Electrodiagnostic findings in myotonic dystrophy: A study on 12 patients

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Abstract

Myotonic dystrophy (DM) is a complex multisystem disease with specific clinical and electrodiagnostic findings. Myotonia can be seen in the distal and proximal muscle groups in upper and lower limbs. There is no established guideline to demonstrate the sensitivity of muscles in the diagnosis of myotonic dystrophy. The aims of this study are to describe common electrodiagnostic findings in patients with DM; and to assess the electrodiagnostic sensitivity of muscles in the diagnosis of DM. In this retrospective study, patients’ age, sex, nerve conduction study findings including common upper and lower limbs nerve functions, and needle examination findings were collected and analyzed. A descriptive analysis (with percentage) was performed on the data obtained from the charts. NCS analysis showed more than half of patients had normal sensory and motor NCS findings. In 11 over 12 patients, sensory NCSs were within normal limits. Only one patient showed abnormal sensory responses. The most common abnormal NCS findings were decreased amplitude with normal latency and normal conduction velocity. The needle analysis showed distal muscles including first dorsal intersosseous, abductor pollicis brevis, tibialis anterior, medial gastrocnemius and peroneal longus muscles are more sensitive in detecting myotonic discharges than proximal muscles including deltoid, triceps, vastus medialis and vastus lateralis. Our findings showed sensory nerve responses were usually within normal limits. The most common NCS abnormality showed decreased motor nerve amplitudes. The needle test showed myotonic discharges were more prominent in the distal muscles in upper and lower limbs.

Introduction

Myotonic dystrophy (DM) is a complex multisystem disease with specific clinical and electrodiagnostic findings.1,2 DM is classified based on the age of onset and clinical characteristics. DM is a genetic disorder and occurs as a result of expansions of the certain trinucleotide on the responsible gene.1,3 A specific electrodiagnostic finding, called myotonia, is seen in the needle part during electromyography (EMG) study. EMG myotonia has characteristic pattern and sound, a typical dive-bomber sound is heard instead of electrical silence while the recording needle inserting into muscle at rest.1,3 Myotonia can be seen in the distal and proximal muscle groups in upper and lower limbs.2 There is no established guideline to demonstrate the sensitivity of muscles in the diagnosis of myotonic dystrophy.

Myotonic dystrophy (DM) is an RNA-mediated multisystemic disorders and has two subtypes, type 1 (DM1) and type 2 (DM2). Different underlying genetic defects are described to cause the DM subtypes. DM1 is caused by a DMPK 3’ untranslated region (3’UTR) CTGexp and DM2 by an intronic CCTGexp in CNBP. Somatic expansion is related to pathological onset but there is no correlation between expansion size and disease severity. Several tissue system involvements are seen DM including skeletal muscle myotonia, weakness/wasting, conduction defect/block, cognitive decline, cataract, hearing impairment, insulin resistance, autoimmune disease, etc.4

RNA toxicity and its related consequences are the main underlying pathogenesis of DM.5 Expanded trinucleotide repeat from the mutant DM alleles cause RNA toxicity. The most common expanded trinucleotide repeat is located in the untranslated region of the dystrophin myotonia protein kinase (DMPK) gene in DM type 1; and in first intron of the ZNF9 (CNBP) gene in DM2. In the DM, the mutant RNA has important role in the pathogenesis of the disease that ending up with no translation in the cell. These repeat expansions may also cause toxic effects on the other genes besides DM1 and DM2 loci.4,6 Given the accumulation of mutant RNAs in the nucleus alteration of RNA-binding protein activity, aberrant splicing and abnormal function of several genes, several abnormalities are seen in the body cells including skeletal muscle chloride channel, insulin receptor, and cardiac troponin. Expanded CUG or CCUG repeats may cause sequestration of Muscle-blind-like (MBNL1)that causing alternate splicing of the B1N1 gene and skipping of muscle-specific exon 11 of B1N1 messenger RNA.6 These RNA changes cause impairment of T-tubule and excitation-contraction coupling. Myotonia, the specific clinical and electrodiagnostic finding of the disease, occurs as a result of skeletal muscle chloride channel dysfunction that engendered by the end of this process.4

In this study, we retrospectively analyzed electrodiagnostic features of patients with adult onset DM-type 1. The aims of this study are 1) to describe common electrodiagnostic findings in patients with myotonic dystrophy; and 2) to assess the electrodiagnostic sensitivity of muscles in the diagnosis of myotonic dystrophy.

