Nursing care for patients with rare diseases on the example of patients with Alternating Hemiplegia of Childhood

Opieka pielęgniarska nad pacjentami z chorobami rzadkimi na przykładzie chorych z Naprzemienną Hemiplegią Dziecięcą

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Abstract

Introduction: Rare diseases are those which occur in 1 person in 2000. Usually they are genetically conditioned. They also comprise of diseases which appear as a result of metabolic, immunological defects, infections and uncommon cancers. Development of science, especially genetics prompted discovery of new gene's mutations. One of them is a mutation of ATP1A3 which is responsible among others for alternating hemiplegia of childhood. It is a disease whose manifestations appear in early childhood and are unpredictable. Strokes can be hemiplegic or tetraplegic. Sometimes they are accompanied by problems with swallowing and breathing.
The aim of the thesis was to compare 2 different cases of patients suffering from AHC, to show problems connected with taking care of them in bio-psycho-socio-spiritual dimensions and to present a model of care on the example of Dorothea Orem's theory.

Materials and methods: The comparative method used in the work compares two cases of patients. Techniques of interview, observation, measurement and documentation analysis were also applied. An interview questionnaire containing 13 questions and an observation conducted in everyday circumstances made it possible to show care problems of patients with AHC. The research was carried out from March 2018 to May 2018. Two cases were juxtaposed: a 10-year-old girl and a 34-year-old man.

Results and conclusions: Patients who suffer from rare diseases have restricted access to diagnosis, cure, rehabilitation and social assistance. They often meet with lack of understanding on the part of society and medical staff who do not have enough knowledge about this issue. By comparing 2 cases it was proved that care problems of these patients are very complicated and vary depending on age, type of stroke, intensity, duration of the disease and family's and society's support. Dorothea Orem's care system seems to be the best one.

Keywords: alternating hemiplegia of childhood, rare diseases, nursing care

Introduction

The presented work was written with a view to raising awareness and sensitising the reader to the problems of people with rare diseases. The advancement of medicine causes the emergence of new medical equipment, medicines and methods of treatment and contributes to the discovery and naming of new syndromes, especially of genetic origin. Such a disease is alternating hemiplegia of childhood, in which there are still many unknowns. Understanding the symptoms of the disease, the mutation of the ATP1A3 gene responsible for this disease and the implementation of the drug Flunarizine seems to be just the beginning to ensure the best quality of life for these patients.

The topic was raised because there have been no publications so far about the rare disease of alternating hemiplegia of childhood in the field of nursing. At the Pomeranian Medical University, the doctoral thesis was defended entitled *Wybrane choroby rzadkie w literaturze medycznej i programach kształcenia przed- i podyplomowego oraz opieka nad chorymi na choroby rzadkie w krajach wysoko uprzemysłowionych. Historia, teraźniejszość i perspektywy* (Selected rare diseases in medical literature and pre- and post-graduate education programmes as well as care for rare diseases in highly industrialised countries. History, present and perspectives) [1]. The interview conducted during own research shows that a master's thesis on physiotherapy at AHC was also written, but due to waiting for the publication of its results, access to it is impossible.

This study compares the cases of two different patients suffering from alternating hemiplegia of childhood, belonging to

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the group of rare diseases. On the basis of the research, nursing diagnoses regarding the bio-psycho-socio-spiritual sphere were made and a care system in accordance with Dorothea Orem's theory was applied.

The object and purpose of the research is to compare two different cases of patients suffering from a rare disease, which is alternating hemiplegia of childhood, to determine their issues in terms of nursing in the bio-psycho-socio-spiritual sphere and to choose a care system according to Dorothea Orem in the care of these patients. To achieve the goals set, dependent and independent variables were defined. The dependent variable is the scope/model of nursing care, the nursing care deficit, and independent variables are the patient's age, his/her condition in the bio-psycho-socio-spiritual sphere, physical fitness during seizures, socio-living conditions (whether he/she lives at home or in the Health Care and Curative Institution), family/community support.

