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A Study of Development of Pseudohypertrophisms in a Patient with Duchenne Muscular Dystrophy

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Abstract

Hypertrophy is the abnormal enlargement of a body part or organ, especially when bodybuilder or athletes take androgenic hormone, likes steroids, androgen, corticoids the muscles can hypertrophy. In Duchenne Muscular Dystrophy (DMD) the Pseudohypertrophisms is found in the patients. The aim of the study is to monitor the development of the pseudohypertrophisms in the DMD patients. The molecular diagnosis and Creatine Phosphokinase test was carried out to confirm the DMD. Anthropometric analysis of enlargement of a body part, organ and muscles were recorded. The collected data was compared with the age matched control participants. The abnormal enlargement of a body muscles and organ were observed in patients. We also collected data regarding the onset of pain in hypertrophic muscles. The DMD is fatal neuromuscular genetic disorder, still no assure cure available on it, though the recent discoveries in genetics give us a wide area of Gene Therapy. The prevention and reduce fatal effect is the only way to control the diseases. Therefore diagnosis in the early stage is important in the DMD. In DMD, the pseudohypertrophisms found in the early stage and it is one of the most primitive symptoms of DMD.

Keywords: Duchenne muscular dystrophy, pseudohypertrophisms, EMG, NCV.

Introduction

Duchenne muscular dystrophy (DMD) is the most common form of muscular dystrophies (MDs) as well as the most frequent muscle disease among children [1, 2] affecting 1:3500 live male births. DMD is a fatal neuromuscular genetic Disorder [2-4] caused by mutations in the DMD gene. The disease is characterised by progressive failure of muscle growth and muscle wasting. The affected individuals show weakness in muscles, beginning from the proximal muscles and gradually progressing towards the distal muscles. The organ or body muscles excessively enlarge due to increased size in the constituent cells and begin to hypertrop. The individuals are also known to have problems in carrying out the motor functions and as the condition worsens, it leads to loss of movement and as the muscle degradation progresses it affects the muscles of vital organs such as the heart, respiratory system and eventually the individual dies due to the disease [5].

Hypertrophy in humans has been attributed to deposition of fat and connective tissue (pseudohypertrophy). Increased muscle mass (true hypertrophy) has been documented in muscle builders.

Materials and Methods

To study the hypertrophy in DMD the anthropometic parameters like are measured Hypertrophy in humans

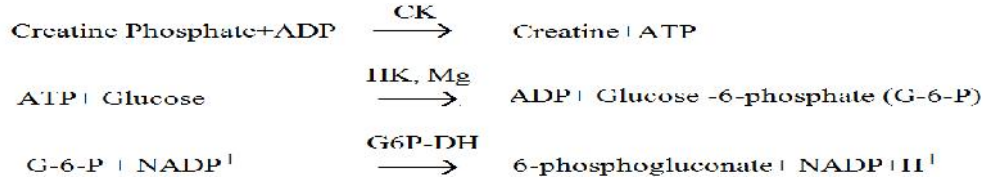
has been attributed to deposition of fat and connective tissue (pseudohypertrophy). Increased muscle mass (true hypertrophy) has been documented in animal models.

Anthropometric analysis

The various anthropometric parameters such as Weight, Height, Arm Span, Chest and circumference of Arms, Thigh and Calf were measured as per the anthropometric standard.

CPK Assessments

A venous blood sample was collected for serum Creatinine Phosphokinase (CPK) levels followed by routine investigations and ultrasonography (USG) of abdomen, transvaginal ultrasound, laparoscopy and/ or laparotomy where appropriate. [6] CPK levels were determined by Kinetic UV Method-NAC Activated.



Nerve Conductance Velocity (NCV)

The sensory and motor nerve conduction velocity and latency in bilateral upper and lower limb nerves was found normal but show slightly reduced amplitude bilateral peroneal nerves. The f-wave study for bilateral median nerve was also normal [7]

Electromyography (EMG)

Deltoid (Left) showed normal insertional activity and reduced amplitude and interference pattern. Medial Head Left Gastrocnemeous also showed reduced insertional activity and reduced amplitude and interference pattern with mild fasciculation, reduced motor units.[8]

Observations and Result

Of the 123 scapula included, 64 belonged to right and 59 to left side. The shape of the GC was found as inverted comma, pear, triangular and oval. The most common shape observed was of pear shaped GC in 69 (56.09%) of 123 scapula. 43 (34.95%) were of inverted comma shape, 8 (6.5%) of oval shape and 3 (2.4%) were triangular.

