Electrodiagnosis of Myotonic Disorders

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INTRODUCTION

Clinical and electrical myotonia is caused by a small group of neuromuscular disorders (Box 1). Myotonia is due to increased excitability of the muscle membrane often caused by dysfunction of muscle ion channels. Clinical myotonia is manifest by incomplete relaxation of muscle following either voluntary muscle contraction or direct muscle percussion. At the bedside, myotonia is clinically demonstrable by slowed muscle relaxation during repetitive hand grip and eye closure or by delayed relaxation of a muscle contraction evoked by tapping various muscles such as the thenar eminence or the finger extensors.

Electrical myotonia is an abnormal spontaneous muscle fiber discharge observed on needle electromyogram (EMG) examination. Electrical myotonia appears as repetitive muscle fiber potential discharges (eg, positive waves or fibrillation potentials) with waxing and waning frequency and amplitude with a firing rate between 20 and 80 Hz (Fig. 1A).9 When played over the audio, myotonic discharges have a characteristic sound of a dive bomber, or in the modern day, an accelerating and decelerating motorcycle engine. Electrical myotonia must be distinguished from neuromyotonia (frequency of greater than 150 Hz with a pinging sound) and chronic repetitive

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KEYWORDS
- Myotonia
- Myotonic dystrophy
- Periodic paralysis
- Nondystrophic myotonia
- Muscle channelopathies

KEY POINTS
- Clinical or electrical myotonia is caused by a small group of neuromuscular disorders including the dystrophic and nondystrophic myotonias.
- Chloride or sodium muscle channelopathies are the causes of myotonia in the dystrophic and nondystrophic myotonic disorders.
- Electrodiagnostic techniques, including needle electromyogram examination, short and long exercise testing, can help distinguish among the various myotonic disorders.
Box 1
Neuromuscular disorders with myotonia

Muscular dystrophies:
- Myotonic dystrophy type 1 and 2
- Myofibrillar myopathies

Muscle channelopathies:
- Nondystrophic myotonia (myotonia congenita, paramyotonia congenita, sodium channel myotonia)
- Hyperkalemic periodic paralysis

Metabolic myopathy:
- Acid maltase deficiency
- Debrancher deficiency
- McArdle disease (myophosphorylase deficiency)

Toxic myopathies:
- Chloroquine/hydroxychloroquine myopathy
- Statin myopathy
- Colchicine myopathy

Endocrine myopathies:
- Hypothyroidism

Inflammatory myopathies:
- Polymyositis
- Dermatomyositis

* Electrical myotonia without clinical myotonia.

Fig. 1. Electrical myotonia. (A) Two-second myotonic discharge in DM1 patient with typical waxing and waning frequency and amplitude. (B) Four-second waning only myotonic discharge in a DM2 patient in which frequency and amplitude decline gradually with no waxing component. (From Logigian EL, Ciafaloni E, Quinn LC, et al. Severity, type, and distribution of myotonic discharges are different in type 1 and type 2 myotonic dystrophy. Muscle Nerve 2007;35:479–85; with permission. Copyright with Wiley InterScience.)
discharges of constant, or less commonly, waning frequency with a machinelike sound.\textsuperscript{9}

This article focuses on electrodiagnosis of the primary myotonic disorders (myotonic dystrophy and the nondystrophic myotonias [NDMs]) as well as the related periodic paralysis (PP) muscle channelopathies.

EVALUATION OF CHANNELOPATHIES

Muscle ion channelopathies alter muscle membrane resting potential, resulting in either muscle hyperexcitability (eg, myotonia) or inexcitability (eg, muscle weakness). In the NDMs, muscle membrane hyperexcitability typically results in muscle “stiffness” during voluntary movement because of delayed skeletal muscle relaxation caused by repetitive muscle fiber action potentials (myotonia) as a result of mutations in the chloride (CLCN1) or sodium (SCN4A) skeletal muscle channel genes.\textsuperscript{10} By contrast, in the PPs, skeletal muscle membrane, inexcitability results in prolonged episodic muscle weakness caused by mutations in the sodium (SCN4A), calcium (CACN1A), and potassium (KCNJ2) channels.\textsuperscript{11} Of note, patients with both hyperkalemic PP and paramyotonia congenita (PC), due to different SCN4A mutations, have myotonia and episodes of skeletal muscle weakness.

