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The needle EMG findings in myotonia congenita

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ABSTRACT

Introduction: Myotonia congenita (MC) is caused by pathogenic variants in the CLCN1 gene coding the chloride channel protein.
Methods: To test the hypothesis that needle EMG could be helpful in distinguishing between the recessive and dominant MC, we performed EMG examination in 36 patients (23 men) aged 4–61 years with genetically proven MC: in 30 patients with autosomal recessive MC *(Becker MC)* and in 6 with autosomal dominant MC *(Thomsen MC)*.
Results: Myotonic discharges were recorded in 95.8% of examined muscles. For the whole MC group we observed a significant positive correlation between parameters of motor unit activity potentials (MUAPs) in vastus lateralis and tibialis anterior muscles and the duration of the disease. Similar correlation for biceps brachii also was found in *Becker MC* subgroup only.
Discussion: EMG could still be helpful in diagnosis of MC and together with provocative tests might be useful in differentiation between recessive and autosomal MC.

1. Introduction

Myotonia congenita (MC) is a hereditary muscle disorder characterized by impaired relaxation of skeletal muscle after voluntary contraction (clinical myotonia) (Colding-Jørgensen, 2005). It is the most common inherited skeletal muscle channelopathy caused by loss of function pathogenic variants in the CLCN1 gene coding for the chloride channel, which result in a relative depolarization of the muscle membrane (Matthews et al., 2010). MC presents as either autosomal recessive form (also known as *Becker MC*) or a less severe autosomal dominant form (called *Thomsen MC*) (Colding-Jørgensen, 2005; Heatwole et al., 2013; Hehir and Logigian, 2013). In both forms muscle stiffness is most pronounced during rapid voluntary movements following a period of rest but improves with repeated activity, which is known as 'warm-up' phenomenon (Heatwole et al., 2013). Electrical myotonia is considered as an evidence of muscle membrane hyperexcitabilty and is one of the key features of myotonia congenita independent of the pattern of inheritance. It is defined as an abnormal spontaneous repetitive muscle fiber discharge with waxing and waning frequency and amplitude with a firing rate between 20 and 80 Hz observed on needle EMG examination (Kimura, 2001; Hehir and Logigian, 2013).

Recessive MC phenotype presents between the age of 4 and 12 years and tends to be more severe than dominant type. It is characterized by moderate to severe myotonia, moderate muscular hypertrophy, depressed tendon reflexes, transient weakness accompanying myotonia, or even permanent weakness and atrophy of forearm and neck muscles with myopathic electromyographic (EMG) and histopathological changes in one fourth to one third of cases (Colding-Jørgensen, 2005; Fialho et al., 2007). It is well established that myopathy may develop in some patients with MC.

In muscle biopsies it may be seen not only as mild, non-specific abnormalities or no abnormalities at all but also as the absence of type 2B fibres, reported in chloride channelopathies (Crews et al., 1976; Meola et al., 2003).

Nowadays, clinical characteristics (Trip et al., 2009a), various protocols of repetitive nerve stimulation patterns (Fournier et al., 2004; Fournier et al., 2006; Tan et al., 2011) or a combination of both (Michel et al., 2007) are used as a guide for focused genetic testing in patients with non-dystropic myotonias (NDM).

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The purpose of our study was to obtain the detailed EMG characteristics of patients with genetically confirmed MC as well as to test the hypothesis that needle EMG could be helpful in distinguishing between recessive and dominant form of MC.

2. Patients and methods

We evaluated data of 36 patients (13 women and 23 men) aged 4–61 years with the genetically proven myotonia congenita. The age at the disease onset varied from 6 months to 19 years. All patients were diagnosed and treated at the Department of Neurology, Medical University of Warsaw. The study protocol was approved by the Bioethical Committee at the Institute of Psychiatry and Neurology (IPiN No 3/2015). All the procedures were in accordance with the standards of the Committee on Human Experimentation at the Medical University of Warsaw and with the Helsinki Declaration of 1975.

2.1. EMG examinations

Electrophysiological studies were performed using Keypoint, Medtronic Functional Diagnostics EMG in 2002–2018 during diagnostic process but prior to genetic confirmation of the disease.

