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Letter to the Editor

Motor unit potential changes in myofibrillar myopathy

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Dear Editor,

Both conventional and quantitative electromyography (Q-EMG) studies have been conducted in relatively large samples in common myopathies such as muscle dystrophies, inflammatory myopathies and mitochondrial myopathies [1,2]. These studies report a rather modest diagnostic sensitivity, with lowest sensitivity values found in mitochondrial myopathies [3]. In contrast, motor unit action potential (MUP) analysis appears very sensitive in sporadic inclusion body myositis, where a decreased mean duration is observed in 94% of the patients [4].

In rare myopathic syndromes, the diagnostic value of Q-EMG remains unknown. In the present study we examined a group of patients with histologically confirmed myofibrillar myopathy (MFM) in order to.

(i) determine the degree and characteristics of quantitative MUP changes.
(ii) investigate possible associations of MUP abnormalities with specific muscle biopsy findings.

We retrospectively reviewed results of muscle biopsies and electromyography from the laboratories of myopathology and clinical neurophysiology of our clinic. 10 patients (5 females, mean age: 54.3 years, range: 24–77 years) having both biopsy proven MFM and Q-EMG workup were identified and included in the study. In 4 patients molecular testing had revealed one of the known MFM mutations (3 subjects had the Desmin (DES) and 1 the Valosin-containing protein (VCP) mutation. Genetic testing was inconclusive in the other cases. All patients were contacted and gave written informed consent for participation in the study which was conducted in accordance with the Declaration of Helsinki and approved by the authors' institution's ethics committee.

Needle EMG was performed with concentric electrodes on a Medtronic (Keypoint 31A02) apparatus. At least 20 MUPs from 3 to 4 insertion points were collected by using a signal decomposition technique of multiple simultaneously active motor unit potentials ("multiMUP"). Normal values for mean MUP parameters as well as for outliers were taken from Bischoff et al. [5]. In muscles with pathologically increased polyphasia, only simple MUPs were used for parameter calculations. In muscles with no (or not abnormally high percentages of) polyphasia, all MUPs were taken into account. After testing for normality, means and standard deviations were used to describe the Q-EMG data. Pearson correlation and χ^2 tests were employed to investigate relations between MUP parameters and histological features.

The complete Q-EMG dataset comprised 31 muscles. Unfortunately, due to the retrospective nature of the study, there was a sampling mismatch in many of the examined muscles, so direct correlation of MUP changes with biopsy findings was possible only in 12 muscles.

Cumulative Q-EMG results from all 31 muscles were suggestive of a myopathic profile with reduced MUP duration, size index (SI = 2 x log10(amplitude) + (area/amplitude)) and thickness. Further analysis conducted for each muscle separately (Table 1), revealed abnormally short MUP duration in tibialis anterior, deltoid and first dorsal interosseous, increased number of phases in vastus lateralis and deltoid, reduced SI in deltoid and first dorsal interosseous and reduced thickness in tibialis anterior, deltoid and first dorsal interosseous. We did not observe neurogenic Q-EMG abnormalities (e.g. increased MUP duration or amplitude).

Regarding muscle biopsy and Q-EMG correlations, the variation coefficient of muscle fibre diameter (determined manually using the Image-Pro Plus 4.5 software) was positively correlated with mean MUP duration, i.e. longer MUP durations were associated with a larger fibre diameter variability (Pearson correlation coefficient = 0.99, $p < 0.05$). It should be noted that here only 11 out of 12 muscles were entered into analysis, since there was one missing value regarding the variation coefficient of muscle fibers of the biceps brachii of subject number 2. A further interesting finding was that the mean diameter of Type 1 muscle fibers was positively correlated with mean MUP amplitude (Pearson correlation coefficient = 0.97, $p < 0.05$). No other significant correlations were observed between MUP parameters and quantitative biopsy findings.

Hence, MFM MUPs exhibited less marked alterations compared to those seen in classical myopathic syndromes such as the muscular dystrophies. The most pronounced changes were observed in the deltoid muscle which exhibited all Q-EMG features typical for myopathy. Myopathic MUP values were also obtained from the first dorsal interosseous and the tibialis anterior muscle. Despite the preponderant proximal muscle involvement in most myopathic syndromes, the

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Table 1
Q-EMG parameters for all studied muscles (n = 31). Abnormal findings are highlighted in bold font if they deviated from the reference limits provided in Bischoff et al. [5], which were available only for tibialis anterior, biceps brachii, vastus laterals, deltoid and first dorsal interosseous. Reference limits according to Bischoff et al. [5] are provided in square brackets. All outliers were abnormal with the respect to the lower normal limit. It should be noted that reference ranges for MUP parameters in our laboratory are very similar to the ranges given in Bischoff et al. [5].

