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Organization of psychological and educational assistance in the rehabilitation of children with severe chronic hereditary metabolic diseases (on the example of mucopolysaccharidosis)

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The paper discloses the contents of correction and pedagogical influence, which was determined according to the severity and duration of the underlying disease, the structure of health problems, the degree of underdevelopment of cognitive activity, type of emotional response, the characteristics of the child's behavior, that had not been associated with age of the child directly. We describe the stages of the remedial educational assistance, the direction of the family, the organization of the process of upbringing and education of the child with inherited metabolic diseases (for example, mucopolysaccharidosis). It is proved that the inclusion of remedial educational assistance in the course of rehabilitation treatment with a team of specialists provides monitoring of the dynamics of mental development which is an important indicator of the health of children; increases the effectiveness of the huge financial expenses for medical treatment due to the socialization of children and preserves mental capacity of family members capable to work.

***Key words**: children with limited health abilities, children with disabilities, special psychological-educational assistance, medical-psychological-pedagogical rehabilitation, health status of children with inherited metabolic diseases, progressive dementia, and children education with severe diseases.*

At the current stage of development pediatrics focuse on finding methods to preserve the life and health of children with inherited metabolic diseases [1, 2]. 18 classes of this kind of disease are known currently. One is a class of lysosomal diseases, which include the clinically polymorphic, relatively heterogeneous group

of MPS - severe disabling disease with a rapidly progressive course and fatal outcome in childhood or adolescence [2-4]. The disease is caused by genetic disorder, multistep process of enzymatic catalysis of glucose amino glycan in the lysosomes (enzymatic cleavage of the carbohydrate moiety of mucous polysaccharides), which leads to the accumulation of nonsolid items in the connective tissue and disrupts its structure [2, 3, 5]. This group includes 10 units with the nature of the various enzymatic disorders and the clinical picture; the designation is made based on letters and numbers: MPS type I, MPS type II, MPS type III, etc. They are all united in the ICD-10 in class 4 under codes E76.0 -76.3 [4, 5]. Symptoms manifest in the first year of life and during early childhood clinic becomes pronounced; without adequate treatment a significant limitation of activity of the child and the rapid death come in [2, 5]. Pathology of metabolism causes a disturbance of the functional state of various organs and systems, which manifests as a symptom complex with a number of common clinical manifestations. In most cases this leads to gross changes in the skeleton, cardiovascular system, respiratory, vision and hearing, as well as a significant distortion of the psyche according to the type of dementia [3, 5]. According to the legislation of the Russian Federation, rehabilitation of children with severe health problems include a complex of medical, psychological, social and economic measures to compensate the possible limitations of life [1, 6]. To stabilize or suspend this pathological process a combination of the following methods are widely spread as comprehensive treatment of this kind of disease: symptomatic therapy, enzyme replacement therapy, stem cell transplantation, bone marrow transplantation, physic therapy, physical therapy, psycho-pedagogical effects [7, 8]. The package of measures to preserve life and health of patients with generalized mucous polysaccharide are legally enshrined in Art 11 "Individual rehabilitation program for persons with disabilities" Federal Law of November 24; 1995 № 181-FZ "Social Protection of Disabled Persons of the Russian Federation"; as well as in Art 55, 66, 67 of the Family Code of the Russian Federation, Russian Federation Law "Education"; and in Art 30 and 61 of the Federal Law «Public Health of the Russian Federation" [1, 2, 9].

Treatment is directed at improving the processes of metabolism, reduction the accumulation of glucose amino glycan in cells in order to enhance the activity of the enzyme deficit, which in turn can slow down the progression of the disease, prolong life and prevent rapid deterioration of physical and neurological status of patients [2, 5]. The literature indicates that even the most modern and expensive treatments, which include a new biotechnological product - a recombinant version of the deficient enzyme does not have a positive impact on the state of the central nervous system which has already held its defeat, and also did not contribute to the restoration of previously accumulated skills and cognitive capacities [2, 3, 8]. The authors attribute this effect to the impermeability of the blood-brain barrier to the drug [2, 5]. Thus, the treatment improves the general condition of the patient, but does not perform the correction of the behavior and response of these children on any type of impact, which greatly complicates the process of clinical management of the patients by physicians, as well as custody and care of loved ones [2, 5].

