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CLINICAL CASE REPORT

Central serous chorioretinopathy and achromatopsia: a case report

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Abstract
Purpose To describe a patient with combined central serous chorioretinopathy and achromatopsia.
Methods Clinical examination, enhanced depth imaging- optical coherence tomography, fundus autofluorescence, fluorescein angiography and electroretinography were used to study a 33-year-old female presented with the complaint of poor vision since childhood in both eyes, which worsened in the left eye (LE) recently.
Results In slit-lamp examination, there was a macular elevation in the LE and macular pigmentary change as well as optic disk pallor in both eyes. Enhanced depth imaging optical coherence tomography revealed central inner/outer segment (IS/OS) disruptions, subretinal fluid and thick choroid. Accessory tests included the full-field ERG with severe reduced photopic response (with relatively normal scotopic responses) and fluorescein angiography (FA), which found distinct leakage points in OD and barely visible hyperfluorescent spots in OS. Based on the history of nystagmus, lifelong stable poor vision, loss of foveal cone thickness with IS/OS disruption and severe reduced photopic response with relatively normal scotopic responses, we determined that the diagnosis was most consistent with achromatopsia (ACHM). On the other hand, OCT and FA findings show the simultaneous occurrence of pachychoroid-related central serous chorioretinopathy in this patient.
Conclusion This case highlights a case of CSC and ACHM.

Keywords Achromatopsia · CSCR · EDI-OCT · Full-field ERG

Introduction

ACHM is a rare autosomal recessive disorder, also known as rod monochromacy, characterized by loss of cone cell function, classically presenting loss of color discrimination, photophobia, infantile pendular nystagmus and reduced visual acuity often less than 20/200. Although signs and symptoms vary depending on the amount of residual cone function, visual acuity is usually stable over time [1–3]

Despite many studies in achromatopsia, to the best of our knowledge, no study has been conducted on the choroidal thickness or pachychoroid spectrum disease. This study aims to report a case of CSC and ACHM and to describe the comprehensive image findings using FA and EDI-OCT.

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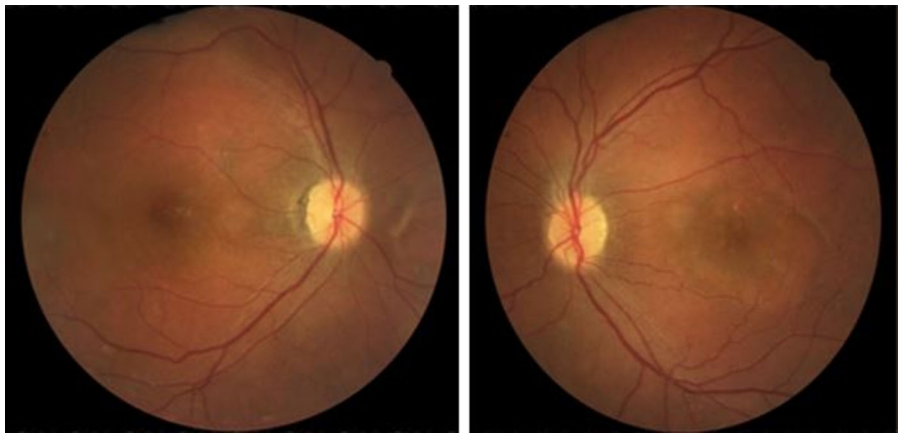

Fig. 1 Fundus photograph of RE shows almost unremarkable findings except for mild macular pigmentary change and disk pallor. An area of mild macular hyperpigmentation and a zone of elevation in the infratemporal of the macula exist in LE

