EWGGD Abstract

Introduction

Saccadic eye movement abnormalities are a consistent clinical feature of neuronopathic disease. Here we evaluated saccadic eye movement parameters using the EyeSeeCam with a view to demonstrating utility of the tool in this cohort. We replicated earlier findings but further identified an unexpected difference in saccadic parameters between patients with specific *GBA1* variants.

Patient & Methods

Sixty patients in the UK with Gaucher disease were recruited; 21 with type 3 disease, 39 with type 1 disease, an opportunistic sample of 35 volunteer healthy controls (HC) were recruited from a single research centre. All underwent clinical examination of eye movements followed by assessment using the video-oculography tool EyeSeeCam with measurement of saccade amplitude, peak velocity, duration, and latency and gain.

Results

We confirmed the saccadic abnormality in patients with type 3 Gaucher disease by comparing mean parameters with healthy controls and type 1 Gaucher patients. On comparison of patients with type 1 Gaucher disease and healthy controls, a significant difference in a range of parameters was identified. Those patients with type 1 Gaucher disease who deviated more than 2 SD from HC means for saccadic velocity or duration were reviewed and found to all share the R463C (p.502) *GBA1* variant. On re-analysis of the whole cohort with consideration of this variant a significant difference in mean peak horizontal velocity at 30° (amongst other parameters), between patients with and without R463C was seen, figure 1.

Conclusions

This unexpected finding suggests there is greater genotype-phenotype correlation than previously observed and further phenotyping of patients with this common variant is warranted to establish the true clinical relevance of the saccadic differences identified.