Issue Rehabil. Orthop. Neurophysiol. Sport Promot. 2021; 34: 7–21. ISSN 2300-0767
DOI: 10.19271/IRONS-000129-2021-34

ORIGINAL ARTICLE

SPINAL MUSCULAR ATROPHY – THE NEEDS OF CHILDREN UNDERGOING PHARMA COLOGICAL TREATMENT AND THEIR FAMILIES AS PART OF THE COMPREHENSIVE CARE SYSTEM IN POLAND -

RDZENIOWY ZANIK MIĘŚNI – POTRZEBY DZIECI LECZONYCH FARMAKOLOGICZNIE I ICH RODZIN W RAMACH SYSTEMU KOMPLEKSOWEJ OPIEKI W POLSCE

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ABSTRACT

Introduction

There is no coordinated care system for patients with rare diseases in Poland.

Aim

The aim of the study was to define the most important aspects of medical, social and professional care under the project titled "Education in a new reality: a comprehensive and long-term model of physiotherapeutic treatment in spinal muscular atrophy" developed by Stowarzyszenie Lwie Serca (Lion Hearts Association).

Material and methods

The study included 27 children with SMA1 and SMA2 treated pharmacologically. Motor development was checked using the CHOP INTEND and HFMSE scales; the body posture was measured with the Bunnell scoliometer and the Rippstein plurimeter, and the range of motion in the joints of the upper and lower limbs was measured. Respiratory and neurologopedic problems were assessed. The social, professional and financial situation of the families was analyzed. Psychological data was collected based on an interview with parents.

Results

Nineteen children were diagnosed with type 1 SMA and 8 with type 2. On the CHOP INTEND scale, the mean number of points was 30.8; in HFMSE 21.7. Disorders in the shape of the chest, spine, hip joints and reduced range of motion were found. 18 children (66.7%) used a ventilator in the invasive and non-invasive mode. Most children had symptoms of dysarthria and stomatognathic disorders. The main psychological problems among parents were severe stress, low mood, and a sense of loneliness.

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Authors reported no source of funding
Authors declared no conflict of interest

Date received: 22nd December 2020
Date accepted: 24th March 2021

Conclusion
In Poland, it is necessary to introduce a system of comprehensive, coordinated care for children with spinal muscular atrophy.

Keywords: spinal muscular atrophy, body posture, developmental scales

STRESZCZENIE
Wstęp
W Polsce brakuje systemu skoordynowanej opieki dla chorych z chorobami rzadkimi.

Cel
Celem pracy było określenie najważniejszych aspektów opieki medycznej, społecznej i zawodowej w ramach projektu „Edukacja w nowej rzeczywistości: kompleksowy i długofalowy model postępowania fizjoterapeutycznego w rdzeniowym zaniku mięśni" opracowanego przez Stowarzyszenie Lwie Serca.

Materiał i metoda
Badaniami objęto 27 dzieci z SMA1 i SMA2 leczonych farmakologicznie. Rozwój motoryczny sprawdzono przy użyciu skal CHOP INTEND i HFMSE; do pomiarów postawy ciała wykorzystano skoliometr Bunnella oraz plurimetr Rippsteina, wykonano pomiar zakresów ruchu w stawach kończyn górnych i dolnych. Oceniono problemy oddechowe i neurologopedyczne. Poddano analizie sytuację społeczną, zawodową i finansową rodzin. Dane psychologiczne zebrano na podstawie wywiadu z rodzicami.

Wyniki
U 19 dzieci rozpoznano typ 1 SMA, a u 8 typ 2. W skali CHOP INTEND średnia liczba punktów wynosiła 30.8; w HFMSE 21.7. Stwierdzono zaburzenia w ukształtowaniu klatki piersiowej, kręgosłupa, stawów biodrowych oraz ograniczenia zakresów ruchu. Z respiratora w trybie inwazyjnym i nieinwazyjnym korzystało 18 dzieci (66,7%). U większości dzieci stwierdzono objawy dyzartrii oraz zaburzenia stomatognatyczne. Główne problemy psychologiczne występujące wśród rodziców to silny stres, obniżenie nastroju, poczucie osamotnienia.

Wniosek
W Polsce konieczne jest wprowadzenie systemu kompleksowej, skoordynowanej opieki dla dzieci chorych na rdzeniowy zanik mięśni.