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Anthropometric analysis

The various anthropometric parameters such as Weight, Height, Arm Span, Chest and circumference of Arms, Thigh and Calf were measured.

Table-1. Anthropometric analysis

S.No	Anthropometric Parameters	Observed	Expected	%. tile	%. type
1	Weight	35.2 Kg	28.1Kg	3rd	72.22%
2	Height	122.5 cm	132.6cm	10th	92.45%
3	Arm Span	125 cm	126.7cm	10th	92.53%
4	Chest	57.5cm	53.75cm	10th	93.65%
5	Arms circumference	R-16.25cm L-16.75cm	17.0cm 17.0cm	10th 25th	95.58% 98.52%

The thigh circumference in DMD patient is on right side 11.2” and on left side 10.9”. Similarly the calf circumference on right side is found to be 9.7” and on left side 8.9”.

CPK

Estimation of the CPK levels in DMD case analysed 8 times during the period of three year, which revealed that the levels ranged from 202 to 15,907 IU/L (mean 5,113±6234 IU/L).

During early stages of disease the CPK levels was significantly elevated. During the course of disease the CPK gradually falls but in late stages it remains above normal.

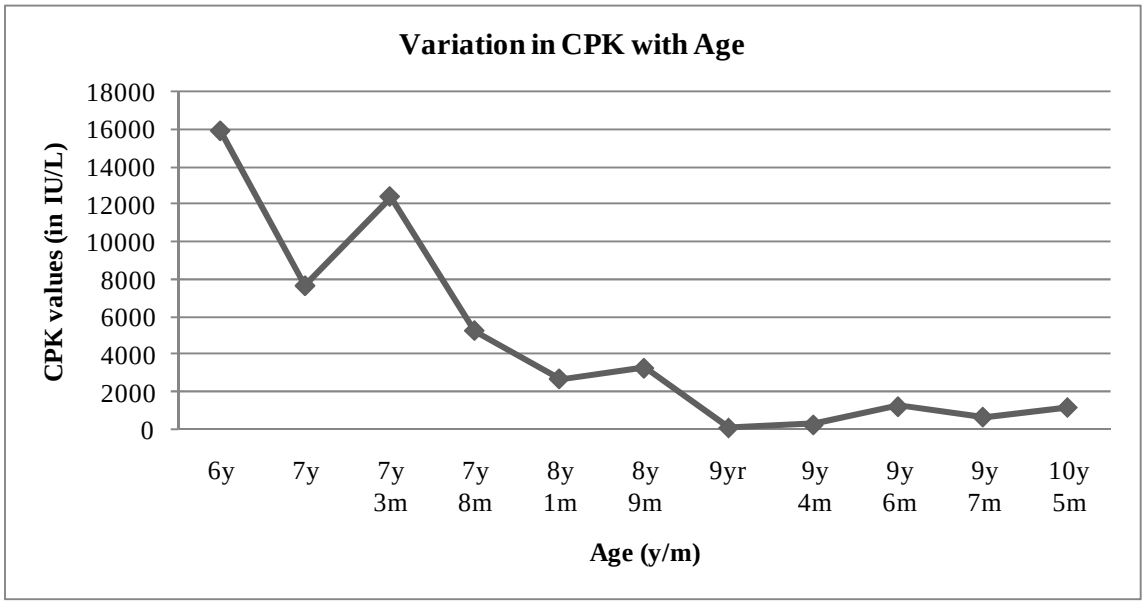


Fig. 1: Variation in CPK with Age

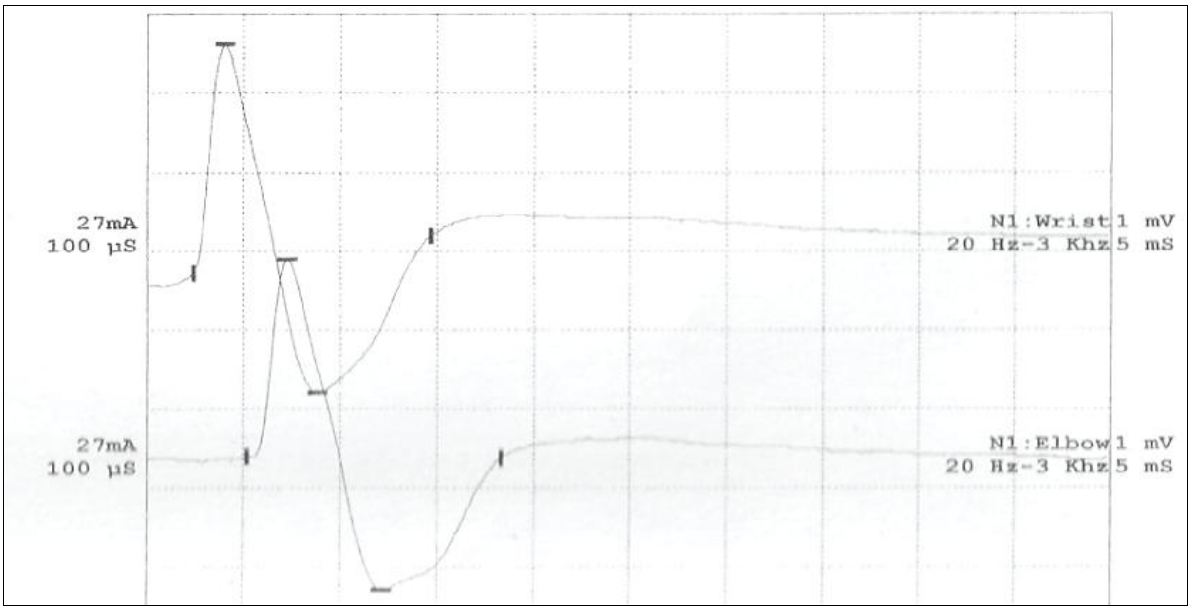
Nerve Conductance Velocity (NCV)

The sensory and motor nerve conduction velocity and latency in bilateral upper and lower limb nerves was found normal but show slightly reduced amplitude in bilateral peroneal nerves. The f-wave study for bilateral median nerve was also normal (Fig. 2, a-h).

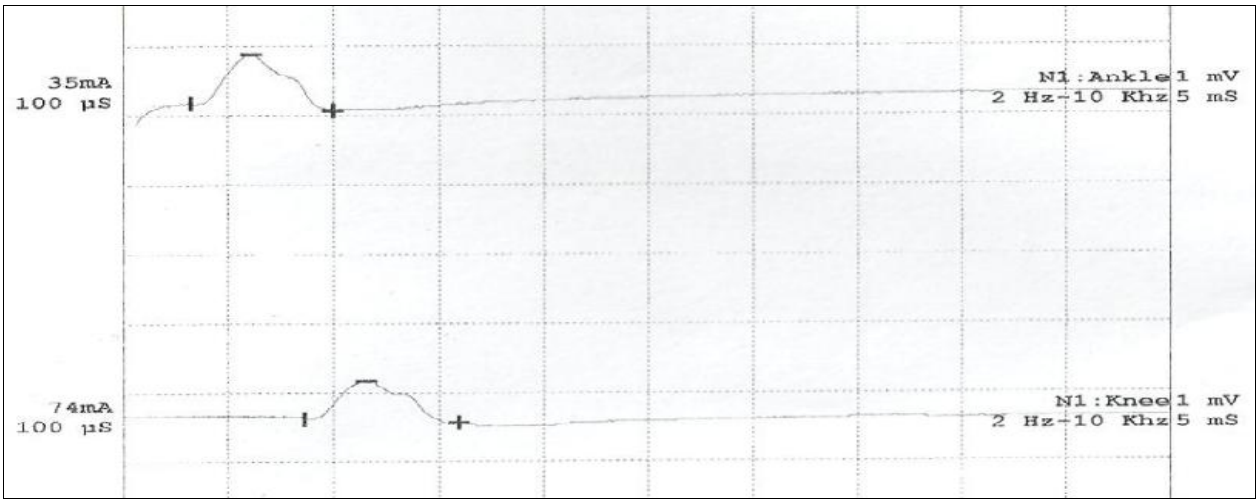
Electromyography (EMG)

Deltoid (Left) showed normal insertional activity and reduced amplitude and interference pattern. Medial Head Left Gastrocnemeous also showed reduced insertional activity and reduced amplitude and interference pattern with mild fasciculation, reduced motor units (Fig. 2, a-h).