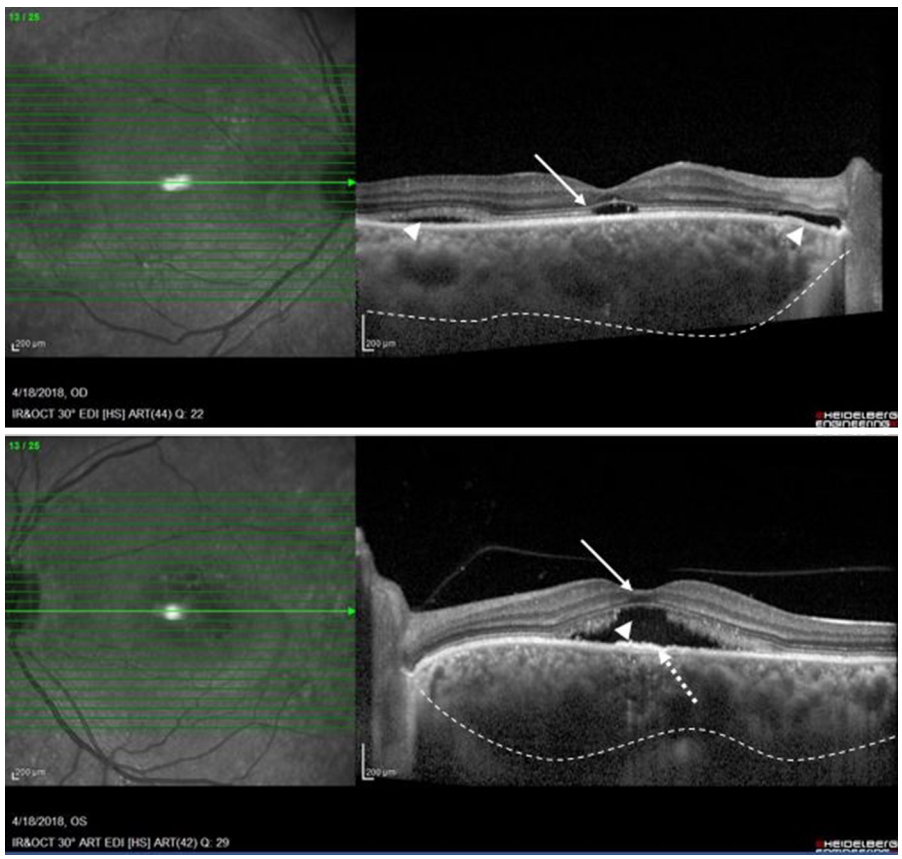

Fig. 2 EDI-OCT demonstrates a sub-foveal hypo-reflective region compatible with IS/OS junction disruption (white arrows), hypo-reflective zone associated with central neurosensory detachment (arrowhead) and shallow irregular detachment of RPE in the optical empty space (dashed arrow). Dilated Haller's vessels and thick choroid (dashed white lines) are demonstrated in both eyes