The most obvious mental disorders are agitation, negativity, and fear of anything new; a change of mood, eating disorders and acts of defecation. International Psychiatric Association groups put them together under the broad term "behavioral and psychiatric symptoms of dementia" [8, 10]. Dementia is understood as diffuse mental functions violations resulting from organic brain damage, manifesting with violation of thinking and memory, as well as disorders of emotion and behavior [3, 10]. In cases of hereditary metabolic disorders like mucopolysaccharidosis a harmful factor has a devastating effect on the CNS continuously, whereby the process is characterized as a progressive, growing steadily manifested by the disintegration of mental functions [2, 10]. The development of progressive dementia is caused to focal or diffuse brain lesions, which is clinically manifested by reduced cognitive abilities, disturbance of orientation in the environment, loss of social skills, that is to say severe neurological symptoms and mental disorders [8, 10]. Such a significant change in neurological status, the psyche and behavior of young patients require timely methods of evaluation and determination of a possible correction [2, 3, and 9]. One of them is a method of psychological-pedagogical influence on the psychological condition of the patient, which also carries the psychological effect of parental support due to their provision of the necessary expertise in matters of education and training of special children [6, 9, and 11]. It was assumed that the inclusion of remedial educational assistance in the treatment of children with serious chronic metabolic diseases (including MPS), along with other (physical therapy, physical and medicinal) species will slow down the impact of social regression skills. A creation of special environment promoting child's cognitive activity and satisfying his/her psychological needs [7, 11, and 12] were planned to achieve this goal.

This study was conducted to examine the specific educational needs of children with mucopolysaccharidosis, and to determine the mechanism of the inclusion of correctional and educational assistance in the rehabilitation process of their health status.

To address this goal it was necessary to determine the methods of psychological and pedagogical examination of the state of the cognitive activity of children in this category. In the process of practical scientific research two techniques have been proven to be effective done by domestic authors: methods of psychological and pedagogical examination of E.A. Strebeleva and methods of examination of nervous and mental development of the G.V. Pantyukhina et al. [13, 14]. With their help information about the features of cognitive activity, emotions and behavior of 11 children aged 4 to 14 years with mucopolysaccharidosis type II and III, with varying duration of its flow were collected and analyzed. By the time of the survey a current level of mental development was set, consisting of their orientation in the world, and in some cases the development of cognitive activity. For definition of potential possibilities of training, effective methods and receptions of the correctional and pedagogical help, a mode of pedagogical loading, a form and duration of occupations we used the method of diagnostic training offered by A.Y. Ivanova — supervision over changes in a condition of mentality and behavior of children at their inclusion in process of systematic

training [9, 11]. For this purpose we have conducted several specially organized developmental activities on a weekly basis, the total amount of which did not exceed five. Between classes parents of children secured skills at home, guided by specially designed for each child's individual educational program. This approach allowed for the 36 weeks of treatment during which children received weekly intravenous infusions of substitutive (total no more than 18 injections) to study the peculiarities of the underlying disease, changes in mental development and behavior of the child. Analysis of the data allowed us to determine the organizational form, the stages, the direction and content of remedial educational assistance in the treatment of children with MPS, improving parents' pedagogical competence in matters of child raising and caring.

Careful analysis of pediatric, neurological, psychological and pedagogical examinations and diagnostic study divides the surveyed children into the three groups, depending on the severity of underlying disease and the depth of mental disorders.

