at a normal rate to compensate for the loss of individual muscle fibers, whereas in neurogenic disorders, the MUAPs fire faster. An EMG is usually normal in steroid or disuse myopathy, both of which are associated with type 2 fiber atrophy; this is because the EMG preferentially assesses the physiologic function of type 1 fibers. The EMG can supplement the clinical examination in choosing an appropriately affected muscle to biopsy.

Imaging Studies Skeletal MRI and ultrasound are increasing utilized to assess the pattern of muscle involvement, which can help in narrowing the diagnosis, and are often more sensitive than the clinical examination and EMG, particularly early in a disease course. For example, there is early predilection of the vastus lateralis and medialis muscles with relative sparing of the rectus femoris muscles on imaging of thigh muscles in patients with inclusion body myositis, and this can be appreciated on imaging prior to weakness being detected on manual muscle testing. MRI can also demonstrate fasciitis when the clinical examination and EMG are normal. Imaging can also be used to help guide what muscle to biopsy in patients with weakness on manual muscle testing and EMG abnormalities only in muscles that are not typically biopsied (e.g., paraspinal or hip girdle). We have found imaging helpful in patients with presumed muscular dystrophy when the muscle biopsy is not diagnostic and genetic testing shows only a variation of unclear significance. In this situation, the pattern of muscle involvement on imaging can support the known pattern of muscle involvement of a specific hereditary myopathy. The cost and availability of MRI preclude routine use in some settings, but ultrasound is more readily available and less expensive.

Genetic Testing This is increasingly available and is the gold standard for diagnosing patients with hereditary myopathies. Next-generation sequencing panels are increasing utilized, but clinicians need to know their limitations; large deletions and duplications can be missed, as can mutations in noncoding (intronic) regions. Furthermore, testing often reveals sequence alterations of unclear significance.