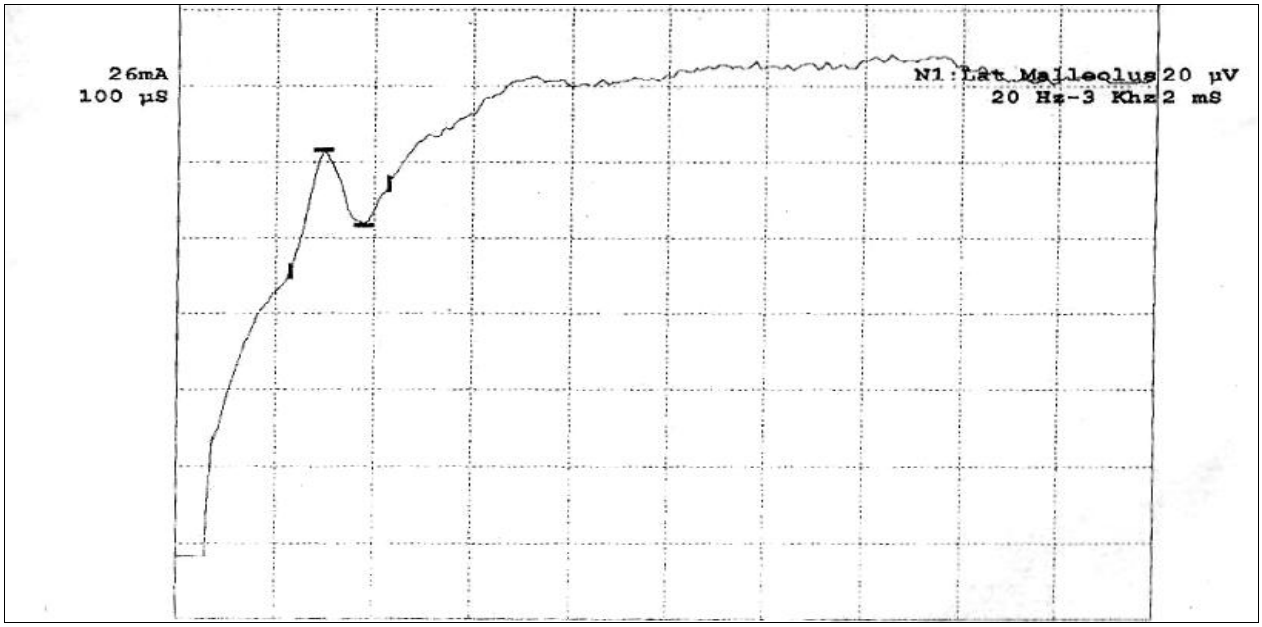
a


27mA
100 µS
N1:Wrist 1 mV
20 Hz-3 Khz 5 mS
27mA
100 µS
N1:Elbow 1 mV
20 Hz-3 Khz 5 mS