Nondystrophic Myotonia

The NDMs include myotonia congenita (MC), PC, and sodium channel myotonia (SCM). The main clinical symptom of NDM is muscle stiffness from myotonia. Some patients also develop weakness and pain.\textsuperscript{10} These conditions must be distinguished from myotonic dystrophy type 1 and 2 (DM1 and DM2), which have significant, extraocular, systemic manifestations. On clinical grounds alone, there is often an overlap among DM2 and NDM. Electrodiagnostic and genetic testing help differentiate among the NDMs and between NDM and DM. When functionally debilitating, myotonia in NDM is treated with sodium channel blockade.\textsuperscript{10,12}

Myotonia congenita

MC presents as either an autosomal recessive form (also known as Becker disease) or a less severe autosomal dominant form (called Thomsen disease.) Both are caused by loss of function mutations in the CLCN-1 chloride channel, resulting in relative depolarization of the muscle membrane.\textsuperscript{10,12} Autosomal recessive MC presents between age 4 and 12 years and autosomal dominant MC presents before age 3 years.\textsuperscript{10} Myotonia typically develops with vigorous voluntary movement after resting; a warm-up phenomenon, that is, improvement in muscle relaxation with repetitive hand grip or rarely eye closure is often described.\textsuperscript{10} Patients with Becker (but not Thomsen) disease may develop transient weakness or “paresis” lasting seconds to minutes that resolves (like the myotonia) with repetitive muscle contractions.\textsuperscript{10}

Paramyotonia congenita (Group 1 SCM)

PC is an autosomal dominant disease due to a gain of function mutation in the SCN4A muscle sodium channel, resulting in relative muscle membrane depolarization.\textsuperscript{10,12} Patients develop not only muscle stiffness due to myotonia but also flaccid muscle weakness when depolarization progresses to membrane inexcitability. Cold temperature and exercise exacerbate myotonia and weakness in this disease.\textsuperscript{10,12} Face and hand muscles are preferentially affected. Most PC patients have paradoxical myotonia of hand grip or eye closure characterized by progressively slower muscle relaxation with repetitive activity, the opposite of the warm-up phenomenon typically seen in the chloride channelopathies.\textsuperscript{10,12} Patients present during the first or second
PC is allelic to hyperkalemic PP and like that condition results in prolonged episodes of muscle weakness.10

**Sodium channel myotonias (Group 2 SCM)**
This group of autosomal dominant diseases is also caused by gain of function mutations of the SCN4A channel similar to PC.10 However, group 2 SCM patients do not typically develop attacks of weakness (as in PC). In contrast to chloride channelopathies, patients with group 2 SCM are often sensitive to potassium, may have eyelid myotonia, lack transient paresis, and more often have pain.10,13 SCM can also be distinguished from PC and chloride channelopathies based on electrodiagnostic testing (see later discussion).14–16

**Electrodiagnosis of NDM**
Commercial genetic testing is available for some of the causative mutations in NDM, but there is a false-negative rate in these conditions as high as 20%.17 Electrodiagnostic studies are extremely helpful in directing genetic testing and also making the diagnosis of NDM in patients with negative genetic testing. Evidence of membrane hyperexcitability (eg, myotonic discharges) is sought with needle EMG examination, whereas evidence of membrane inexcitability (eg, drop in motor response amplitude) is investigated with long and short exercise testing. Needle EMG reveals diffuse myotonic discharges in proximal and distal muscles in NDM10,12,14,18; at times the number of myotonic discharges can be so substantial that evaluation of voluntary motor unit potential morphology and recruitment is not possible. In addition to myotonic discharges, low-amplitude (100–600 µV), high-frequency (150–250 Hz) discharges resembling neuromyotonic discharges have recently been observed in some NDM patients.16 Occasionally, repetitive firing of muscle fibers following a single stimulus termed “postexercise myotonic potentials” can be observed as delayed lower amplitude motor responses following the compound motor action potential (CMAP) during performance of routine nerve conduction studies.14 Postexercise myotonic potentials are described in both SCN4A and CLCN1 mutations.14