Materials and methods

The case study and comparative methods were used in the study. Techniques of interview, observation, measurement and documentation analysis were also applied. The research tools were: an original interview questionnaire, observation sheet, measurement sheet and documentation (discharge reports, results of medical consultations and medical records of illnesses, results of laboratory and imaging examinations, psychological and pedagogical opinions, health certificates, school certificates). The interview questionnaire consisted of 13 questions and personal data. The measurement sheet included direct measurements (height, body weight, temperature, heart rate, blood pressure, breaths) and indirect (BMI) measurements. The observation sheet contained data identifying the patient through name, date, duration and place of observation, and description of nursing observations. The author conducted an interview with the president of the Polish Association for Persons with AHC and participated in the EUROPLAN III conference – Poland for Rare Diseases, which took place on 14–15 December 2017 in Warsaw.

Organisation and conduct of research

In Poland there are about 20 patients with alternating hemiplegia of childhood associated in the Polish Association for Persons with AHC. According to information from the president of said association, some patients who have clinical symptoms corresponding to AHC (or their legal guardians) have not agreed to a genetic test. Not everyone suffering from this disorder entered the ranks of the Association. Unfortunately, there is no reliable source of information on how many people in Poland suffer from alternating hemiplegia of childhood.

The study was conducted by analysing cases of two patients suffering from alternating hemiplegia of childhood. These persons and their guardians agreed to the survey in written and oral form.

Case No. I: girl, 10 years old, residing in the Silesian Voivodeship; case No. II: male, 34 years old, residing in the Lesser Poland Voivodeship.

The research was conducted from March 2018 to May 2018 in the domestic environment of patients with alternating hemiplegia of childhood. Interviews were collected from the mother and father of the girl, and in the case No. II from the patient and his mother. An analysis of documentation (discharge reports, results of medical consultations and medical records of illnesses, results of laboratory and imaging examinations, psychological and pedagogical opinions, health certificates, school certificates) was carried out, which allowed determining the current course of the diagnosis and treatment process until receiving the correct diagnosis. Direct and indirect measurements were also carried out, the results of which were within the norm. Observation of patients allowed for the assessment of their bio-psycho-social status, verification with previously conducted interviews and establishing nursing diagnoses.

The activities carried out were aimed at forming complete knowledge and materials needed to obtain nursing diagnoses and comparing both cases of disorder.

The patients participating in the research differed in: age (10 and 34 years), sex (female, male), origin (city, village), education (primary, secondary), mental fitness (light impairment, no mental disability).

They were linked by meeting the clinical criteria and due to the result of a genetic test confirming the mutation of the ATP1A3 gene of 13 May 2015 performed in the laboratory in Göttingen, which confirmed the diagnosis of alternating hemiplegia of childhood.

Results

The conducted research allowed for establishing common care problems. These are:

Nursing issue No. 1

Danger of injury to the osteoarticular and neuromuscular system during an attack of hemiplegia and/or tetraplegia

Purpose of care: prevention of injuries, possibly reduction of their consequences

Nursing interventions:

1. Ensuring safety by taking the patient by the arm during hemiplegia to prevent any fall or positioning during tetraplegia, preventing breathing and swallowing difficulties.
2. Taking the wheelchair when leaving the house, apartment.
3. Talking with the family about the patient's limitations and the scope of support necessary for him/her.
4. Adapting the bed to the needs of the patient, eliminating protruding, dangerous elements.

5. Accompanying the patient with presence and interest.
6. Providing the right footwear (avoiding footwear that is too loose and tight, footwear with a rubber tread that prevents slipping is recommended).
7. Recommending the use of safeguards for angular elements in the apartment (table, cabinets).
8. If possible, removing floor mats and rugs from the patient's surroundings, or securing them with anti-slip underlays.
9. Encouraging the patient to use the help of another person.
10. Informing the family about proper behaviour during a possible fall. One should not shout or show non-verbal fear at that moment, as this may increase the stress on the suffering person.
11. Recommendation to fix the handles in the bathroom and toilet, which the patient will be able to hold, as well as to provide non-slip mats in the bathtub and shower. A special stool can be placed there.

Nursing issue No. 2

Swallowing and breathing difficulties during an attack of hemiplegia and/or tetraplegia

Purpose of care: prevention of aspiration, opening of airway, ensuring safety

Nursing interventions:

1. Observation of the patient and accompanying him/her.
2. In case of alarming symptoms (tachycardia, tachypnoe, cyanosis), calling a doctor, resuscitation team, emergency medical team or perform resuscitation actions.
3. Positioning the patient in a half-high or high position with a slightly bent neck to prevent aspiration of saliva into the respiratory tract.
4. Providing fresh air.
5. Providing aid in eating meals between attacks.
6. Implementing a rich and energetic diet.