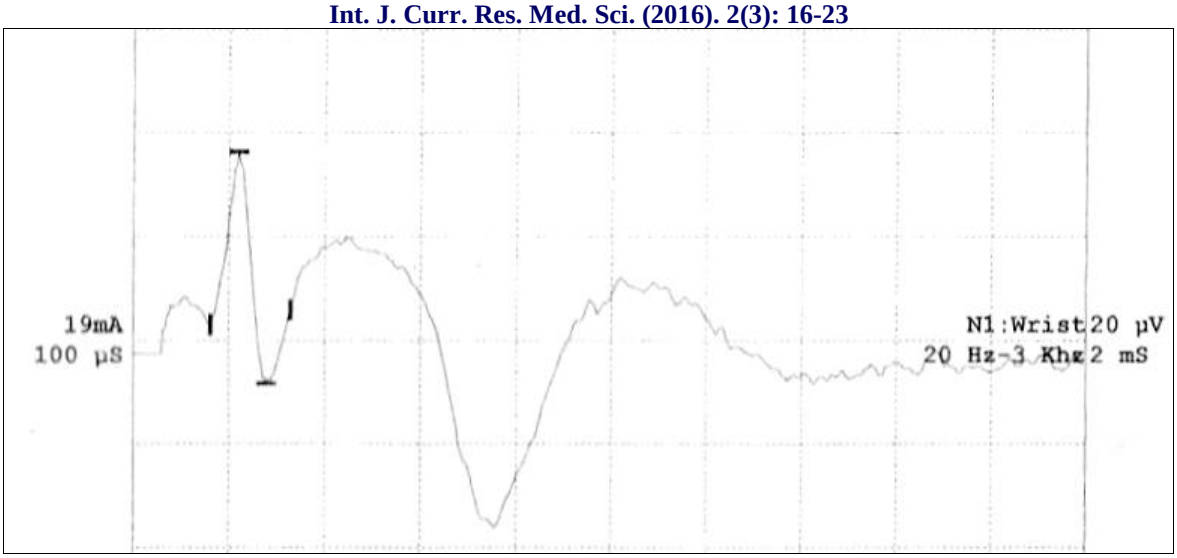
b


35mA
100 µS
N1:Ankle 1 mV
2 Hz-10 Khz 5 mS
74mA
100 µS
N1:Knee 1 mV
2 Hz-10 Khz 5 mS

