"She had genetic testing—but no follow-up"

PHYSICIAN: **Christine Kay, MD,** Vitreoretinal Associates, Gainesville, FL, and Affiliate Assistant Professor of Ophthalmology at the University of South Florida, Tampa

PATIENT: **Emily,** age 5, was referred to me by a pediatric ophthalmologist to confirm a suspected diagnosis of IRD.

Referral for genetic testing: Emily's symptoms started her first year of life when her parents noticed she wasn't properly focusing on things. In 2014, she was referred to an academic institution 4 hours away. There, specialists performed an ophthalmic exam and full-field electroretinogram (ERG). The report listed a suspected variant of Leber congenital amaurosis (LCA), a rare genetic disorder marked by reduced visual acuity and night vision, nonrecordable ERG and nystagmus, among other symptoms. The family was advised to have genetic testing but their insurance wouldn't cover it, so a university-based nonprofit lab ran Emily's genetic panel for free.

After that, it was unclear what happened, but records show the test sample was processed with the results back 5 months later. However, Emily's family never got the report. Maybe the lab never faxed it or the academic institution couldn't reach the family. Regardless, time went by and the family likely got caught up in daily life. In 2017, the parents took Emily to a pediatric ophthalmologist and asked if he could help them get the test results. Because he wasn't the ordering physician, these records could be difficult to obtain, but he told the family he'd try and, in the meantime, referred them to me.

Follow-up and treatment: I saw Emily at age 5—more than 2 years after she had genetic testing. I was finally able to get her results from the nonprofit lab: They were positive for two mutations in *RPE65*, which are linked to LCA and retinitis pigmentosa.

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In recent years, the landscape for IRDs has changed dramatically; there are now multiple clinical trials and one FDA-approved treatment for a genetically inherited retinal condition. In Emily's case, both the timing and her genetic results were fortuitous. A phase 3 clinical trial had recently been completed showing safety and efficacy of a gene therapy (voretigene neparvovec-rzyl) to treat her biallelic mutation, and in 2018, Emily was successfully treated. Today, her ability to navigate in dim light is much better—and her father was overjoyed when, for the first time, they could participate in her school's annual father-daughter dance.

Considerations: This case underscores the critical importance of timely genetic testing and reporting of results. It's the doctor's responsibility to ensure this happens because, in reality, patients often don't follow up; they're waiting for a phone call. Fortunately, it's gotten much easier. We work with the Foundation Fighting Blindness (*FightingBlindness.org*), which offers a free genetic testing and counseling program through My Retina Tracker® (*MyRetinaTracker.org*). Any eye specialist can refer qualified patients, with results available in 4 to 6 weeks. (Other options are also available; see p. 6.) By signing up for the program, patients can also find out if they're eligible for any gene therapy trials. Given that we now have one approved therapy and more on the horizon, genetic testing should be the standard of care. It's medically required to appropriately diagnose patients with an IRD, and in cases like this one, treatment may exist!