Słowa kluczowe: rdzeniowy zanik mięśni, postawa ciała, skale rozwojowe

Introduction and aim
International standards of care for individuals with spinal muscular atrophy (SMA) involve collaboration of specialists in many fields of medicine (Mercuri *et al.* 2018). This is due to the course of the disease and various symptoms such as muscle weakness (Iannacone *et al.* 2000; Kroksmark *et al.* 2001; Merlini *et al.* 2004; Febrer *et al.* 2010; Kaufman *et al.* 2012; Montes *et al.*, 2014; Wadman *et al.* 2018), respiratory disorders (Chng *et al.* 2003; Bach *et al.* 2012; Bach *et al.* 2017, Wijngaarde *et al.* 2020), chewing and swallowing disorders (van den Engel-Hoek *et al.* 2008; van den Engel-Hoek *et al.* 2009; Kruse *et al.* 2020), osteoarticular deformities (Rodillo *et al.* 1989; Robinson *et al.* 1995; Fujak *et al.* 2013;

Stępień *et al.* 2020), contractures (Wang *et al.* 2004; Fujak *et al.* 2010; Stępień *et al.* 2020) and the resulting limitations of functioning in everyday life.

There is no coordinated care system for patients with rare diseases in Poland, including persons with SMA. The Rare Diseases Plan, under development for several years, has not yet been implemented. People suffering from SMA and their families are forced to look for specialists on their own. This situation leads to delays in the treatment process and frustration for patients and their loved ones. Reaching specialists with appropriate competences requires a lot of time and money.

For two years, pharmacological treatment of patients with SMA has been reimbursed in Poland. The use of pharmacological treatment gives a chance to improve the functional state and quality of life of sick individuals, especially children. However, it is necessary to take into account the dynamics of individual developmental changes resulting from the introduced pharmacological treatment and to follow the guidelines of care. Worldwide recommendations for the care of patients with SMA include: stretching, positioning aimed at symmetrical loading and preventing deformations, breathing stimulation, strengthening exercises, and training of changes in position and gait (Mercuri *et al.* 2018). The interventions used should be individually tailored to the functional state of persons who cannot sit by themselves (NS – not sitters), sit (S – sitters) or can walk (W-walkers) (Mercuri *et al.* 2018).

The non-medical goal of treatment should be education for life in society and preparation for pursuing a profession, which is the basis of social and professional rehabilitation. The treatment process should also include taking care of the sick person's family. Unfortunately, in Poland, support for the families of sick individuals is very limited. For years, patient organizations have been striving for the possibility of support by assistants of the disabled, adapting educational institutions to the needs of children, respite care and psychological support.

The aim of this study was to define the most important aspects of medical, social and professional care that could be used to develop a system of comprehensive care for children with SMA treated pharmacologically and their families.

The above goal was implemented under the "Education in the new reality: a comprehensive and long-term model of physiotherapeutic treatment in spinal muscular atrophy" project developed by the Lion Hearts Association, financed by the Civic Initiatives Fund for 2014–2020. The study was approved by the Senate Bioethics Committee at Józef Piłsudski University of Physical Education (SKE 01-28/2019).

Materials and methods

Participants

In accordance with the assumptions of the project, 27 children with genetically confirmed spinal muscular atrophy and their families were qualified for the study. The condition for inclusion in the project was a diagnosis of SMA1 and SMA2, confirmed by genetic testing before pharmacological treatment. Families participated in the project without incurring the costs of visits, accommodation and meals. The only expense was the cost of commuting, which was lower than the standard fares for separate journeys to 5 specialists invited to participate in the study. All parents were informed about the purpose of the study.

Methods

The project assumed two meetings of a child and the child's parents with three physiotherapists, a neurologopedist and a psychologist at a 10–12 month interval in three cities in Poland. During the first meeting, therapeutic goals and the procedures were defined. During the second examination, the effects of the therapeutic procedure were assessed and further goals were set. The meetings were attended by physiotherapists responsible for the child's daily therapy. Participation in the meetings was also made possible for neurologopedists and special educators

willing to collaborate. Rehabilitation management specialists also participated in the project.

Before the study, parents of the children completed a questionnaire with questions about anthropological, demographic, health and social data. Psychological data was collected based on an interview with parents. Additionally, participants' medical records were analyzed, including x-rays of the spine and hip joints.

