UDC 616.832-009.54

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Application of ICF classification system in functional assessment of children with spinal muscular atrophy

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Modern Pediatrics. Ukraine. (2022). 2(122): 50-54. doi 10.15574/SP.2022.122.50

For citation: Nazar OV, Hurzhii OI, Martyniuk VV, Moiseienko RO. (2022). Application of ICF classification system in functional assessment of children with spinal muscular atrophy. Modern Pediatrics. Ukraine. 2(122): 50-54. doi 10.15574/SP.2022.122.50.

Spinal muscular atrophy (SMA) is a rare genetic inherited neuromuscular disease that requires a comprehensive multidisciplinary approach to diagnosis and treatment.

Purpose — to provide a review of scientific works on comprehensive care for children with SMA according to the principles of the International Classification of Functioning, Disability and Health (ICF), standardized tools for assessing the functional status of the patient.

Materials and methods. The article presents the data of the literature review on the use of ICF principles in the organization of care for children with SMA, the use of standardized tools for assessing the functional status of the patient.

Review and discussion. The implementation of the ICF in daily practice helps to improve the rehabilitation aid to children with SMA. Using the philosophy of the ICF, physicians began to comprehensively assess the health of the child, to estimate the functionality of the child using standardized tools, to form rehabilitation goals in accordance with the functional capabilities of the child, to determine environmental factors which do not allow the child to fully realize their rehabilitation potential.

Conclusions. ICF makes it possible to determine of all aspects of the life of a child with SMA and to determine the impact of environmental factors on its level of functioning. According to ICF principles it is necessary to use standardized tools to assess the health status and the functional activity of a child with SMA, for evaluating the effectiveness of rehabilitation.

No conflict of interests was declared by the authors.

Key words: ICF, spinal muscular atrophy, rehabilitation, functioning.

Застосування системи класифікації ICF для оцінювання функціонального стану дітей зі спінальною м'язовою атрофією

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Спінальна м'язова атрофія (СМА) — рідкісне генетичне наслідкове нервово-м'язове захворювання, що потребує комплексного міждисциплінарного підходу до діагностики й лікування.

Мета — навести огляд наукових робіт щодо комплексної допомоги дітям зі СМА відповідно до принципів Міжнародної класифікації функціонування, інвалідності і здоров'я (ICF), із застосуванням стандартизованих інструментів оцінки функціонального стану пацієнта.

Матеріали та методи. Наведено дані огляду літератури щодо застосування принципів ICF в організації допомоги дітям зі СМА, використання стандартизованих інструментів оцінки функціонального стану пацієнта.

Огляд та обговорення. Запровадження ICF у повсякденну практику дає змогу поліпшити реабілітаційну допомогу дітям зі СМА. Застосовуючи філософію ICF, лікарі почали комплексно оцінювати стан здоров'я дитини, її функціональні можливості за допомогою стандартизованих інструментів, формувати реабілітаційні цілі відповідно до функціональних можливостей дитини, визначати фактори навколишнього середовища, що не дають змоги дитині повною мірою реалізувати власний реабілітаційний потенціал.

Висновки. ICF дає змогу визначити всі сторони життя дитини зі СМА, а також виявити вплив факторів навколишнього середовища на рівень її функціонування. Відповідно до принципів ICF, слід застосовувати стандартизовані інструменти для оцінювання стану здоров'я і функціональної активності дитини зі СМА, а також для оцінювання ефективності реабілітації.

Автори заявляють про відсутність конфлікту інтересів.

Ключові слова: ICF, спінальна м'язова атрофія, реабілітація, функціонування.

Introduction

Spinal muscular atrophy (SMA) is a rare genetic inherited neuromuscular disease that is also one of the most devastating neurological diseases in childhood. Infants and children with SMA suffer from progressive muscle weakness, with the development of severe muscle atrophy and flaccid paralysis of skeletal muscles and respiratory muscles caused by degeneration of motor neurons in the spinal cord and brain stem [15].