The first group included four children (one preschooler, one student with MPS type I and two preschool children with MPS type II) that were observed during treatment with stabilization of the underlying disease; the body of the child responded positively to the replacement therapy, whereby the regression of psychological and cognitive abilities had been slowed down. It manifested as a slight decay of the previously formed mental functions, a gradual increase in mnemonic difficulties, and emotional liability in passive or violent manifestations of negativism, reduced efficiency and difficulty in performing accurate motor acts, and significant speech impairment. All children, regardless of their age, had the thinking at the level of visual-motor; cognitive activity was sharply reduced, they didn't experience the needs for a new study. They couldn't spend their time on playing; actions with toys were stereotyped and manipulative in nature, taking into account the properties of the object and its purpose. The arbitrariness of action has been retained, but exhausted within 10-15 min, after which the activity in the environment has become chaotic and destructive. Speech regulated their actions, they understood and met the requests of adults, simple grammar phrases were used in communication. The dictionary was extremely poor, tired children were passing to the more elementary modes of communication: gestures, facial expressions and tone of voice.

The second group included three patients (two preschool children with MPS II and type III, a boy with MPS type I) who had a gradual deterioration in overall health and psyche with significant setback to the level of elementary practical orientation in the environment. Activity was involuntary, the new space was scared and excited for them, the emotional state was labile. The explosive emotion was changed into a complete passivity. Transitory interest in toys could cause a desire for self-movement in space accompanied by poorly coordinated walk with legs wide apart for balance. Moving in this way, they examined the room aimlessly, thus satisfying the innate human need for activity. They couldn't calculate the distance to the object and the speed of movement on their own, often stumbled, hit the barriers and fell. Caught the toy, children immediately threw it without any

interest in its investigation, skill orientation to its sensory properties and quality has been lost. Speech was not controlled, they understood a few words, and responded to the tone. They informed an adult about their needs by shouting, changing behavior and violent differentiated emotional reactions.

The third group consisted of four children with various types of MPS (a teenager, a pupil and two preschool children), whose health and psychological state was rapidly deteriorating because of severe, rapidly progressing disease course. The process of disintegration of the psyche could be characterized as a progressive dementia. The survey results revealed a gross violation of cognitive activity to a level of indicative reactions to the impact of sensory stimuli. The behavior of the children were prone to agitation, there were affective disorders, disinhibit ion of instincts, impaired walking, inability to self-service and control of biological needs. The view of new toys aroused excitement, the desire to seize her, but the child couldn't perform a coordinated motor act without adults' help. At the time of acquisition a toy children had positive emotions, facial expressions of pleasure, and separate sounds of rapturous tones. They did not commit actions with the subject, doing some waving motions, causing the sound a toy lost and trying to find it did not produce anything, children became indifferent. Self-activity of children during the day could be characterized as passive with a tendency to excitation. However, with the help of a special pedagogical influence was possible to enhance the children from previous ability of orientation in the environment, the skill of the social means of communication (voice, facial expressions) to communicate the feeling of hunger and pain, his/her social desires and preferences.

Thus, we found that regardless of the severity of the disease, its consequences in the form of secondary and tertiary violations of development, children were in need of constant health status monitoring, careful selection of drug therapy and regime of rehabilitative treatment load, as well as the establishment of special educational conditions in the treatment due to life limitations, which manifested itself in the inability to self-adapt to new conditions, the implementation of social contacts and acquisition of skills.

Significant differences in the pathogenesis and progression of the disease, the level of mental development defined the necessity to determine an individual approach to the selection of content, methods and techniques of remedial teaching. Analysis of individual educational programs and training revealed that the remedial teacher working with each group of children had different goals, which were determined by the severity and duration of the underlying disease, the structure of health problems, the degree of underdevelopment of cognitive activity, especially emotional response and behavior, while they were not directly associated with the type of illness and age of the child.

Thus, for the children of the first group goal of correctional and pedagogical influence was to create conditions for enhancing the needs and the skill for self-usage of previously acquired skills, social behaviors and communicative forms of interaction with others for the gradual accumulation of new experience.

