Research Article

Pupillometric findings in ATTRv patients and carriers: results from a single-centre experience

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Abstract

Introduction

Hereditary transthyretin amyloidosis (ATTRv) is a treatable multisystemic disease with great phenotypic heterogeneity. Among extra-neurological features, pupillary abnormalities have been reported, either related to amyloid deposition in the eye or to a progressive autonomic neuropathy.

Objective

To evaluate the role of automated pupillometry, a non-invasive and rapid test able to provide objective and reproducible data on pupil size and reactivity, as a marker of disease severity in late-onset ATTRv patients.

Patients and methods

We performed automated pupillometry on a cohort of ATTRv patients and pre-symptomatic TTR mutation carriers and compared results to healthy controls. An exhaustive clinical and instrumental evaluation was performed on all enrolled subjects.

Results

A statistically significant difference in most pupillometry parameters was found in ATTRv patients as compared to both carriers and healthy controls. Moreover, in ATTRv patients, we found a significant correlation between many pupillometry findings and disease duration, as well as widely accepted clinical scales and investigations (NIS, Sudoscan from feet, and Norfolk QoL-DN questionnaire).

Conclusions

We suggest pupillometry may play a role as a reliable and non-invasive biomarker to evaluate ATTRv disease severity and monitor its progression.

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