SMA requires a comprehensive approach to diagnosis and treatment, optimal control of the course of the disease requires the cooperation of many healthcare professionals [19]. All patients diagnosed with SMA should be referred to a specialized clinic / centre for neuromuscular diseases, where they can receive multidisciplinary care of: a pediatric neurologist (examination, coordination of multidisciplinary care, determination of indications for gene therapy), consultation with a pulmonologist (functional assessment of respiration, determination of indications for respiratory support), a gastroenterologist (optimal nutrition of a child with SMA, management of dysphagia, indications for the installation of gastrostomy), an orthopedist (prevention of orthopedic complications, orthosis, prevention osteoporosis), a doctor of physical and rehabilitation medicine, a physical therapist, an ergotherapist (for the

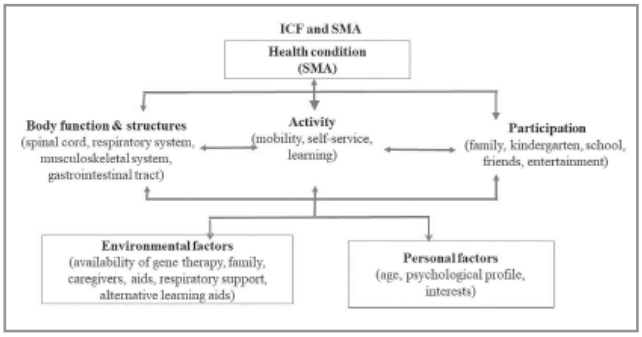

Fig. ICF and SMA

provision of rehabilitation services, the formation of a home rehabilitation program, the selection of auxiliary means), a geneticist (families and adults with SMA should also be provided with advice on genetic issues) [6,15,19].

Specialists should work in an interdisciplinary team and take into account not only the patient's medical needs, but also take into account and respect their social, cultural and spiritual needs. The International Classification of Functioning, Disability and Health (ICF) is the classification, in terms of which we can cover all the patient's problems caused by SMA, which allows doctors to diagnose these problems in time, assess the level of functionality and rate of disease progression, select medical treatment and rehabilitation interventions [21,22].

Purpose of the study — to provide a review of scientific works on comprehensive care for children with SMA according to the principles of the International Classification of Functioning, Restriction of Life and Health (ICF), standardized tools for assessing the functional status of the patient.

Materials and methods

The article presents the data of the literature review on the use of ICF principles in the organization of care for children with SMA, the use of standardized tools for assessing the functional status of the patient.

Review and discussion

The ICF makes it possible to accurately reflect the health status of children with varying degrees of functional impairment. According to this classification, patient health indicators are presented in the form of various categories (which are indicated by a code) of the main components of the ICF, such as body functions and structures, activities and participation. Disease and disability are not a purely biomedical problem, the determination of the impact of environmental factors on the level of functioning of the patient and social adaptation is also reflected in the categorical profile of the ICF [26]. Sometimes environmental factors play a crucial role in improving a child's level of functioning. If we take the example of a patient with SMA, it is the availability of pathogenetic gene therapy that will prevent the progression of the disease and death; the availability of respiratory support equipment will extend the quality and life expectancy of the child; multifunctional strollers improve patient mobility. In such cases, the support of the state and local communities plays a very important role, which has reformatted the approach to providing care to children with disabilities from a biomedical model of care to a biopsychosocial model.

Thus, the ICF classifies both various health indicators (such as respiration, muscle strength, gait, self-care) and indicators that are related to health (education, transportation, availability of medicines, auxiliary means, rehabilitation services, various social interactions) (fig.) [16,20].

Over the last decade, the world has been creating multidisciplinary teams of specialists to form basic categorical sets of assessment for patients with the most common diseases, such as cerebral palsy and autism in children. There is no basic set approved by the World Health Organization for children with SMA. At the same time, a clinical case of evaluating the effectiveness of gene treatment was published, where the authors presented the management of SMA and evaluation of the effectiveness of treatment in accordance with the ICF, a categorical profile was formed [21].

It is very important to determine the degree of violation of the structure and function, activities and participation for each profile category, using standardized assessment scales. Until recently, the clinical assessment of the functional capabilities of a patient with SMA was reduced to «standing / not standing, sitting / not sitting, turning over from back to stomach / not turning over, holding the head / not holding». The quality of movements was not taken into account. It is impossible to standardize such indicators and evaluate their changes in dynamics.

