IRDS AROUND THE WORLD

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There are disparities in health care access present in every country around the world. But sometimes, the contrast in access is stark, particularly in the case of rare conditions. In this issue, focused on rare and inherited retinal diseases (IRDs), we highlight the progress that has been made in genetic testing and IRD clinical trials. Hossein Ameri, MD, PhD, and Luciano C. Greig, MD, PhD, who are practicing in Los Angeles, note that identifying gene mutations has become a part of routine clinical care "thanks to the low cost and widespread availability of gene panel testing." (Check out *The Latest in Gene Therapy Clinical Trials for IRD* for their excellent rundown of the robust pipeline of therapeutics under investigation.)

The examples of genetic testing used in clinical practice can be found in this issue's *Retina Detectives: Mystery Cases*. The article is a wonderful demonstration of the challenges of making a clinical diagnosis for IRDs—for example, when polled on social media, only 18% of retina specialists got the diagnosis correct for case no. 1. It's also fun to test your diagnostic acumen and look over some very interesting clinical images. If you want to know *how* to order genetic testing, Jennifer Huey, MS, CGC, and Debarshi Mustafi, MD, PhD, share their tips and tricks in *Order Genetic Testing Like a Pro*.

However, on the other side of the world, it's a different story. Paul Runge, MD, has been volunteering in Ukraine and was surprised to find himself treating an unusually high number of patients with IRDs (see *Global Retina Care: The Ukraine Experience*). He agrees that genetic testing *should* be routine, but it's neither widespread nor low cost in Ukraine. In fact, "there is no genetic testing available in our area, and I have had families travel to Italy or Poland for genetic testing," he explained in an interview. Now that a confirmed genetic diagnosis could provide access to a clinical trial or even treatment options, "it is essential that genetic testing be made available to our patients," he added. This is just one of many somewhat surprising hurdles he is working to address with Ukrainian colleagues.

In other countries, such as those in West Africa, the most advanced technology available for an eye examination may be only a vision chart, penlight, and direct ophthalmoscope (read more in *Global Retina Care: Uveitis in West Africa*). These tools are useful for identifying cataracts, corneal scarring, and other anterior segment findings, but they can limit a clinician's ability to diagnose early signs of another relatively rare group of conditions, different types of uveitis, which is prevalent in this population. Steven Yeh, MD, and Jessica G. Shantha, MD, MSc, have been working in Sierra Leone with Lloyd Harrison-Williams, MD, and Jalikatu Mustapha, MD, to meet this challenge head-on, particularly as it relates to patients with Ebola virus disease.

It's disparities such as these that keep us humble and remind us that, although we have come so far in the field of retina, there is much left to do. In the United States and Europe, even with multiple clinical trials moving forward for retinitis pigmentosa, Leber congenital amaurosis, Usher syndrome, and other IRDs, many other rare diseases affect patient populations so small that a clinical trial isn't feasible. Those patients deserve our best, too, and Drs. Ameri and Greig call upon regulators to one day update the regulatory framework so that researchers can more easily adapt successful gene delivery platforms to provide custom treatment options for rare mutations.

Only time will tell which, if any, gene therapies will make it to market for various IRDs. In the meantime, we can work toward a much more tangible goal: improving access to genetic testing, regardless of our geographical location. As several experts make clear in this issue, a genetic diagnosis can guide patient care, reduce the risk of complications, and, for some, offer hope of treatment. And hope is what it's all about with IRDs. ■