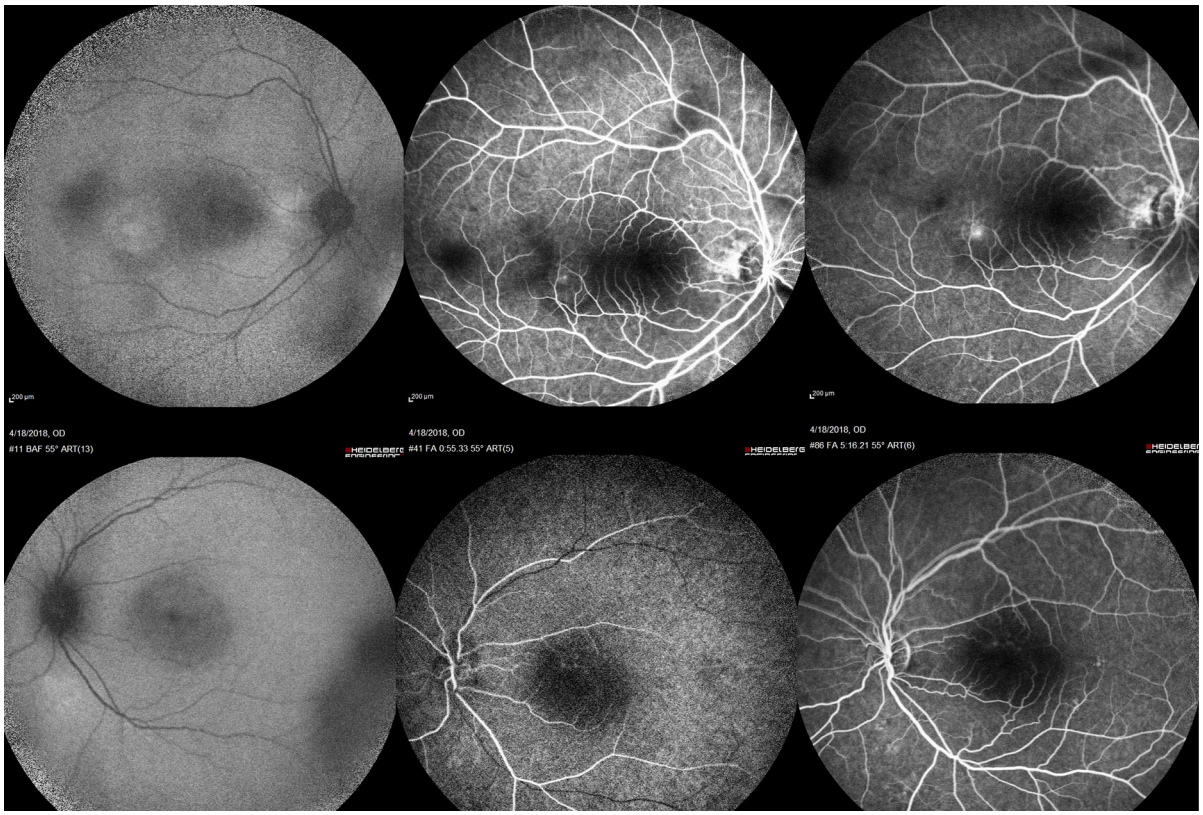

Fig. 3 Fluorescein angiography (FA) shows pinpoint leakage in OD which increases in size and intensity as the angiogram progresses and barely visible hyperfluorescent spots in OS

Case presentation

A 33-year-old woman presented with the complaint of continued poor vision and photophobia which worsened in her left eye for two months. She had no history of systemic disease and had not undergone ocular surgery. Ocular examination revealed pendular nystagmus. The best-corrected visual acuity (BCVA) was 20/200 for both eyes with no significant refractive error. The results of anterior segment examination were unremarkable. Color vision testing with both saturated and desaturated Farnsworth D-15 revealed a complete absence of color discrimination with no associated color defect pattern. Color fundus photography showed macular mottling, reduced choroidal vascular marking in the posterior pole, mild optic disk pallor in both eyes and macular subretinal fluid in the LE; otherwise, her results were relatively normal (Fig. 1).

The EDI-OCT of the macula revealed narrowed outer nuclear layer and ellipsoid zone loss in subfoveal area in each eye, as well as a central neurosensory detachment in the LE and paracentral detachments (temporal to the fovea and peripapillary) in the right eye (RE). OCT also revealed thick RPE in both eyes and shallow irregular pigment epithelial detachment (PED) with a double reflective layer, double layers sign (DLS) directly below the optical empty space in the LE. It also showed a thickened sub-foveal choroid of 419 μm in the right eye and 415 μm in the left eye as well as relatively large choroidal vessels directly below the SRF area in both eyes (Fig. 2).

Fundus autofluorescence (FAF) demonstrated lower intensity corresponding to the area of subretinal fluid in the left eye. Fluorescein angiography (FA) showed pinpoint leakage in OD, which increased in size and intensity as the angiogram progressed and barely visible hyperfluorescent spots in OS, although OCT revealed a larger detachment in OS (Fig. 3). Full-field ERG showed severe reduced photopic responses while the scotopic waves were relatively normal. We used the MonPackOne system