Nursing issue No. 3

Muscle pain during a spastic attack

Purpose of care: alleviation of pain

Nursing interventions:

1. Monitoring the nature, location and intensity of pain (on a VAS scale from 1 to 10).
2. Observing verbal and non-verbal (e.g. facial expressions) signs of pain as well as factors strengthening and alleviating pain.
3. Increasing the pain threshold by providing massage and changing position.
4. Administering painkillers according to the individual medical order sheet.
5. Assessing the effectiveness of analgesic treatment and modifying it in the absence of desired effects.
6. Using passive (abduction, adduction, limb stretching, rotation, straightening, bending) or active exercises depending on the patient's abilities.
7. Placing in a comfortable position with the use of facilities (wedge pillows, pneumatic pillows, sponge foundations, feather pillows, pillows filled with silicone, foamed polystyrene, gel of various shapes).
8. Combining performed procedures.
9. Ensuring comfortable underwear that does not restrict movement.
10. Assisting in carrying out everyday activities.
11. Ensuring comfort and safety of the surroundings.
12. Eliminating environmental factors affecting the increase of pain sensations such as: loud conversations, telephone, etc.

Nursing issue No. 4

Fatigue due to prolonged attack

Purpose of care: enabling rest and regeneration

Nursing interventions:

1. Observing the patient and monitoring his/her vital signs (temperature, blood pressure, pulse, saturation).
2. Documenting measurements.
3. Informing about the necessity to report alarming symptoms.
4. Creating favourable conditions for rest (silence, peace, room ventilation).
5. Combining procedures – performing them efficiently, accurately and gently.
6. Assisting in undertaking everyday activities.

Nursing issue No. 5

Deficit in coping with the demands of everyday life (food, toilet, defecation, independent movement)

Purpose of care: reduction of the deficit, fulfilling the necessary needs arising from daily activities

Nursing interventions:

1. Assessing restrictions in terms of undertaking self-care (assessment of physical fitness, assessment of the severity of depressive states).
2. Assisting in the performance of daily activities depends on the patient's condition and his/her abilities (hygiene and body care, preparation and consumption of meals).
3. Planning facilities in the apartment (handrails, shower chair, bathtub and shower handles, as well as training the patient, how to use orthopaedic equipment or a wheelchair) that increase the independence of the patient, and thus improve his/her self-esteem and mood.
4. Allowing the patient to perform activities for as long as he/she needs to make full use of his/her current fitness.
5. Encouraging independence.
6. Encouraging and providing opportunities for rest.
7. Assisting in performing active, passive, resistance and isometric exercises – adjusting activity to capabilities.
8. Assisting in changing the position in bed.
9. Educating the family, guardians on how to perform a full

body hygiene, lift and change positions, use facilities, change bed linen, exercise, massage, serve meals, implement a diet, on the necessity to provide assistance in carrying out activities that may be difficult for the patient.
10. Assessing patient's fitness (e.g. using the Barthel scale).
11. Assessing the effectiveness of actions taken.

Nursing issue No. 6

Increased demand for sleep and rest
Purpose of care: enabling sleep and rest
Nursing interventions:
1. Providing opportunities for sleep and rest.
2. Creating conditions that facilitate relaxation (silence, peace, room ventilation, comfort in bed).
3. Combining procedures.
4. Enabling sleep for as long as necessary.
5. Observing the patient during sleep and rest.

Nursing issue No. 7

Difficulties when concentrating and learning
Purpose of care: increasing the possibility of concentration, raising the level of knowledge
Nursing interventions:
1. Assisting in learning by completing tasks together.
2. Encouraging training in memorising skills, e.g. through educational games, rebuses, crosswords.
3. Strengthening the patient's self-esteem at the present stage of education, showing positive sides, encouraging contact with peers.
4. Repeating tasks multiple times until they are completed.
5. Writing down tasks on a sheet of paper.

Nursing issue No. 8

Emotional lability, lowered mood, malaise, negativity.
Purpose of care: improving mood, increasing self-awareness of the emotional state, discharge of negative emotions
Nursing interventions:
1. Accompanying and conversing with the patient.
2. Kindly addressing, strengthening self-esteem through righteous praise, recognising small successes.
3. Assisting in making contact with a psychologist.
4. Talking with the family about providing support and the way to accompany the patient.
5. Mobilising the patient for social activity and contacts.
6. Talking about the essence of the disorder, its symptoms and effects.
7. Encouraging to verbalising fears such as disability or pain.
8. Identifying support groups (Polish Association for Persons with AHC, local groups for people with disabilities) and encouraging participation in their lives.

