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BRIEF COMMUNICATION

Optic disc drusen prevalence in the retinitis pigmentosa population

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INTRODUCTION

The reported prevalence of optic disc drusen (ODD) ranges from 0.3–2.4% in the normal population [1]. ODD can be visible or buried and this feature may partly explain the variation in prevalence data [1]. Furthermore, the assessment technique can contribute to the rate at which ODD are identified; autofluorescence (AF), OCT, B-scans, and histopathology are more sensitive in detecting buried ODD than clinical examination alone [1]. A recent study of ODD in the normal population involving histopathology put the prevalence of ODD at the higher end of the range at 1.8% [2].

Retinitis pigmentosa (RP) has been associated with ODD and the prevalence of ODD in an RP population ranges broadly from 3–80% [3, 4]. The higher rates in this range, however, were identified in small populations affected by the less common RP subtypes of preserved para-arteriole RPE & Usher's syndrome [3, 5]. More recent studies of ODD in the RP population have shown rates of 2.95–3.6% [4, 5]. This retrospective study was undertaken to further investigate the prevalence of ODD using AF in an RP population.

METHODS

A database of RP patients at Save Sight Institute, confirmed by electrophysiology between 2015 to 2020, was retrospectively examined. Clinical notes, fundus photography, and AF were assessed in each patient for ODD. Two clinicians examined fundus photographs for ODD as defined by focal optic disc hyperfluorescence on AF. Genetic testing results were not available for all patients but were included when possible.

RESULTS

A total of 200 patient files were reviewed with 5 excluded as AF images were unavailable. There were a total of 101 women (51.8%) and 94 men (48.2%). The median age of patients was 38 (range: 7–84) years. Six patients (3.1%) had ODD of which two had bilateral ODD. Four cases (66.7%) of ODD were male and two (33.3%) were female. Three cases were X-linked with an identified RPGR gene and three cases did not have genetic testing completed. Fifteen patients with Usher syndrome were examined and none demonstrated ODD. No individuals had preserved para-arteriole RPE RP.

DISCUSSION

Our RP population showed an ODD rate of 3.1% which is consistent with the lower range of ODD prevalence. These results are more in keeping with the normal population prevalence of ODD within an RP population. Despite using AF, an identification method that would increase our detection of buried drusen, our results remained close to the lower range of ODD prevalence in RP. Our cohort of Usher's syndrome patients showed no cases of ODD which is not consistent with their reportedly high rates (7–35%) of ODD [5]. The X-linked RPGR associated RP cases made up 50% of our ODD cases. Currently, there is no established significant difference in ODD prevalence between autosomal dominant, autosomal recessive, or X-linked RP [1]. The variation in ODD prevalence across studies may be explained by particular subtypes of RP having stronger genetic associations with ODD. The link with RPGR is worth further investigation.

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AUTHOR CONTRIBUTIONS

NR was responsible for data collection and processing, interpreting and analysing results, and writing the report. CF was responsible for study design, interpreting, and analysing data and results, and providing feedback on the report. JG was responsible for study design, interpreting and analysing data, and providing feedback on the report.

COMPETING INTERESTS

JG acts as a consultant for Novartis. There are no other disclosures to report.

ADDITIONAL INFORMATION

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