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Motor unit reorganization in progressive muscular dystrophies and congenital myopathies

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ABSTRACT

The aim of this study was to analyze motor unit reorganization in different types of progressive muscular dystrophies and congenital myopathies.

The study population consisted of patients with genetically verified progressive muscular dystrophies: Duchenne (DMD) ($n = 54$), Becker (BMD) ($n = 30$), facio-scapulo-humeral (FSHD) ($n = 37$), and Emery–Dreifuss (E-DD) ($n = 26$). Patients with probable limb-girdle dystrophy (L-GD) ($n = 58$) and congenital myopathies ($n = 35$) were also included in the study. Quantitative EMG recordings were obtained from 469 muscles. Muscle activity at rest and during slight voluntary and maximal muscle contraction was analyzed. The motor unit activity potential (MUAP) duration, amplitude, area, size index (SI), polyphasicity, and the presence of "outliers" were evaluated.

Diminished values of MUAP parameters and decreased maximal amplitude of maximal muscle contraction were recorded most frequently in DMD and mainly in the biceps brachii muscles. SI was the most frequently changed EMG parameter. "Outliers" with amplitude below the normal range were recorded more frequently then a decreased mean MUAP amplitude (what could indicate a very high sensitivity of this EMG parameter). Pathological interference pattern was recorded in 34.7% of biceps brachii and in 21.2% of rectus femoris muscles. In FSHD, decreased MUAP duration and SI and pathological interference pattern with low amplitude were recorded most frequently in the tibial anterior and deltoid muscles.

The presence of potentials with reduced parameters is a result of decreasing motor unit area (reduced number and size of muscle fibers), while high amplitude potentials recorded in BMD and E-DD could indicate a slow and mild course of disease and muscle regeneration.

1. Introduction

1.1. *Progressive muscular dystrophies*

Progressive muscular dystrophies (PMD) are a group of congenital muscle disorders with different clinical symptoms, course, and prognosis. Some forms of PMD are associated with defects in structural proteins connected to the sarcolemma (dystrophin, various sarcoglycans, dysferin, merosin) or are directly associated with abnormalities of nuclear membrane proteins (emerin, lamin A/C). Common features of protein defects in PMD and congenital myopathies (CM) result in

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muscle weakness and wasting, although some differences in their phenotypes are also observed [1–9].

1.2. *Diagnosis of progressive muscular dystrophies*

Genetic testing is considered the only reliable diagnostic criterion in neuromuscular disorders but this method is rarely the first line of laboratory tests and needs guidance from other methods. Electrophysiological tests could be the first key tool for the diagnosis of primary muscle diseases, especially in limb-girdle dystrophy, and they remain very important in the evaluation of disease progression and muscle dysfunction [10–13].

1.3. *Electromyography in progressive muscular dystrophies*

By EMG, the criteria for myopathy in primary muscle diseases are most commonly fulfilled as decreased values of single motor unit action potentials (MUAPs), an increased percentage of polyphasic potentials, and a pathological interference pattern at maximal muscle activation [14–21]. In addition to short and low MUAPs, characteristic for myopathy, potentials with an increased amplitude and prolonged duration are also observed and their origin has not been sufficiently explained yet. Contribution of a neurogenic factor to reorganization of a myopathic motor unit has been discussed in the literature [16,22–25].

1.4. *Aims of the study*

The aims of the study were:

- to analyze EMG recordings obtained in progressive muscular dystrophies (PMD), including Duchenne (DMD), Becker (BMD), limb-girdle (L-GD), facio-scapulo-humeral (FSHD), and Emery–Dreifuss (E-DD) types, and in congenital myopathies (CM);
- to compare EMG data in two dystrophinopathies, quickly progressing DMD and a more benign, slowly progressing BMD;
- to compare EMG data in a dystrophinopathy (BMD) and in a nucleopathy (E-DD), both with a slow course of disease and different localization of muscle structural lesions.

2. Material and methods

2.1. *Characteristics of patients*

Two hundred and forty patients (186 M; 54 F, mean age 17.5 years) were recruited to the study at the Department of Neurology, Medical University of Warsaw, and a written informed consent was provided by all participants.

The study population consisted of genetically verified patients with four progressive muscular dystrophies: DMD, BMD, FSHD, and E-DD. In addition, patients with probable L-GD and with CM after combining clinical status data and biopsy findings were included in the study group (Table 1).

2.2. *EMG studies*

Electromyographic (EMG) recordings were obtained from 469 muscles (186 biceps brachii (BB), 219 rectus femoris (RF), and additionally in the FSHD group also 34 tibial anterior (TA), and 30 deltoid (DD) muscles) (Table 1).

Strength of the examined muscles was assessed using the MRC scale (0–5, with 0 indicating no action, and 5 indicating normal muscle strength), and muscle atrophy was assessed using a 0–3 scale (0 – no atrophy, 3 – marked atrophy).

Muscle activity was recorded during routine EMG examinations using a concentric needle electrode (DCN37 type) with 0.07 mm^2 uptake area, 0.46 mm diameter and 37 mm length. The Keypoint system (Medtronic) was used to evaluate EMG recordings. EMG recordings were registered at muscle rest and during slight voluntary (according to the multi-MUAP method) and maximal muscle contractions.

At rest, spontaneous activity was analyzed, including pseudomyotonic discharges, positive sharp waves, and fibrillations.

During voluntary muscle contraction (10–20% of maximal muscle contraction), automatic quantitative evaluation of single motor unit potentials (MUAPs) was performed, and the evaluated parameters included duration, amplitude, area, size index (SI), and polyphasicity [26–28]. The mean values of these parameters and the number of outliers (minimum 3 single MUAPs) out of the normal range were calculated [29]. Mean results were compared to the reference values according to Bischoff and Stålberg [30,31] used in our laboratory. The mean

Table 1 – Characteristics of patients.

Diagnosis	Sex		Age years (range)	Number of muscles			
	M	F		Biceps brachii	Rectus femoris	Deltoid	Tibial anterior
Duchenne muscular dystrophy	54		6 ± 2 (4–15)	27	29		
Becker muscular dystrophy	30		13 ± 6 (3–24)	35	53		
Fascio-scapulo-humeral dystrophy	23	14	28 ± 16	35	37	34	30
Emmery–Dreifuss dystrophy	26		18 ± 6	21	23		
Limb-girdle dystrophy	31	27	19 ± 11 (12–57)	42	55		
Congenital myopathies	22	13	14 ± 11	26	22		
Overall	186	54	17.5	186	219	34	30