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A CLINICAL CASE OF PHYSICAL THERAPY OF A CHILD WITH MULTIPLE SULFATASE DEFICIENCY

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Ключові слова: *орфанне захворювання, реабілітація, терапевтичне втручання, масаж, фізична терапія, вертикалізація*
Ключевые слова: *орфанное заболевание, реабилитация, терапевтическое вмешательство, массаж, физическая терапия, вертикализация*

Abstract. A clinical case of physical therapy of a child with multiple sulfatase deficiency. Grygus I.M., Nagorna O.B., Nesterchuk N.E., Nogas A.O., Podoliaka P.S., Gamma T.V. *The article deals with the problems of physical therapy in children with multiple sulfatase deficiency (MSD). The purpose of the study presented in the article was to substantiate the need to form a structured, personalized comprehensive rehabilitation program for children with multiple sulfatase deficiency. The objectives of the study were to analyze the literature on the topic of the study, to study the clinical phenotype of multiple sulfatase deficiency and potential complications of this pathology, to justify the use of physical therapy in children with multiple sulfatase deficiency. Research methods: analysis of scientific and methodological sources of domestic and foreign authors, pedagogical observation, collection of anamnestic information of the patient. The publication discusses the clinical phenotype of genetic pathology and possible potential complications of this orphan disease, strategic vectors of an individual rehabilitation program. The description of a clinical case of late infantile form of the disease is presented. The effectiveness of a six-months' implementation of the physical therapy program is being investigated. For rehabilitation examination of children with MSD, it is proposed to use testing of children with psychomotor disorders. The technique of massage, the appropriateness of verticalization and orthotics, sensory enrichment of the environment of a child with this genetic pathology are revealed. Exercises of therapeutic physical culture are proposed, they will help to maintain postural control, support ability and functioning of arms, legs, hand-eye coordination, and help prevent diseases of the bronchopulmonary system. It is noted that MSD has been insufficiently studied not only from the standpoint of a treatment strategy, diagnostic algorithms for clinical multisystem manifestations, but also requires attention to analyze the effectiveness and efficiency of the system of modern rehabilitation technologies for children with this diagnosis.*

Реферат. Клинический случай физической терапии ребенка с множественным дефицитом сульфатазы. Григус И.М., Нагорная О.Б., Нестерчук Н.Е., Ногас А.А., Подоляка П.С., Гамма Т.В. *В статье рассматриваются проблемы физической терапии детей с множественным дефицитом сульфатазы (МДС). Целью исследования, изложенного в статье, было обоснование необходимости формирования структурированной персонализированной комплексной программы реабилитации детей с множественным дефицитом сульфатазы. Задачами исследования предполагался анализ литературных источников по теме исследования, изучение клинического фенотипа множественного дефицита сульфатазы и потенциальных осложнений данной патологии, обоснование применения средств физической терапии детям с множественным дефицитом сульфатазы. Методы исследования: анализ научно-методических источников отечественных и зарубежных авторов, педагогическое наблюдение, сбор анамнестической информации пациента. В публикации рассматриваются клинический фенотип генетической патологии и возможные потенциальные осложнения*

данного орфанного заболевания, стратегические векторы индивидуальной программы реабилитации. Представлено описание клинического случая поздней инфантильной формы заболевания. Исследуется результативность шестимесячной реализации программы физической терапии. Для реабилитационного обследования детей с МДС предлагается применять тестирование детей, имеющих психомоторные нарушения. Раскрывается методика массажа, целесообразность вертикализации и ортезирования, сенсорного обогащения среды ребенка с этой генетической патологией. Предлагаются упражнения лечебной физической культуры, которые будут способствовать сохранению постурального контроля, опороспособности и функционирования рук, ног, зрительно-моторной координации, помогут предотвратить заболевания бронхолегочной системы. Отмечено, что МДС недостаточно изучена не только с позиции лечебной стратегии, диагностических алгоритмов клинических многосистемных проявлений, но и требует внимания анализ результативности и эффективности системы современной реабилитационной терапии детей с данным диагнозом.

In the world, more than 300 million people suffer from one or more rare diseases. Much of orphan diseases are diagnosed at an early age, in 35% of cases, they are the cause of death of infants under one year, in 10% – children under 5 years of age, in 12% – aged 5 to 15 years. An extremely rare disease – multiple sulfatase deficiency (MSD) – neurometabolic disorder, inherited in an autosomal recessive form needs particular attention [1, 4, 5, 6].

