Background

Inherited retinal diseases (IRD) are a leading cause of blindness in the working-age population. Although research into new treatments is underway, their translation into approved treatments has been limited by the lack of sensitive, reliable, and patient-relevant visual function outcome measures. The standard clinical visual assessments, particularly visual acuity, are poorly suited to detecting early or functionally meaningful vision changes in IRD. There has therefore been a desperate need to identify and validate alternative visual function measures that better reflect disease progression and treatment effects.

Aims and objectives

The primary aim of this study was to assess the clinical utility of degeneration-targeting visual function tests in patients with IRD. Specific objectives were to evaluate the reliability, reproducibility, and discriminatory ability of selected visual function tests; to compare these measures with standard assessments and patient-reported outcomes; and to explore patient experiences of testing to inform recommendations for research delivery and clinical care.

Methods

This was a cross-sectional observational study conducted at a specialist tertiary referral eye hospital. Eighty-one participants were recruited, comprising 40 patients with genetically confirmed IRD and 41 age-matched healthy controls. Participants completed a series of visual function assessments, including standard visual acuity, low luminance visual acuity, alternative acuity charts, mesopic microperimetry, scotopic microperimetry, and validated patient-reported outcome measures. A subset of participants completed repeat testing to assess test–retest reliability and reproducibility. A qualitative component was undertaken using semi-structured interviews with a purposively sampled group of participants to explore experiences, acceptability, and perceived burden of testing. Following early review of the qualitative data and feedback from the Patient and Public Involvement (PPI) group, an inductive qualitative approach was adopted to generate patient-centred recommendations for improving research delivery and clinical care.

Results

The study demonstrated that mesopic microperimetry showed greater sensitivity and discriminatory ability than visual acuity tests and scotopic microperimetry in patients

with IRD. It also showed more reliable performance than scotopic microperimetry. No greater benefit was found with the alternative acuity tests compared to standard visual acuity. Findings highlighted the importance of test standardisation and protocol refinement to improve consistency and interpretability for use in clinical trials. Qualitative findings identified key factors influencing patient experience, including test duration, fatigue, communication, and the need for rest breaks during complex assessments. These findings informed practical recommendations to improve the accessibility, acceptability, and feasibility of visual function testing in both research and clinical settings.

Outputs and impact

The study has generated 10 peer-reviewed publications (with two more in peer review and 1 more in development) and several national and international conference presentations targeting optometrists, ophthalmologists, allied health professionals and commercial stakeholders. The outputs provide an evidence base to support improved outcome measure selection, standardisation, and patient-centred testing approaches in inherited retinal disease research. Collectively, the findings have relevance for future clinical trial design, regulatory decision-making, and clinical practice, with the potential to support more robust evaluation of emerging sight-saving therapies.