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Updated review: optical coherence tomography findings of the pachychoroid disease spectrum

The ocular choroid consists of 5 distinct layers: innermost Bruch's membrane, capillaries of the choriocapillaris, Sattler's layer housing medium-sized blood vessels, Haller's layer containing larger-sized vessels, and the suprachoroidal space. Pachychoroid disease spectrum (PDS) was first described by Warrow et al.[1] in 2013 as a group of chorioretinal disease entities characterized by a thickened choroid exceeding 300 μm and leading to exudative and neovascular complications. However, improved multimodal imaging, including extended depth imaging optical coherence tomography (EDI-OCT), has helped characterize PDS as a spectrum of ocular phenotypes all sharing 3 common characteristics: engorged Haller's layer vessels, Sattler's layer compression, and choriocapillaris attenuation.[2] Recent evidence also has linked choriocapillaris hyperpermeability and choroidal anastomoses secondary to vortex vein obstruction as the key factors in PDS development, but the unequivocal etiology of the spectrum has yet to be concluded.[3]

Because PDS is primarily a choroidal vessel anomaly, indocyanine green angiography is extremely useful in demonstrating choroidal hyperpermeability.[4] However, limited access to indocyanine green angiography makes strong knowledge of multimodal imaging findings important in accurately diagnosing PDS variants. This is crucial because treatment of PDS entities, such as peripapillary pachychoroid syndrome, can mimic common retinal pathologies such as diabetic macular edema but do not require repeated intravitreal anti—vascular endothelial growth factor (anti-VEGF) injections.[5] The purpose of this brief communication is to highlight the key imaging features of common multimodal imaging modalities for the 6 PDS variants in order to improve diagnostic accuracy and minimize patient morbidity.

Central Serous Chorioretinopathy

Central serous chorioretinopathy (CSC) is characterized by dysfunction of the retinal pigment epithelium (RPE), detachment of the neurosensory retina, and RPE atrophy. The serous detachment of the neurosensory retina is typically at the macula and can be associated with pigment epithelial detachments (PEDs). Symptoms include blurred vision, metamorphopsia, central scotoma, micropia, and dyschromatopsia. EDI-OCT will demonstrate subretinal fluid and serous PEDs, along with increased choroidal thickness in both eyes (Fig. 1C). In chronic CSC, outer retinal hyperreflective foci with "shaggy borders" can be present, which represent degradation of the outer photoreceptors. Fibrinous subretinal hyperreflective material can be seen on OCT imaging in severe CSC cases, including those induced by pregnancy or steroid use. Fundus autofluorescence (FAF) typically shows hypo-autofluorescence corresponding to the area of subretinal fluid, whereas fluorescein angiography demonstrates leakage in either ink-dot (more common) or smoke-stack patterns. FAF is crucial in the diagnosis of chronic CSC, with the classic "guttering" pattern demonstrating subretinal fluid settling in the inferior retina. The acute form of CSC is usually self-limited, with complete resolution of the subretinal fluid typically seen within 3—6 months.

Pachychoroid Pigment Epitheliopathy

Pachychoroid pigment epitheliopathy (PPE) was first demonstrated by Warrow et al.[1] in 2013 after reporting 9 cases that had CSC characteristics but lacked subretinal fluid. One of the major features of this entity is the enlarged pachyvessels with no history of subretinal fluid. Other clinical findings include reduced fundus tessellation, small PEDs, and RPE abnormalities due to underlying pachyvessels. PPE may represent forme fruste CSC. The key on EDI-OCT images is similar features to CSC but lack of subretinal fluid (Fig. 1D). FAF will show granular hypo-autofluorescent areas mixed with stippled hyper-autofluorescence.

Focal Choroidal Excavation

Focal choroidal excavation (FCE) is characterized by focal regions of atrophy in the choriocapillaris without evidence of scleral ectasia or posterior staphyloma. Patients are often asymptomatic but may report mild metamorphopsia or generalized blurred vision. Fundus examination is generally unremarkable, with subtle findings of yellowish white lesions in an area of pigmentary change. OCT imaging is crucial to demonstrate the downward depression of an intact RPE secondary to focal attenuation of choriocapillaris and secondary collapse of outer retinal tissue. OCT imaging demonstrates 2 unique FCE states, conforming and nonconforming FCE (Fig. 1A). Photoreceptors in conforming FCE maintain contact with RPE because this layer falls posteriorly through the attenuated inner choroid, whereas photoreceptor tips in nonconforming FCE appear detached from the RPE with a hyporeflective space between photoreceptor outer segments and the RPE layer. FAF demonstrates focal hypo-autofluorescence due to atrophic RPE. EDI-OCT remains the crucial modality to diagnose FCE through inner choroid attenuation, RPE downward depression, and focal pachyvessel enlargement.