Today, the literature describes a bit of data on the clinical phenotype of MSD, the main complications of the underlying disease. Therefore, the treatment protocols and directions of rehabilitation of patients with a diagnosis of MSD are not sufficiently elaborated. An important aspect of the physical therapy program of such children is to form a rehabilitation system at home.

Consequently, the purpose of our study was to justify the need for the development of a structured personalized complex of rehabilitation of children with a multiple sulfatase deficiency.

MATERIALS AND METHODS OF RESEARCH

The study introduces analysis of scientific and methodological sources of domestic and foreign authors, systematization of clinical manifestations of MSD in the age aspect, differentiation of individual pathologic states and the features of this genetic disease. An analysis of the patient's video archive was performed.

RESULTS AND DISCUSSION

According to ICD-10, multiple sulfatase deficiency corresponds to E 75.2 from the group of lysosomal diseases of accumulation [2], MeSH – D052517, MIM – phenotype number # 272200, indicated in the classification of phenotypical genetic relations in the National Center for Biotechnological Information of the United States (National Center for Biotechnological Information, NCBI). In recent materials it is noted that 75 publications are generally offered today. According to their data, 143 MSD patients with a total number of 53 unique mutations SUMF1 are considered. 75 clinical signs and 11 clinical groups of signs [8] were detected.

The clinical picture of MSD combines symptoms of various sulfatase deficits and resembles a late infantile form of metachromatic leukodistrophy (250100) or mucopolysaccharidosis (MPS6; 253200) [3]. According to the age of the clinical debut, the newborn, late infantile and young subtypes of the disease are distinguished [5]. Experts emphasize the dependence of the first clinical manifestations of the MSD and the perspective of the course in accordance with the age. This genetic disease has no gender discrimination, the incidence of lesion of men and women is the same [5].

Clinical management of patients with this genetic pathology is poorly elucidated. A number of genetics, neurologists, pediatricians study this pathology [6, 8]. In the professional literature, the simultaneous symptom complex does not have a clear diagnostic algorithm and, accordingly, differention with other pathologies, the result of which is motor disorders.

According to publications, MSD treatment does not exist. The therapy of the main problems is mostly symptomatic palliative.

Our rehabilitation program is based on previously published studies in relation to the clinical course of the disease, a wide range of changes in the body. We did not find a specific rehabilitation assessment of children with MSD. Therefore, the survey means are tests and scales, typically used for children with disorders of motor functions [2, 3, 5].

Clinical case. Boy, aged 3 years 2 months. Late infantile type, severe MSD form. The diagnosis was made in the Center of orphan diseases "Okhmatdit" of the Ministry of Health of Ukraine. The basis for our examination and pedagogical observation was a biopsychosocial model of rehabilitation of international classification of functioning.

Structures and functions. *Anamnestic information*: According to parents and on the basis of analysis of photographic materials it is obvious that the initial development of the child was in accordance with the stages of typical development. In the period of independent walking at the age of 1.5-2.5 years, walking was atactic, genu recurvation. On examination: the child can only sit with his legs

on the mother's arms or on a chair with the help of one of the parents. The motor age of the child corresponds to 6 months. According to Gross Motor Function Classification (hereinafter GMFCS) according to Palisano, Rosenbaum, Bartlett & Livingston, 2007, in relation to the age of two to four years, the level of V motor development – physical defects limit voluntary control and the ability to maintain antigravitation of the head and postural control. All spheres of motor function are limited. Functional restrictions in sitting and standing mode are not compensated completely. It is necessary to use adaptive equipment and auxiliary technologies, no ability to move on their own even on a cart.

Disturbance of postural control. The muscle tone in the abductor muscles is elevated, poor mobility in the ankle joint more in the right: according to a modified Ashworth scale, muscle tone corresponds to 1 point. There is no visual-motor coordination. Functioning of hands is not productive, chaotic. The skin at the time of the review is thickened, scalded, expressed marble. The distal parts of the extremities are cold. Deformation of the chest is in the form of thickening and expansion of the 10th, 11th and 12th pairs of rib arcs. Complaints on defecation troubles: difficulty, once every three days, needs laxatives per os and per rectum. Frequent respiratory diseases.

