WHEN RARE IS RELATIVE


When you specialize in inherited retinal diseases (IRDs), it can be easy to lose perspective of how rare these conditions are. Those of us who manage IRDs see these patients frequently and routinely spot clinical signs of retinitis pigmentosa (RP), classic Stargardt disease, and Leber congenital amaurosis (LCA). However, even these more common conditions do not present to all retina practices often. After all, RP has a prevalence of approximately one in 4,000; that's only 110,000 patients in the United States.[1] Compare that to the nearly 20 million patients diagnosed with AMD.[2]

When we step away from our IRD clinics and engage with the retina community, we are reminded how uncommon these diagnoses are. However, the latest news and conferences bring IRD research and innovation into the spotlight. During one session at this year's Atlantic Coast Retina Club, half of the case presentations were IRDs. In the first half of 2024, 16% of the news stories that ran on Eyewire+ were focused on IRDs; since January, at least 11 companies have announced updates to their IRD clinical trial programs.[1,3-15]

With the advent of gene and cell therapies, IRDs have become an important focus for researchers. Although voretigene neparvovec-rzyl (Luxturna, Spark Therapeutics) for *RPE65*-associated LCA is the only approved genetic therapy for an IRD, many companies are working their way through clinical trials for various inherited conditions.

This issue of *Retina Today* is an ode to the innovation happening in the IRD space. Cristy A. Ku, MD, PhD, and her team at Baylor provide a roundup of gene therapy trials (nearly 30!), while Ishrat Ahmed, MD, PhD, and Mandeep S. Singh, MD, PhD, summarize novel approaches, such as stem cell therapy, optogenetics, and retinal prosthesis. Elizabeth Kellom, MS, CGC, and Kimberly Stepien, MD, explore various genetic testing approaches and what to do if the results are inconclusive. Jonathan F. Russell, MD, PhD, and his colleagues at the University of Iowa discuss managing IRD patients who present with complications, and Kevin C. Allan, MD, PhD, and Alex Yuan, MD, PhD, look at various imaging techniques. Shima Dehghani, MD, and Stephanie M. Llop, MD, highlight clinical trials in uveitis, another rare retinal condition with a robust therapy pipeline—they touch on 16 ongoing phase 3 and 4 trials. Finally, a can't-miss article in this issue is a mystery case compilation to test your diagnostic acumen.

Although the IRD patient population is small compared with other retinal conditions, the research on novel therapeutic approaches is not. Patients with an IRD may feel isolated because of their diagnosis, yet hundreds of researchers and clinicians are working hard to bring new therapies to market. *Rare*, in this case, is a relative term. For those of us working in the IRD space, it doesn't feel as if their condition is all that rare with so much buzz about genetic testing, imaging, monitoring, and (one day) treatment. We should share this enthusiasm and research momentum with our patients; if nothing else, it helps them find hope—and perhaps a clinical trial and potential cure. ■

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- Financial disclosure: Clinical Trial Support (Beacon, Biogen/Nightstar, Editas, Foundation Fighting Blindness, Sanofi); Consultant (4D Molecular Therapeutics, AGTC, Aldebaran, Aldeyra, Alia, Alkeus, Arrowhead, Ascidian, Atsena, Beacon, Biocryst, BlueRock, Coave, Dompé, Editas, Endogena, Foundation Fighting Blindness, FREST, GenKore, Ingel, Horama, Iveric Bio/Astellas, J-Cyte, Janssen, Kala, Kiora, Nacuity, Ocugen, Ora, Prime Editing, ProQR, PTC, PYC, Ray, RestoreVision, Regenexbio, Roche, Sparing Vision, SpliceBio, Spotlight, Thea, Theranexus, ZipBio); Data and Safety Monitoring Board (Akous, Gensight); Equity (Aldebaran, Atsena, Endogena, EnterX, Ingel, Kiora, Nacuity, Ocugen, ZipBio)