EMG results were obtained for the following muscles: biceps brachii (BB), first interosseous dorsalis (FID), vastus lateralis or vastus medialis (VL) and tibialis anterior (TA). The reports were evaluated for description of spontaneous activity (SA) and morphology of motor unit action potentials (MUAPs) in each examined muscle. Muscle strength was assessed using the 6-grade Medical Research Council (MRC) scale and muscle atrophy using a 2-grade scale (no atrophy – 0, muscle atrophy present – 1). Additionally, three distal upper limbs (UL) muscles: flexor carpi ulnaris (FCU), abductor digiti minimi (ADM) and extensor digitorum communis (EDC) were screened only for the occurrence and intensity of SA, especially myotonic discharges (MD). Abnormal SA was categorized as fibrillation potentials (fib.), positive sharp waves (PSW), complex repetitive discharges (CRD) and MD using the criteria set forth by the American Association of Electrodiagnostic Medicine (2001). The frequency of SA types was graded on an ordinal 5-graded scale proposed by Kimura (2001). Based upon the frequency and amplitude, the MD and CRD were classified using the scale proposed by Hanisch et al. (2014).

Motor unit action potentials (MUAPs) were recorded during routine EMG examinations using concentric needle electrodes with the uptake area of 0.07 mm^2. The parameters of a single MUAP including the amplitude, duration, area, size index (SI) and the recruitment pattern were estimated as previously described (Nojszewska et al., 2018). We categorized MUAPs as myopathic (low amplitude and short duration with interference pattern) and "pseudo-neurogenic" (high-amplitude and/or long-duration, often polyphasic MUAPs with a reduced recruitment pattern). We also analysed the incidence of polyphasic MUAPs. EMG results were compared with the normal values using the method described previously (Bischoff et al., 1994).

Nerve conduction studies were performed unilaterally with supramaximal surface stimulation on the median, ulnar, peroneal and sural nerves using standard methodology as described previously (Nojszewska et al., 2018).

2.2. Statistical analysis

Collected data was recorded in the Microsoft Excel 2010 spreadsheet. The data set has been analysed with SAS 9.2 software. The descriptive analysis included: means, standard deviations, medians, lower and upper quartiles. In the first step, the correlations between variables using Spearman's rank correlation coefficients have been calculated. Correlation coefficients (r) were calculated with statistical significance at the level $p < 0.05$. Several non-parametric tests have been used in the analysis, like: U-Mann-Whitney, Kruskall-Wallis (for multiple comparisons), Wilcoxon (comparison of two dependent paired samples of data). A multivariate analysis of logistic regression has been used to find independent factors that influence each MUAP's parameters changes.

Table 1
Comparison of clinical and electrophysiological findings between the *Becker MC* (n = 30) and *Thomsen MC* (n = 6).

Characteristics	*Becker MC*	*Thomsen MC*
Total no of patients	30	6
Gender (F:M)	11:19	2:4
Mean age of onset, yrs (range)	8.1 ± 4.2 (0.5–19)	10.4 ± 5.7 (2.5–19)
Mean age of examination, yrs (range)	29.4 ± 15.4 (4–61)	25.7 ± 11.1 (16–47)
Mean disease duration, yrs (range)	21.3 ± 16.8 (0–56.5)	15.3 ± 7.3 (6–28)
Tested muscles (n) full EMG/MD search	104/153	23/38
BB	30	6
FID	26	5
VL	28	6
TA	20	6
FCU + ADM + EDC (only MD search)	49	15
Number of muscles where myopathic MUAPs were registered (n; %)	22 (21.1)	7 (30.4)
BB	6 (20.0)	3 (50.0)
FID	9 (34.6)	2 (40)
VL	1 (3.6)	1 (16.7)
TA	6 (30)	1 (16.7)
Number of muscles where *"pseudo-neurogenic"* MUAPs were registered (n; %)	4 (3.8)	4 (17.4)
BB	1 (3.3)	1 (16.7)
FID	0 (0)	1 (20.0)
VL	2 (6.7)	1 (16.7)
TA	1 (3.3)	1 (16.7)
Occurrence of MD (n; %)	152 (99.3)	31 (81.6)
BB	30 (1 0 0)	5 (83.3)
FID	26 (1 0 0)	4 (80.0)
VL	27 (96.4)	5 (83.3)
TA	20 (1 0 0)	5 (83.3)
ADM	16 (1 0 0)	4 (80.0)
EDC	16 (1 0 0)	4 (80.0)
FCU	17 (1 0 0)	4 (80.0)

MC: myotonia congenita; BB: biceps brachii; FID: first interosseous dorsalis; VL: vastus lateralis; TA: tibialis anterior; MUAP: motor unit action potential; SI: size index; ampl.: amplitude; mm: muscles; NCS: nerve conduction study; CMAP: compound muscle action potential.

