REHABILITATION PROGRAMS FOR PEOPLE WITH NEUROMUSCULAR DISORDERS

PROGRAM REHABILITACIJE ZA BOLNIKE Z ŽIVČNO-MIŠIČNIMI BOLEZNIMI

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Key words: *neuromuscular diseases; rehabilitation programs*

Abstract – *Neuromuscular diseases are inherited, chronic, degenerative and progressive. The main characteristics of neuromuscular diseases are: muscular weakness, contractures, scoliosis, respiratory insufficiency, cardiac affection, nutrition disturbances, dependence on the help of others, possible social isolation and physiological problems. Appropriate rehabilitation programs should influence all mentioned characteristics. A special unit for rehabilitation of patients with neuromuscular diseases within the Institute for rehabilitation of the Republic of Slovenia was established in 1993 at the initiative of the Muscular Dystrophy Association of Slovenia. The main aim of this establishment was to try to guide the patient and his family through the course of the disease. The article describes the work of the mentioned unit. Different clinical rehabilitation programs for people with neuromuscular diseases are presented and some research results of the unit are mentioned.*

Ključne besede: *živčno-mišične bolezni; rehabilitacijski programi*

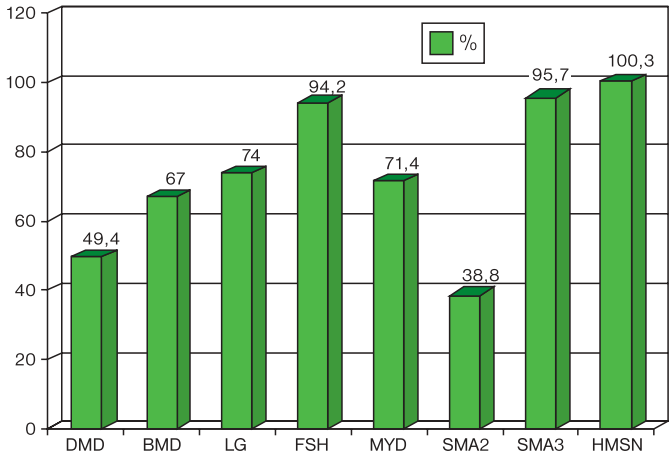
Izvleček – *Živčno-mišične bolezni so dedne, kronične, degenerativne in progresivne. Glavne značilnosti živčno-mišičnih bolezni so mišična oslabelost, kontrakture, skolioza, dihalna insuficienca, prizadetost srca, motnje prehranjevanja, odvisnost od tuje pomoči, možna socialna izolacija in psihološke težave. Z ustrezno rehabilitacijo je potrebno vplivati na vse omenjene značilnosti. Posebna služba za rehabilitacijo bolnikov z živčno-mišičnimi boleznimi je bila ustanovljena na Inštitutu Republike Slovenije za rehabilitacijo v letu 1993 na pobudo Društva mišično obolelih Slovenije. Glavni namen ustanovitve je bil, da bi bolnike in njihove družine »vodili« skozi bolezen. Članek opisuje delo omenjene službe. Opisani so različni klinično rehabilitacijski programi za bolnike z živčno-mišičnimi boleznimi ter nekateri raziskovalni rezultati.*

In my lecture I would like to present different programs, which we practice in our unit for rehabilitation of patients with neuromuscular diseases within the Institute for rehabilitation of the Republic of Slovenia. This unit was established in 1993 at the initiative of the Muscular Dystrophy Association of Slovenia. The main aim of this establishment was to try to guide the patient and his family through the course of the disease.

Neuromuscular diseases are inherited, chronic, degenerative and progressive. Once diagnosis is determined numerous questions arise that we address through our work. We accompany and advise the patient through the course of disease in a different way than his/her family. We have to consider the level of the patient's physical disability, his intellectual capability and his environment. And it is very important to account for the different life periods (childhood, puberty, adolescence, and adulthood). We must be aware of critical periods in the guidance of patients, which are the following: when a child can't start walking, the loss of walking capability, respiratory failure and acceptance of a ventilator, serious cardiac affection and death. One of the most important aims of our work is that every patient, regardless of disability level, must grow up and learn to live on his own. This is sometimes very difficult because being dependent on the help of others can be a real curse.