Over the last few years, we have been seeing very important advances in the pathogenetic (gene) treatment of SMA. An objective assessment of clinical symptoms and the degree of physical activity restriction allows to analyze the effectiveness of drug treatment of a patient with SMA. It makes it possible to quantify rehabilitation measures (in scores), indicate the areas of their

immediate development, increase the maximum outpatient stage of the disease.

Taking these criteria into account, several scales have been developed in the world to assess the motor activity of patients with spinal muscular atrophy, depending on the age and type of SMA. Some of these tests contain identical points, which is quite logical when evaluating neuromuscular activity. All scales provide for retesting after 6–12 months to quantify changes in the patient's condition. The choice of scales depends on the type of SMA, age and functional status of the patient.

The Bayley scales (Bayley Scales of Infant and Toddler Development) assess large motor skills in children with developmental delays aged 1 to 42 months [1].

The CHOP-INTEND (Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders) scale was specially developed by the Children's Hospital of Philadelphia to assess the motor skills of debilitated children in the first year of life [19,22]. This scale consists of 16 points, each of which has the following rating options: 0 — no answer, 1 — minimal answer, 2 — partial answer, 3 — almost complete answer, 4 — full answer level. Motor activity is evaluated in 7 domains: spontaneous movements; hand grip; head movements; hip adductors; turning over; flexion of the shoulder, elbow, hip; Galant reflex. Without treatment, children with type 1 of SMA experience a steady decline in CHOP-INTEND scale over time. The overall CHOP-INTEND score correlates with patients' need for respiratory support. Patients with SMA requiring BiPAP received lower scores on this scale [8,9].

HINE-2 scale (The Hammersmith Infant Neurological Examination, section 2) evaluates the development of a child's motor skills in 8 domains. It is used in children aged 2 to 24 months. The number of scores on this scale is from 0 to 26 [5]. With its help, the doctor quickly and effortlessly has the opportunity to assess the compliance of motor skills with the child's age. Patients with type 1 of SMA without treatment have a low score on the HINE-2 scale and do not increase over time. No patient with this diagnosis has a chance to learn to turn over or sit without support in the absence of specialized treatment. Therefore, achieving higher scores on the HINE-2 scale indicates the effectiveness of therapy [5].

The MFM (Motor Function Measure) scale evaluates physical activity in older patients. It allows to assess the degree of disease progression, as well as to detect even a slight positive trend in the background of treatment [2,23,25]. It evaluates the severity and progression of neuromuscular diseases in 3 domains: distal motor functions; axial and proximal motor functions; standing and moving positions. Each point of the scale has the following assessment options: 0 — the movement does not initiate or the ascending position cannot be maintained; 1 — partially completes the exercise; 2 — performs the exercise with compensations, slowly or clumsily; 3 — completes the exercise according to the standard scheme. The total amount of points is interpreted as: 85–96 — mild form of SMA; 65–85 — moderate one; 30–65 — severe one; 0–35 — very severe form of SMA.

The MFM scale can be used in a wide range of SMA patients and allows to conduct a full assessment of movement functions. If the amount of MFM points is converted to a scale from 0 to 100, a healthy person can get 100 points. Instead, patients with type 2 and 3 of SMA never get a score higher than 70 points without treatment. Given the progressive nature of SMA, stabilization of movement functions, or any improvement in MFM-32 is considered to be an important result. Improving the score by ⩾3 points is considered to be a significant clinical achievement. Important advantages of the scale are the ability to use in patients with different SMA phenotypes and different functional status (walking, non-walking), as well as the ability to fully assess motor function in the range from small movements to global motor skills [2,23,25].

The Modified Hammersmith scale (Expanded Hammersmith Functional Motor Scale (HFMSE), consisting of the HFMS (Hammersmith Functional Motor Scale) and some elements of the GMFM (Gross Motor Function Measure) scale is used to assess the degree of activity restriction of patients with SMA older than 5 years old [14,17].

