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Early Detection of Retinal Abnormalities with Spectral-Domain Optical Coherence Tomography and Ultra-Wide Field Autofluorescence

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• SD-OCT • Fundus autofluorescence • Rabin cone test • Hereditary retinal disease • Stargardt • Retinitis pigmentosa • Central serous chorioretinopathy • Acute zonal occult outer retinopathy (AZOOR)

Key points

- Spectral-domain optical coherence tomography (SD-OCT) performs in vivo high-resolution imaging of the retinal layers. Abnormalities of the outer retina invisible to ophthalmoscopy are often revealed using this technology.
- Fundus autofluorescence (FAF) imaging provides an en face topographic map of lipofuscin in the outer retina using a fundus camera to detect the integrity of the outer retina.
- Ultra-wide field FAF allows imaging of 200° of the retina in each photograph and detects abnormalities missed using standard fundus photography fields.
- Abnormalities in FAF patterns permit detection of diseases of the outer retina that would otherwise be missed by ophthalmoscopy and standard fundus photography.

INTRODUCTION

For decades, direct and indirect ophthalmoscopic examination of the retina and standard fundus photography using a color fundus camera with 20°, 30°, and 50° fields of view have been the mainstay of fundus examination. Additional

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testing modalities, such as fluorescein angiography (FA), allowed for the detection of vascular abnormalities, also with the use of standard 50° fields of view with a fundus camera. However, technologies introduced in the first decade of the twenty-first century, have opened up a whole new way of detecting diseases of the macula and retina, invisible to the older examination techniques. Optical coherence tomography (OCT), using laser light to penetrate the retina, enabled the eye care practitioner to view retinal layers in real time to better detect and follow diseases of the retina [1]. In addition to disorders of the outer retina, which are highlighted in this article, inner retinal abnormalities, such as attenuation of the retinal nerve fiber layer (RNFL), and the ganglion cell complex in glaucoma, have proven remarkably important. Fundus autofluorescence (FAF) imaging, introduced into clinical practice in the second decade of this century, enable eye care practitioners to view en face images that demonstrate abnormalities in the outer retina not appreciated by ophthalmoscopy or fundus photography [2]. The advent of wide-field imaging systems allowed for viewing almost the entire retina in 1 or 2 photographs, as opposed to painstakingly piecing together a composite photograph of multiple 50° views of the retina. The result is the improved detection of disease, progression of disease, and diagnosis of myriad retinal disorders [3].

This article reviews select hereditary and acquired retinal diseases that are either invisible or nearly invisible with ophthalmoscopy and fundus photography. In addition to the improved detection of disease, these technologies demonstrate retinal changes over time and patterns of abnormalities that aid understanding of the course of many of these retinal conditions. The diseases that may be missed with standard ophthalmoscopy and fundus photography fall into 2 broad categories: hereditary and acquired.

HEREDITARY RETINAL DISEASES

Hereditary retinal diseases include retinitis pigmentosa (RP) and its various subtypes, which include RP sine pigmento (without pigment), sector RP, pericentral RP, peripapillary RP, syndromic RP (Usher syndrome and Laurence-Moon-Bardet-Biedel), Bietti retinal dystrophy, benign flecked retina, fundus albipunctatus (a form of congenital stationary night blindness), cone dystrophy, cone-rod dystrophy, Stargardt disease (STGD), central areolar choroidal dystrophy, achromatopsia, and Best vitelliform disease. This brief article includes 3 young siblings with STGD and a case of atypical RP.

ACQUIRED RETINAL DISEASES

Acquired retinal diseases include macular and peripheral drusen, age-related macular degeneration (AMD), central serous chorioretinopathy (CSCR), polypoidal choroidal vasculopathy, pattern dystrophies of the retinal pigment epithelium (RPE), angioid streaks, toxoplasmosis, retinoschisis, retinal detachment, diffuse unilateral subacute neuroretinitis, acute zonal occult outer retinopathy (AZOOR), and optic disc drusen. This article includes cases of AZOOR, CSCR, retinoschisis, and retinal detachment.