The main characteristics of neuromuscular diseases are: muscular weakness, contractures, scoliosis, respiratory insufficiency, cardiac affection, nutrition disturbances, dependence on the help of others, possible social isolation and physiological problems. Through our rehabilitation programs we try to influence all the above-mentioned characteristics. These programs are carried out at the Institute for Rehabilitation of the Republic of Slovenia and at »Dom Dva Topola« in Izola on the Slovenian part of the Adriatic Coast. The Dva Topola building is located at the seaside and is surrounded by a beautiful park. Everything, including the beach, is especially adapted for wheelchair users. In Dom Dva Topola we perform programs of so-called »renewal« – restorative rehabilitation. During the summer more than 400 patients are treated, each for at least a fortnight. At the Institute in Ljubljana, however, complex programs of rehabilitation are performed throughout the year; but here we can only accept a relatively small number of patients.

Assessment of the functional state. Before we start with rehabilitation programs for individual patient we have to know about his functional state. We have developed a test protocol consisting of 10 tests to assess accurately and objectively the functional state of patients and to follow the progression of disease (1). By comparing the estimates for simple and complex functional activities we can calculate so called quotient of adaptation-rehabilitation which gives us the possibility to assess the degree of patient's adaptation-rehabilitation and his need for rehabilitation programs (a quotient higher than 1 means good while lower than 1 means bad degree of rehabilitation).



DMD Duchenne muscular dystrophy
BMD Becker muscular dystrophy
LG limb-girdle muscular dystrophy
FSH facioscapulohumeral muscular dystrophy
MYD myotonic dystrophy
SMA2 spinal muscular atrophy type 2 (intermediate)
SMA3 spinal muscular atrophy type 3 (juvenile)
HMSN hereditary motor and sensory neuropathy

Figure 1. *Average values of forced vital capacity (FVC) as a percentage of reference values in single diagnostic groups.*

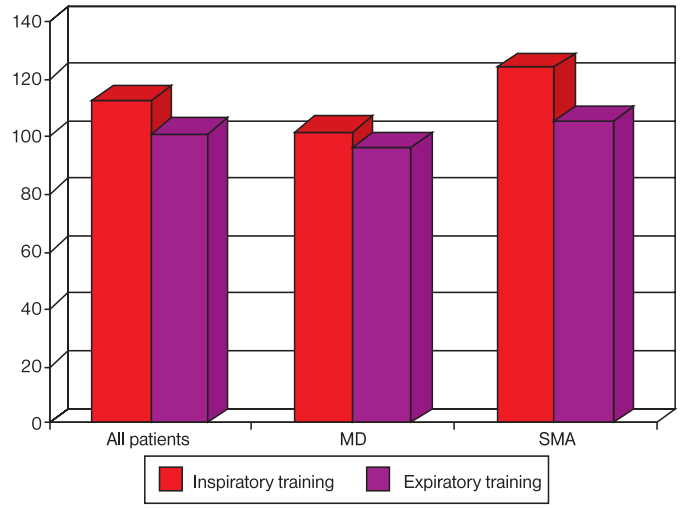


Figure 2. *Changes of values of forced vital capacity (FVC) at the end of the program of inspiratory and expiratory exercises, expressed as a percentage of starting values before the beginning of the program of respiratory exercises measured in sitting position for all patients and separately for the subgroup of patients with muscular dystrophy (MD) and for the subgroup of patients with spinal muscular atrophy (SMA).*

Muscular weakness. In order to maintain muscular strength it is necessary to perform active or active assisted exercises; passive exercises are insufficient for this purpose. Active exercises should be dynamic and isometric, they must be performed daily. In childhood these exercises should be introduced as a game to avoid the feeling of pressure. We encourage exercises in water and, if possible, swimming; especially the breaststroke to strength trunk and neck extensor muscles. In Izola patients can swim in the sea or in a pool with seawater. They are properly assisted down a ramp on a special wheelchair into the water. Exercises in water are useful because of physical effects of water. It is essential that patients do not to get too tired through these exercises and this is especially true for patients with Duchenne muscular dystrophy (DMD) for whom active exercises can be questionable (2).

