# KEEP UP WITH THE IRD PATIENT STANDARD OF CARE

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With many gene therapies under evaluation in clinical trials for rare retinal conditions and one FDA-approved retinal gene therapy, genetic testing for inherited retinal disease (IRD) has become increasingly important for us in the clinic. In fact, our editors (along with the AAO) would argue that offering or facilitating genetic testing for monogenetic IRD patients is now a part of the standard of care. Hower, genetic testing for IRDs remains a complex and nuanced process that requires significant time and expertise to execute properly.

It also takes time and training to review and synthesize the clinical trial data for retinitis pigmentosa, Stargardt, Usher syndrome, choroideremia, Leber congenital amaurosis, and any number of other rare retinal conditions—whether it's natural history studies or investigations of novel therapies.

All this effort on the clinician's part is necessary to ensure they can properly advise patients with an IRD on potential clinical trials, which differs widely depending on the patient's genetic diagnosis and stage of disease.

Now, recently released FDA guidance adds further nuance to the field. The draft guidance is designed to help developers use existing scientific and regulatory knowledge to more efficiently bring promising gene therapies to patients. The document acknowledges, "The ability to leverage prior knowledge to expedite product development may be particularly helpful in the context of GE [genome editing] products intended to treat rare diseases, many of which may be serious and life threatening."[1]

Needless to say, IRD is not an easy field, and it's not for everyone. However, all retina specialists see patients with suspected or diagnosed IRD at some point or another, and knowing how to care for these patients is imperative.

To that end, this issue is designed to help bolster your clinical acumen regarding the management of IRDs, including genetic testing, the challenges of specific patient populations, diagnosing rare IRDs, and clinical trials for patients with IRDs. Sidney A. Schechet, MD, and his team provide a high-level look at genetic testing in the clinic, noting that it can take up to 15 years for some patients to receive a correct diagnosis.[2]

Other articles touch on managing IRDs in children (by Ramiro Maldonado, MD, and his colleagues at Duke) and the promise of gene-agnostic therapies (by an international team of researchers from Turkey and New York, led by Stephen H. Tsang, MD, PhD). Breaking from the clinical side of things, Melissa Yuan, MD, and Ahmad Al Moujahed, MD, PhD, discuss the challenge of IRD clinical trial endpoints, including the search for endpoints that better assess visual changes in patients with low vision. Lastly, several teams came together to provide a challenging mystery case article that will test your diagnostic acumen.

Whether genetic testing, clinical characterization of disease progression, and clinical trial candidacy is part of your practice or not, it's certainly something you must have a general understanding of to deliver a positive message and facilitate the best referral to an IRD specialist. We hope this issue boosts your confidence in understanding some of the complexities in caring for patients with an IRD and adds to the armamentarium of your clinical practice. ■

1. Leveraging prior knowledge in the development of human gene therapy products incorporating genome editing. FDA. June 2026. Accessed June 10, 2026. tinyurl.com/decww9f7
2. Shah AH, Park E, Luke T, Xu Q, Jewell A, Couser NL. The role of genetic testing in avoiding diagnostic delays in inherited retinal disease. [published online ahead of print April 27, 2024]. *Retin Cases Brief Rep.*

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