Medical Retina

FUNDUS FLAVIMACULATUS, A MORE BENIGN AND INFREQUENT VARIANT OF ABCA4 RETINOPATHIES

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PURPOSE: Consider Fundus Flavimaculatus (FF) as a more benign phenotypic variant of ABCA4 retinopathies (ABCA4-R) due to its relevance in genetic counseling.

METHODS: Case report

RESULTS: A 35-year-old patient complained of blurred vision. Best-corrected visual acuity was 20/20 in both eyes. Eye fundus showed yellowish flecks widely scattered over the posterior pole and mid-periphery with macular sparing, which were hyperautofluorescent on fundus autofluorescence. Optical coherence tomography revealed hyperreflective deposits at inner part of retinal pigment epithelium (RPE) and fluorescein angiography demonstrated a hyper/hypoautofluorescent speckled pattern without dark choroid sign. Full-field electrorretinogram showed loss of cone function and multifocal electroretinogram showed a decreased response at paramacular area. Genetic testing identified two pathogenic variants in the ABCA4 gene with an autosomal recessive inheritance pattern, confirming the diagnosis of FF as an ABCA4-R variant.

CONCLUSIONS: Stargardt disease and FF belong to a heterogeneous group of hereditary disorders caused by mutations in the ABCA4 gene. ABCA4-R have a wide clinical spectrum, and several inherited macular dystrophies have phenotypic similarities that could make clinical diagnosis challenging. FF is characterized by the presence of flecks across the posterior pole, few areas of RPE atrophy and preserved visual acuity associated with foveal sparing, as in our patient. FF has characteristically a late-onset of the disease and slower visual impairment. Multimodal imaging and electrophysiology provide additional information to help support diagnosis. Currently, there is no treatment for FF, but early diagnosis based on clinical features, multimodal imaging, and molecular genetics is essential for genetic counseling.