In working with children of the second group the goal was defined as an extension of current cognitive skills, as well as consolidation of self-orientation in the

environment through simple, previously formed habits. In our opinion, this should provide the child the opportunity to exercise for longer rational interaction with the social environment, have a sense of security and independence in his/her.
The purpose of correctional and pedagogical impact of children of the third group was the creation of conditions to meet the inherent requirements of sensory stimuli and communication with others, providing emotional and physical comfort. Remedial educational process with this group of patients was based on the type of formation of behavioral stereotype so that a child could use, regardless of the communication partner and changes in the environment, conditions of stay. Summary of the results taken 1 year and a half allowed us to determine the ability to turn technology into pedagogical rehabilitation of children with MPS types. Remedial teaching for children with MPS was carried out in three stages, each of which consisted of several interrelated parts. The number and sequence of stages in the treatment of each of the child remained unchanged. However, due to significant "fluctuations" in the health and psyche of children's time intervals each stage might vary.
The first stage is a diagnostic (rate definition and characteristics of the psychological development of children) comprised two parts.
The first part is the study of the child’s history, analysis of his/her anamnestic data, and conduction of psychological and pedagogical examination. For the diagnosis of mental development of young children the technique GV Pantyukhina et al. Psychological and pedagogical examination of all the others carried out by the method of E. Strebelevoy was used. Given the significant differences in cognitive development, the nature and severity of behavioral problems, special emotions in the process of asking children with MPS, the use of additional sensory stimuli (novelty, color, sound, tactile contact), large and strong gaming aids should be used to provide assistance through sharing of some action, do more pauses in a survey to find time to rest.
The second part consisted of the diagnostic phase of training, the duration of which was not more than three weeks. This technique was used by us to identify potential opportunities for children to master new learning with the help of the "zone of proximal development", which could not be done during the initial examination. Diagnostic studies allowed to smooth the negative reaction of children to contact with unfamiliar adults, to help them adapt to the new learning environment and the rules of interaction with the teacher, many times to produce the same job, make learning new material through practical use over a certain time segment for two or three classes. Parents of the children secured skills by organizing similar developmental activities at home at least twice a day. The results obtained in the second phase of the information collated and subjected to rigorous analysis, and establishment of the level of mental development of children at the start of treatment was possible.
The second stage called the impact of remedial teacher was consisted of four interrelated parts.
The first part was designed to determine the form, mode and content of remedial educational assistance.

The second phase involved the development of the individual program of training and education, which was compiled separately for each child (a set of exercises, methods and techniques, teaching toolkit, which were appropriate for teachers' use in the classroom and parents at home in their daily lives to improve the quality of social adaptation of children).

The third part of the stage was devoted to the immediate implementation of educational assistance, and expressed in the form of individual sessions with the child. The theoretical framework that we have implemented was remedial teaching process of L.S. Vygotsky that "while there is a defect with the psychological trend in the opposite direction compensatory possibilities to overcome it are found, and they come to the fore in the development of the child and should be included in the educational process as it is the driving force" [11]. Remedial educational assistance was carried out in two directions: the creation of conditions for the correction of both general and specific abnormalities in the development, the formation of new, more complex levels of interaction with reality in the process of leading and typical activities. Weekly developmental studies were conducted only in the morning before the infusion or within two days after it, at least no more than two times per week they were organized in the form of a game, and stimulated an arbitrary orientation and exploratory activity of children in the exercise of practical action with objects on the background of positive emotional contact with an adult. Assignments and didactic materials were selected for this task affecting multiple analyzers simultaneously, carrying an element of novelty and feasible complexity. In the exercise, the teacher-pathologist sought compensatory possibilities to intensify the child's body, to restore lost cognitive and psychomotor skills of the child.

The fourth and final part of the second stage was devoted to parents' teaching how to use special educational technology throughout the day. Practical exercises with relatives occurred in the form of special teacher who demonstrated methods and techniques of cognitive and social activity of the child; emotional and developmental ways of communicating with his/her baby.

