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ARVO Annual Meeting Abstract | June 2022

Clinical, structural, and functional characteristics of *RPE65* mutation related Leber congenital amaurosis in a Mexican cohort

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Investigative Ophthalmology & Visual Science June 2022, Vol.63, 617 – A0332. doi:

Abstract

Purpose : To describe the clinical, structural, and functional features of *RPE65* mutation related Leber congenital amaurosis (LCA) in a Mexican cohort.

Methods : A descriptive, observational, and transversal study was carried out. Twelve unrelated patients with molecularly and clinically diagnosed *RPE65*-LCA were included. Visual function studies included best-corrected visual acuity (BCVA), refraction testing, Farnsworth-Munsell D-15 test, full-field electroretinography (ERG), Full Field Stimulus Threshold test (FST), Goldmann kinetic visual field, chromatic perimetry, and microperimetry. In addition, spectral domain optical coherence tomography (SD-OCT) and autofluorescence (AF) imaging (488 nm and 532/633 nm) were performed in all patients. Measures of central tendency were utilized to analyze the data.

Results : Twenty-four eyes of 12 patients (ages 10-32) were analyzed. The BCVA ranged from 20/50 (snellen) to light perception and the average was of 1.09 ± 0.75 (logmar). There were no detectable rod and cone ERGs in all patients. By FST (analyzed in 17 eyes), 82% (n=14) presented a rod- and cone-mediated function while the remaining 18% (n=3) presented only a cone-mediated function. The extent of kinetic fields varied widely regardless of the age and only 25% (n=6) of the eyes detected a III4e stimulus (range of 5-70 degrees of remaining central vision). In the chromatic perimetry, the mean global deficit of rod- and cone-mediated function was of 46.8 ± 9.1 dB and of 31.0 ± 7.1 dB respectively. Absent (n=20) or minimal (n=4) AF was observed in both short and long wavelength AF images. On average, the horizontal extension of the ellipsoid zone (EZ) was of 3366 ± 3047 µm and the subfoveal outer nuclear layer (ONL) thickness was of 53 ± 29 µm. In the microperimetry (evaluated in 8 eyes), the preferred retinal loci were situated mainly in the fovea-parafovea and superior perifovea.

Conclusions : Accurate diagnosis of *RPE65*-LCA remains the foremost goal since it allows to stage the degree of retinal degeneration, estimate the prognosis, and tailor the treatment addressing the characteristics of each patient. Our study contributes to the understanding of the genotype-phenotype correlations of this disease in the Mexican population and accentuates the importance of undertaking a multimodal evaluation to identify patients who may benefit from gene therapy.

This abstract was presented at the 2022 ARVO Annual Meeting, held in Denver, CO, May 1-4, 2022, and virtually.