Materials and Methods

This is a retrospective and descriptive study to aim to analyze clinical and electrodiagnostic features of patients with DM-type 1 whom previously seen in Carilion Clinic neurology clinic, Roanoke, VA. This study was approved by the local Institutional Review Board (IRB). Twelve charts of patients who were previously clinically diagnosed with DM type 1 and underwent electrodiagnostic tests in Carilion Clinic neurology outpatient clinic were retrospectively reviewed. The type of DM was made based on typical clinical findings and family history. Seven of these patients had genetic confirmation as well.

A routine electrodiagnostic (EDx) study, including nerve conduction studies (NCS) using standard laboratory techniques was performed by one board certified electromyographers. Limb’s temperature was maintained at 32°C at the dorsum of the
hand and foot. The NCS was performed by
the same physician and an average of the
differential nerve responses was used during
data analysis. The NCS were performed
under a uniform protocol using a Caldwell
EMG machine that included bilateral medi-
al and ulnar motor responses; bilateral radial,
medial and radial sensory responses; bilateral
sural and superficial sensory responses; and bilateral peroneal
tibial motor responses in 11 subjects,
only lower NCS was performed in one sub-
ject. Routine needle upper EMG studies
included examination of deltoid, biceps, tri-
ceps, pronator teres, first dorsal interosseous (FDI), and aductor pollicis
brevis (APB) muscles; and the needle lower
EMG studies included vastus medialis
(VM), vastus lateralis (VL), tibialis anterior
(TA), peroneal longus (PL), and medial gas-
trocnemius (MGast) muscles. The upper
and lower limbs needle tests were per-
formed in 11 subjects and only the lower
limb needle test was performed in one sub-
ject. Interpretation of the EMG was per-
formed according to accepted guidelines in
order to minimize inter-rater reliability.
During interpretation, myotonic discharges
were categorized from 1+ to 4+ scales
based on amplitude, frequency, and prol-
ongation.

Patients’ age, sex, nerve conduction
study findings including common upper
and lower limbs nerve functions, and needle
examination findings were collected and
analyzed. A descriptive analysis was per-
formed on the data obtained from the charts.

Table 1. This table demonstrates the summary of myotonic discharge findings of the patients in upper and lower limbs. Myotonic discharges are categorized from 1+ to 4+ scales based on amplitude, frequency, and prolongation.

| Age | Sex | Deltoid | Triceps | Biceps | PT. | FDI | APB | VL | VM | TA | PL | MedGast |
|-----|-----|--------|---------|--------|-----|-----|-----|-----|-----|-----|-----|---------|
| Pt 1 | 37 M | 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ | 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ |
| Pt 2 | 49 F | 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ |
| Pt 3 | 31 F | Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt | Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt |
| Pt 4 | 41 M | 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ |
| Pt 5 | 78 F | 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ |
| Pt 6 | 65 M | 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ |
| Pt 7 | 68 F | 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ |
| Pt 8 | 36 M | 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ |
| Pt 9 | 64 F | Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt |
| Pt 10 | 59 M | 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ 1+ |
| Pt 11 | 46 F | Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt Slt |
| Pt 12 | 65 M | 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ 2+ |

Pt: Patient, R: Right, L: Left, PT: Pronator Teres; FDI: first dorsal interosseous, APB: Aductor Pollicis Brevis, VL: Vastus Lateralis, VM: Vastus Medialis, TA: Tibialis Anterior, PL: Peroneus Longus, MedGast: medial gastrocnemius, Slt: Silent.

Results

Charts of twelve patients with diag-
nosed with adult onset myotonic dystrophy
were reviewed. The mean of age was
53.3±15.2 years. Of the subjects, five were
male; and seven were female. Twelve
patients were clinically and electrodia-
tagnostically were diagnosed with DM-type 1.

