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Measures of Function and Structure to Determine Phenotypic Features, Natural History, and Treatment Outcomes in Inherited Retinal Diseases

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Abstract

Inherited retinal diseases (IRDs) are at the forefront of innovative gene-specific treatments because of the causation by single genes, the availability of microsurgical access for treatment delivery, and the relative ease of quantitative imaging and vision measurement. However, it is not always easy to choose a priori, from scores of potential measures, an appropriate subset to evaluate efficacy outcomes considering the wide range of disease stages with different phenotypic features. This article reviews measurements of visual function and retinal structure that our group has used over the past three decades to understand the natural history of IRDs. We include measures of light sensitivity, retinal structure, mapping of natural fluorophores, evaluation of pupillary light reflex, and oculomotor control. We provide historical context and examples of applicability. We also review treatment trial outcomes using these measures of function and structure.