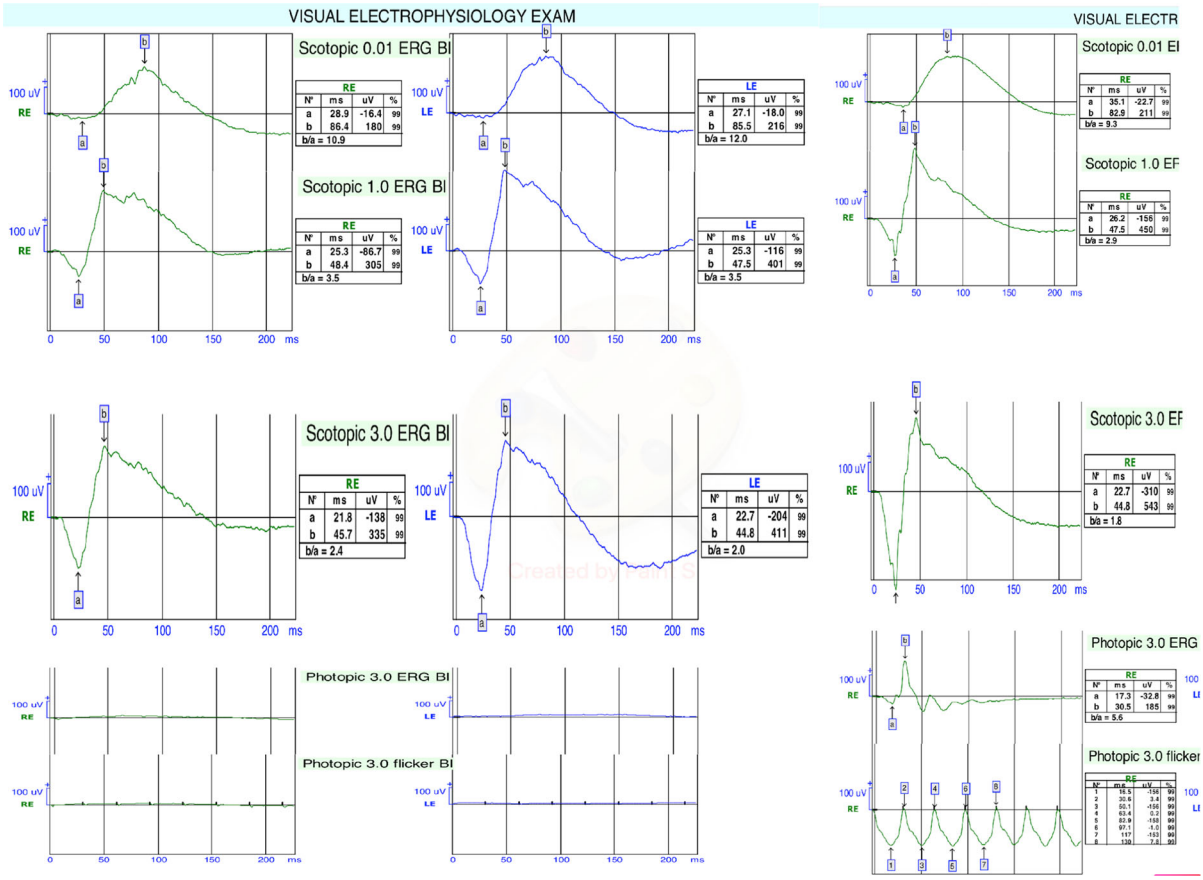

Fig. 4 Full-field electroretinogram (Full-field ERG) shows severe reduced photopic responses and relatively normal scotopic responses (left and middle columns: right and left eye in ACHM patient, right column: normal full-field ERG)

(Metrovision, Perenchies, France). Recording electrodes were scleral contact lenses, and full-field ERG was recorded according to the International Society for Clinical Electrophysiology of Vision (ISCEV) protocol [4] (Fig. 4).

At the third-month follow-up, the EDI-OCT demonstrated complete remission of the serous detachment on both eyes. The patient was satisfied with the improvement in left eye vision, but BCVA had not changed. (Fig. 5)

Discussion

The diagnosis of ACHM can be made through clinical examination, family history, visual acuity test, nystagmus and color vision evaluation, full-field ERG and OCT finding. ACHM patients generally exhibit lifelong stable poor vision, nystagmus, loss of foveal cone thickness often with IS/OS disruption and severe reduced photopic ERG with relatively normal scotopic waves [2, 5, 6].

As the patient had an extinguished cone response, cone-rod dystrophy was a potential differential diagnosis. Although genetic testing is the definitive method of distinction between them, it was not possible in our setting. Therefore, we determined that the diagnosis was more consistent with ACHM based on the history of stable nystagmus and poor vision, OCT finding and relatively normal rod responses.

On the other hand, the FA and OCT findings showed a transient accumulation of subretinal fluid, which suggested the diagnosis of CSC as a reason for the recent visual complaint. Also, shaggy photoreceptors in the OCT and hypoautofluorescent area corresponding to subretinal fluid in the FAF indicate that the SRF has recently occurred. Interestingly, there is no shaggy appearance in the optical empty space in