In general, the pattern and location of electrical myotonia does not distinguish among the NDM disorders,14 but the long and especially the short exercise test results can be helpful in this regard. Postexercise recording of serial CMAPs evaluates the functional consequences of ion channel mutations. A long-exercise protocol was initially described, which shows 80% sensitivity in PP and 15% to 30% sensitivity in NDM.19 Subsequently, a short-exercise protocol was developed that has been shown to be more sensitive for the detection of NDMs in general and useful in differentiating among the individual NDM disorders.

**Short-exercise protocol**
In the short-exercise protocol, serial CMAPs are recorded from the abductor digiti minimi (ADM) after supramaximal stimulation of the ulnar nerve at the wrist. Supramaximal CMAPs are recorded at baseline and then following 10 seconds of sustained contraction of the ADM. Additional CMAPs are recorded 2 seconds after exercise and then every 10 seconds for a total of 60 seconds; this protocol is repeated 3 times.14 Postexercise CMAP amplitudes are compared with the patient’s preexercise baseline CMAP amplitudes. The short-exercise protocol is easy to perform and causes minimal patient discomfort when compared with other electrodiagnostic tests such as prolonged repetitive nerve stimulation.

The short-exercise protocol has a high sensitivity of 100% (83%–100%) in PC, 83% (53%–100%) in MC, and 60% in SCM.12,14,15 Repeating the protocol with limb cooling
(to 20–25°C) improves the sensitivity in MC (70%–100%) and exaggerates the abnormality observed in PC.\textsuperscript{15,16}

Three patterns are observed in patients with NDM, Fournier I, II, and III.\textsuperscript{10,14,15} Patients with PC exhibit Fournier pattern I with a decrement in CMAP amplitude (19%–40% less than preexercise baseline CMAP), which persists for 60 seconds and which increases with subsequent trials and with cooling (Fig. 2).\textsuperscript{14,15} This decrement reproduces the weakness in PC patients that develops with exercise, especially with muscle cooling.

Patients with MC commonly exhibit Fournier pattern II with an initial postexercise CMAP decrement, which repairs by 60 seconds and is less pronounced on subsequent trials (see Fig. 2).\textsuperscript{14} The Fournier pattern II is also observed in some patients with DM1 and DM2 whose myotonia, like MC, is due to chloride channel dysfunction.\textsuperscript{10} At room temperature, a subset of MC patients (primarily dominant MC) exhibit Fournier pattern III without a postexercise decrement or sometimes an initial increment following exercise with return to baseline (see Fig. 2).\textsuperscript{14} Fournier III is also observed in normal controls and most patients with SCM.\textsuperscript{14,15} However, repeating the short exercise test with cooling reveals the Fournier pattern II in 91% to 100% of patients with recessive MC and in 14% to 71% of patients with dominant MC.\textsuperscript{15} Rewarming the limb and repeating the test may identify a small group of additional patients with MC.\textsuperscript{16}

Most patients with SCM exhibit the Fournier pattern III at room temperature, similar to normal controls. With cooling, a subset of SCM patients exhibits the Fournier pattern I, similar to PC.\textsuperscript{15} In one series, combining the clinical symptom of eye closure myotonia (historical or on clinical examination) with the Fournier pattern III distinguished SCM from MC with a specificity of 94% and sensitivity of 82%.\textsuperscript{16} Although helpful in the evaluation of hyperkalemic PP, the long exercise test (LET) adds little to the diagnostic evaluation of NDM. The LET has a sensitivity of 25% in MC and 89% in PC.\textsuperscript{12,20}