Contractures. To prevent the development or aggravation of contractures it is necessary to stretch contracted muscles. Stretching should be performed at least twice a day. For patients who are still able to stand or walk the best way of stretching is standing or walking, we recommend to them that they be in a standing position for at least 3 hours a day (3, 4). Wheelchair-bound patients are regularly put in an erect position with the use of tilt table or different standing frames. For proper stretching of contractures we invented a special device (5). The device enables stretching of contractures in different joints at the same time (at ankles, knees, hips and elbows). During stretching the device also enables the performance of active exercises. In our study we included 10 boys with DMD; 4 of them were daily using this device for one year, 6 of them didn't. We measured the degree of contractures at different joints at the beginning and at the end of the study. In the treated group of boys the average increase of contractures in all joints was much lower than in the control group. We generally don't recommend surgical treatment of contractures, especially in wheelchair-bound patients because the contractures quickly develop again.

Scoliosis. To prevent the development of spinal deformity we recommend active exercises for patients during the early stage of disease. When scoliosis is already developed we advise spinal surgery, as soon as possible, especially for patients with DMD and spinal muscular atrophy type 2 (SMA2). For patients who have problems with sitting and who can't sustain spinal surgery we provide proper spinal orthosis or an especially made seat. We generally don't recommend spinal orthosis as a preventive measure at all; because of passive stabilization of the spine the paraspinal muscles degenerate even more quickly.

Respiratory problems. Respiratory capabilities are tested in each patient at least once a year. Figure 1 shows the averages values of forced vital capacity (FVC) as a percentage of reference values for more than 400 patients with different forms of disease. The lowest average value is in patients with SMA2. We followed also the changes of FVC over a 10-year period in patients. The decline of FVC was the biggest in patients with DMD while in patients with SMA2 there was practically no decline of FVC during the 10-year period (1).

We advise all patients from the very beginning to perform respiratory exercises, especially inspiratory. There are many reports about the effects of respiratory exercises in patients with neuromuscular diseases, some of them report improved respiratory capabilities others find no effects (6–11). We studied the effects of respiratory muscle training on the respiratory capabilities of 29 children (12 with DMD and 17 with SMA). Each patient was subjected for 2 six-weeks periods of training: inspiratory muscle training and expiratory muscle training. The sequence of these two periods was random for each patient. FVC and some other parameters were measured before and following each six-week period. Figure 2 shows that the average values of FVC were increased following inspiratory muscle training whereas there was no change after the expiratory counterpart. The increment of values for FVC was statistically significant after inspiratory muscle training only for children with SMA; for children with DMD there was no statistically important increase of FVC (12).
We recommend that children with NMD be all vaccinated (many physicians/general practitioners are still afraid of vaccinating children with neuromuscular diseases). In the later stages of disease when respiratory insufficiency is already deve-

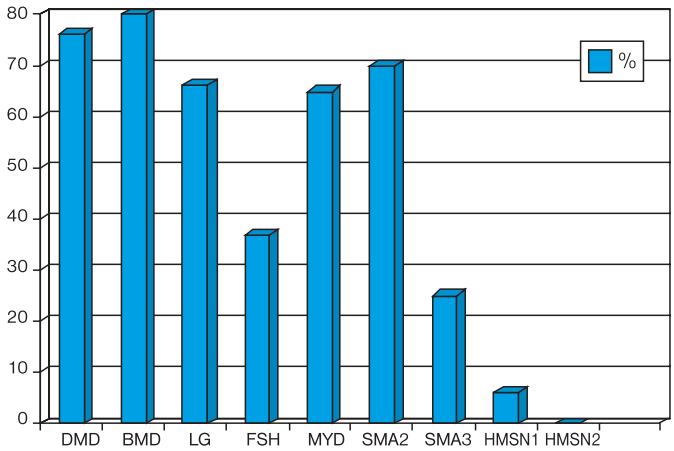


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Figure 3. *Percentage of patients with electrocardiographic (ECG) changes per diagnostic group.*

