The New Zealand Genetic Frontotemporal Dementia Study (FTDGeNZ): Baseline retinal characteristics

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BACKGROUND

The key to combating dementia is early detection. Prospective longitudinal studies to search for pre-clinical dementia are required.

- Genetic FTD provides an opportunity to identify pre-clinical markers because the causative mutation can be identified decades before expected symptom onset.

- FTDGeNZ is one of the largest, single-family longitudinal FTD studies internationally.

- OCT imaging has been shown to be a marker of disease progression in other neurodegenerative disorders.

- There are reports of retinal layer thinning in patients with sporadic FTD, and individuals with pre-symptomatic FTD caused by progranulin mutations.1,3

- Cerebral hypoperfusion has been documented in patients with FTD.

OBJECTIVE

Identify retinal biomarkers of pre-clinical FTD in a longitudinal cohort study of a kindred with genetic FTD caused by a tau mutation.

METHODS

- Genotyping of FTDP-17 cohort:
  - Isolated DNA from 500 µl buffy coat (Genetra Purgene Blood Kit, Qiagen)
  - Sanger sequencing (ABI 3130XL Genetic Analyzer) using custom primers
  - Validated using a TaqMan Genotyping Assay specific to SNP IDrs6375101

- Carrier participants pre-symptomatic at baseline, based on neurological and neuropsychological examination

- Comprehensive neuro-ophtalmic examination including SD-OCT (Zeiss Cirrus™) and OCT-A (Angioplex®)

- Support vector machine (SVM) image analysis of vessel density and vessel distribution

- One eye of each participant used in analysis

RESULTS

- Included in analysis were 6 carriers and 18 non-carrier controls.

- Both groups had similar baseline ophthalmic and refractive characteristics (Table 1).

- There were no statistically significant differences between carriers and non-carriers for peripapillary and macular OCT and OCT-A measures (Tables 2 and 3).

- These findings were consistent across age-matched paired T-test analysis

- SVM analysis was not sufficient to determine the genetic status of participants.

REFERENCES


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