Motor development was assessed using the CHOP INTEND scale and the Hammersmith Functional Motor Scale – Expanded (HFMSE) applied in studies of persons with SMA (Mercuri *et al.*, 2006; Glanzman *et al.*, 2011). The CHOP INTEND scale was used in children with SMA 1, and the HFMSE scale for SMA 2.

During the examination, body posture was assessed and the range of motion in the joints of the upper and lower limbs was measured. The body posture measurements were performed with the use of the Bunnell's scoliometer and the Rippstein plurimeter, which were used in previous studies of children with SMA (Stępień *et al.*, 2019; Stępień *et al.*, 2020).

To assess the function of the respiratory system, measurements of the chest volume during the inhalation and exhalation phase at three levels of the chest were used. The respiratory tract, cough strength and voice volume were also tested on a four-point scale developed by one of the authors of the project.

The neurological evaluation included an analysis of the stomatognathic system (structure and functions of the structures of the oral cavity and the facial skeleton) (Milewski *et al.* 2017) and the functions of the hearing and speech organs.

Psychological data was collected based on an interview with parents. Meetings with the psychologist initially took place in small groups of mothers and fathers. After a year, the sessions were conducted with the psychologist alone, because parents reported the need for an individual conversation, they wanted to receive psychological support in areas they were embarrassed to talk about in front of other session participants.

The collected information was classified into the following categories:

1. Medical condition: health condition, specialist care, pharmacological treatment, functional status, musculoskeletal system, breathing, speech, swallowing, mental status, nutrition, orthopedic supplies.
2. Social, professional and financial situation of families.

Results

The study involved 27 children, including 11 girls and 16 boys (age 5.3 ± 3.6, weight 15.6 ± 8.7, height 104.1 ± 22.0) with spinal muscular atrophy. Additionally, 51 parents (mothers aged 36.5 ± 4.8, fathers aged 36.9 ± 4.2) participated in the activities under the project. The families came from different regions of Poland.

Health condition

Nineteen children were diagnosed with type 1 SMA and 8 with type 2. The first symptoms of the disease appeared in a given group on average at the age of 5.1 ± 4.5 months. The disease was diagnosed in the study group at the mean age of 8.8 ± 6.7 months, the earliest diagnosis was made after two weeks of life, and at the latest at 22 months of age. Only in the case of one child physiotherapy in the first year of life had not been implemented.

Eighteen participants were treated with Spinraza, 4 with Risdipam, 3 were treated with Branaplam, one participant had Spinraza replaced with Risdiplam, and one child was treated with Spinraza after genetic treatment. In the period from the introduction of pharmacological treatment, 40.7% of parents reported improvement in respiratory functions (breathing, coughing, blowing the nose), 33.3% reported improvement in gross motor skills (e.g. head movements, turning, sitting, moving in a lying or sitting position), 40.7% of children improved their upper limb movements, and 20.0% of their lower limb movements, 36% of parents reported improved appetite and weight gain and postural control, 12% reported improved swallowing, better sleep quality and less fatigue, and 8% better pass motion.

19 patients (70.4%) had subluxation/dislocation of one or both hip joints. Also in 19 individuals (70.4%) scoliosis was diagnosed. In the last year, 16 participants (59.3%) had 1–4 respiratory infections requiring treatment. Two children had anti-reflux treatment in the past.

Specialist care

The children were looked after by doctors of various specialties, as well as physiotherapists, neurologopedists and orthotic specialists (Table 1). On average, children attended 15.5 ± 21.4 visits to specialist doctors a year. Six families (23.1%) considered access to specialist doctors as easy, 13 (50%) as difficult, and the remaining families perceived access to specialists as varied. All of the children benefited from free physical therapy, which was provided by the National Health Fund (66.7%), a kindergarten or school (14.8%), as part of respite care and home ventilation (14.8%) or in some other form. Additionally, all children participated in non-reimbursed physiotherapy. The mean duration of physiotherapy per week in the study group, both under reimbursed and private care, was 5.3 ± 1.7 hours. 40.7% of parents found the physiotherapists taking care of their children as excellent, 48.1% as good, others had different experiences. 92.6% of parents believed that physiotherapists should get additional training with specialists who have experience in working with patients with SMA.

Functional status

13 of the respondents could not sit independently (NS), 13 were able to sit without support (S), and one girl walked alone.

17 children were examined using the CHOP INTEND scale. The average number of points was 30.8 (2–56), while the maximum score in the scale was 64 (each of the 16 items could be scored from 0–4 points). This is on average 48.1% of the maximum abilities, and in the case of the most fit child – 87.5%.

