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Relationships between clinical data and quantitative EMG findings in facioscapulohumeral muscular dystrophy

Dystrofia twarzowo-łopatkowo-ramieniowa – korelacje elektrofizjologiczno-kliniczne

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Abstract

Background and purpose: In recently published reports, electrophysiological findings were analysed, in some facioscapulohumeral muscular dystrophy (FSHD) cases without genetic disease confirmation. In several reports, some electrophysiological findings were described, not specific for myopathy. The aim of study was to analyse electrophysiological findings in a genetically homogeneous FSHD group to find possible relationships between electromyography (EMG) abnormalities and clinical symptoms.
Material and methods: 37 patients with genetically proven FSHD (23 men and 14 women) aged 7-58 years (mean 28.8 years) were studied. Electromyographic examinations were done according to a uniform scheme for FSHD. Quantitative EMG examination was performed in vastus lateralis, tibialis anterior, deltoid and biceps brachii muscles.
Results: There was no correlation between clinical features and electrophysiological findings. EMG confirmed myopathic changes in all patients with most advanced changes in tibialis anterior and deltoid muscles. Some of these changes were unspecific for myopathy and the degree of their intensity differed in particular muscles. The most advanced changes were observed in the tibialis anterior and deltoid muscles. The usefulness of the size index for myopathic processes assessment was confirmed. Analysis of so-called outliers

Streszczenie

Wstęp i cel pracy: W dotychczas publikowanych doniesieniach analizowano zjawiska elektrofizjologiczne u chorych z dystrofią twarzowo-łopatkowo-ramieniową (*facioscapulohumeral muscular dystrophy* – FSHD), niekiedy bez genetycznego potwierdzenia rozpoznania. W niektórych badaniach wykazano obecność nieswoistych dla miopatii zmian elektrofizjologicznych. W pracy podjęto próbę oceny zmian elektromiograficznych (EMG) w genetycznie homogennej grupie chorych na FSHD. Przeanalizowano zmiany elektrofizjologiczne i wyłoniono ewentualne korelacje elektrofizjologiczno-kliniczne w FSHD.
Materiał i metody: Materiał stanowiła grupa 37 pacjentów z genetycznie potwierdzonym rozpoznaniem FSHD (23 mężczyzn i 14 kobiet) w wieku 7–58 lat (średnia wieku: 28,8 roku). Badania EMG przeprowadzono wg jednolitego schematu. Wykonywano ilościowe badanie EMG mięśni: obszernego bocznego uda i piszczelowego przedniego, naramiennego, dwugłowego ramienia.
Wyniki: Nie wykazano korelacji pomiędzy stopniem nasilenia objawów klinicznych a parametrami EMG. Badania pozwoliły na potwierdzenie miopatycznego charakteru zmian. Zmiany były nieswoiste, a stopień ich nasilenia różny w poszczególnych mięśniach. Największe zmiany obserwowano w mięśniu piszczelowym przednim i naramiennym.

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for motor unit activity potential parameters did not show any new data for evaluation of the myopathic process. Myopathic changes in our material were not as advanced as those described in classical dystrophies. Histopathological examinations of skeletal muscle were normal in about 1/3 of patients.

Conclusions: We established that myopathic changes are clearly present in FSHD, with different degrees of intensity, most pronounced in tibialis anterior and deltoid muscles. There was no correlation between electrophysiological findings and clinical features. The size index provided the highest motor unit potential diagnostic sensitivity in FSHD.

Key words: myopathy, facioscapulohumeral muscular dystrophy, electromyography, motor unit potential, size index.

Potwierdzono przydatność wskaźnika wielkości w ocenie procesu miogennego, analiza tzw. *outliers* dla parametrów potencjałów jednostki ruchowej nie wniosła nowych danych. Zmiany miopatyczne w analizowanym materiale nie były tak zaawansowane jak te w klasycznych dystrofiach. Badanie histopatologiczne mięśnia szkieletowego w ok. 1/3 przypadków było prawidłowe.

Wnioski: W FSHD badania elektrofizjologiczne potwierdzają miopatyczne uszkodzenie mięśni, o różnym stopniu nasilenia w poszczególnych mięśniach. W największym stopniu zmiany obecne są w mięśniu piszczelowym przednim i naramiennym. Parametry elektromiograficzne nie wykazują korelacji ze stopniem zaawansowania objawów klinicznych. W opracowaniu autorów największą diagnostyczną czułość dla zmian miopatycznych wykazywał wskaźnik wielkości potencjałów czynnościowych jednostek ruchowych.

Słowa kluczowe: miopatia, dystrofia twarzowo-łopatkowo-ramieniowa, elektromiografia, potencjał czynnościowy jednostki ruchowej, wskaźnik wielkości.

Introduction

Facioscapulohumeral muscular dystrophy (FSHD), one of the most common muscular dystrophy variants [1], was first described in the middle of the 19th century [2]. The inheritance is autosomal dominant and the genetic defect results from deletion of D4Z4 tandem repeats at the 4q35 locus [3]. The molecular pathology is very complicated and not clearly understood [4].

Clinical features are very characteristic, and the distribution of the weakness is quite unique. Facial weakness is evident in limited movements of lips; patients are unable to whistle or inflate their cheeks. Scapular winging and characteristic appearance of the shoulder girdle, so-called triangular shoulders, is typical. Lower limb weakness affects mainly distal muscles, especially the anterior tibialis muscle, which, if evident, may suggest the neurogenic process. Contrary to other dystrophies, asymmetry and selectivity of muscle involvement are very characteristic for FSHD.

Before molecular tests became available, the diagnosis of FSHD was based on the clinical grounds, family history and electromyography (EMG) findings. At that time, FSHD diagnosis in some cases was difficult to make, due to relatively high frequency of atypical and subclinical cases, also due to intra- and interfamilial clinical variability of the disease.

The results of EMG in FSHD were generally assessed as myopathic [5,6]. In some patients, however, EMG changes atypical for myopathy were described. Changes atypical for myopathy included increased amplitude of single motor unit potentials (MUPs) and spontaneous activity at rest [7]. One of the causes of atypical electrophysiological findings recorded in FSHD could be the presence of inflammatory changes seen in a muscle specimen [8-11]. Munsat even suggested the existence of an FSHD inflammatory variant and showed the presence of fibrillations at rest, which he interpreted as a neurogenic sign [12]. It is now known that fibrillations and positive sharp waves may be recorded not only in neurogenic but also in myopathic processes, especially in the acute stages of myositis and in quickly progressive muscular dystrophy [13,14]. There are no current data available on the frequency of so-called inflammatory changes in FSHD muscles in patients with the diagnosis confirmed by molecular tests.

The aim of the study was electrophysiological characterisation of FSHD, especially in the view of recent reports. We intended to assess the relationship between clinical and electrophysiological findings and discuss appropriate selection of particular muscle and MUP parameters to be tested. We also planned to assess the electrophysiological changes in subclinical cases of FSHD.