

2007 Research Days Abstract Form – Department of Ophthalmology – UNIFESP/EPM

2. SCIENTIFIC SECTION PREFERENCE (REQUIRED): Review the Scientific section Descriptions. Select and enter the two-digit Code for the one (1) Section best suited to review your abstract
NO

3. PRESENTATION PREFERENCE (REQUIRED) Check one (1)
(a) Paper
 (b) Poster

4. The signature of the First (Presenting) Author. (REQUIRED) acting as the authorized agent for all authors, hereby certifies.
 That any research reported was conducted in compliance with the Declaration of Helsinki and the UNIFESP Ethical Committee*

Carolina Ramos
 Signature of First

- Scientific Section Descriptions
- (OR) ORBIT
 - (PL) OCULAR PLASTIC SURGERY
 - (RE) RETINA / VITREOUS
 - (RX) REFRACTION-CONTACT LENSES
 - (NO) NEURO-OPHTHALMOLOGY**
 - (TU) TUMORS AND PATHOLOGY
 - (ST) STRABISMUS
 - (UV) UVEITIS
 - (LS) LACRIMAL SYSTEM
 - (LV) LOW VISION
 - (CO) CORNEA / EXTERNAL DISEASE
 - (GL) GLAUCOMA
 - (RS) REFRACTIVE SURGERY
 - (CA) CATARACT
 - (US) OCULAR ULTRASOUND
 - (TR) TRAUMA
 - (LA) LABORATORY
 - (BE) OCULAR BIODESIGNING
 - (EP) EPIDEMIOLOGY
 - (EF) ELECTROPHYSIOLOGY

Deadline: 29/10/2007

FORMAT:
 Abstract should contain:
Title, Name of Authors, Name of other authors (maximum 6), Purpose, Methods, Results, Conclusions.
 Example: ARVO (1.10 x 1.70) Abstract Book

1. FIRST (PRESENTING) AUTHOR (REQUIRED)
 Must be author listed first in body of abstract

() R1 () R2 () R3
 () PG0 **(X) PG1** () Estagiário () Tecnólogo () PIBIC

__Ramos__ Carolina__ do Val Ferreira

Last Name First Middle

__Neuro-oftalmologia__ 1352/05
 Service (sector) N° CEP

LARGE OPTIC DISCS MAY MAINTAIN MUTATION CARRIERS AS UNAFFECTED IN AN EXTENDED BRAZILIAN PEDIGREE WITH LEBER'S HEREDITARY OPTIC NEUROPATHY

Carolina V.F. Ramos ¹, Piero Barboni ², Valerio Carelli ³, Anna Maria De Negri ⁴, Federico Sadun, Adriana Berezovsky ¹, Rafael W. Cinoto ¹, Rubens Belfort Jr ¹, Alfredo A. Sadun, Solange R. Salomão

1 Department of Ophthalmology, Federal University of São Paulo, UNIFESP, São Paulo, Brazil; 2 Fondazione G.B. Bietti -IRCCS, Roma, Italy; 3 Dipartimento di Scienze Neurologiche, Università di Bologna, Bologna, Italy; 4 Azienda San Camillo -Forlanini, Rome, Italy; 5 Ospedale San Giovanni, Tivoli, Italy; 6 Doheny Eye Institute, University of the Southern California, Los Angeles, CA, USA.

Purpose: To investigate retinal nerve fiber layer thickness (RNFL) and optic nerve head (ONH) morphology in a 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 (LHON-affected), 48 LHON unaffected mutation carriers (LHON-carrier) and 78 controls (26 off maternal lineage spouses and 52 descendants of males who do not carry or inherit respectively the LHON mutation). All individuals underwent optical coherence tomography (OCT) measurements including RNFL thickness and ONH analysis.

Results: The RNFL thickness analysis showed a significant increase in the temporal and inferior quadrants as well as in the 360° average in the LHON-carrier compared to controls. This RNFL thickening was particularly significant in male LHON-carrier. LHON-affected had a drastic reduction of RNFL thickness in all measurements. ONH topographic analysis showed a significantly larger optic disc area and vertical disc diameter in LHON-carrier compared to LHON-affected and controls, whereas LHON-affected did not show a statistical difference with controls.

Conclusions: Our study confirms the previously reported pattern of RNFL changes in LHON. Further, we show that anatomical conformation of ONH may be an important determinant of disease penetrance, large optic discs being a putative protective factor. If these data, obtained in a single large family, are a general feature in LHON, the ONH investigation by OCT may have a prognostic significance for risk of being affected.