

### Transcript Details

This is a transcript of an educational program. Details about the program and additional media formats for the program are accessible by visiting: <https://reachmd.com/programs/neurofrontiers/diagnosing-muscle-disorders-when-to-use-biopsy/26391/>

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## Diagnosing Muscle Disorders: When to Use Biopsy

### Announcer:

Welcome to *NeuroFrontiers* on ReachMD. On this episode, we'll discuss peripheral nerve and muscle biopsies with Dr. Aziz Shaibani. Not only is Dr. Shaibani a Director of the Nerve and Muscle Center of Texas, board certified in neurology, neuromuscular medicine, and neuromuscular pathology, and Clinical Professor at Baylor College of Medicine, but he also presented a session on this exact topic at the 2024 American Association of Neuromuscular and Electrodiagnostic Medicine Annual Meeting. Let's hear from him now.

### Dr. Shaibani:

The muscle biopsy is a very valuable tool for the diagnosis of muscle disorders. However, over the last 20 years, there have been advances in our genetic and biochemical understanding of muscle disease so that many genetic and serological testings were introduced that minimized the need for a muscle biopsy. Therefore, the purpose of this session will be to explain the moving indications of muscle biopsy.

So the major indications of muscle biopsy today are not just to identify if the patient has a muscle disease, but also to provide more specific information. For example, patients with myopathic presentation—like proximal symmetrical muscle weakness, especially with elevated muscle enzymes and abnormal EMG—if you don't find an answer from the genetic or serological testing, then you need to do a muscle biopsy to see if there are some morphological features that would help you in making the specific diagnosis. Or, if your genetic testing provides some variants of unknown significance, they are not listed as pathogenic, and what is labeled as variants of unknown significance may be significant. It is just a matter of time. It takes, sometimes, a couple of years before these are listed as pathogenic, so you do a muscle biopsy to look for morphological features to confirm the significance of that variants of unknown significance. Also, muscle biopsy is useful for mitochondrial disorders as the muscle tissue is more sensitive than the blood for the detection of mitochondrial abnormalities. I still would do a blood test, and if it is negative and I still strongly suspect mitochondrial myopathy, then muscle biopsy would be the way to go. Other indications may include, for example, metabolic myopathies. For someone with exercise-induced muscle cramps, dark urine, and genetic testing that is not conclusive, the muscle biopsy is very useful to look for accumulation of metabolic products, such as lipids or glycogen. If we suspect vasculitis, inflammation of the blood vessels of the tissue, then adding a piece of a muscle to the nerve would increase the sensitivity of pathology to vasculitis.

Now, there's one caveat here that we see many patients who are referred for elevated CPK, but they have normal exam, normal EMG. Should we do a muscle biopsy? Isolated hyperCKemia poorly predicts the outcome of muscle biopsy. We have published about 750 cases of muscle biopsy to find out the determinants of the outcome, and we found that if the CPK was elevated—regardless of how elevated it was, and without weakness and EMG findings—then the probability of finding specific myopathic features is probably 10 percent, and so this will have to be evaluated by the physician who's going to do the biopsy. Also, it is important to know which stains to apply for which disorders and to use the electron microscope if needed to have more details because there are many stains. Some of them are just regular histological stains. Some of them are enzyme histochemistry. Some of them are immunological stains. You have to deploy the right stains for the right condition to maximize your diagnostic outcome.

### Announcer:

That was Dr. Aziz Shaibani discussing his session at the 2024 American Association of Neuromuscular and Electrodiagnostic Medicine Annual Meeting that focused on peripheral nerve and muscle biopsies. To access this and other episodes in our series, visit *NeuroFrontiers* on ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening!