
REHABILITATION PROGRAMS AND CUSTOMIZED APPROACH TO CARE FOR A CHILD WITH LESCH - NYHAN

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Abstract: As regards the problems of patients and medical specialists related to the rare diseases, EU gives directions of more awareness, transparency and personalized approach in the field of the healthcare, as well as creation of rehabilitation programs for the rare diseases. In this connection, the EUROPLAN Project gives recommendations the good paramedical practices to be coordinated in order to service the specific needs and to secure rehabilitation for patients with rare diseases. Present publication presents the results of the performed rehabilitation treatment of a child suffering the Lesch-Nyhan syndrome. The disease Lesch-Nyhan Syndrome is caused by flaws of a gene in the X chromosome. As in the cases of hemophilia, women usually do not have symptoms, because they have two X chromosomes, but men suffer it. The illness is caused by the lack of an enzyme called hypoxanthine-guanine phosphoribosyltransferase (HGPRT). This enzyme is decisive for the recycling of live cells. The cells are normally decomposed by DNA in various parts and two of them (adenine and guanine) should be broken down by HGPRT.

The supporting role of the kinesiotherapy is substantial with this disease. It is focused on influencing the way of life by developing compensatory behavioral reactions in the conditions of present neurological and behavioral deficit.

The impairment in the motor development of our patient appeared at the age between the 6th and the 18th months after birth, when involuntary movements started, showing extrapyramidal dysfunctions. The illness is demonstrated also by retention of the height and very slow sexual maturation. The neurological and cognitive functions suffer. The choreoathetotic neurological disorders exclude the possibility the child to learn how to walk and that is why it is in a wheel-chair with protective means – splints and belts. The basic sensor functions were within the norm, but the motion ones were demonstrated with grave dystonia.

In view of the insufficient research works on the problem in our country and the existing differences in the literature, we interpret the results as pilot ones. However, an impression is created that the methods implemented by us improved the functional condition of the sick child as after an individual procedure, as well as after the training period. Children with disabilities in Bulgaria are subject of attitudes and practices inherited from the past, when most of them had been isolated, protected and taken care by their families or by the state. A big share of that group continue to be isolated and unable to go out of their homes due to the still inaccessible environment, as well as to the moods and attitudes of the society towards their problems.

Keywords: Lesch-Nyhan syndrome, multidisciplinary approach, rehabilitation, kinesiotherapy

1. INTRODUCTION

The rare diseases are life threatening or chronic incapacitating illnesses with lesser propagation and high degree of complexity. The total number of people suffering rare diseases in the EU is between 27 million and 36 million. These patients are particularly isolated and vulnerable.

EU appeals for more awareness and transparency as regards the problems of patient and medical specialists related to the rare diseases, as well as for personalized approach in the field of health cares and support for the creation of rehabilitation programs for the rare diseases. One of the recommendations under EUROLAN Project is the good paramedical practices to be coordinated in order to service the specific needs and to secure rehabilitation for patients with rare diseases.

In view of improving the quality of life and extending the survival term of such patients, the efforts of an interdisciplinary team are needed for rendering adequate, timely and personalized health cares, where the supporting rehabilitation share is substantial.

The objective of the present publication is to show the results of the supporting therapy and to report the results of the performed rehabilitation treatment of a child with Lesch-Nyhan Syndrome, the main goal of which is improvement of patient's general condition and prophylactics of the contractures. The supporting role of the kinesiotherapy is substantial with this disease. It is focused on influencing the way of life by developing compensatory behavioral reactions in the conditions of present neurological and behavioral deficit.

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There is no definitive treatment of the disease yet and its nature leads to grave functional deficit, which brings forth many psychological, social and health problems. The hardest is the problem of the self-service, eating and independent movement of the sick. At that very direction the efforts of the team are aimed in view of securing and bettering the quality of life.

In 1964 Michael Lesch and William Nyhan described for the first time a syndrome, which is characterized by neurological and behavioral disorders – *choreoathetosis*, spasticity, hyperreflectivity, mental retardation, aggressive and impulsive behavior, as well as permanent and grave self-aggression.