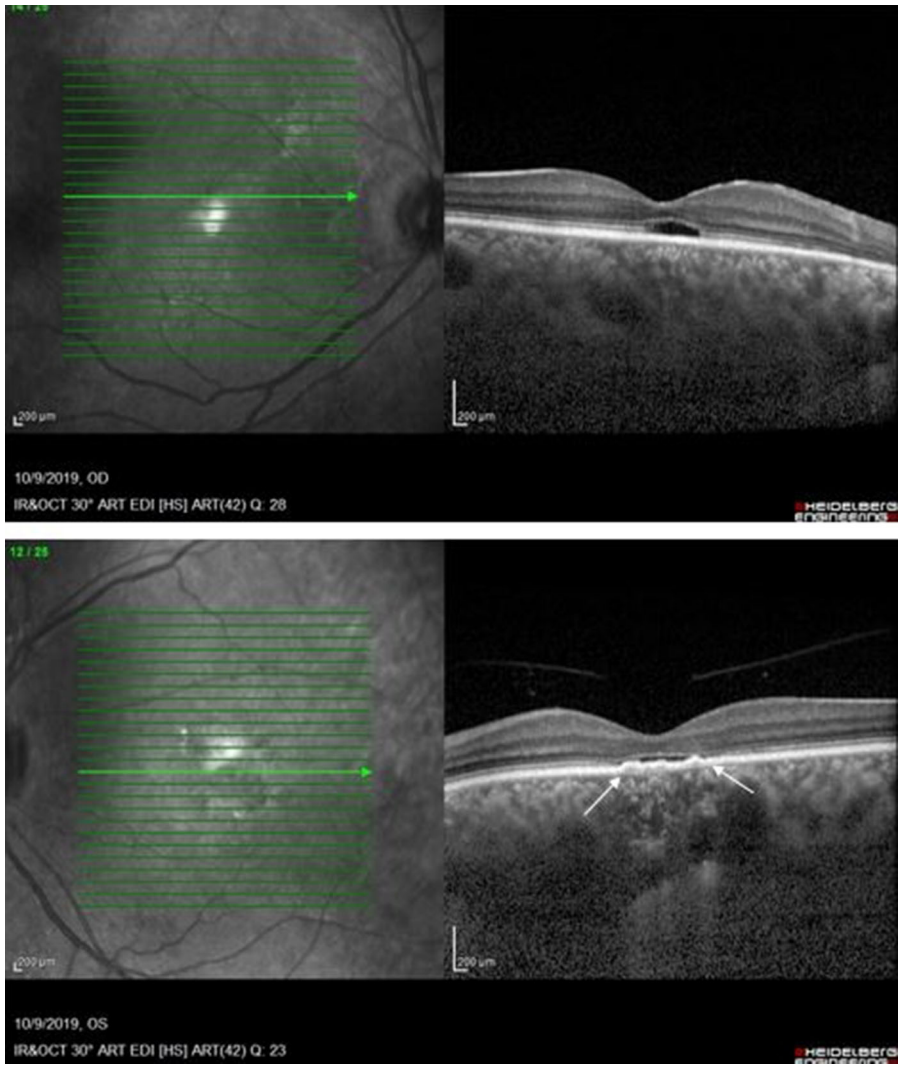

Fig. 5 EDI-OCT demonstrates the complete remission of SRF in both eyes and also shows low lying irregular PED (DLS) in the optical empty space in the left eye (between arrows)

OCT (Fig. 2, bottom). Indeed, according to Greenberg's staging [7], our patient is in stage 3 or 4 of ACHM. Based on this staging and other studies, the cone cell dysfunction appears to be higher at higher stages and probably we have a complete lack of cone cells in the optical empty space (OES) in stages 3 and 4, making us unable to see shagginess. However, at the adjacent region we have a combination of remaining partially functional cone cells and relatively normal rod cells that are responsible for the shagginess of the EZ line in our patient [2, 7].

The EDI-OCT of the macula revealed thick RPE in both eyes and shallow irregular PED directly below the optical empty space in the LE. It also showed a pachychoroid and relatively large choroidal vessels directly below the SRF area in both eyes.

Pachychoroid spectrum refers to a group of clinical entities characterized by an attenuation of the choriocapillaris and dilated choroidal veins which are associated with progressive RPE dysfunction. These entities include pachychoroid pigment epitheliopathy (PPE), CSC, pachycoroid neovasculpathy (PNV) and polypoidal choroidal vasculopathy (PCV). In patients with CSC, choroidal hyperpermeability is believed to increase hydrostatic tissue pressure. The reason for subretinal leakage is assumed to be a micro-rip of the

RPE secondary to increased choroidal pressure overwhelming the physical resistance of the RPE [8, 9].

At the third-month follow-up, the EDI-OCT demonstrated complete remission of the serous detachment on both eyes. The patient was satisfied with the improvement in left eye vision, but BCVA had not changed. It might be due to various measurement methods of vision, which were performed by two different persons, or differences in the measurement conditions, such as room light, monitor light or diurnal variation. However, the patient subjectively reported the improved vision after resolving the subretinal fluid in OCT.

In conclusion, this study aims to report a case of CSC and ACHM and to describe the comprehensive image findings via FA and EDI-OCT.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Informed consent Informed consent was obtained from all participants included in the study.

Patient consent The patient has consented to the submission of the case report for submission to the journal.

Statement of human rights All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Declaration of Helsinki and its later amendments or comparable ethical standards.

Statement on the welfare of animals This article does not contain any studies with animals performed by any of the authors.

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