3. Results

The pathogenic variants in the CLCN1 gene known to cause autosomal dominant MC (*Thomsen MC*) were confirmed in 6 cases (4 men and 2 women, mean age at the time of EMG examination 25.7 ± 11.1 years) and autosomal recessive form of MC (*Becker MC*) in 30 cases (19 men and 11 women, mean age at the time of EMG examination 29.4 ± 15.4 years). Clinical characteristics of patients are presented in Table 1.

Despite marked imbalance of number of MC patients in subgroups we found no statistically significant differences between *Becker MC* and *Thomsen MC* subgroups in regard to the mean patient age at the time of EMG examination and the disease onset, as well as gender distribution. Clinically persistent weakness was not found in *Thomsen MC* and was found only in 5 patients with *Becker MC* (4 men), and was most evident in distal limb muscles. In these patients the duration of symptoms was longer than 52 years (except one female patient with disease duration 38 years) and the onset of the disease was before the age of 7 (median 6.5 years).

Altogether we examined 191 muscles (70 proximal, 68.6% upper limb). The number of muscles with abnormal SA in the whole group was 183 (95.8%). In all patients only MD were recorded (details

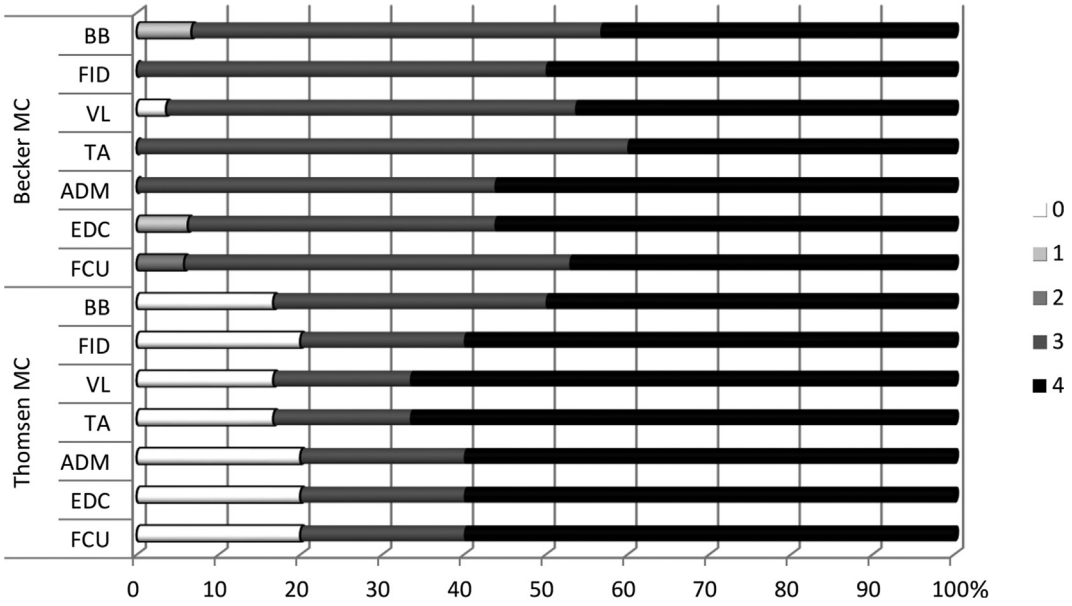

Fig. 1. Kimura scale scores' distribution in different muscles in patients with *Becker* (n = 30) and *Thomsen* (n = 6) MC. MC: myotonia congenita; BB: biceps brachii; FID: first interosseous dorsalis; VL: vastus lateralis; TA: tibialis anterior; FCU: flexor carpi ulnaris; ADM: abductor digiti minimi; EDC: extensor digitorum communis.

presented in Table 1). In the whole group of patients with MC the myotonic discharges can be described as common (grade: 3+) or widespread (grade: 4+) using Kimura scale. The grade (3+) was seen a bit more often in *Becker MC* whereas grade (4+) in *Thomsen MC* especially in distal limb muscles (FID and TA) where these differences reached the statistical significance ($p < 0.05$). All details are shown in Table 1 and Fig. 1.

Based upon the frequency and amplitude, the MD in our MC patients were classified as type 1 (classical waxing and waning MD) or type 2 (waning only MD). In *Becker MC* subgroup the type 2 of MD was more frequently registered, especially in proximal limb muscles (BB and VL). In this subgroup of patients we also saw a mixed pattern of MD (both types of MD present in one muscle) more often in BB and VL than in other muscles. All details are shown in Table 1 and Fig. 2.

The mean values of MUAP parameters as well as distribution of MUAP subtypes in all patients with myotonia congenita as well as in subgroups of patients with *Becker and Thomsen MC* are presented in Table 2.

When mean values of MUAP's parameters: amplitude, duration and SI obtained from MC patients were compared to the normal values adopted in our EMG laboratory no statistically significant differences were found. For the whole MC group we observed a statistically significant positive correlation between longer disease duration and higher values of MAUPs parameters. In VL a moderate positive correlation was seen between the duration of the disease and the amplitude and area of MUAPs ($r = 0.52$, $p < 0.001$ and $r = 0.5$, $p < 0.05$ respectively), and a mild positive correlation with SI of MUAPs ($r = 0.38$; $p < 0.05$). For MUAPs parameters in TA and the disease duration these positive correlations were even stronger (amplitude $r = 0.69$, $p < 0.001$; area $r = 0.61$, $p < 0.001$ and SI $r = 0.51$, $p < 0.05$). In

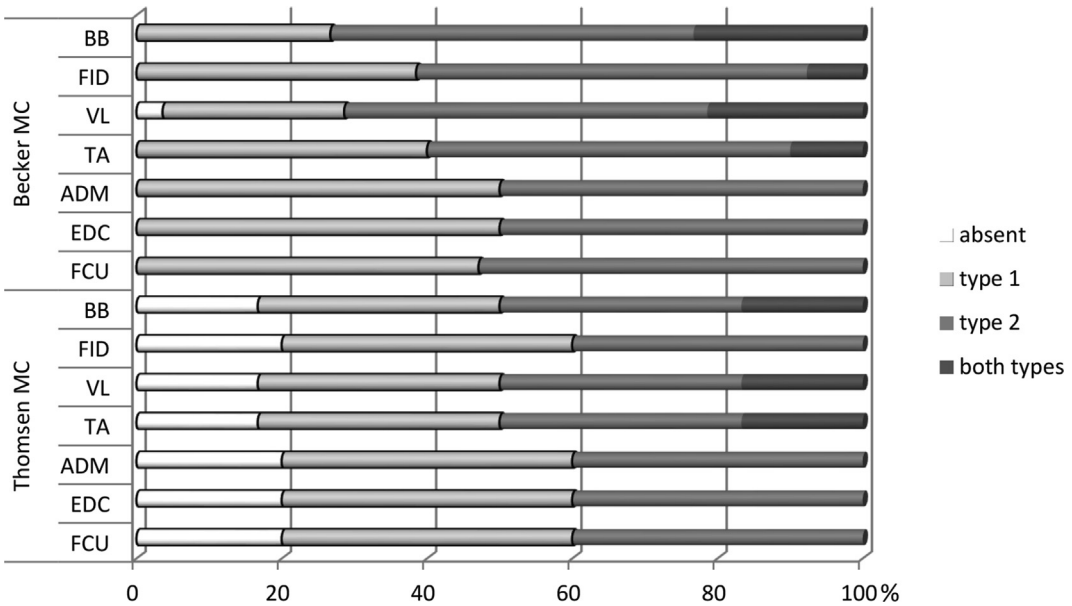

Fig. 2. Distribution of MD types in different muscles in patients with *Becker* (n = 30) and *Thomsen* (n = 6) MC. MC: myotonia congenita; BB: biceps brachii; FID: first interosseous dorsalis; VL: vastus lateralis; TA: tibialis anterior; FCU: flexor carpi ulnaris; ADM: abductor digiti minimi; EDC: extensor digitorum communis; type 1: classical waxing and waning MD; type 2: waning only MD.

Table 2
Comparison of electrophysiological data in the whole group of patients with MC (n = 36) and the subgroups with *Becker MC* (n = 30) and *Thomsen MC* (n = 6).

Characteristic		Whole MC group	Becker MC	Thomsen MC
BB	Muscle examined, n	36	30	6
	Strength (points); median (range)	4.25 (3–5)	4 (3–5)	4.5 (4–5)
	MUAP ampl. (μV); median (range)	448.5 (272–1151)	448 (272–1151)	616.5 (227–901)
	MUAP duration (ms); median (range)	9.35 (6.3–11.8)	9.15 (6.3–11.1)	9.8 (7.8–11.8)
	MUAP area (μV/ms); median (range)	656 (291–1605)	641 (291–1605)	786.5 (327–1284)
	MUAP SI (median, range)	0.66 (−0.07–1.46)	0.65 (−0.07–1.46)	0.8 (−0.02–1.4)
	Polyphasic MUAPs (%); median (range)	5 (0–15)	5 (0–15)	5 (0–14)
FID	Muscle examined, n	31	26	5
	Strength (points); median (range)	4.0 (2–5)	3.5 (2–5)	4 (3–5)
	MUAP ampl. (μV); median (range)	617 (328–1921)	609 (328–1024)	691 (497–1921)
	MUAP duration (ms); median (range)	8.75 (6.7–10.17)	8.8 (6.7–10.1)	8.5 (7.3–9.8)
	MUAP area (μV/ms); median (range)	783 (427–1873)	781 (427–1334)	878 (452–1873)
	MUAP SI (median, range)	0.77 (0.19–1.8)	0.79 (0.19–1.8)	0.74 (0.22–1.51)
	Poliphasic MUAPs (%); median (range)	5 (0–41)	5 (0–32)	8 (0–41)
VL	Muscle examined, n	15	9	6
	Strength (points); median (range)	5.0 (3–5)	4.75 (3–5)	5 (5–5)
	MUAP ampl. (μV); median (range)	648 (405–1870)	659 (405–1657)	557.5 (474–1870)
	MUAP duration (ms); median (range)	10.7 (8.5–13.5)	10.7 (8.5–13.5)	11.25 (8.8–11.9)
	MUAP area (μV/ms); median (range)	955 (514–2799)	967 (514–2545)	841.5 (708–2799)
	MUAP SI (median, range)	1.05 (0.0–1.65)	1.06 (0.0–1.65)	1.03 (0.63–2.0)
	Polyphasic MUAPs (%); median (range)	5 (0–36)	5 (0–36)	2.5 (0–15)
TA	Muscle examined, n	26	20	6
	Strength (points); median (range)	4.5 (3–5)	4.25 (3–5)	4.75 (4–5)
	MUAP ampl. (μV); median (range)	696 (298–2695)	696 (298–2695)	724.5 (407–1522)
	MUAP duration (ms); median (range)	10.3 (7.5–12.5)	10.3 (7.5–12.5)	10.3 (7.7–11.5)
	MUAP area (μV/ms); median (range)	868 (417–4157)	1010 (417–4157)	800.5 (460–1891)
	MUAP SI (median, range)	1.03 (0.27–2.26)	0.64 (0.27–2.26)	0.82 (0.29–1.54)
	Polyphasic MUAPs (%); median (range)	14 (0–40)	14 (0–40)	19 (12–23)

MC: myotonia congenital; BB: biceps brachii; FID: first interosseous dorsalis; VL: vastus lateralis; TA: tibialis anterior; MUAP: motor unit action potential; SI: size index; ampl.: amplitude; mm: muscles; n: number.

Becker MC subgroup we also found a statistically significant moderate positive correlation between the duration of the disease and amplitude, area and SI of MUAPs in BB ($r = 0.41$, $p < 0.05$; $r = 0.42$, $p < 0.05$ and $r = 0.40$, $p < 0.05$ respectively) and VL ($r = 0.58$, $p < 0.05$; $r = 0.53$, $p < 0.001$; $r = 0.40$, $p < 0.05$ respectively). The correlation between MUAPs parameters in TA and disease duration in this subgroup was even stronger (amplitude $r = 0.74$, $p < 0.001$; area $r = 0.61$, $p < 0.05$ and SI $r = 0.49$, $p < 0.05$). We also observed in this subgroup of patients a correlation between the area of MUAPs in BB and the disease duration, but this did not reach a statistical significance ($p = 0.0511$). In *Thomsen MC* we could not find such correlations, probably due to small number of patients in this subgroup. No statistical correlation was found between age and gender of the patients and MUAP's parameters.