The third stage was control. The first part has been implemented on a quarterly basis and a controlling psycho-pedagogical study was required for the timely assessment of psychological status, the effectiveness of teacher effects, the dynamics of cognitive development, and pace of the formation of psychological achievement; fixing the characteristic changes in behavior and emotions, making the necessary changes in the educational process.

The second part of the third stage was organized every six months in order to clarify the expert team of child health, discuss and share with family decisions about tactics in future regenerative therapies (therapies, the content of rehabilitation programs, special educational sessions).

The proposed approach to remedial educational assistance in the complex rehabilitation of children in health care facilities enables close monitoring team of professionals for changes in health status and mental status of children with severe chronic hereditary metabolic diseases in patients receiving replacement therapy, and high-quality educational evaluation. It helps make a differentiated selection of

remedial teaching methods and techniques in a systematic practice with a child on a specially designed individual program of education and training, as well as implement a humanistic approach to the presentation of the parents of a new specialized information (medical, educational, psychological) about the health of your baby, necessary types of medical and educational assistance, the forecasts of its psycho-physical development, improve the efficiency of huge material costs for the treatment of these children by raising their level of socialization and the psychological state of working family members.

In conclusion, based on analysis of data obtained during the two-year study, it is necessary to draw the following conclusions:

- Rapidly progressive and severe course of illness requires immediate use of complex medical, psychological and pedagogical rehabilitation methods since the detection of hereditary metabolic diseases in the child, that is, during early childhood should be done;
- The process of disintegration of mental functions in children with severe chronic diseases of metabolism may be suspended only if somebody uses a complex of modern methods of rehabilitation therapy: symptomatic (palliative) and enzyme replacement, physical therapy, physical therapy, remedial teaching and counseling;
- For one year of comprehensive treatment of mucopolysaccharidosis types I and III in children older than 3 years significant changes in health status of patients were not observed, which may be regarded as the stabilization of the disease;
- The severity of the collapse of the cognitive activities and mental processes depends on the type and duration of disease, time of initiation of enzyme replacement therapy;
- By virtue of bilateral destruction of the organic matter of the brain and its pathways preserved compensatory capacity of the organism and therapeutic action of enzyme replacement therapy is not enough even for a partial restoration of normal functioning (minimal positive changes in mental and physical development of children in a partial restoration of broken skills and abilities are the only evidence slowing the rate of dementia, mental functions);
- Minimal positive changes in the volume and quality of motion, pain reduction were observed after six months of comprehensive rehabilitation with the use of enzyme replacement therapy;
- The systematic application of a significant number of drug and non-drug methods to influence the child's body in the first place helped to restore the most basic, life supportive body, elementary functions (gestural-mimic), communication skills, and arbitrary behavior;
- The minimum recovery of sensation, awareness of their own ability and skill needs of the biological information of others with signs of their biological needs is an indication that the nature of the violation of the exchange have a devastating effect on brain structure: striking white matter of the brain and disrupt the afferent system, which in turn , shows the disintegration of mental functions, motor skills and sensitivity, including the control of defecation and urination;
- Total defeat of the brain, pathways accompanied by a decrease of sensitivity and inability to control feelings of hunger and satiety. This circumstance requires a

careful selection of a balanced diet, taking into account height-weight performance and allergic status of the child, strict eating regime, education of parents in child care issues with the IPU and care for it;
- The need for children with MPS in a systematic high-tech medical care indicates a need to establish in Russia a system of state hospitals for long-term rehabilitation of a sick child in the process which could take an active part of his family.
The results showed that the metabolic disorder leads to a significant restriction of activity in the form of rapid distortion of mental, behavioral disorders, loss of skills at all for children with this kind of disease without an exception. This, in turn, becomes an obstacle to their socialization; it is extremely difficult to process the education and care for them. Without the creation of special training conditions, children with severe chronic metabolic diseases are much faster than the previously formed loose psychological achievement, so physicians need to include educational technology in the rehabilitation process of their health in order to improve the quality of life.

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