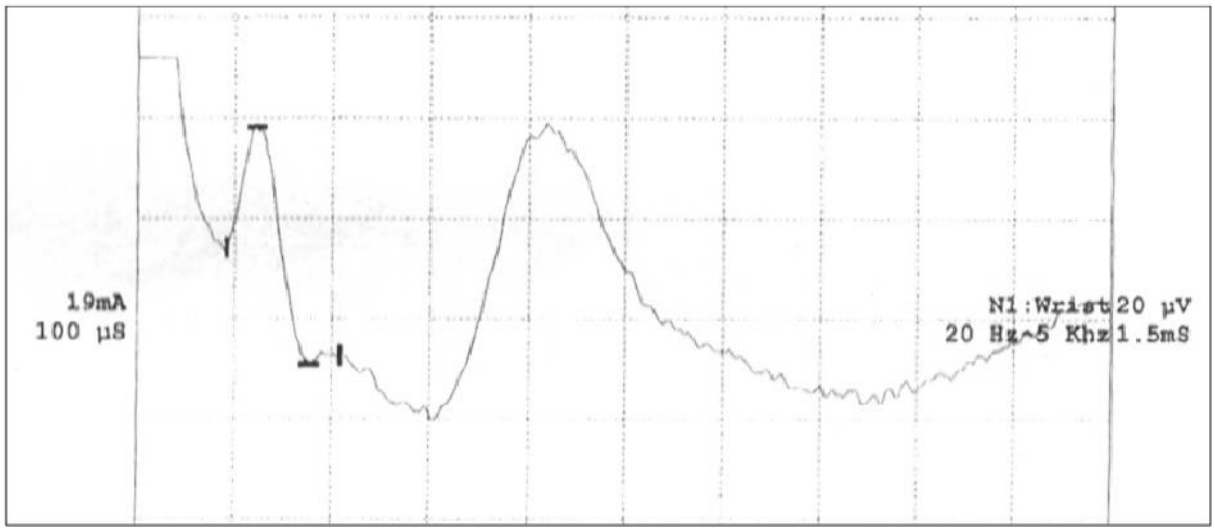
c


26mA
100 µS
N1:Lat Malleolus 20 µV
20 Hz-3 Khz 2 mS

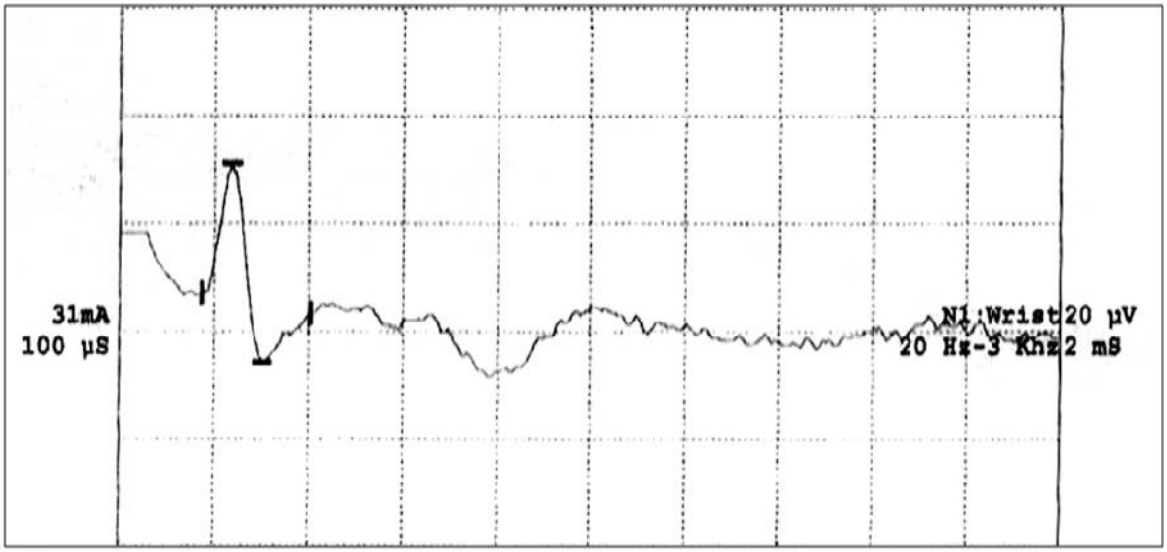
d


19mA
100 µS
N1:Wrist 20 µV
20 Hz-3 Khz 2 mS

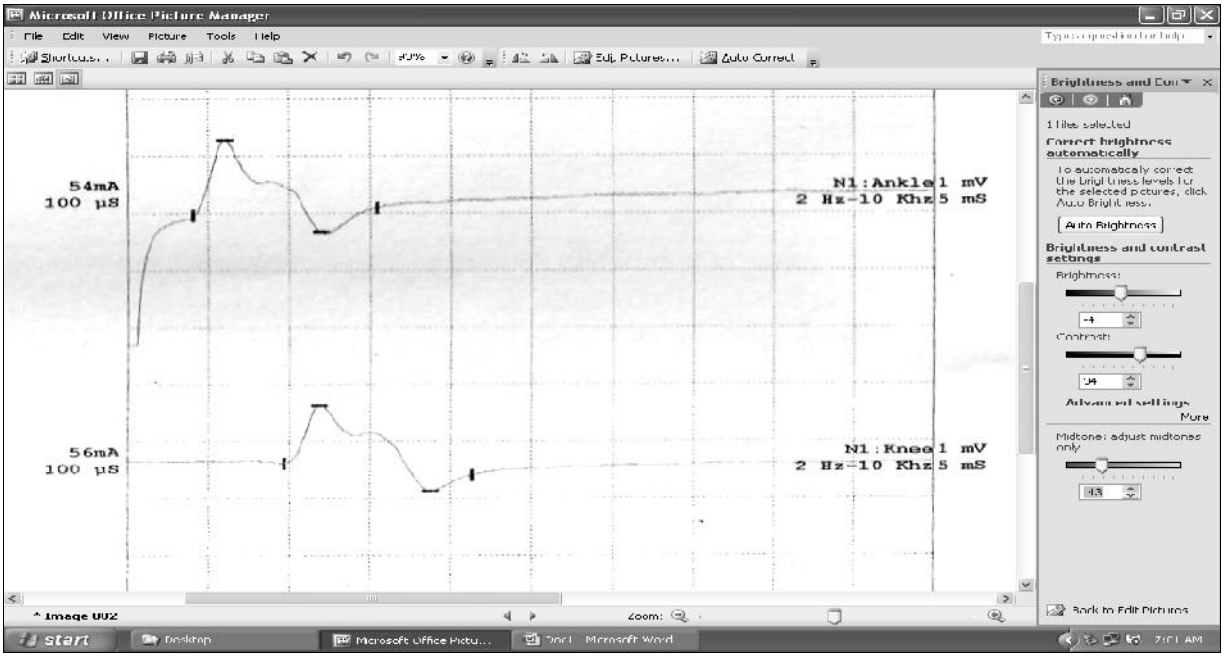
e


19mA
100 µS
N1:Wrist 20 µV
20 Hz-5 Khz 1.5mS

f


31mA
100 µS
N1:Wrist 20 µV
20 Hz-3 Khz 2 mS

g



h

Fig. 2: Nerve Conductance Velocity (NCV) and Electromyography (EMG) of the sensory and motor nerve (a-h).