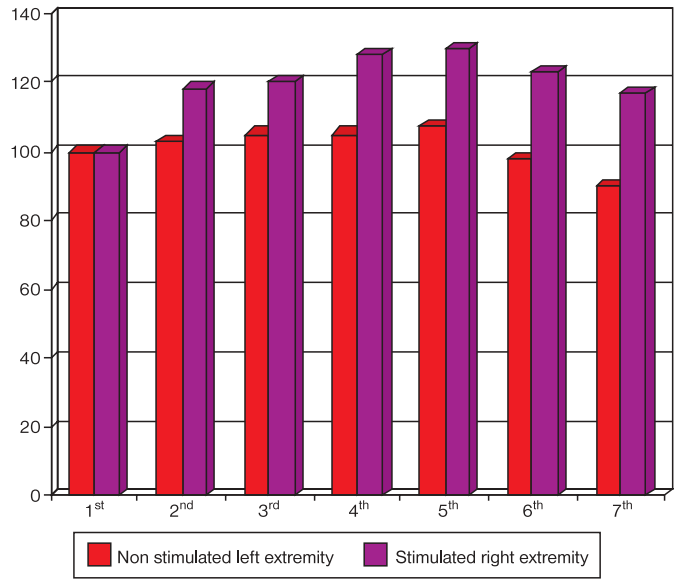

Figure 4. *Histograms of average torques in the ankle at maximum voluntary contraction in the direction of dorsal flexion of the foot for the left nonstimulated and the right stimulated extremity, expressed as a percentage of starting values before stimulation (1st): for a group of 5 children who stimulated their right tibialis anterior muscle for 9 months. Three measurements (2nd, 3rd, and 4th) were made after 1, 2, and 3 months of stimulation; the other 3 measurements (5th, 6th, and 7th) were made after 5, 7, and 9 months of stimulation.*

loped, we perform procedures for cleaning airways, especially during respiratory infections: percussion, postural drainage and coughing. We recommend drinking enough liquid and proper air humidity. Regarding coughing procedures we apply a manually assisted method and use a coughing machine – insuflator/exsuflator. We have had very good results with the latter. For patients with the most serious respiratory problems we use assisted ventilation. All expenses connected with assisted ventilation are covered by health insurance.

Cardiac problems. The cardiac function of each patient is annually examined by electrocardiography (ECG). If there is something wrong echocardiography is applied and, if necessary, appropriate therapy is introduced. In our study we investigated the status of the cardiovascular system in 227 patients with different forms of disease. As shown in Figure 3 an assessment of the frequency of ECG changes has been made for each group of patients. The highest percentages were registered in patients with Becker muscular dystrophy (BMD); these were followed by patients with DMD, SMA2, limb-girdle muscular dystrophy (LG), those with myotonic dystrophy (MYD), those with facioscapulohumeral muscular dystrophy (FSH), and, finally, those with SMA3 (13).
Most of patients, and especially those who are wheelchair-bound, have problems with hypostatic edema, mostly in the ankles and have signs of bad peripheral circulation. We treat hypostatic edema using mechanical lymph drainage. Special »boots« are put on the patient's legs and an apparatus intermittently blows air into them. The results are satisfactory; after the procedure the edema are temporarily diminished and patients have a relaxed feeling in their legs. For bad peripheral circulation the manual massage is used which is beneficial also for contracted muscles.

Proper sitting. It is extremely important especially for children to sit properly in their wheelchairs. We prescribe wheelchairs with great caution (14). The patient should sit upright; the lumbar part of the spine should be in slight hyperlordosis. The seat surface (cushion) should be slightly inclined backwards and it must have anchorage for the pelvis. The upper limbs should be leaned on arm supports; both shoulders must be in the same level, and the elbows being flexed at 90 degrees. The feet must be on foot support with an angle of 90 degrees in the ankles if possible. If necessary we use side supports for the trunk, head support, different pads to prevent oblique position of the pelvis and some other additions.

