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MULTIMODAL IMAGING CHARACTERIZATION OF DIFFERENT PHENOTYPES OF ABCA4-RELATED RETINOPATHY

Oral

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Purpose:

To analyse, by means of multimodal imaging, the retinal and choroidal features of different phenotypes of ABCA4-related retinopathy, including cone dystrophy (COD), cone-rod dystrophy (CORD), Stargardt disease (STGD), late-onset Stargardt (LO-STGD) and fundus flavimaculatus (FFM)

Methods:

Our study was designed as observational and cross-sectional. We included 46 consecutive patients (92 eyes) with genetically-confirmed ABCA4 retinopathy. Patients were assigned to the different ABCA4 phenotypes on the basis of clinical and electrophysiology data. They underwent visual acuity, fundus photography, optical coherence tomography, blue-light and near-infrared autofluorescence and 6x6-mm swept-source optical coherence tomography angiography. The following variables were analyzed: central subfoveal and choroidal thickness (CST, SFCT) and atrophy according to the Classification of Atrophy Meeting on OCT; the extent of retinal pigment epithelium (RPE) atrophy and the shape of retinal flecks using autofluorescence; choriocapillaris flow deficits (FID) on SS-OCTA

Results:

Overall, 13 patients were affected by COD, 3 by CORD, 10 by STGD, 13 by LO-STGD and 7 by FFM. Mean age was 45.8 ± 18.7 years, while mean VA was 51.7 ± 23.5 letters. CORD patients had the worst vision, whereas FFM and LO-STGD patients had higher VA (all $p < 0.05$).

Thirty-five patients had RPE atrophy (5.9 ± 8.4 mm²) which was larger in LO-STGD eyes ($p < 0.05$). Flecks were detected in 85 eyes (93%) with resorbed flecks more frequent in LO-STGD and CORD. Lastly, patients with LO-STGD had significantly lower SFCT and larger choriocapillaris FID than other groups ($p < 0.05$) with the exception of CORD ($p = 0.12$).

Conclusions:

The phenotypes of ABCA4 retinopathy show different clinical and imaging characteristics. COD and STGD patients show mostly photoreceptors' alterations, while CORD and LO-STGD patients carry significant RPE and choriocapillaris damages. These might be important outcome measures to consider for future interventional studies in ABCA4 retinopathy.