Decreased values of mean MUAP parameters (more than 2 SD compared to controls), resulting in their categorization as myopathic MUAPs, were obtained in 22.8% of muscles in the whole MC group. When divided in two subgroups the incidence of myopathic MUAPs in *Becker MC* was 21.1% (22/104 muscles) and in *Thomsen MC* subgroup 30.4% (7/23 muscles). Myopathic MUAPs were seen most often in distal limb muscles in the former group (34.6% in FID; 20% in TA) and in the upper limb muscles in the latter group (50% in BB and 40% in FID).

In contrast, "pseudo-neurogenic" MUAPs were registered only in minority of patients with MC (in 6.3% of muscles in the whole MC group). They were more commonly registered in the *Thomsen MC* subgroup independent of the strength of the muscles. In *Becker MC* subgroup all "pseudo-neurogenic" MUAP were registered in older male patients with persistent weakness (n = 4), but only in full strength muscles. In weak muscles myopathic or less commonly normal MUAPs were found. In the woman with persistent weakness all weak muscles were assessed as myopathic.

The mean parameters of the motor and sensory nerve conduction studies were within normal limits.

4. Discussion

The only type of abnormal SA registered in our patients was MD. It was found in 95.8% of examined muscles, in almost 100% muscles of patients with *Becker MC* and in about 80% muscles in *Thomsen MC* subgroup. The difference in intensity of MD in distal limb muscles (FID and TA) between these subgroups was statistically significant.

Our findings from the whole MC group are generally in agreement with the previously published data, where needle EMG revealed diffuse MD in proximal and distal muscles in patients with clinical diagnosis but without genetic confirmation of NDM (Michel et al., 2007; Saperstein, 2008). In some cases MD were so prevalent that evaluation of voluntary MUAP morphology and recruitment was not possible (Hehir and Logigian, 2013). Various studies described myotonic discharge characteristics such as amplitude and frequency in different myotonic diseases. In few of them MD characteristics were used to differentiate between diseases type. However, often the disease was not confirmed genetically (Torbergsen et al., 2003; Drost et al., 2015). In general the former reports supported the hypothesis that the pattern and location of MD does not distinguish among the NDM disorders, especially between sodium and chloride channelopathies (Fournier et al., 2004). However Tan et al. (2011) described a low amplitude but high-frequency (150–250 Hz) discharges as a hallmark of paramyotonia congenita (PMC) – a sodium channelopathy. Most recently a systematic quantitative analysis of the MD characteristics in genetically confirmed patients with NDM was conducted by Drost et al. (2015). They observed that the first interdischarge interval (IDI1) of the rectus femoris muscle was the best variable to discriminate chloride from sodium channelopahies. A prominent difference at the onset of the myotonic trains in these two groups of NMD was assumed to be an effect of different pathophysiology underlying both disorders. To the best of our knowledge a systematic analysis of occurrence, intensity and morphology of MD in patients with genetically proven myotonia cogenita has not been reported yet.

In the whole MC group we found a statistically significant moderate to strong positive correlation between the duration of the disease and the parameters of MUAPs (amplitude, area and SI) in VL and TA muscles. Additionally when *Becker MC* subgroup was analysed we found a moderate positive correlation between the disease duration and the amplitude, area and SI of MUAPs in BB too. Myopathic MUAPs were obtained in more than 20% of muscles in the whole MC group. The incidence of myopathic MUAPs were different it two subgroups of MC patients – they were seen most often in distal limb muscles in *Becker MC* and in the upper limb muscles in *Thomsen MC* group. In contrast, "pseudo-neurogenic" MUAPs were registered only in minority of patients with MC. In *Becker MC* subgroup they were registered only in men with persistent weakness, but exclusively in full strength muscles. In our study weakness of distal limb muscles (especially FID) in neurological examination was found in 16.67% of patients with *Becker MC* (80% of them were men). In these patients the duration of symptoms was the longest and the onset of the disease was before the age of 7 years. Almost all weak muscles were evaluated as myopathic.

Our observations are in accordance with data from most recent radiological (ultrasound and MRI) studies where evidence for structural muscle changes in MC as well as for relationship between severity of myopathy and duration of exposure to the symptom were established (Trip et al., 2009b; Morrow et al., 2013). Nowadays it is clear that muscle damage can occur in the non-dystrophic myotonias including myotonia congenita but its pathomechanism and frequency are unknown (Matthews et al., 2010). In the study conducted by Trip et al. (2009b) echo intensity was significantly increased (what seemed to be caused by muscle changes such as fat and fibrosis) in all muscles, except for the rectus femoris muscle. The greatest ultrasound abnormalities were detected in forearm flexors of patients with chloride channelopathy and men. The muscles changes seemed to increase with age and degree of muscle use. They reported also that the muscle echo intensities were negatively correlated with the corresponding range-of-motion which may be the result of muscle shortening due to pathological processes in muscles. The range-of-motion in the work of Trip et al. (2009b) was significantly lower in chloride channelopathies.