10 children (8 with SMA2 and two with SMA1) were tested using the HFMSE scale. Two children, despite the diagnosis of SMA1, were also tested using the HFMSE scale due to very good motor development as a result of early treatment (one child walking, the other sitting and crawling). The average number of points was 21.7 (4–54) out of 66 points (33 items rated on a scale of 0–2). Thus, the examined children obtained an average of 32.9% of the maximum score, and the most fit child – 81.8%.

None of the examined children obtained the maximum number of points, which indicates the need for physiotherapy aimed at improving the functional status in the entire study group.

Musculoskeletal system

Most of the respondents showed disorders in the shape of the chest, spine, hip joints, and limited range of motion in the hip joints. Detailed information on individual body structures is provided in Table 2.

A total of 14 individuals (51.9%) showed reduced range of motion in the shoulder, elbow or wrist joints, and 22 (81.5%) individuals showed reduced range of motion in the hip, knee or ankle joints.

All participants of the study required the implementation of appropriate physiotherapeutic interventions due to disorders of the musculoskeletal system.

Breathing

10 SMA1 patients and 1 SMA2 patient were supported by NIV (Non-invasive ventilation) in the BiPAP (biphasic positive airway pressure) mode and 7 children were supported by invasive ventilation in the PSV (pressure support ventilation) and PCV (pressure controlled ventilation) modes. In total, 18 children (66.7%) used a ventilator in the invasive and non-invasive mode.

The average time of using the ventilator in the group of 7 children with tracheostomy was 22.9 hours/day (6 children used the ventilator 24 hours/day, 1 child used the ventilator for 16 hours/day). The average time of using a ventilator in a group of 11 children supported by NIV ventilation in BiPAP mode was 11.6 hours/day. Of these, 10 children were connected to the ventilation for 12 hours/day, and 1 child for 6 hours/day.

Table 1. Number of children using the care of individual specialists.

Specialist	N (%)
Physiotherapist	27 (100%)
Orthotic specialist	24 (88.9%)
Neurologist	23 (85.2%)
Pediatrician	22 (81.5%)
Orthopaedist	21(77.8%)
Neurologist	18 (66.7%)
Anesthesiologist, rehabilitation doctor	17 (63.0%)
Dentist	14 (51.8%)
Ophthalmologist	10 (37.0%)
Gastroenterologist	8 (29.6%)
Cardiologist	7 (25.9%)
Psychologist	6 (22.2%)
Dietician	5 (18.5%)
Laryngologist/otolaryngologist	4 (14.8%)
Nephrologist, pulmonologist, surgeon	2 (7.4%)
Allergist, orthodontist, dermatologist, SI therapist	1(3.7%)
Other	1 (3.7%)

Note: N – number of participants using specialist care; % – percentage of the study group.

Table 2. Disorders of selected body structures and functions in the SMA group.

Disorders of body structures and functions	N	%
Motor development disorders according to CHOP INTEND	17	100%
Motor development disorders according to HFMSE	10	100%
Oblique position of the pelvis (above 2°)	23	85.2%
Increased torso rotation angle (above 3°)	22	81.5%
Abnormal shape of the chest (asymmetry greater than 3°)	18	66.7%
Reduced ranges of cervical rotation	17	63.0%
Reduced ranges of flexion in the shoulder joints	11	40.7%
Reduced ranges of extension of the elbows	9	33.3%
Reduced range of motion in the wrist joints	12	44.4%
Reduced range of extension in the hip joints	17	63.0%
Reduced range of extension in the knee joints	20	74.1%
Reduced extension in the ankle joint	8	29.6%
Reduced mobility of the chest during breathing	22	81.5%
Reduced volume of voice	17	63.0%
Ineffective cough	20	74.1%
Dysarthria	24	88.9%
Swallowing disorders	14	51.9%
Stomatognathic disorders	20	74.1%
Abnormal pressure in the ear	13	48.1%
Scoliosis	19	70.4%
Hip subluxation	19	70.4%

Note: N – numer of participants with selected impairments or activity limitations; % – percentage of the study group.

77.8% of parents used an ambu bag in their daily breathing training. 60.4% of parents used an aspirator during the daily care of a child.