Activity and participation. *Anamnestic information*: The child could amuse himself, attended a kindergarten, at the age of 1.5 years he knew letters, animals' names, simulated animals' sounds, repeated the movements of his parents. At the time of inspection of the child's leisure he sits on the arms of one of the parents. Slightly reacts to the toys. The child's mother takes hand of the boy to take a toy trying to amuse him. Emotionally unstable. Slightly reacts to the sounds and verbal message. Communication is episodic in the form of visual contact. Spoon-feeding on the arms of the mother, but it is planned to transfer to grinded food from the bottle. **Factors of the external environment** - a child on the parents' hands during leisure and meals, daytime, night sleep, on the go.

Parents' request – "to do anything."

Figure 1 presents a patient with MDS at the first course of physical therapy.

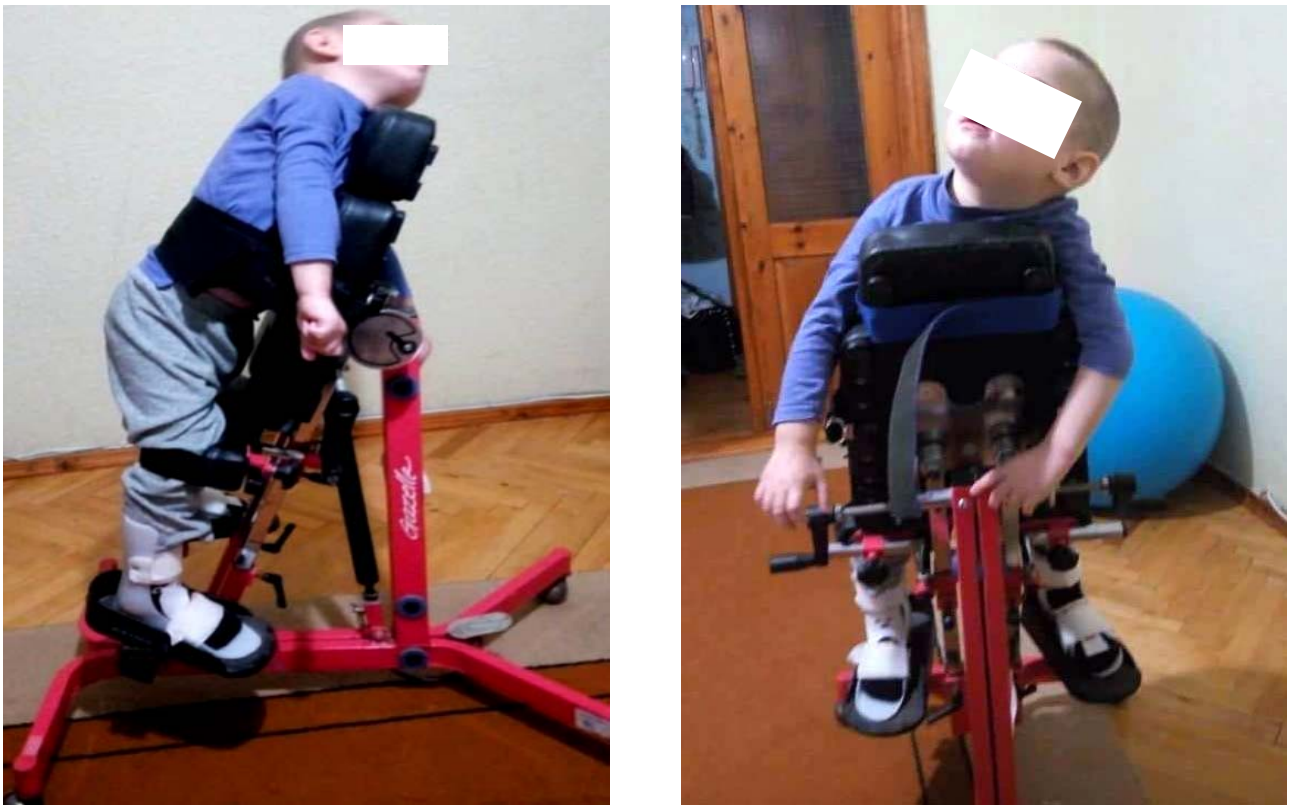

Fig. 1. Patient with MDS at the first course of physical therapy

The task of rehabilitation intervention: to modify the environment to an independent child's stay; prevention of potential clinical complications: to prevent further deformation of the chest, legs,

spine, taking into account the forecast of deteriorating bone density and local spastic tone of leg muscles and with a tendency to hypotonic tone of the muscles of hands and back.