	Tibialis anterior (n = 8)	Biceps brachii (n = 5)	Vastus lateralis (n = 5)	Deltoid (n = 4)	1st dorsal interosseous (n = 2)	Rectus abdominis (n = 2)	Orbicularis oris (n = 2)	Biceps femoris (n = 1)	Thoracic paraspinal (n = 1)	Orbicularis oculi (n = 1)
Mean Duration (ms) [reference limits for mean], [reference limits for outliers]	**7.3** [9.3–13.8], [4.6–18.4]	7.9 [7.1–12.7], [4.2–16.4]	9.8 [7.9–15.5], [4.6–21.6]	**6.8** [7.8–13], [162–1531]	**6.1** [6.8–12], [4–18]	6.8	3.4	8.7	6.8	3.4
Mean outliers (n) for Duration	2.2	**3.5**	0.5	**7.0**						
Mean Amplitude (μV) [reference limits for mean], [reference limits for outliers]	610.0 [194–1174], [158–1572]	441.8 [206–666], [178–1414]	761.0 [206–1165], [172–1954]	475.5 [330–770], [162–1531]	740.5 [258–1246], [188–2301]	362.0	212.0	334.0	333.0	242.0
Mean outliers (n) for Amplitude	1.4	**4.3**	0.3	**3.0**						
Polyphasia (%)	13.5	18.5	**36.8**	**27.5**	12.0	22.5	12.0	2.0	10.0	8.0
Mean SI [reference limits for mean], [reference limits for outliers]	0.7 [0.57–1.77], [−0.397–2.463]	0.2 [−0.01–1.31], [−0.539–2.053]	1.1 [0.46–2.02], [−0.478–2.916]	**0.2** [0.23–1.47], [−0.478–2.916]	**0.0** [0.22–1.74], [−0.912–2.281]	0.2	0.0	0.5	0.0	−0.8
Mean Thickness (ms) [reference limits for mean], [reference limits for outliers]	**1.2** [1.21–2.1], [0.575–2.81]	1.1 [1.06–1.86], [0.564–2.093]	1.6 [1.26–2.18], [0.602–3.11]	**1.0** [1.12–2], [0.648–2.943]	**0.5** [1.06–1.86], [0.564–2.093]	1.2	0.9	1.5	1.0	0.6

present data from biceps brachii and vastus lateralis muscles exhibited little or no abnormalitiy. This probably reflects the phenotypic variability of muscle weakness distribution in MFM which is predominantly distal or mixed (proximal and distal) in many patients [6]. In agreement with previously published reports, fibrillation potentials and positive sharp waves were uncommon (7 out of 31 muscles), while myotonic discharges were more frequent (12 out of 31 muscles).

Comparisons of histological features with Q-EMG parameters yielded two significant findings. First, there was a positive correlation between muscle fibre diameter variability seen in muscle biopsy (pooled for type I and type II fibers) and duration of MUPs. This is in agreement with published computer simulations of MUPs with varying combinations of duration, amplitude and number of phases which show that MUPs with the same number and equal mean diameter of fibers develop longer durations if they exhibit larger diameter variability [7]. The second finding indicates that type I muscle fibers of small diameter are more likely associated with small amplitude MUPs. It reflects the fact that MUP amplitude scales with the distance from the tip of the electrode and the diameter of the closest muscle fibre. A size reduction of the latter produces a lower action potential amplitude. It should be kept in mind that the low effort at which the MUPs are collected, probably introduces a sampling bias towards type 1 muscle fibers. The attempt to correlate MUP parameters with qualitative, partly MFM specific, histopathological characteristics failed to reveal any statistical correlation.

Overall, the presence of only a few significant Q-EMG -biopsy correlations might be partly caused by the small number of the included patients or the phenotypic inhomogeneity of the MFM syndrome as such. Nonetheless, in the few studies of the more common myopathies that correlated Q-EMG to histology, there were significant discrepancies between MUP features muscle biopsy [8,9].

Clearly, recruitment of more cases with this rare myopathy will require multicenter collaborations and that would allow testing electrophysiological features in specific, genetically defined, MFM subgroups.

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