Only 5 (18.5%) participants used the thoracic breathing path, while 8 (29.6%) were able to breathe only through the abdominal path. Chest circumference measurements showed that as many as 22 (81.5%) children had limited chest mobility. A 1.5 cm difference between the inhalation and exhalation circumference was assumed as the norm. Only one boy (3.7%) had effective cough.

Speech

Most of the children had symptoms of dysarthria and stomatognathic disorders (Table 2). 11 children (40.7%) had ankyloglossia in the

form of a shortened sublingual frenulum, and 6 (22.2%) had a shortened frenulum under the upper lip (frenulum labii superioris).

Nearly half of the examined children had problems with abnormal pressure in the middle ear (Table 2), 5 children had obstruction in one ear canal, and 2 children had obstruction of two canals, which may result in conductive hearing loss. Only 10 children (37.0%) had normal voice volume, while 7 participants (25.9%) had a significantly reduced voice volume.

Swallowing

85.2% could swallow, but overall more than half of the participants had dysphagia (Table 2). 70.4% were able to consume solid foods. 14.8% of the children were fed with a tube or PEG (gastrostomy). 37.0% reported the occurrence of bringing up food, 1 child choked. The suckling reflex did not occur in the past in 6 children (22.2%).

Mental state of parents

First group consultations, then individual consultations highlighted many problems faced by project participants on a daily basis. The most outstanding issues reported by parents included: severe stress, low mood after receiving the child's diagnosis, feeling of loneliness among the closest family and friends, emerging and increasing conflicts between spouses over time, social withdrawal. Parents reported a strong focus on the sick child at the expense of other family members and a decline in taking care of their own needs. They complained of chronic mental and physical fatigue, neuralgia appearing more and more frequently, and a lack of psychological support.

In the interview, 26 families noticed the need for psychological support. 7 families (27%) participating in the project systematically use psychological advice in their place of residence.

Orthopedic supplies

All participants used rehabilitation equipment. 18 children (66.7%) were verticalized, 18 (66.7%) were wearing a corset, 17 (63%) were recommended orthoses, and 5 children (18.5%) used a stabilization belt. In addition, parents invested in home furnishings tailored to the needs of children, equipment for breathing support and airway clearance, communication systems, and ramps.

Social, professional and financial situation of families

26 families participated in the project. Two children qualified for the study came from one family. Eight families raised only one child. In 13 families there were two children, and in the remaining ones parents raised three, four (1 family) or five children (1 family). 8 children attended school, 6 went to kindergarten, and the remaining 13, due to their age, stayed at home.

92.0% of mothers and 62.5% of fathers had completed higher education, 8.0% of mothers and 29.2% of fathers had secondary education, while 8.3% of fathers were less qualified.

Physical support for families (*care*)

Nine families (34.6%) did not have any support in childcare. The remaining 17 families benefited from the help of various individuals in caring for the child. One family indicated that the help was sporadic, 8 families benefited from the help of family members, 1 family asked for help from friends, in the case of 3 families, nurses were involved, 3 families used the help of a nanny, and 1 family was supported by an employee of a public institution. Among the families benefiting from support, the frequency of received assistance varied – from incidental to daily assistance.

Financial support

Among 26 families, 96.1% benefited from financial support due to the child's disability. 11 families did not receive a care allowance due to resignation from work, in the remaining families a mother received an allowance (53.8%). 23.1% of families financed their own child care. 51.8% of the surveyed families had problems with obtaining funds for rehabilitation.

12 families were familiar with support programs for disabled individuals and their families, 14 did not have such knowledge. Information on possible family support was obtained through: the community of individuals with disabilities (92.3%), the Internet (69.2%), institutions (19.2%) or the press (3.8%).

46.1% of families benefited from programs implemented in the gmina (commune). These programs included: early development support, specialist care services and respite care.

The costs of treating children for the parents were high (Table 3). In total, the average monthly fees amounted to PLN 3304.7 net, which was approximately 87% of the expected net national average in 2020 (approximately PLN 3800).

Table 3. Average costs of treatment and rehabilitation of children with SMA in Poland.