Rehabilitation program included exercise therapy, massage, verticalization, orthosis, sensory saturation of the environment and lasted for 6 months.

Massage was used to peel and remove the dead cells of the epidermis; improvement of the excretory, respiratory, thermoregulatory function of the skin; normalization of the functional state of the nervous system; increase in the inflow of arterial blood to the area being massaged and surrounding tissues, enhancing fermentation processes. By combining mentioned mechanisms of influence, we identified the main massage techniques: reflex-segmental massage, action on all segments of the skeleton. Influence is deep, warming up, of moderate intensity with correction of spasticity in the lower extremities. Its duration in the preparatory period (1-3 days) is 40-50 minutes, in the main period (4-10 days) – 50-60 minutes, daily. Areas being massaged: legs, hands, stomach, chest, intercostal muscles, back (paravertebral zones, tender zones), collar zone. Massage techniques: stroking, deep grinding, deep kneading, non-perforated, intermittent vibration, passive movements in the joints. The skin after massage becomes pink, warm, elastic. This approach was performed in one rehabilitation session that lasted, on average, over an hour.

In order to prevent the deformation of the lower extremities during verticalization, active orthoses DAFO were put on the child's knee-ankle joints, foot. Verticalization of a child in the vertical static position in the anterior and then in the posterior vertical one with an angle of inclination was used, as well as specialized chair for playing and eating.

The complex use of physical exercises envisaged conditioning ones in the form of passive exercises for the upper, lower extremities, mobilization of joints, static and dynamic respiratory exercises. Exercises were performed from different preparatory positions. In particular, on the fitball in the position on the back, the traction for the forearms for the promotion of grouping, sitting, strengthening muscles of the abdominal press was performed. In order to activate the muscles of the back, shoulder girdle, the recovery of the hands, control over the position of the head, exercises on the fitball in pronate position were performed: planch on the ball with palms, forearms, with legs on the floor. Thus, the child was at an angle to the support, which facilitated the exercise. To form the transfer of body weight, "steps" were performed with the hands on the floor: the child was lying on his stomach on a fitball. To improve postural control, balance training, coordination of movements, exercises were performed in a sitting position on a fitball: jumping, swinging from side to side, in a circle. To restore visual-motor coordination, maintaining the functioning of the hands, exercises were performed in a standing position in the verticalizer by the method of "hand in hand" – exercises with the ball "hit the target", "catch the balloon", follow the flashlight on toys.

Lying on the "blade" helped to strengthen the muscles of the back, the ability of the forearm, visual tracking of objects.

The recommended exercises are designed for daily performance both in classes with a physical therapist and during the day with one of the parents. Distracted physical activity during the day provided a moderate load and prevented the child's fatigue.

For the purpose of sensory enrichment, musical compositions with major character from cartoons, bright sound toys were used. Criteria for assessing the effectiveness of physical therapy – curb the deterioration of muscle tone, motility, orthopedic disorders, emotional and volitional disorders, respiratory diseases.

Structure and functions. Muscle normotony was restored after 10 first-year massage sessions. The volume of movements in the ankle joint has been restored. Visually, the deformation of the chest has significantly decreased, which, according to parents, is also noted by close relatives. The translational automatisms were partially restored, which is manifested by the control over the position of the body in the verticalizer. The condition of the skin – warm, clean, elastic, with physiological turgor. During the therapeutic support, which fell on the autumn-winter-spring period, the child never had diseases of the respiratory system. In a child, the act of defecation occurs every other day, sometimes daily, the use of oral laxatives is situational, and the use of a potty has significantly improved the child's quality of life. At the same time, the expression of primitive reflexes, in particular the asymmetric cervical tonic reflex and the Moro reflex, is periodically observed.

Figure 2 shows patient in the end of investigation.

Factors of the external environment. Spoon-feeding is carried out only in a specialized chair. Sleeping in a separate baby crib. Calmly responds to standing in a vertical system for 40 minutes twice a day, favourite amusement in this position - ball game, playing with toys, viewing cartoons.