The NCS analysis showed normal sen-
sory and motor NCS in seven subjects.
Reduced amplitudes were detected in bilat-
eral median motor, and right tibial motor
nerves in the patient #1; in bilateral median
motor, ulnar motor, tibial motor, and left
median motor nerves in the patient #5; in
bilateral medical motor, bilateral tibial
motor, bilateral ulnar motor, and bilateral
gastrocnemius nerves in the patient #6; and
in right peroneal motor, bilateral tibial, and
left ulnar nerves in the patient #7. The NCS
of Patient #12 showed decreased amplitude
in left peroneal motor nerve with decreased
conduction velocity (CV); decreased amplit-
udes in bilateral tibial motor nerve with
prolonged latency and decreased CV, and
absent sensory responses in bilateral super-
ficial peroneal and bilateral ulnar nerves.

The needle assessment of upper limbs
showed deltoid and triceps muscles were
negative for myotonic discharges in three
subjects; and biceps and PT muscles were
negative for myotonic discharges in one
subject. Distal upper limbs muscles includ-
ing FDI and APB were most active muscles
for myotonic discharges and were present
in all 11 patients. The needle assessment
of lower limbs in 12 patients showed VL and
VM muscles were negative for myotonic discharges in five patients; and TA and PL
muscles were negative for myotonic dis-
charges in one patient. Distal lower limbs
were more active for myotonic discharges
in TA, PL, and MGast muscles. In two
patients, MGast were silent, but in these
patients MGast were atrophic and fibrotic.
The needle findings are summarized in
Table 1.

Discussion

EMG findings are a key element in the
diagnosis of myotonic disorders. Myotonic
potentials are one of the most specific
potentials seen on needle EMG test. Myotonia is often easier to detect on EMG
test than on neurological examination.3,7 In
this study, we aimed to determine the com-
mon NCS findings and electrodiagnostic
sensitivity of upper and lower limbs’ mus-
cles in the diagnosis of myotonic dystrophy.
This is the first study to make in the litera-
ture to assess EDx findings in myotonic
dystrophy.

NCS analysis showed more than half of
patients had normal sensory and motor NCS
findings. In 11 over 12 patients, sensory
NCSs were within normal limits. Only one
patient showed abnormal sensory respons-
es. The most common abnormal NCS find-
ings were decreased amplitude with normal
latency and normal conduction velocity. The
diminished amplitude of motor nerves may be related to muscle wasting or motor
axonal damage. Sensory nerves functions
are usually within normal limits in patients
with myotonic dystrophy. There is no previ-
ous study to compare our findings. Our
results showed peripheral neuropathy is
uncommon in myotonic dystrophy.

Myotonic discharges are spontaneous
discharges with a waxing and waning of
amplitude and frequency.1,3 The needle
analysis showed distal muscles are more
sensitive in detecting myotonic discharges
than proximal muscles in both upper and

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lower limbs. In fact, the needle test of proximal muscles can be within normal limits with silent spontaneous activities in patients with myotonic dystrophy. In two cases, the needle test was unusually silent in MedGast muscles but MedGast muscles were atrophic and fibrotic in those cases. These findings are expected in the clinical context of adult onset myotonic dystrophy since distal muscles are generally more affected than proximal muscles.

EDX test is most important diagnostic test in myotonic disorders. Providers may diagnose myotonic disorder with EMG test for clinically subtle cases. DM 1 patients typically have distal motor weakness with associated clinical and electrical myotonia. In our series, distal muscles showed more prominent myotonic discharges than proximal muscles. In some cases, medial gastrocnemius muscle was silent for myotonia in advances cases that may be related to muscle wasting and fibrosis. Myotonia may be detected in clinically asymptomatic proximal muscles.

Conclusions

This study shows distal muscles are more sensitive to demonstrate myotonia in DM1 patients. However proximal muscles should be assessed as well. Sensory nerve responses are usually normal in DM1 patients. Decreased motor amplitude is the most common NCS findings that may be related to muscle wasting or motor axonal damage. EMG test is key test in diagnosis of myotonic disorder, particularly in clinically subtle cases.

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