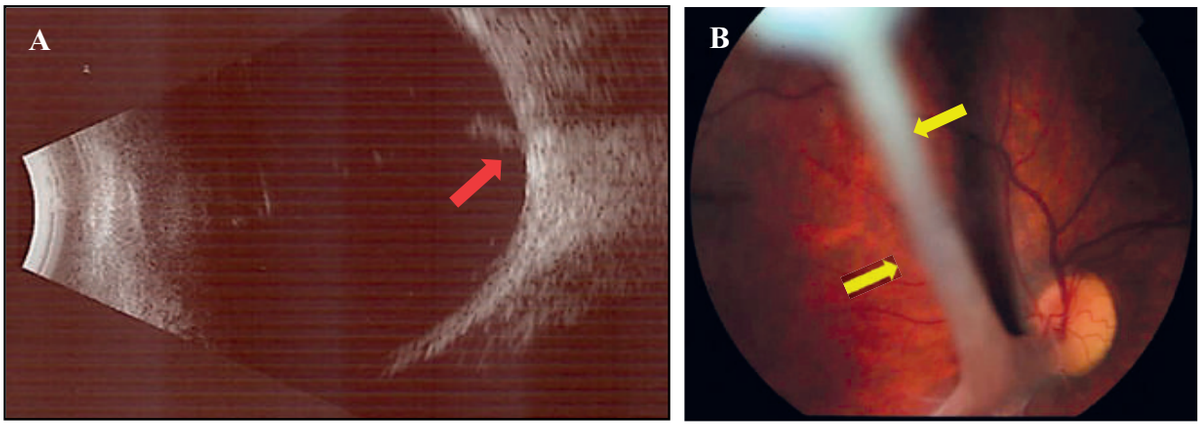

Fig. 4. (A) B-scan ultrasonography of the left eye demonstrating a hyperechoic structure in the vitreous extending from the optic nerve head towards the lens (red arrow). (B) Colour image showing more complete structure of the stalk as compared to the B-scan.

of glaucoma and a retrolental vascular membrane. The posterior subtype of PHPV is characterized by a remnant vascular stalk arising from the optic nerve head into the vitreous. This subtype may be associated with other developmental abnormalities of the retina, optic nerve head, macula and vitreous [1].

Treatment. Depending on age, early detection, extent of ocular involvement, complications (e.g. glaucoma and progressive retinal detachment, microphthalmos), there is a wide range of treatment for PHPV. Management of the anterior PHPV includes observation, medical treatment or surgery. Posterior and combined PHPV, which may be associated with progressive retinal detachment or angle-closure glaucoma, is an indication for surgery. Post-operation visual rehabilitation includes correction of refractive error and treatment of amblyopia. Management of PHPV involves collaboration of paediatric ophthalmologist, vitreoretinal specialist and an orthoptist [2].

Discussion. We present a case of PHPV involving anterior and posterior segments of the eye. The condition was first described by Algernon Reese in 1955 [3]. PHPV usually presents as unilateral condition (90%). However, there is a group of patients (10%) with bilateral manifestations. About 14% of patients have associated neurological abnormalities [1]. Based on embryogenesis of the eye, the hyaloid vascular system starts to form in the first trimester of gestation. In the third trimester, the embryonic hyaloid artery undergoes regression. If the involution process of the primary vitreous and hyaloid vasculature fails by the time of birth, the foetal vasculature can persist [4, 5]. The pathogenesis of this condition remains unknown although it has been recently proposed that a combination of abnormal regulation of apoptosis and atypical timing of gene expression with irregular levels of growth factors (VEGF, bFGF and angiopoetin-2) could result in PHPV [6].

The common clinical features of PHPV are leukocoria, microphthalmos, cataract, glaucoma and retinal detachment. It has been previously reported that the most common diagnostic procedures for confirming PHPV are: B-scan ultrasonography, computed tomography (CT), magnetic resonance imaging (MRI), visual evoked potentials (VEP), fluorescein angiography and molecular genetic analysis [7]. Optical coherence tomography (OCT) has not been applied so far to illustrate the topographical structure of the macular in PHPV. However, this investigation could have an important diagnostic value for further management of patients with PHPV.

Learning points. PHPV is a rare, congenital, usually unilateral eye condition occasionally associated with systemic abnormalities such as Norrie's disease. Diagnostic slit lamp biomicrography, retinography, anterior and posterior OCT and B-scan ultrasonography are of value.

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ПЕРВИННЕ ПЕРСИСТУЮЧЕ ГІПЕРПЛАСТИЧНЕ СКЛОПОДІБНЕ ТІЛО: ДІАГНОСТИКА ТА ЛІКУВАННЯ

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Первинне персистуюче гіперпластичне скловидне тіло (ППГСТ), також відоме як персистуюча вакуляризація зародка ока, є рідкісною вродженою аномалією, яка характеризується призупиненням інволюційного процесу гіалоїдної васкуляризації.

Ми використали мультимодальний підхід для отримання оптичних зображень за допомогою ретинальної камери, біомікроскопії, оптичної когерентної томографії та ультразвукового обстеження.

Поєднання цих діагностичих методик дозволило нам продемонструвати детальну анатомію та патофізіологію ППГСТ, провести диференціальну діагностику, а також визначити тактику лікування та спостереження в період реабілітації.

Ключові слова: *первинне персистуюче гіперпластичне скловидне тіло, оптична когерентна томографія, мікрофтальм*.

ПЕРВИЧНОЕ ПЕРСИСТИРУЮЩЕЕ ГИПЕРПЛАСТИЧЕСКОЕ СТЕКЛОВИДНОЕ ТЕЛО: ДИАГНОСТИКА И ЛЕЧЕНИЕ

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Первичное персистирующее гиперпластическое стекловидное тело (ППГСТ), также известное как персистирующая васкуляризация зародыша глаза, – редкая врожденная аномалия, которая характеризуется приостановлением инволюционного процесса гиалоидной васкуляризации.

Мы использовали мультимодальный подход для получения оптических изображений при помощи ретинальной камеры, биомикроскопии, оптической когерентной томографии и ультразвукового обследования.

Сочетание этих диагностических методик позволило нам продемонстрировать детальную анатомию и патофизиологию ППГСТ, провести дифференциальную диагностику, а также определить тактику лечения и наблюдения в период реабилитации.

Ключевые слова: *первичное персистирующее гиперпластическое стекловидное тело, оптическая когерентная томография, микрофтальм.*

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