In the muscle MRI study by Morrow et al. (2013) fatty infiltration in T1-weighted sequences was found in 30% (128/420) of thigh muscles and in 37% (94/252) of calf muscles in patients with NDM. The changes were categorized as mild extensive in 14% and marked in 19% for both calf and thigh sequences. In the calf there was a relative sparing of TA. The second obtained parameter – STIR hyperintensity, which represents muscle oedema caused most probably by toxic, metabolic or inflammatory changes, was not identified in any of the thigh images but was identified in the calf images in 90% (19/21) of patients, most commonly in the medial gastrocnemius muscle (15/21), followed by lateral gastrocnemius muscle (9/21) and TA muscle (5/21). The authors described also a potential MRI hallmark of chloride channelopathy – the presence of a hyperintense stripe in medial gastrocnemius, which was present in 10/11 CLCN1 patients (Morrow et al., 2013). There was also a significant correlation between overall MRI involvement and age despite the diversity in phenotype and genotype. The correlation was stronger for the recessive MC. In another study Kornblum et al. (2010) reported no abnormalities in 3 patients with recessive MC using whole body MRI including T1w, T2w and fat-supressed T2w, but it was a small study in young MC patients. The main discrepancy between these two MRI studies probably resulted from differences in age as myopathies in NDM have been mainly reported in patients over 65 years (Colding-Jørgensen, 2005; Matthews et al., 2010; Raja Rayan et al., 2010).

Our results in association with radiological data support the hypothesis that the muscle changes in MC could be exacerbated by duration of the disease especially by exposure to the symptom. On one hand, the cumulative afterdepolarization could give rise to the self-maintaining activity. On the other hand large depolarization (of 10–20 mV) may force enough sodium channels into the inactive state as to render the membrane temporarily inexcitable. This may explain a transient weakness observed in patients with recessive MC (Colding-Jørgensen, 2005). This prolonged high intracellular sodium concentration in MC could induce muscle fiber damage, which might subsequently lead to functional changes in terms of weakness and contractures (Trip et. al., 2009b). The second possible explanation is mechanism documented in healthy muscles after exercise. The changes in muscle following exercise resulted from a number of proposed mechanisms including water shift from intra- to extracellular spaces, increase in extracellular or vascular fluid volumes, and increase in proportion of "free" water to macromolecular "bound" water (Ababneh et al., 2008). Considering that myotonia represents abnormally prolonged muscle contraction, similar mechanisms might cause the muscle changes observed in NDM patients (Morrow et al., 2013).

Our observation regarding morphology of MUAPs is slightly different than previously published data. In the majority of former published reviews MUAPs were obtained as normal in *Thomsen MC* and myopathic or normal in *Becker MC* (Colding-Jørgensen, 2005; Heatwole et al., 2013) but mostly the diagnosis of MC was made based on the clinical features, inheritance patterns and electrophysiological exercise stimulation tests. To our knowledge no systematic quantitative analysis of MUAPs' parameters in genetically proven MC has been published yet. The presence of myopathic MUAPs in *Becker MC* is not surprising since atrophic distal limbs and anterior neck (specifically sternocleidomastoids) muscles on physical examination were described (Colding-Jørgensen, 2005).

The new finding in our study is a positive correlation of MUAPs parameters such as amplitude, area and SI in BB, VL and TA with duration of the disease, which in some cases (in our material with the longest disease duration) led us to obtaining *"pseudo-neurogenic"* MUAPs but only in older men with persistent weakness, in full strength muscles exclusively. We could not exclude, that sex related factors might play an important role in compensatory mechanism, which counteracts with progression of muscles' weakness in the course of MC.

In conclusion, this study supports the hypothesis that needle EMG examination could still be helpful in diagnosis of myotonia congenita and together with exercise and cold stimulation tests be useful in differentiation between recessive and autosomal form of MC.

Ethical publication statement

We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

Declaration of Competing Interest

None of the authors has any conflict of interest to disclose.

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