Expenses	**Annually**	**Monthly**
Specialist consultations (without physiotherapy)	1355.4	112.9
Physiotherapeutic consultations	15442.3	1286.9
Travel costs to specialists	1465.4	122.1
Costs of commuting to the hospital (pharmacological treatment)	3280.8	273.4
Orthopedic supplies	16756.4	1396.4
Travel costs for orthopedic supplies	1356.1	113.0
Total costs	**39656.4**	**3304.7**

Discussion

The literature describes the symptoms of patients with spinal muscular atrophy very thoroughly, but much less is known about the course of the disease in children treated pharmacologically. Information collected from parents confirmed that children with SMA treated with pharmacological treatment require multidisciplinary care, in which doctors specializing in various fields of medicine, physiotherapists, speech therapists and neurologists, nutritionists, orthotic specialists and psychologists are involved.

The study group comprised children of various age, functional status and pharmacological treatment used. The varied age of children and their motor abilities required differentiated assessment in the form of one of two validated scales: CHOP INTEND and HFMSE (Hammersmith Functional Motor Scale – Expanded) (Mercuri *et al.*, 2006; Glanzman *et al.*, 2011).

The CHOP INTEND test, designed for weak infants, takes into account both the features of motor development taken from other scales and those developed by the authors of the test. All motor features included in CHOP INTEND were tested in a group of SMA1 patients (Glanzman *et al.*, 2011). Having a good research tool at their disposal, subsequent authors analyzed the results of the CHOP INTEND scale in 122 patients with SMA 1 (Pane *et al.*, 2018). In their research, they showed that CHOP INTEND scores in patients over 2 years of age were generally lower than those in younger children. Their results also confirm that after the age of 2, patients with an early onset of the disease and those with the most common phenotype (1.1/A and 1.5/B) who survive, generally have tracheostomy and gastrostomy, and very low CHOP scores. Greater variability is found in those with the mildest phenotype who previously showed head control.

The extended HFMSE scale is designed and widely used for patients with SMA 2 and 3. The items tested on the scale were carefully selected by clinicians after careful observation and evaluation of many SMA patients (Mercuri *et al.* 2006). The HFMSE assessment elements have proven to be extremely useful in clinical practice as a rehabilitation assessment tool as well as in clinical trials to determine disease progression (Kaufmann *et al.* 2012; Mercuri *et al.* 2016). One of the challenges of using the scale is that patients with SMA constitute a clinically very heterogeneous group; and even if we limit ourselves to the type 2 and 3 phenotypes whose functional domains are covered by HFMSE, the clinical

picture still varies from seated patients to patients who are able to complete almost all of the 33 items on the scale.

Mazzone *et al.* compared two scales, HFMSE and MFM, also intended for patients with SMA 1 and 2. They showed that HFMSE is more targeted at stronger SMA patients, while MFM is more sensitive to detecting changes in function in weaker patients, taking into account axial and upper limb function. However, as the authors claim, the choice of the scale should depend on the inclusion criteria and the expected changes (Mazzone *et al.*, 2014).

In our own examinations, the CHOP INTEND scale for SMA 1 was selected to assess motor development, and the HFMSE scale for SMA 2. In our own examinations, 27 patients were analyzed, including 19 with SMA1 and 8 with SMA2. Patients with SMA 3 did not take part in the examinations. 17 children were examined using the CHOP INTEND scale, with the mean number of points being 30.8 (2–56). Two children, despite the diagnosis of SMA1, were examined with the HFMSE scale due to very good motor development as a result of early treatment (one child walking, the other sitting and crawling).

10 children were examined using the HFMSE scale, with the average number of points 21.7 out of 66 possible. As previously shown by Mercuri *et al.* patients with spinal muscular atrophy are heterogeneous groups, and this was noticed both in the assessment of motor development performed on the CHOP INTEND and in the HFMSE scales (Mercuri *et al.* 2012).

An analysis using functional scales showed that all children in the group studied by us require physical therapy aimed at improving their functional state. Due to the varied functional status, the treatment process should include interventions tailored to the needs of individual participants.

Musculoskeletal system

The examination of the musculoskeletal system showed that all examined children require medical care due to the deformities and the reduced range of motion. Over 80% of children showed reduced range of motion in the joints of the lower limbs, and over 50% in the joints of the lower limbs. Contractures in the knee and hip joints were the most common. In addition, the majority of children showed reduced rotation of the neck. In the studies conducted so far, many authors have pointed out contractures in the upper and lower extremities (Fujak *et al.* 2010; Wang *et al.* 2004) in patients with SMA. It has also been shown that the ranges of neck rotation in children with SMA not treated pharmacologically are smaller than in healthy children and increase with age (Stępień *et al.* 2020). Due to the prevalence of contractures in the SMA population, the care guidelines for individuals with SMA include stretching exercises that should be performed regularly (Mercuri *et al.*, 2108). It is also advisable to use verticalization, which can reduce the risk of contractures in the hip joints (Mercuri *et al.*, 2018). Counteracting contractures is also important due to the relationship shown in research between the range of motion in the joints and functional capabilities (Salazar *et al.* 2018).