\begin{figure}
\centering
\includegraphics[width=\textwidth]{fig2.png}
\caption{Short exercise test. (A) Transient decrease in CMAP amplitude in myotonia congenita. (B) Fournier pattern III: initial increment in CMAP with return to baseline. No change with subsequent tests. (C) Fournier pattern II: initial CMAP decrement postexercise with return to baseline on subsequent trials. (D) Fournier pattern I: decrement of CMAP amplitude postexercise, which worsens on subsequent trials. (Modified from Fournier E, Arzel M, Sternberg D, et al. Electromyography guides toward subgroups of mutations in muscle channelopathies. Ann Neurol 2004;56:650–61; with permission. Copyright with Wiley InterScience.)}
\end{figure}
**Repetitive nerve stimulation**

Prolonged repetitive stimulation of the ADM has been studied in NDM. In the typical protocol, the ulnar nerve is stimulated at the wrist at 10 Hz for 10 seconds; the test is repeated after 5 minutes rest with 10 Hz stimulation for 5 seconds.\(^\text{18}\) Similar to the short exercise test, repetitive stimulation is repeated with cooling. At room temperature, patients with autosomal recessive MC and DM exhibit a decrement in CMAP amplitude that is more pronounced with cooling.\(^\text{18}\) Patients with PC exhibited a decrement only with cooling.\(^\text{18}\) In one series, repetitive nerve stimulation identified two-thirds of patients with recessive MC compared with one-third in the short exercise test.\(^\text{18}\) A later trial showed no added diagnostic information with repetitive stimulation when added to the short exercise test.\(^\text{16}\) Repetitive stimulation may be useful in identifying patients with recessive MC who exhibit Fournier III pattern on short exercise testing. However, this test causes significant patient discomfort compared with the short exercise test.

**Directed genetic testing**

Over 100 missense, nonsense, insertion, deletions, and splice site mutations have been identified in the CLCN1 channel gene on chromosome 7q35. More than 40 mutations are described in the SCN4A gene on chromosome 17.\(^\text{10}\) Only a portion of these mutations can be evaluated commercially. In addition, genetic testing can be costly and in some cases not covered by the patient’s medical insurance policy.

Fournier and colleagues\(^\text{14,15}\) propose directed genetic testing based on the results of the short exercise protocol and repetitive nerve stimulation. Patients with the Fournier pattern I should be checked for SCN4A mutations. Patients with the Fournier pattern II should be screened for CLCN1 mutations and if negative SCN4A mutations as a small subset of SCM patients exhibit this pattern. Fournier II patients with negative testing for both CLCN1 and SCN4A should be screened for DM1 and DM2. Patients with the Fournier pattern III who exhibit eyelid myotonia should be screened for SCN4A; those patients without eyelid myotonia should be screened for CLCN1.\(^\text{16}\) The 10-Hz repetitive nerve stimulation protocol may help distinguish between SCM and MC in patients with negative genetic testing; this may prove useful if mutation-specific therapies are developed.\(^\text{18}\)

**Periodic Paralysis**

The PPs are autosomal dominant muscle channelopathies that result in muscle inexcitability and recurrent attacks of flaccid paralysis. They include hypokalemic PP, hyperkalemic PP, Andersen-Tawil syndrome (ATS), and thyrotoxic PP. Most weakness episodes are transient and are not life threatening; most affected patients develop persistent mild proximal weakness over time that is not linked to frequency or severity of attacks.\(^\text{11,12}\) Patients with ATS may develop life-threatening cardiac arrhythmias.\(^\text{11}\) Patients with PP are treated with a combination of lifestyle modification to avoid triggers and medical therapy with carbonic anhydrase inhibition with acetazolamide (125–1000 mg/d) or dichlorphenamide (50–200 mg/d).\(^\text{11}\)

