P.7.18
Effects of upper extremity exercise training on respiratory function and quality of life in children with Duchenne Muscular Dystrophy

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To investigate the effects of trainings including two different types of exercises for upper extremity (aerobic and strengthening) on respiratory function and quality of life in children with Duchenne Muscular Dystrophy (DMD). 24 children (8–15 years of age) with DMD whose upper extremity functional status were between Grade 1–3 according to the Brooke Upper Extremity Functional Classification included in the study. Children were divided into 2 groups randomly as study (N = 12) and control group (N = 12). T-Shirt wearing time (s) which is one of the timed performance tests were recorded. Peak Expiratory Flow (PEF) were recorded by using Microlife PF 100 – Asthma Management System. Peak Expiratory Flow (Microlife AG, Switzerland) electronic respiratory muscle strength measuring device. Quality of life of children were assessed with The Pediatric Quality of Life Inventory (PedsQL)-Neuromuscular Module Turkish version (child and parent forms). Children in the study group received 45-min exercise sessions with electronic arm ergometer, 3 days a week for 8 weeks. Strengthening exercises for upper extremity were given as home program to control group during 8-weeks. Assessments were repeated after trainings. The time elapsed for T-Shirt wearing decreased after training in study group (mean ages 9.5 ± 1.4 years) compared to control group (mean ages 9.3 ± 1.4 years) (z = −2.281; p < 0.05). There was a statistically significant increase in PEF values after training in study group (z = −3.059; p < 0.01), while no difference in control group (z = −1.844; p > 0.05). There were negative, strong correlation between PEF-T-Shirt wearing time (r = −0.629; p < 0.05) and positive, moderate correlation between PEF-PedsQL-child form (r = 0.590; p < 0.05). In our study, it was concluded that upper extremity aerobic exercise training has positive effects on respiratory performance and quality of life of DMD children.

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P.7.19
Effects of upper extremity exercise training on upper extremity endurance in patients with Duchenne Muscular Dystrophy

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To investigate the effects of 8 weeks upper extremity aerobic and strengthening exercises on upper extremity endurance in children with Duchenne Muscular Dystrophy (DMD). 24 children with DMD who have similar physical characteristics between 8 and 15 years of age included in the study. Children randomly divided into two groups. 12 DMD children whose functional status were between Grade 1 and 3 according to the Brooke Upper Extremity Functional Classification were included in each study and control group Minnesota Hand Dexterity Test (MHDT) was used to assess the upper extremity endurance and hand dexterity of children. Children in the study group received 45-min exercise sessions with electronic arm ergometer, 3 days a week for 8 weeks. 8-weeks home program including strengthening exercises for the upper extremity were given to the patients in control group. The number of elbow flexion–extension were recorded before and after each exercise sessions in study group. Assessments were repeated after 8 weeks training and results were compared within and between groups. There were statistically significant decrease in unilateral placing (z = −2.354) and bilateral turning (z = −2.786) parameters of MHDT in study group (mean ages 9.5 ± 1.4) while only bilateral turning (z = −2.354) in control group (mean ages 9.3 ± 1.4) after training, compared to baseline (p < 0.05). There were statistically significant increase in the number of elbow flexion–extension after training (1st week mean: 4.4 ± 5.3; 8th week mean: 8.9 ± 7.8) (p < 0.05). Positive effects of both two different types of exercise trainings (aerobic and strengthening) on muscle endurance were shown in our study. It is thought that, increase in endurance might lead to increase in speed of performance in daily living activities related to upper extremity. This may also lead to toleration of fatigue for patients with neuromuscular diseases.

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P.7.20
Effects of dynamic arm training on trunk muscle strength and lower extremity functions in children with Duchenne Muscular Dystrophy

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To investigate the effects of dynamic arm training on trunk muscle strength and lower extremity functions in children with Duchenne Muscular Dystrophy (DMD). Manual muscle testings of back extensors and abdominal muscles were performed to 12 DMD children who are still ambulant and between 8 and 15 years of age. North Star Ambulatory Assessment (NSAA) was performed to determine the ambulatory status of children. Children were trained with RECK MOTOMed viva 2 model arm training device during 8 weeks, 3 days a week, 45 min in each session. Trunk muscle testing and NSAA were performed before and after 8 weeks training and compared to baseline assessments. There were not any statistically significant difference of muscle strength (back extensors, abdominal and total trunk) before and after training (p > 0.05). NSAA scores showed a statistically significant increase after training (NSAA scores – before training: 17.5 ± 8.6; after training: 20.2 ± 8.8) (p < 0.05). This study shows that, aerobic training which only applied upper extremities has positive effects on lower extremity functions in DMD.

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ALPHA-DYSTROGLYCANOPATHIES

P.8.1
Muscle biopsy findings in Limb Girdle muscle Dystrophy 2I (LGMD2I)

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Reduction of LARGE expression in different types of muscular dystrophies other than dystroglycanopathy

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Mutations in the genes coding for putative or demonstrated glycosyltransferases or other proteins involved in alpha-dystroglycan (ADG) glycosylation pathway result in the failure of alpha-dystroglycan to be properly glycosylated and lead to genetic forms of muscular dystrophy, collectively termed as dystroglycanopathies. An important enzyme which is involved in maintaining muscle cell viability, known as LARGE, has been shown to participate in O-mannosyl phosphorylation of ADG. It could also act as a bifunctional glycosyltransferase and allow ADG to bind laminin-G domain containing ligands. Additionally, transient over-expression of LARGE enzyme demonstrated a marked increase in hyper-glycosylation of ADG and a corresponding increase in high affinity binding to several extracellular matrix ligands. To date, it has not been investigated whether LARGE enzyme affects the basic pathogenic mechanisms of muscular dystrophies, except for dystroglycanopathies and the influence of LARGE gene expression in different types of muscular dystrophies is not known. In this study, the expression level of LARGE and ADG immunofluorescence were examined in skeletal muscle biopsies from 26 patients with different forms of muscular dystrophy (i.e. DMD, BMD, calpainopathy, sarcoglycanopathy, dysferlinopathy, and merosin and collagen VI deficient CMDs) and correlation with different histopathological findings was investigated. We detected reduced expression level of LARGE gene in different types of muscular dystrophies, partly correlating with the severity of dystrophic changes, but we did not find any significant relationship between reduction of LARGE expression and ADG hypoglycosylation. Our results suggest that LARGE enzyme might have another function in skeletal muscle fibers that is probably distinct from adding a critical sugar chain onto ADG.

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