In the study group, more than 65% of children experienced scoliosis, deformity of the chest or abnormal position of the pelvis. In the past, there have been numerous reports of spinal and thoracic deformities in pharmacologically untreated children with SMA (Rodilo *et al.* 1989; Robinson *et al.* 1995; Fujak *et al.* 2013; Stępień *et al.* 2020). The results of our research indicate that despite the introduced pharmacological treatment, measures should be taken to counteract the emergence and increase of spine and chest deformities in children with SMA.

The obtained results confirmed the opinions of other authors that children with SMA often have abnormal hip joints (Rodillo *et al.* 1989; Robinson *et al.* 1995; Fujak *et al.* 2013; Stępień *et al.* 2020). Hip dislocation or subluxation in more than 70% of respondents is still a challenge for doctors and physiotherapists.

Breathing

Respiratory failure causes mortality in spinal muscular atrophy types 1 and 2. The result of the weakness of the respiratory muscles is ineffective cough and reduced ability to clear the lower respiratory secretions, underdevelopment of the lungs and the chest wall, and respiratory depression (Hull *et al.* 2012, Wijngaarde *et al.* 2020). Respiratory care for patients with spinal muscular atrophy is essential for their survival and quality of life (Wang *et al.* 2007; Hull *et al.* 2012; Mercuri *et al.* 2018).

Most of the studied children, despite pharmacological treatment, showed respiratory disorders and used ventilation, the effectiveness of which is emphasized in the literature (Bach *et al.*, 2017). Over 80% of participants had chest mobility disorders, which may be related to the breathing pattern and the characteristic shape of the chest (LoMauro *et al.* 2018). Our results confirm the necessity of using respiratory physiotherapy in children with SMA1 and SMA2 treated pharmacologically.

Speech and swallowing disorders

Most of the children studied in the project had dysarthria. Dysarthria is defined as a symptom or a group of disorders in the implementation of the process of speaking on a neurological basis. Pathological changes characteristic of dysarthria include speech implementation, and in particular they distort articulation, may cause pathological changes in the value of the oral and nasal sounds, cause changes in intonation, accent, and speech rate, which are often accompanied by shortened phonation time. In individuals with dysarthria, there may be respiratory changes (shortness of breath, loud inhalation, exhalation) and voice changes, e.g. voice volume (Gatkowska 2012; Mirecka 2012).

The literature deals with swallowing disorders (dysphagia) occurring in individuals with SMA (van den Engel-Hoek *et al.* 2008; van den Engel-Hoek *et al.* 2009; Kruse *et al.* 2020). Aspiration is a dangerous symptom of swallowing disorders. This means that food/fluid enters the larynx below the vocal folds and into the airways, which can cause pneumonia. Dysphagia usually accompanies dysarthric speech disorders, and properly planned therapy allows to alleviate the symptoms of both swallowing and speech disorders. Muscle weakness in the mouth, throat and esophagus in SMA makes swallowing difficult. One of the potential causes of dysphagia may be inappropriate head control (van den Engel-Hoek *et al.*, 2008). Swallowing speech disorders diagnosed in our group of children confirm the need to implement multidisciplinary care in this area.

The speech and swallowing disorders observed in the participants of the study indicate the need for them to be looked after by doctors of various specialties, neurologopedists and physiotherapists.

Social, professional and financial situation of families

The situation of families, where one of the members faces disability, is complicated and often requires social support. Social support for the family of a disabled child is related to various areas of family life and concerns four basic systems: economic support system, service system, social support system and emotional support system (Radachoński, 1991).

Meetings with parents and guardians of children suffering from SMA gave an insight into the mental and social functioning of this specific group. Parents reported severe stress and depressed mood. The stress experienced by the parents was caused primarily by the constant anxiety about the child's condition. The smallest signs of illness, such as temperature or coughing, resulted in immediate involvement of parents in providing help. Such activity was very often associated with long journeys, waiting in hospitals and a lack of understanding on the part of the medical staff. The caregivers talked about social withdrawal, chronic mental and physical fatigue, emerging pain ailments.

