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SPECTRAL-DOMAIN OPTICAL COHERENCE TOMOGRAPHY ANALYSIS IN SYNDROMIC AND NONSYNDROMIC FORMS OF RETINITIS PIGMENTOSA DUE TO USH2A GENETIC VARIANTS

Oral

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Purpose:

This study aimed to analyze macular structure by using spectral-domain optical coherence tomography (SD-OCT) in a cohort of patients affected by autosomal recessive retinitis pigmentosa and Usher syndrome, due to genetic variants in USH2A gene, and to correlate optical coherence tomography (OCT) parameters with functional and genetic data.

Methods:

The subjects of this study were 92 patients, 46 syndromic (Usher syndrome type IIa [Ush2]) and 46 nonsyndromic (autosomal recessive RP [arRP]), with clinical and genetic diagnosis of USH2A-related retinal dystrophy, who underwent a complete ophthalmic examination and spectral-domain OCT analysis. The study focused on evaluating the differences between the 2 groups in the following parameters: best-corrected visual acuity (BCVA), ellipsoid zone (EZ) width, presence of epiretinal membrane (ERM), and cystic macular lesions (CMLs). Variants in USH2A gene were divided into 3 categories, according to the expected impact (low/high) at protein level of the different variants on each allele.

Results:

BCVA and EZ width were significantly lower in Ush2 than in arRP patients ($p<0.0001$ and $p=0.001$). ERM was detected in 34.8% (16/46) of arRP patients and in 65.2% (30/46) of Ush2 patients ($p=0.003$). CML was detected in 17.4% (8/46) of arRP patients and 30.4% (14/46) of Ush2 patients ($p=0.14$). The allelic distribution was statistically different ($p=0.0003$) by dividing the 2 diseases: for Ush2 patients it was 45.7% (high/high), 39.1% (low/high) and 15.2% (low/low); for arRP patients it was 8.7% (high/high), 56.5% (low/high), and 34.8% (low/low). The severity class of the variants significantly affected visual acuity and EZ width parameters.

Conclusions:

Retinal disease, as evaluated by means of SD-OCT, shows more advanced degeneration signs in the syndromic than the non-syndromic form of retinal dystrophy related to USH2A gene. However, since the 3 allelic profiles can be found in both Usher and RP patients, other factors must necessarily play a determining role.