( ) R1 ( ) R2 ( ) R3 ( ) PG0 (X) PG1 ( ) Estagiário ( ) Tecnólogo ( )
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Service (sector) Neuro-Ophthalmology Nº CEP

Large optic discs may maintain mutation carriers as unaffected in an extended Brazilian pedigree with Leber's hereditary optic neuropathy. AUTHORS: Carolina do V.F. Ramos, MD, Piero Barboni, MD, Valerio Carelli, MD, PhD, Costantino Bellusci, MD, Giacomo Savini, MD, Anna Maria De Negri, MD, Federico Sadun, MD, Adriana Berezovsky, PhD, Celina Tamaki, PhD, Milton N. de Moraes Filho, MD, Rafael Cinoto, BA, Paula Y. Sacai, COMT, Hevillin M. de P.P. Miura, COMT, Rubens Belfort Jr, MD, PhD, Alfredo A. Sadun, MD, PhD, Solange R. Salomao, PhD., et al. **Purpose:** To investigate retinal nerve fiber layer thickness (RNFL) and optic nerve head (ONH) morphology in a very large family from Brazil with Leber's hereditary optic neuropathy (LHON) carrying the 11778/ND4 mutation. **Methods:** We enrolled 139 individuals belonging to the previously reported Brazilian SOA-BR LHON pedigree, divided in the following subgroups: 13 LHON affected patients, 48 LHON unaffected mutation carriers and 78 controls (off pedigree spouses and descendants of males who do not carry or inherit respectively the LHON mutation). The RNFL thickness (3.4) and fast optic disc scan protocol by optical coherence tomography (OCT) were used. **Results:** The RNFL thickness analysis showed a significant increase in the temporal and inferior quadrants as well as in the 360° average in the unaffected carriers group compared to controls. This RNFL thickening was particularly significant in unaffected male carriers. Affected individuals had a drastic reduction of RNFL thickness in all measurements. ONH topographic analysis showed a significantly larger disc area and vertical disc diameter in unaffected carriers compared to controls, whereas LHON affected patients did not show a statistical difference with controls. Rim area was also larger in unaffected carriers and reduced in affected patients compared to controls. Gender stratification in unaffected carriers showed that significance for ONH measurements was mantained in males and lost in females. **Conclusions:** Our study shows for the first time that anatomical conformation of the optic nerve head may be an important determinant of disease penetrance and large optic discs are a protective factor. This is more significant for males. We propose that if these data, obtained in a single large family, are a general feature in LHON, the study of optic nerve head by OCT may have a prognostic significance for risk of being affected.