Other very frequently reported problems were: growing conflicts in marriage and lack of ties, slow emotional distancing, increasing gaps in spending free time together, defective division of marital duties, and the disappearance of intimate life. Spouses reporting a crisis very often mentioned a completely different vision of living in a family with a sick child from that represented by the "other half".

The meetings with parents showed how important psychological support is in the lives of families with children suffering from SMA. Talking to a psychologist can be considered extremely important due to the fact that the expression of suppressed emotions can have a therapeutic effect. Unexpressed emotions often contribute to physical symptoms such as chronic headaches, stomach discomfort, muscle tension or high blood pressure. Expression of accumulated pain and other suppressed emotions usually brings tremendous physical and emotional relief (Schneider *et al.* 2002).

Psychological and emotional problems may undoubtedly be related to the difficult financial situation of families and the limited possibilities of their own development. In the families participating in the project, the mother more often resigned from work to look after the child. It is worth noting that most of the mothers had higher education. The reason why the mother quit her job to care for the child has not been investigated. On the basis of literature and research on the situation of women taking care of children with disabilities, the following reasons are determined: conventional issues (one person secures the family financially, the other one looks after the home), the cultural aspect, the emotional needs of parents, and the amount of income (a person earning more remains professionally active) (Komorowska 2015).

Caregivers of children with SMA confirmed that the cost of providing for a child is high. In 2018, the cost of raising one healthy child up to the age of 18 was approx. PLN 190–210 thousand, while two children PLN 350–385 thousand. In the case of families diagnosed with SMA, the costs resulting from the special needs of children with SMA, such as visits to specialists, rehabilitation, orthopedic supplies, aids and medical supplies, should be added. At the same time, it should be noted that apart from financial costs, the family also incurs time costs related to travel to centers where children with SMA are treated.

Collected data on the cost of caring for a child with SMA showed that families spend over 80% of the average salary on providing adequate care.

96.1% of the families benefited from financial assistance offered by the state, but nearly half of them (46.2%) admitted that they did not understand the financial support system. This situation may create a risk that families do not make use of all the financial support available to them. Institutions that are responsible under Polish law for providing financial aid to disabled individuals and their carers were indicated as a source of information by only 19.2% of families. Over 50% of the surveyed families had problems with finding funds for medical care for their children. The above data indicate that in Poland the costs of care are largely shifted to families with a disability.

In Poland, in recent years, the government has launched programs to support families in caring for a disabled child. For many reasons, including financial ones, the programs, in the opinion of parents, are not implemented in every commune. Only 46.2% of the surveyed families used the programs financed by the local government.

In 2019, the respite care program was launched. In 2020, as part of the Solidarity Fund, Poland allocated PLN 30 million to the respite care program for family members or carers of individuals with disabilities. Unfortunately, the program is not implemented in every commune (it requires 20% of its own contribution from the commune). According to the information obtained from families, only 4 of them had the opportunity to benefit from support under this program.

Due to the inability to use systemic support, some families do not have any support in caring for their children, some of them use the

help of the closest family, and the rest finance the assistance from their own resources.

Providing greater social support to the family as part of caring for a disabled person and increasing the possibility of using free services as part of rehabilitation, pedagogical therapies and psychological support would significantly improve the condition of the patient and his/her relatives. Providing such a model would allow patients with SMA and their caregivers to fulfill social roles comparable to those of families who do not struggle with the disease on a daily basis.

There are some limitations to this study. Part of the data was obtained on the basis of a questionnaire that gave parents freedom of expression. However, taking into account the multifaceted nature of the project, it can be concluded that the obtained results provide valuable information about the medical condition of children with SMA receiving pharmacological treatment and about the social, professional and economic situation of families raising a disabled child.

Conclusions

1. In Poland, it is necessary to introduce a system of comprehensive, coordinated care for children with spinal muscular atrophy. The care system should include procedures adapted to the medical, social and professional situation of patients and their families.
2. The rehabilitation process, taking into account the care of doctors, physiotherapists, speech therapists, psychologists, orthotics and other specialists, should be based on a detailed examination and adapted to the functional state of the child and his/her age.
3. It is important to increase the economic aid from the state in financing medical care and orthopedic supplies for children with SMA and psychological support for families.
4. The care system for children with SMA and their families should enable parents to start working and achieve their personal goals.

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