**Hypokalemic periodic paralysis**

Hypokalemic PP is an autosomal dominant muscle channelopathy with incomplete penetrance in women.\(^\text{11,12,21–23}\) Prevalence is estimated at 1 per 100,000.\(^\text{11}\) Sixty percent of cases are due to mutation in the calcium CACNA1S channel and 20% are due to SCN4A mutations.\(^\text{11}\) Patients develop episodes of flaccid focal or generalized weakness that typically spare respiratory and facial muscles. Attacks last for hours or days and usually begin in the first or second decade.\(^\text{11}\) Attacks are most common on waking in the morning and may be triggered by prolonged rest after
exercise or a carbohydrate rich meal. Weakness is typically associated with low serum potassium; attacks can often be aborted with potassium administration. Thyrotoxic PP is also related to hypokalemia; thyroid-stimulating hormone and free T4 should be evaluated in all cases of suspected hypokalemic PP because patients with this condition often have minimal manifestations of thyrotoxicosis. They do not have a known calcium or sodium channelopathy, but there is genetic data to implicate potassium channel mutations. In addition, patients without channelopathies can develop a subacute proximal myopathy in the setting of severe hypokalemia (serum K <2.5 mEq/L).

**Hyperkalemic periodic paralysis**

Hyperkalemic PP is an autosomal dominant SCN4A channelopathy that is allelic to PC and typically begins in the first decade. Attacks of weakness are usually shorter than those in hypokalemic PP (hours) and are associated with elevated levels of potassium in 50% of cases. The remaining 50% have normal potassium levels but are believed to have a relative elevation of potassium level within the normal range, resulting in weakness. Weakness is often triggered by rest after exercise, stress, and fatigue. Unlike hypokalemic PP, electrical myotonia is appreciated in 50% to 75% of cases. In addition, during attacks of weakness, CMAP amplitude may be decreased or absent as in other causes of PP. Needle EMG examination discloses decreased insertional activity, increased fibrillation potentials/positive waves, and increased polyphasic motor units.

**Andersen-Tawil syndrome**

ATS is a disorder that combines PP, ventricular arrhythmias, and skeletal anomalies. It is caused by mutations in the inward rectifying muscle potassium channel KCNJ2. Prevalence is estimated to be one-tenth of hypokalemic PP. Episodes of weakness are triggered by rest after exercise and stress. Cardiac abnormalities range from prolongation of the corrected QT interval to ventricular ectopy and runs of ventricular tachycardia. Skeletal anomalies include small mandible, hypertelorism, syndactyly, clinodactyly, broad nose, and short stature.

**Electrodiagnosis of PP**

Similar to NDM, commercial genetic testing for PP is incomplete and in some cases prohibitive because of cost or lack of insurance coverage. Electrodiagnostic testing is helpful in guiding genetic testing and in distinguishing between PP and other forms of weakness. Some patients with hyperkalemic PP show an initial increment during the short exercise protocol. However, the short exercise test does not have significant diagnostic value in PP as most patients show no abnormalities. The LET is more useful in these patients. Some patients with hyperkalemic PP exhibit electrical myotonia; this finding is not described in hypokalemic PP.

**Long exercise test**

LET is also typically performed stimulating the ulnar nerve at the wrist and recording the ADM motor responses. Supramaximal ADM responses are recorded at baseline, throughout 5 minutes of exercise, and then more than 45 to 60 minutes following exercise; investigators vary on the frequency of recordings following exercise. Patients with PP are expected to show postexercise drop in motor response amplitude or area corresponding to the clinical symptom of flaccid paralysis.