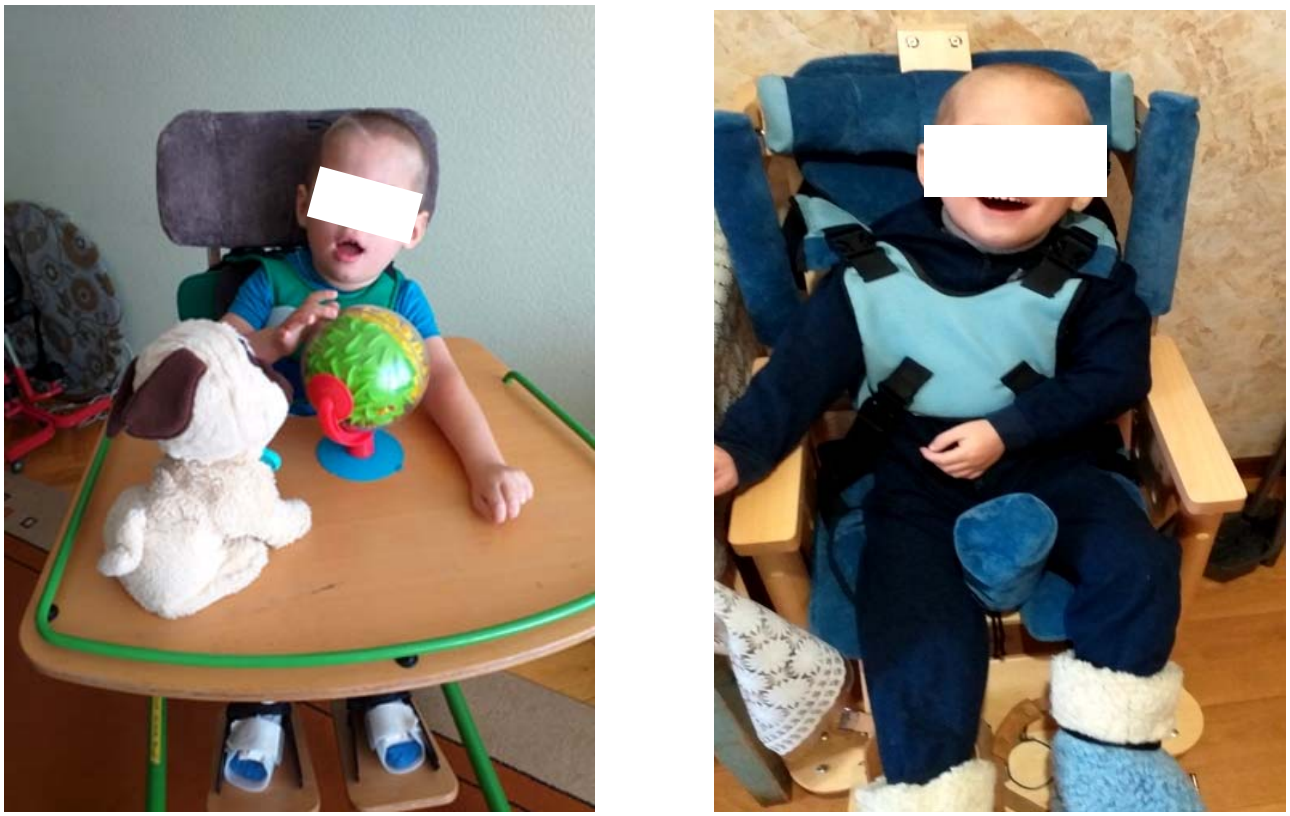

Fig.2. Patient with MSD after the course of physical therapy

CONCLUSIONS

1. Analysis of literary sources showed insufficient information on timely diagnosis and program of therapeutic supervision of children with multiple sulfatase deficiency. It was established that the clinical course of this orphane disease is characterized by a rapid deterioration of psychomotor, speech, cognitive levels. Differentiation of the phenotype and the detection of genuine diagnosis and thus management strategy, requires careful study, clarification and coordination. To date, there are no clinical guidelines for the therapy of children with a multiple sulfatase deficiency. The introduction of a comprehensive program of physical therapy of the patient with MDS resulted in the improvement of the child's condition and the quality of life both of a child and the family, which was expressed in control over the tone of muscles, settling of bowel movement, prevention of respiratory diseases, absence of cutaneous problems, settling of leisure.

2. Given the orphane disease, non-sufficient experience of therapy of such children, require careful selection of means of rehabilitation and ergotherapy, as well as an analysis of the results of rehabilitation intervention and the search for effective means of physical therapy.

Conflict of interest. The authors declare no conflict of interest.

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