The disease Lesch-Nyhan Syndrome is caused by flaws of a gene in the X chromosome. As in the cases of hemophilia, women usually do not have symptoms, because they have two X chromosomes, but men suffer it. The illness is caused by the lack of an enzyme called hypoxanthine-guanine phosphoribosyltransferase (HGPRT). This enzyme is decisive for the recycling of live cells. The cells are normally decomposed by DNA in various parts and two of them (adenine and guanine) should be broken down by HGPRT.

The lack of this enzyme becomes the reason the two components to accumulate and form uric acid, which provokes irritation of the cells and formation of stones in the kidneys. Individuals suffering the Lesch-Nyhan Syndrome tend to bite their lips, tongue and fingers. Although the injuries are minimal, there are cases of graver ones. This behavior is so frequent that it was called “self-cannibalism syndrome”. The pathogenesis of the neurological and behavioral symptoms is unknown. The simplest explanation is that the uric acid affects the cells and the sick bite their most sensitive tissues, gaining pains such as insects’ stings, scratches, etc.

There is another theory that the uric acid causes insufficiency of dopamine in the brain. The neurophysiologic researches show substantial disorders in the dopamine in the basal ganglia. When this syndrome appears, however, dopamine could be released in the brain under the effect of the processes in the cells and the enzymes, which make the patients suffer much more. Many physicians have found out that this disease is opposite to the Parkinson disease. While patients with Parkinson cannot perform activities, which are rational, the victims of the Lesch-Nyhan Syndrome cannot be controlled. At present there is no remedy for the illness, but there is treatment for the symptoms. Women with family anamnesis of the syndrome should be examined before and during the pregnancy – whether they carry the defect gene.

Choreoathetotic movements are observed with children having this syndrome, which are characterized by quick and sudden movements typical for chorea and slower spastic movements of the body characteristic for the athetosis.

2. MATERIALS AND METHODS

Subject of our research is an eight years old boy initially wrongfully diagnosed as suffering cerebral palsy (CP) and treated in the clinic of Prof. Chavdarov in Sofia. Due to its individual progress and specific symptomatics, the disease remained unrecognized and after two years kinesiotherapy course the patient was discharged as unpromising and unsusceptible to the therapy. The correct diagnose was made two years later after a genetic test of the mother provoked by the fact that this is the second boy with similar symptoms she gave birth to. After the examination of the patient, the Lesch-Nyhan Syndrome was proven. Immediately after making the diagnose, the child was included in a special program for children with special needs at the “Mihail Lakatnik” School in Burgas. The rehabilitation course is continuing for five years by applying a multidisciplinary approach. A kinesiotherapist, a nurse, a speech and language therapist and a resource teacher participated in the rehabilitation team.

Due to the lack of sufficient data from clinical researches of patients with the Lesch-Nyhan Syndrome, the authors think that the kinesiotherapy methods applied by them in combination with highly professional nurse cares play substantial role for the improvement of the quality of life and extension of the term of survival with that patient.

It is show that the systematic application of analytically selected and purposeful physical activity improves the muscle tone, the muscle strength and endurance of the upper and lower limbs, as well as the motion volume in the joints, related to maintenance of the normal daily activity of the patients. The changes in the condition of the sick person in time impose also modifications of the kinesiotherapy program. Its directions are determined mainly by the graveness of the mobility deficit. The place of kinesiotherapy in the rehabilitation program for children with physical injuries is well known, but its effect on the mobility independence of children with Lesch-Nyhan Syndrome has not been studied enough yet.

The impairment in the motor development of our patient appeared at the age between the 6th and the 18th months after birth, when involuntary movements started, showing extrapyramidal dysfunctions. The illness is demonstrated also by retention of the height and very slow sexual maturation. The neurological and cognitive functions suffer. The choreoathetotic neurological disorders exclude the possibility the child to learn how to walk and that is why it is in a

wheel-chair with protective means – splints and belts. The basic sensor functions were within the norm, but the motion ones were demonstrated with grave dystonia. There were also behavioral problems demonstrated by grave self-aggression. Urological and joint complications were observed, such as heavy contractions in the shoulder, elbow, hip, knee and ankle joints. The damage of the deep sensibility hindered the reproduction of passive motions and impossibility of performing the physiological synergies of the motions. The *choreoathetosis (quick involuntary movements in the proximal and distal joints with wormlike motions in the fingers)*, the *torsion dystonia (slow fit motions of the torso and the neck)* were combined with double athetosis.

