

# MONTHLY Feature

## The Challenge and Promise of Genetic Testing



Over the past 15 years, genetic testing for neuromuscular diseases has expanded exponentially. As scientists continue to identify the precise proteins, mutations, and transcripts or nucleotides fueling certain conditions, the number of tests that can accurately pinpoint them in an individual has mushroomed. It's an exciting time to be working in the field, to be sure, but it's also overwhelming.

"We're moving so quickly, not only in treatment paradigms but in the mutations being detected and the number of tests available," says Cheryl Smith, MD, neurologist at J.W. Ruby Memorial Hospital in Morgantown, W.Va., and assistant professor at West Virginia University Rockefeller Neuroscience Institute. "It's a phenomenal thing to watch, but it can become a labor of love trying to keep up with it all."

Knowing which tests are available and which are applicable in an individual case is a constant struggle for clinicians who see patients with confirmed or suspected neuromuscular diseases.

*"We're moving so quickly, not only in treatment paradigms but in the mutations being detected and the number of tests available."*

*— Cheryl Smith, MD*

Here are some best practices for navigating this frenetically fertile landscape.

**1. Find a genetic counselor.** If you're fortunate enough to have genetic counselors on staff, lean on them heavily. Genetic counselors are up to date on which testing panels are appropriate for which symptoms and

# MONTHLY Feature

conditions, which tests are free, and the most effective strategies for navigating insurance companies when tests aren't free. "If you have access to a good genetic counselor, it's golden," Dr. Smith says. "They can become a clinician's right hand."

Clinicians without genetic counselors on staff can often connect to one via external resources, like the providers of genetic testing panels.

**2. Connect with test providers.** The organizations that develop and provide genetic tests have a thorough grasp on the specific purposes of their panels — and how to pay for them. Representatives at Perkin Elmer, Catalyst, Invitae, Nationwide Children's, and other organizations can break down the purpose, applications, testing locations, and funding options surrounding their individual panels. This is crucial in geographically isolated areas such as Dr. Smith's rural West Virginia territory, where if were it not for free, conveniently located testing, some patients may be forced to go without.

**3. Retest patients.** If you have patients who were tested prior to the late 1990s, consider retesting them. This is especially true for limb-girdle muscular dystrophy (LGMD), Becker muscular dystrophy (BMD), and Duchenne muscular dystrophy (DMD), for which misdiagnoses are not uncommon. "We're finding older individuals who were diagnosed decades ago with LGMD who, as it turns out, have BMD," Dr. Smith says. Also consider retesting patients who previously received negative or inconclusive results, as new tests provide greater chances of getting a genetically confirmed diagnosis.

**4. When in doubt, refer to a specialist.** "In my area of the world, primary care physicians, and even general neurologists, really can't be up to date on all the testing available," she says. "If you can refer patients to a neuromuscular disease specialist, the chances that they



will get properly tested and diagnosed go way up."

Against this complicated backdrop of testing options, clinicians must also weigh risks and patient concerns. "One of the biggest concerns for patients getting tested is whether they should get long-term care coverage before getting tested," Dr. Smith says. "I have some patients who simply want to know their diagnosis, period, and others who would prefer to establish long-term care insurance before diagnosis."

Dr. Smith advises that having an accurate diagnosis can help patients and their families better prepare for the future, but ultimately it's a patient's call.

Another common concern is whether to test family members. "When a result comes back pathogenic, it's crucial to talk to the patient about what that means in terms of genetic inheritance," she says. "What's recessive versus autosomal-dominant, what does this mean for their kids, etc."

This is another case in which genetic counselors can play an important role in helping patients and their loved ones understand test results and what it means for their families.

The benefits of genetic testing far outweigh the risks and concerns. A genetically confirmed diagnosis gives physicians vital information for targeting therapies to an individual's disease and can lead to better treatment. It may also open doors to participating in clinical trials and other research that works toward treatments and cures.

"Some of these treatments are just now coming up from the lab and haven't gone to clinical trials yet, so it becomes more imperative to get these patients diagnosed so that they'll be ready for trials, medications, and therapies as soon as they're FDA-approved," Dr. Smith says.

To view MDA's full suite of information on genetic testing, please visit <https://gateway.on24.com/wcc/experience/elitemda/1962960/2162296/genetic-testing-initiative>. Continue to check back for regularly updated content.

Image: iStock.com/malengpass