https://doi.org/10.1016/j.jcjo.2022.06.014
ISSN 0008-4182

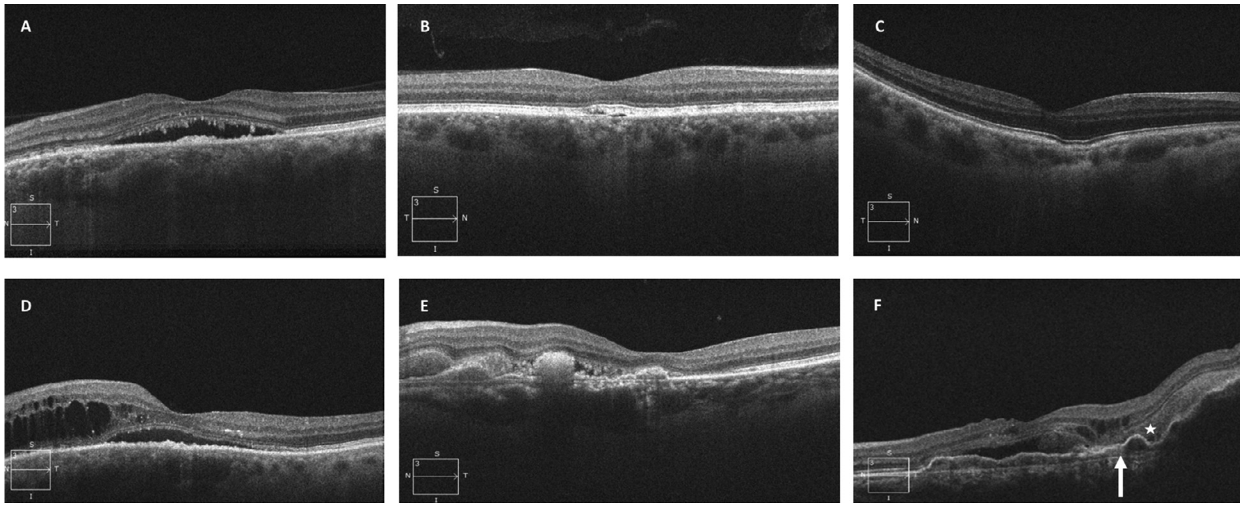

Fig. 1—Enhanced depth imaging optical coherence tomography of (A) central serous chorioretinopathy demonstrating serous detachment of the neurosensory retina, with or without pigment epithelial detachment, and underlying pachyvessel enlargement; (B) pachychoroid pigment epitheliopathy demonstrating retinal pigment epithelium disruption and underling pachyvessel enlargement; (C) conforming focal choroidal excavation demonstrating focal pachyvessel engorgement, choriocapillaris attenuation, retinal pigmented epithelium (RPE) atrophy, and posterior displacement with normal neurosensory retinal layers with photoreceptors maintaining adherence to the RPE; (D) peripapillary pachychoroid syndrome demonstrating choroidal folds, pachyvessel enlargement, choriocapillaris attenuation, and intraretinal fluid at the nasal macula respecting the fovea; (E) pachychoroid neovasculopathy demonstrating shallow, irregular separations of the RPE from Bruch's membrane with double-layer sign and hyperreflective foci corresponding to type 1 choroidal neovascularisation; (F) polypoidal vasculopathy demonstrating peaked pigment epithelial detachment, thumbprint sign, notch sign, double-layer sign, double-hump sign, and enlarged Haller's vessels. *Arrow identifies double-hump sign in polypoidal choroidal vasculopathy; star identifies peaked pigment epithelial detachment in polypoidal choroidal vasculopathy.

Peripapillary Pachychoroid Spectrum

First described in 2018, peripapillary pachychoroid spectrum (PPS) is characterized by peripapillary intraretinal fluid with underlying pachyvessels localized to the nasal macula. Severe cases may be complicated by central intraretinal and subretinal fluid. Patients with PPS may be mistaken for refractory diabetic macular edema patients because of persistent cystoid macular edema. EDI-OCT demonstrates choroidal thickness in the nasal macular regions associated with peripapillary cystoid macular edema (Fig. 1B). This contrasts with CSC, where choroidal thickening is common at the central or temporal macula. FAF demonstrates mottled hypo-autofluorescence in the peripapillary region with or without choroidal folds. Fluorescein angiography occasionally demonstrates disc leakage but more commonly shows speckled hyperfluorescent window defects. A crowded optic disc appearance is typical of PPS, and this disorder can be misdiagnosed as papillitis or uveitis.[1]

Pachychoroid Neovasculopathy

Q2 Initially described by Pang and Freund in 2015, Pachychoroid neovasculopathy (PNV) is characterized by type 1 choroidal neovascularization in the absence of subretinal fluid or macular degeneration features in the contralateral eye. However, pachydrusen, which are larger, more irregularly shaped and more sparsely distributed than typical soft drusen, may be present in either eye. Patients with PNV typically present with reduced visualization of choroidal features or reduced tessellated fundus. Distinctive features are pigment epithelium elevations with subfoveal neurosensory detachments in association with type 1 neovascularization. These elevations on EDI-OCT appear as shallow, irregular separations of the RPE from Bruch's membrane, which appears as a double-layer sign on top of the areas of pachyvessels (Fig. 1E). These areas present as speckled hypo-autofluorescence surrounded by a hyper-autofluorescent ring on FAF. The response to anti-VEGF treatment in PNV is variable, with some eyes requiring combined treatment with photodynamic therapy.

Aneurysmal Type 1 Neovascularization/Polypoidal Choroidal Vasculopathy

Polypoidal choroidal vasculopathy was described initially in 1982 as a recurrent serosanguineous maculopathy associated with orange nodules located around the peripapillary region. EDI-OCT is crucial in the diagnosis; various hallmark OCT features can be identified and include: engorged Haller's vessels, peaked PEDs, thumbprint signs within peaked PEDs with hyperreflective rings and internal hyporeflective lumen, PED notch sign, double-hump sign of an adjacent PED due to extravasation of fluid from the initial peaked PED, and double-layer sign between the RPE and Bruch's membrane (Fig. 1F). Saccular dilations are also visible on en-face OCT as hyperreflective lesions at the level of the RPE. Current treatment of PCV is anti-VEGF monotherapy or combined with photodynamic therapy based on recent pivotal trials.

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Originally received Aug. 17, 2021. Final revision Apr. 21, 2022. Accepted Jun. 23, 2022.

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Footnotes and Disclosure

The authors have no proprietary or commercial interest in any materials discussed in this article.