Nursing issue No. 9

Inability to cope with stress (screaming, crying, aggression)
Purpose of care: assistance in constructively dealing with stress
Nursing interventions:
1. Assisting in obtaining psychological consultation.
2. Teaching relaxation e.g. by listening to music, having fun, watching a film.
3. Teaching to relieve unpleasant feelings – anger, aggression through, for example, work, solitude and shouting.
4. Talking about the patient's emotions and behaviour.
5. Making the patient aware of the effects of inappropriate behaviour (sadness, fear, separation) on others.

Nursing issue No. 10

Threat of another hemiplegia and/or tetraplegia attack
Purpose of care: ensuring safety
Nursing interventions:
1. Talking to the patient about the factors that can trigger an attack (physical activity, emotions, stress, fatigue, trauma, bright light, warmth, cold or bathing) and the need to avoid them.
2. Informing the patient about the necessity to report alarming symptoms.
3. In the event of imminent attack, taking a safe position that prevents injury.
4. Observing the patient.
5. Ensuring that a company and assistance will be provided as needed.

Nursing issue No. 11

No understanding from the public and medical staff due to a lack of knowledge about the disorder and its sequelae.
Purpose of care: provision of safety, emotional support
Nursing interventions:
1. Conversing with the patient about the current knowledge about rare disorders.
2. Encouraging the patient to talk about his/her disorder in public.
3. Providing information about AHC to medical staff and acquaintances.
4. Strengthening the patient's self-esteem.

Nursing issue No. 12

Possibility of contagious disease as a result of skipping preventive vaccinations
Purpose of care: prevention of infection, early detection of signs of an infectious disease
Nursing interventions:
1. Observing the patient (skin, appearance) and monitoring vital signs (heart rate, blood pressure, temperature, saturation, breaths).
2. Documenting measurements.
3. Collecting material for laboratory tests in accordance with

the individual order sheet (morphology, ESR, biochemical tests, specialised tests).
4. Avoiding contact with patients with infectious diseases and staying in places at increased risk, e.g. hospitals with an infectious profile.
5. Caring for hand hygiene and washing food products.
6. Informing the patient about the need to report alarming ailments.

Nursing issue No. 13

Difficulties with speech and communication with the people in the surrounding

Purpose of care: establishment of contact, ensuring safety

Nursing interventions:

1. Encouraging the patient to speak.
2. Showing kindness, support, dedicating as much time as needed to understand the patient.
3. Accompanying and observing him/her during an attack.
4. Educating family and significant people about communication difficulties, encouraging patience, empathy and kindness.

Obviously, the above problems do not fully cover the difficulties of patients with alternating hemiplegia of childhood and ways of dealing with them, so nursing issues in individual cases have been identified, and so:

Case No. I

Nursing issue No. 1

Possibility of acid-base balance disorders caused by kidney problems

Purpose of care: early detection of acid-base balance disorders

Nursing interventions:

1. Observing the patient's condition and monitoring his/her vital signs (temperature, blood pressure, heart rate, saturation, breaths – number, nature, smell),
2. Documenting measurements.
3. Encouraging regular examinations at the Nephrology Outpatient Clinic.
4. Administering medication in accordance with the individual Order Sheet.
5. Ensuring a proper diet and fluid supply.

Nursing issue No. 2

Possibility of pharmacotherapy complications – side effects of flunarizine

Purpose of care: early detection of pharmacotherapy complications

Nursing interventions:

1. Observing the patient for possible side effects: excessive sleepiness and fatigue, increased appetite and weight, insomnia, anxiety, dyspeptic symptoms: heartburn, nausea, stomach ache, dry mouth, muscle pain, rash, galactorrhoea, depression, extrapyramidal symptoms (movement, gait disorders, tremor and muscular stiffness).
2. Educating the patient and his/her family about the possible complications of pharmacotherapy.
3. Informing about the necessity to report any alarming ailments.
4. Contacting a doctor in case of adverse effects.

Nursing issue No. 3

Difficulties when moving due to clubfeet

Purpose of care: ensuring safety

Nursing interventions:

1. Observing the patient.
2. Assisting in moving in the state of fatigue, in the event of an attack.
3. Providing comfortable, professional footwear.
4. Ensuring the possibility of using a wheelchair when leaving the apartment.
5. Encouraging regular visits to the orthopaedic clinic.