Technical aids. To improve quality of life and enable patients to live more independently we try very hard to get the most appropriate technical aids for our patients from health insurance or other state institutions. The situation regarding electric wheelchairs is quite good. Electric wheelchairs are prescribed and paid for in consideration of a patient's degree of disability; the greater the disability, the greater the health insurance supports. Patients who are employed or attend school can get a new wheelchair every three years, others, every five years. The situation is much worse regarding special equipment required for using computers or driving a car, for the most part, patients themselves must pay for such equipment.

Dependence on the help of others. We have published a guidebook on how to help NMD patients (lifting, transferring, turning around, washing, dressing, feeding, etc). Clear illustrations with descriptions demonstrate how to help efficiently and in a manner, which is safe for both patient and care-giver (15).

Nutritional disturbances (obesity, malnutrition, constipation etc.). To prevent these disturbances we recommend balanced nutrition with enough proteins and vitamins, if necessary we prescribe laxatives.

Pain. Cryomassage and electrotherapy are mostly used to alleviate pain. Sometimes pain can be relieved by cautious passive exercises. We try to avoid, as much as possible, the use of analgesics.

Social and psychological assistance. A psychologist and a social worker are included in the team at our unit for rehabilitation of patients with neuromuscular diseases. At the Dom Dva Topola patients have numerous social contacts, they associate with other patients, with nursing staff, with civil soldiers and volunteers from all parts of the world who help there.

Electrical stimulation. Based on experiments by Gerta Vrbova (16) where she shows that it is possible to transform fast muscle fibres into slow ones using low frequency electrical stimulation, and, on the basis of reports that slow muscle fibres are more resistant to dystrophic process than fast fibres (17), we started a program of low frequency electrical stimulation for boys with DMD (18). The right tibialis anterior muscles of five boys with DMD were stimulated for nine months, the left side served as a control. The muscles were stimulated with 8 Hz for one hour twice a day. Figure 4 shows the average torques in the ankle at maximum voluntary contraction in the direction of dorsal flexion of the foot for the left non-stimulated and for the right stimulated extremity, this is expressed as a percentage of starting values before stimulation (1st). Three measurements (2nd, 3rd and 4th) were made after 1, 2 and 3 months of stimulation; the other three measurements (5th, 6th and 7th) were made after 5, 7 and 9 months of stimulation.
We can conclude from our study that although dystrophic muscles are incapable of retaining the strength that became obvious after some months of stimulation, it seems that the weakening of the muscles can be retarded to a certain extent through prolonged application of low frequency electrical stimulation (18).

Coencime Q10. CoQ10 is an essential part of the mitochondrial respiratory chain and can be found in every cell of the body (19). The best-known functions of CoQ10 are its involvement in electron transfer and its antioxidative role in the mitochondria (20, 21). In this way CoQ10 helps to provide energy stores to the cell in the form of ATP and gives protection from free radicals to the mitochondria (19).
There are some reports about beneficial effects of CoQ10 treatment in patients with neuromuscular diseases (22); they also report that low concentrations of CoQ10 in the serum of patients can be increased by taking CoQ10. We have performed a double blind study of CoQ10 treatment in 50 patients with different forms of muscular dystrophy and SMA. The study lasted for 8 months. Each patient received CoQ10 (400 mg a day) for 4 months and placebo for 4 months. The sequence of these two periods was random for each patient. Measurements of selected parameters were done at the beginning of the study and at the end of each 4-month period. We measured several biomechanical parameters and analyzed blood samples. After treatment with CoQ10 there were no statistically significant changes in biomechanical parameters (23) but from blood sample testing we found a statistically significant fall of cell CO_2 and of blood sugar. Most of the patients reported improvement of physical well-being and most of them correctly guessed the period during which they received CoQ10 or placebo.

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