The scale consists of 33 points, each of which has the following assessment options: 0 — no answer; 1 — partial answer; 2 — full answer. The ability to turn over independently, sit, hold your head, get up and lie down from a sitting position, crawl, stand, take steps, run and jump is evaluated.

The RULM scale (Revised Upper Limb Module) is used to assess the motor function of the upper extremities in children and adults with SMA, including the transition from a walking state to a non-walking one. Movements in the shoulder and elbow joints, wrist and hand are evaluated. The scale consists of 20 sections, and the maximum possible number of points is 37. Each point of the scale has the following assessment options:

Table

Tests Recommended in Different Functional Types of SMA

Scales	Non-sitters (type 1)	Sitters (type 2 / non-ambulant type 3)	Ambulant (type 3)
Motor development			
HINE	■		
Functional scales			
CHOP-INTEND	■		
HFMSE		■	■
RULM		■	■
MFM		■	
Measurement of endurance			
6MWT			■

0 — cannot perform; 1 — partially performs movement; 2 — movement in full. Improving the score by 2 points is considered to be a clinically significant result. The scale is designed specifically for SMA involving patients and caregivers and is easy to use. The scale indicators reflect conscious functional actions from everyday life that are of clinical significance for patients, their caregivers and clinicians. These are manipulations with objects (hold a cup, pencil, draw a line, press a button, open a plastic container), lifting and moving your hand to the side, holding the weight, etc. It allows you to monitor changes in motor function, including the transition from an outpatient state to a non-outpatient one [12,13].

The 6-Minute Walk Test assesses physical endurance based on the distance covered in 6 minutes in patients with type 2/3 of SMA. The age and height of children affect the control range [24]. The result of this test correlates with the results of the Hammersmith score.

Recently, it is recommended to use scales in accordance with the functional types of SMA (table) [4,18].

Depending on the level of functioning of the patient, an individual rehabilitation program is developed, and rehabilitation interventions and auxiliary tools are selected, the ICF intervention table is filled in, where need to indicate which interventions will be used and who will conduct these interventions [10]. A multidisciplinary team is formed. At the end of the rehabilitation cycle, which a child with SMA can have up to 6 months, a second assessment is carried out using standardized assessment scales [10,11,15].

In addition, the problems faced by the patient are identified, namely accessibility to medical and social services, gene therapy, inclusive training, provision of auxiliary means, barrier-free environment. It is very important to make political decisions at the level of society, region and state regarding the accessibility of a child with SMA to gene therapy, which prevents further disease progression and is an integral part of the health of a child with SMA [3,7]. In addition, access to such medical services as physical therapy, ergotherapy, auxiliary means (respiratory support devices, multifunctional strollers) is important [6,15,19].

Realizing the importance of the impact of environmental factors on the health status of a child with SMA, public parent organizations continue to take initiatives to local self-government bodies to reduce their negative impact on the rehabilitation potential of a child with SMA.

Taking into account all the components of the ICF in organizing the provision of assistance to children with SMA in Ukraine, several projects were initiated: creation of a working group on translation and linguistic adaptation into Ukrainian of the clinical protocol for assistance to children with SMA (Ministry of Health of Ukraine); creation of centers for neuromuscular diseases with a multidisciplinary team; support of patients by local communities for financial support for the purchase of gene therapy drugs; approval of the action plan «National strategy for creating a barrier-free space in Ukraine until 2030».

Conclusions

CF makes it possible to determine of all aspects of the life of a child with SMA and to determine the impact of environmental factors on its level of functioning.

It is thanks to the ICF that specialists and parents have the opportunity to take a comprehensive approach to assessing the health status, develop an individual rehabilitation program and improve the

system of medical care and social protection of a child with SMA together with local communities.

According to ICF principles it is necessary to use standardized tools to assess the functional activity of a child with SMA, the effectiveness of rehabilitation interventions. Different tools of assessment may be recommended for children depend on ages and functionalities.

No conflict of interests was declared by the authors.

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Стаття надійшла до редакції 15.11.2021 р., прийнята до друку 06.03.2022 р.