Case No. II:

Nursing issue No. 1

Decreased self-esteem, caused by the diagnosis of alternating hemiplegia of childhood and its consequences

Purpose of care: strengthening self-esteem, personal worth, provision of information about the disorder

Nursing interventions:

1. Accompanying the patient during hospitalisation, stay at home.
2. Talking about fears, actively listening to the patient, supporting him, showing empathy, acceptance, encouraging to verbalise expectations about the future.
3. Educating about activities that increase physical and self-service fitness, encouraging active participation in the rehabilitation process.
4. Encouraging cooperation with a therapeutic team, contact with a psychologist, psychotherapist.
5. Assisting in finding solutions for the best functioning with disability.
6. Talking with family and guardians about their and the patient's fears, anxieties.
7. Assessing the effectiveness of actions taken.
8. Showing people with disabilities and how they cope (e.g., Nick Vujicic), referring to authorities, e.g. Fr. Józef Tischner.
9. Observing the patient, his/her behaviour and emotional state.
10. Providing news on the latest scientific developments regarding AHC.
11. Assisting in preparing a realistic and integrated activity plan, assisting in administering medications, physical therapy, in choosing the right way to deal with the disorder.

12. Attempting to find positive achievements together with patients, highlighting small successes, explaining the reasons for a bad mood.
13. Encouraging new contacts and maintaining old acquaintances, socialising as much as possible, devoting free time to interests.
14. Encouraging contact with family and significant people.

Nursing issue No. 2

Limitation of social contacts due to the change of external appearance

Purpose of care: enabling contact with people in the surrounding, increasing self-esteem

Nursing interventions:

1. Conversing with the patient and his/her family.
2. Encouraging contact with family, acquaintances and support groups.
3. Informing about the possibility of participating in forums, associations, rehabilitation trips.
4. Looking together for reasons why social contacts are limited.
5. Creating the possibility of contact with a psychologist.

Nursing issue No. 3

Uncertainty, fear over the possibility of another attack

Purpose of care: reduction of fear, ensuring safety

Nursing interventions:

1. Providing support by talking about uncertainty, fear, letting the patient verbalise these feelings, showing empathy.
2. Reflecting together on how to reduce these feelings, e.g. by avoiding stimuli that trigger an attack (physical activity, emotions, stress, fatigue, trauma, bright light, heat, cold or bathing), alleviating its effects.
3. Encouraging him/her to inform people about alarming symptoms.
4. Assisting in contact with a psychologist.
5. Encouraging conversations with significant people.

Discussion

Alternating hemiplegia of childhood is not only a rare disease, but it can be classified as ultra-rare because it occurs at a frequency of 1/1000000. It is puzzling that there is too little access to information on rare disorders and social knowledge, especially among medical staff. As Machaczka [34] describes, general practitioners and specialists underestimate the symptoms reported by patients suffering from a rare disorder due to lack of time, resources and knowledge. This results in an increased number of consultations, incorrect diagnoses and a delay in making the correct diagnosis. Studies conducted on two cases of patients with AHC indicate that both they and their parents had to go to many specialists, various hospital departments, not only domestic but also abroad. In the second case, the mother also consulted people involved in alternative medicine. Identification always began with a diagnosis: epilepsy. Neither anti-epileptic treatment nor its subsequent modifications had any effect. The respondents unanimously emphasise that, in their opinion, most of doctors did not listen to them and ignored the symptoms they reported. This delayed making the right diagnosis: in the first case it took 7 years, and in the second case up to 31 years. Perhaps these long searches could be avoided if the clinical criteria described in 1993 by Bourgeois [15] were used, and then collected as criteria for the diagnosis of recurrent hemiplegia of childhood by the International Classification of Headache Disorders in 2013 [23,29]. As Gergont et al. [18] write, the lack of mutation of ATP1A3, a gene discovered in 2012 by scientists from Duke University that is responsible for AHC, does not exclude alternating hemiplegia of childhood. Skoczylas [1] in his doctoral dissertation and Kieć-Wilk [35] rightly note that it is necessary to constantly update information and educate medical staff, because knowledge about disorders, including rare disorders, is gradually expanded. Previously mentioned Gergont et al. [18] suggest that the AHC problem may affect many people, but due to poor diagnostics the issue seems to be minimal. And so there are about 20 people associated in the *Association* for Persons with *AHC*.