During the LET, baseline supramaximal CMAPs are typically measured every 10 seconds for 1 to 2 minutes to establish a stable baseline. The patient is then exercised for 5 minutes with periodic, short-rest periods every 15 seconds. CMAPs are
recorded after each minute of exercise. Some investigators then record postexercise CMAPs every minute for 5 minutes followed by every 5 minute CMAPs for 40 to 45 minutes.14 Others recommend postexercise CMAPs every 1 to 2 minutes for 30 to 45 minutes.19 In the initial version of the LET, decrements in amplitude and area were measured from the maximal CMAP obtained during or immediately after exercise.19,20 Subsequent investigators have measured decrement from the baseline CMAP obtained before exercise.14 One study shows the methods to be similar, although use of the preexercise baseline may be less sensitive in hyperkalemic PP.16

Among the small cohorts studied, the LET has a sensitivity of 80% to 90% in both hypokalemic PP and hyperkalemic PP.12,14,19,20 In one series of ATS, the LET was demonstrated to have a sensitivity of 80% to 100%, which improved to 100% if CMAP area was examined alone.16

Normal controls typically show a small increment in amplitude and area (10%) following exercise and a subsequent decrement of 15% in amplitude and area compared with maximal-exercise CMAP (never more than 30%).19,20 An abnormal decrease is defined as 40.9% for amplitude and 48% for area (2 SDs).19,20 When measured from preexercise baseline, an abnormal decrement in CMAP amplitude is defined as greater than 20% of baseline.14

Two typical LET patterns are observed in PP, which may help to distinguish between calcium and sodium channel mutations.14 Patients with a sodium channel mutation often exhibit Fournier pattern IV (Fig. 3) manifest by an increment in CMAP amplitude/area with exercise followed by a decrement in amplitude/area 40% to 80% of baseline (dependent on method); maximal decrement is observed between 30 and 45 minutes postexercise.14,16,20 Fournier pattern V manifest by a maximal decrement in area/amplitude after 20 to 40 minutes is more typical of calcium channel-related PP; this decrement may or may not be preceded by an initial increment in CMAP amplitude/area.14,16,20 However, there is some cross over between pattern IV and V in hyperkalemic and hypokalemic PP, which may be explained by the proportion of hypokalemic PP patients who have an SCN4A mutation.14 Pattern V is most typical in ATS.16

**Directed genetic testing**

Genetic testing in PP can be guided by a combination of clinical presentation, serum potassium measurement during an attack, and the LET. Patients with skeletal anomalies and cardiac symptoms should be screened for ATS. In the absence of distinctive clinical or serum K abnormalities, one strategy is to screen patients exhibiting Fournier pattern IV for CACNA1S first and if normal SCN4A. Similarly, patients with Fournier pattern V could be screened for SCN4A first and if normal CACNA1S.

**EVALUATION OF MYOTONIC DYSTROPHY**

DM1 and DM2 are inherited disorders of skeletal muscle that result in progressive weakness similar to other muscular dystrophies. The presence and pattern of weakness, clinical and electrical myotonia, and extramuscular manifestations distinguish the myotonic dystrophies from NDM and other forms of dystrophy.

**Myotonic Dystrophy Type 1**

DM1 is an autosomal dominant progressive muscular dystrophy due to an unstable trinucleotide repeat (cytosine-thymine-guanine [CTG]) on chromosome 19q.27 DM1 exhibits anticipation with more severe disease in subsequent generations who inherit trinucleotide repeats of increasing length.27,28 Clinically, the age of onset of disease is inversely proportional to the CTG repeat length.27
DM1 patients have a classic pattern of facial weakness, mild ptosis, bulbar weakness, and distal motor (finger flexors/ankle dorsiflexion) weakness with associated clinical and electrical myotonia. Myotonia is typically absent for the first year in infantile onset disease. Myotonia is usually manifest by difficulty relaxing handgrip and with percussion of thenar or finger extensor muscle groups.

DM1 causes significant extramuscular disease, which distinguishes it from NDM. Patients typically develop a combination of early cataracts (before 50 years old), cardiac arrhythmias, psychological dysfunction/cognitive dysfunction, sleep disorders, gastrointestinal irritability, various neoplasms, and glucose intolerance.27 Disabling myotonia can be treated with sodium channel blockade (eg, mexiletine) and many of the systemic manifestations necessitate referral to other subspecialists.

