S5.003 - Retinal and Visual Cortex Damage and Higher Order Visual Dysfunction in Carriers of E46K Mutation of alpha-Synuclein Gene

April 16, 2016, 1:30 - 1:45 PM

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Disclosures

Abstract
Objective: To identify retinal and visual cortex imaging variables associated to higher order visual dysfunction (HOVD) in carriers of E46K mutation of alpha-synuclein gene (E46K-SNCA), dementia with Lewy bodies (DLB) and Parkinson's disease (PD).

Background: E46K-SNCA is a rare genetic model of LB disease with early and prominent visual and cognitive manifestations. HOVD is associated to the development of dementia in LB spectrum disorders, which have proven damage of the retina and visual cortex.

Methods: Cross-sectional evaluation of sex/age matched 48 participants: n=7 E46K-SNCA [4 motor parkinsonism, 1 autonomic neurovascular symptoms and 2 asymptomatic], n=19 PD, n=3 DLB and n=19 healthy controls (HC).

Measures: 1) Retinal OCT (Spectralis): bilateral average thickness of macula and its layers; 2) Brain MRI (3T): visual and frontal cortex NAA levels and thickness; 3) Neuropsychological measures for global cognition and HOVD (visual attention, processing speed and memory and visuoperceptive and visuoconstructive tasks) 4) PD clinical features.

Results: Compared to HC and PD, symptomatic E46K-SNCA and DLB had statistically significant poorer results in all HOVD measures, in macula thinner ganglion cell-inner plexiform layer complex (GCIPL), inner nuclear layer (INL) and outer nuclear layer (ONL) and thicker photoreceptors outer segments layer (PROSL) and in visual cortex lower absolute NAA levels. Analyzing all patients together, higher atrophy of INL and ONL, thicker PROSL and lower NAA levels in visual cortex were significantly associated in patients (and not in HC) to poorer performance in HOVD measures.

Conclusions: HOVD is associated to a specific damage of retina and visual cortex in LB spectrum disorders. This injury is stronger in E46K-SNCA and DLB than in PD, which may support the existence of an agressive PD phenotype more purely driven by LB pathology.

Study support: MJFF (RRIA 10189) and ISCIII (PI14/00679)