Kinesiotherapy tasks:

1. Suppression of the primitive tonic reflexes;
2. Suppression of the pathological synergies and sinkineses;
3. Suppression of the vicious positions of the limbs;
4. Retraining of the residual tonic reflexes;
5. Turning the voluntary movements into physiological synergies;

Basic principles of the kinesiotherapy at each stage were:

1. Relaxation of the body muscles before the application of the passive exercises and stretching.
2. The kinesiotherapy was performed in accordance with the ontogenetic development of the healthy child motor activity.
3. The curative exercises should be obligatorily combined with respiratory exercises.
4. Each exercise should be obligatorily accompanied by verbal instructions.

3. KINESIOTHERAPY MEANS

In order to reduce the increase muscle tone we applied initially spot massage on the biologically active points under S.A. Bartfold (quoted after K.A. Semenova, 1976). According to their location, we were looking for relaxation of the pectoral muscle, facilitating the contractions of the abdominal muscles, relaxation and facilitating the contractions of the sternocleidomastoid muscle, relaxation and facilitating the motions of the entire upper limb, as well as facilitating the contractions of the scapula muscles and the extensors in the shoulder joint. By means of the massage of biologically active points located on the lower limb, we stimulated – relaxation and facilitating the extension and abduction of the foot sole and extension of the big toe, the extensors of the foot sole and the toes, relaxation of the calf muscle and facilitating the plantar flexion, relaxation of the knee joint flexors and facilitating the abduction and the external rotation of the hip joint. Stimulating the points on the upper limb we aimed at relaxation of the fingers and the thumb, relaxation and facilitating the supination, the flexors and the internal rotators of the armpit, the extension in the elbow joint and the fingers' flexion.

In order to generally reduce the increase muscle tone we used the “embryo posture” proposed by B. and K. Bobat (1956). We applied also exercises for suppression of the asymmetric neck tonic reflexes, exercises for stimulation of the extensor synergy of the upper limbs, exercises for stimulation and formation of the Landau standing (extensor) reflex, curative exercises for facilitating the torso motions and for the abdominal muscles – tonifying massage by all possible techniques, special spot massage, lying on the back on a big ball combined with tonifying massage. In order to reduce the rigidity and to increase the general mobility we applied stretching from various initial positions. It is done very slowly with continued pressure on the tendons of spastic or extrapyramidal muscles or groups of muscles capable of passive volume of motions in the relevant joint, immediately after or between the applications of thermotherapy (in our case by heated to 38°C gel muffers).

We performed the manual relaxation of the increased muscle tone immediately after relaxing massage and it consisted of carrying out passive exercises due to the lack of active motions. Besides the mechanical impact, the stretching of the muscles is improving their excitability and blood supply and through the functional exercising of the limbs, the joint linking apparatus is protected against inactivity and degeneration.

The passive relaxing is done slowly, by gradual increase of the motion volume and rate. The passive motions should be performed freely without pressure along the motion axes from the proximal to the distal joints and to repeat the functions of the muscles engaged in the relevant motion. Exercises too strenuous in view of the patient's condition are not applied.

4. DISCUSSION

In view of the insufficient research works on the problem in our country and the existing differences in the literature, we interpret the results as pilot ones. However, an impression is created that the methods implemented by

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us improved the functional condition of the sick child as after an individual procedure, as well as after the training period. In view of the scares experience we take the chance with certain reserves to assume that the range of methods tested and applied by using the selected by us ones in the work with children suffering grave neurological diseases provokes better reactions in the extrapyramidal part of the central nervous system. Taking into consideration that the extrapyramidal nerve pathways and structures control the coordination and the fine motions, we can assume that these methods have good influence on the available rigidity and coordination, which is confirmed by the occurring, although very small, improvements in the general condition of the patient. The intimate mechanisms, however, could be hardly explained at this stage.

Children with disabilities in Bulgaria are subject of attitudes and practices inherited from the past, when most of them had been isolated, protected and taken care by their families or by the state. A big share of that group continue to be isolated and unable to go out of their homes due to the still inaccessible environment, as well as to the moods and attitudes of the society towards their problems (Stancheva – Popkostadinova, 2006).

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