Myotonic Dystrophy Type 2

DM2, aka proximal myotonic myopathy, is also an autosomal dominant inherited progressive muscular dystrophy due to a CCTG expansion on chromosome 3q.27 Anticipation is also observed in DM2. In contrast to DM1, patients with DM2 exhibit more prominent proximal hip and shoulder weakness and present later in life (mean
age onset fourth/fifth decade). Pain is reported by more than 50% of patients with DM2, and myotonia is less obvious in DM2 than DM1 on clinical and EMG examination.

DM2 is also associated with extramuscular disease. Early development of cataracts, cardiac conduction abnormalities, and endocrine abnormalities are typical. Gastrointestinal and cognitive dysfunctions are atypical in DM2. In contrast to DM1, a congenital form of the disease is not observed in DM2.

**Diagnosis of Myotonic Dystrophy**

In most patients with DM1 and DM2, the diagnosis is made at the bedside and the EMG laboratory based on the pattern of weakness, presence of myotonia, and extramuscular features. The diagnosis is confirmed with genetic testing that is clinically available and abnormal in 100% of patients with DM1 and 99% of patients with DM2.

The role of electrophysiology is to confirm the presence of myotonic discharges that may not be obvious on clinical examination. This is particularly the case for DM2 patients in whom grip and percussion myotonia may be absent or subtle. Differences in muscle histology are described in DM1 and DM2, but muscle biopsy is rarely needed to confirm the diagnosis.

**Muscle Biopsy**

Similar to other muscular dystrophies, muscle biopsy in DM1 and DM2 typically shows variation in fiber size, rounded atrophic fibers, increased central nuclei, increased connective tissue, and fatty replacement of muscle on H&E and trichrome staining of frozen tissue. Occasional moth-eaten fibers are observed with reduced nicotinamide adenine dinucleotide - tetrizolium reductase (NADH-TR) staining. Nuclear clumps, not typical of other dystrophies, are observed in both types of myotonic dystrophy. DM1 patients generally show preferential atrophy of type 1 fibers, whereas DM2 patients show predominantly type 2 atrophy. The finding of nuclear clumps amidst typical dystrophic histologic changes should prompt workup for DM1 and DM2. The additional presence of selective muscle fiber type atrophy (type 1 fiber atrophy in DM1 and type 2 fiber atrophy in DM2) can potentially help guide the order of genetic testing.

**Electrodiagnostic Testing**

Widespread electrical myotonia on needle EMG is the electrophysiologic hallmark of myotonic dystrophy, but it is more easily evocable in DM1 than DM2 and tends to be “waxing and waning” in DM1 and “waning” in DM2. Moreover, a subset of patients with DM2 exhibits only subtle “waning” myotonia; these waning discharges can easily be misclassified as fibrillation potentials, chronic repetitive discharges, or nonsustained increased insertional activity (see Fig. 3). Electrical myotonia is most prominent in distal limb muscles in both DM1 and DM2. However, myotonia is more prevalent in proximal leg muscles in DM2 than DM1. There is a similar occurrence of thoracic paraspinal myotonia in both conditions. The authors recommend sampling an array of proximal and distal extremity muscles as well as thoracic paraspinal muscles in patients with suspected myotonic dystrophy. The presence of only waning myotonia should prompt genetic testing for DM2 before DM1. Finally, rare patients with DM2 are described without electrical myotonia. Therefore, genetic testing for DM2 may still be indicated in patients with suspicious clinical or histologic features of myotonic dystrophy in the absence of electrical myotonia.

As discussed in the previous section, patients with DM1 and DM2 may show Fourner pattern II on short exercise testing, similar to patients with chloride channel
myotonia.\textsuperscript{10} Patients exhibiting Fournier pattern II with negative genetic testing for mutations of CLCN1 and SCN4a should have genetic testing for DM1 and DM2.

**SUMMARY**

Clinical and electrical myotonia is caused by a small group of neuromuscular disorders. Electrodiagnostic testing is used to confirm the presence of myotonia and membership in this group of diseases, to distinguish the specific cause of myotonia, and to guide additional diagnostic testing and treatment.

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