Discussion

Serum creatinine kinase (CK) level is the commonly used marker to test for muscular dystrophy, although not specific for DMD, nevertheless CK levels tend to decrease substantially in patients with gross muscle wasting. This, together with relevant clinical profile such as disease history and muscle biopsy results determine the diagnosis and possible course of action.

Beckmann and his colleagues in West Germany have compared this new method with the standard CPK technique and found it to be reliable. They have favored the introduction of a screening programme and in their initial survey [9,10] of 16 520 newborn infants have detected 5 boys with preclinical muscular dystrophy which would give an incidence of 1 in 1700 male births. [11]

The Gilboa and Swanson (1976) highlighted that the wide range of CPK in normal newborns. In some the level was up to 10 times the normal [12]. They have noted the lower level in cord blood than in capillary blood and a peak level in the infant during the first 24 hours after delivery. They wisely suggest that any screening for muscular dystrophy should be postponed beyond the immediate newborn period.

Evidence of proximal muscle weakness or high concomitant serum CPK levels will greatly increase the probability of an occult muscular dystrophy or inflammatory myopathy. Serum transaminase levels may seem quite high, particularly in boys with DMD who are ambulant. Finding of Mcmillan et al suggest that the ALT values up to 22.6 times the upper limits of normal is consistent [13].

Bayram et al [14] observed significant body composition changes occur in patients with DMD. Anthropometric and multifrequency bioelectrical impedance analyses measurements show good correlation between motor function scales. These results may also be helpful to evaluate the effects of new treatment strategies.

Mok et al [15] estimates Total Body Water (TBW) which was 66% of expected values obtained from reference data in boys (16, 17). The muscle mass was decrease by 25% from normal values (18) reduced muscle mass may contribute to decreased TBW (75% muscle water content), the correlation between these 2 variables was reported to be weak (19, 20). In fact, TBW is made of intracellular and extracellular water. Muscle mass correlates with intracellular water and total body potassium and declines at a rate of 4%/y, which reflects the progression of the disease (21-23). At variance, exchangeable sodium and extracellular water grow with age, which suggests the replacement of muscle by connective tissue, as observed by histologic experiments in DMD (24). Because children with DMD had less TBW, the apparent normal weight observed in these children can be misleading, as it reflects excess body fat.

Paganoni 2013 [25], EMG is performed in a patient with a dystrophinopathy, it typically reveals increased insertional and spontaneous activity in the form of fibrillation potentials and PSWs, along with brief, small, polyphasic MUAPs with early recruitment. In the end stages of the disease, however, when muscle is replaced by connective and fatty tissue, the insertional activity is reduced and a mixed population of short and long duration MUAPs might be appreciated, reflecting the chronicity of the disease process [26,27].

Szajewska and Kopeć (2008) found the structural changes in Electromyographic pattern in Duchenne and Becker muscular dystrophy, the decrease of amplitude, area and MUAPs duration also progressed. These pattern reflect in our EMG study [26].

Needless to say that spinal collapse in the sitting position in patients with DMD does nothing but increase the restrictive respiratory deficit, thus compromising VC severely. That could be the reason why surgery does not increase life expectancy in patients with DMD according to most of the literature [27-33]. Therefore, it has turned out that the only important goal of spinal surgery in DMD is to improve the quality of life of these patients.

Conclusion

From this study we can conclude that up till now still no cure is available on DMD, The prevention and reduce fatal effect is the only way to control the disease. Therefore early diagnosis is important in the DMD. The pseudohypertrophism found in the early stage is one of the most primitive symptoms of DMD. In DMD, significant increase is found in CPK levels. The Amplitude of Nerve Conductance Velocity (NCV) is reduced in bilateral peroneal nerves. Electromyography (EMG) study shows reduced insertional activity, reduced amplitude and interference pattern with mild fasciculation and reduced motor units.

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