Establishing the correct diagnosis was possible due to the determination of the parents and, in the second case, the patient himself, who did not acknowledge the information that it was epilepsy or subsequent incorrect diagnoses. In their search they relied mainly on Internet sources and the press (Newsweek). Liburai et al. [3] also indicate in their research that the most important source of information about a rare disorder for patients and their guardians are the Internet, Facebook, internet forums, specialist physician and foreign language sources. This indicates the necessity for new solutions to ensure effective diagnosis and better coordination of care in terms of rare disorders.

The diagnosis of alternating hemiplegia of childhood, as the name implies, is most often made in the case of children. Since it has not been shown that the disease can be inherited, the authors Gergont et al. [18] in their 2014 publication suggest that other family members should be tested. In these cases, no one in the family had AHC. Only the father of the man who went through an ichemic stroke in February this year is suspected to have a genetic disorder. This requires further verification.

Alternating hemiplegia of childhood is paroxysmal in nature. It has an unpredictable and variable course. As Kansagra [15] writes in his study, the clinical picture is complicated and depends on many factors. Sometimes a disorder may affect a person who does not meet the clinical criteria described by the International Classification of Headache Disorders in 2013 [23,29], because e.g. the first symptoms start after 18 months of age. In the group of participants, the first symptoms occurred at 4 and 8 months of age, the attacks of hemiplegia affected both

sides or the whole body, the presence of intellectual deficits was noted and other disorders (epilepsy, and also, in the case No. I, metabolic diseases) were excluded. The attack was interrupted by sleep. The frequency of seizures is variable and depends on factors such as lifestyle and medication. Simmons and Gergont et al. [15,18] noted that neuroleptic drugs in this group of patients do not have a therapeutic effect. Mediocre results were also obtained using amantadine, acetazolamide, chlorhydrates or benzodiazepines. They highlighted the effect of Flunarizine on reducing attacks and improving the quality of life of AHC patients. Flunarizine is used to prevent migraine and balance disorders. In the examined girl, when Flunarizine was included in the therapy (from 2015), the frequency and duration of attacks clearly decreased, despite the fact that the child's mother is considering stopping the use of this drug and ceasing treatment of her child. The man participating in the research took Flunarizine for 3 months and due to his indisposition, after consulting a doctor, he stopped further treatment. It is too short a period to comment on the therapeutic effectiveness of this drug. Perhaps the treatment with Flunarizine should be attempted again to assess its effects. This is justified by seizures that occur quite often in his case – 3–5 a week and about 5 tetraplegic in a month, which in comparison with the discussed girl is almost twice the value. The more so due to the fact that, according to Gergont et al. [18], the frequency and duration of attacks should decrease with age. Clearly, this can also be due to a bad lifestyle and not avoiding triggers such as: stress, emotions, physical activity, fatigue, warmth, cold or bathing. Wong et al. [33] also examined a Chinese girl treated with corticosteroids. During the four-week therapy, the symptoms of the disorder were stopped, but after it ended they returned again. Finding an effective drug for AHC requires further research and encouraging pharmaceutical companies to produce orphan drugs, as was done in the United States in 1983, as mentioned by Machaczka [34].

The ketogenic diet seems to have a beneficial effect. Schinarizi [24] described the case of a patient whose epileptic seizures and typical attacks of alternating hemiplegia of childhood stopped due to this diet. This still requires confirmation in further research. The patients participating in the research and their guardians heard about the ketogenic diet, but were sceptical about it, not believing in its effects, even though they never tried to use it. This approach to the issue may result from a low level of knowledge on the subject, from a small amount of scientific research confirming the effectiveness of this diet or the associated financial expenses. The consequences of introducing a ketogenic diet in a 10-year-old girl entering puberty also raise doubts.

AHC not only causes changes in the human body, but also carries intellectual, emotional and social deficits. The presence of intellectual deficiencies is confirmed by the research of numerous scientists: Winczewska-Wiktor et al. [29], Terwindt [23], Kansagra et al. [15], Gergont et al. [18]. The girl participating in the research attends a special school and her mental retardation is approx. 3 years. In the case of the man, no intellectual disability was found, he was able to pass the secondary school final examinations and obtain the title of an electrical and computer technician. To a large extent, he owes the above to his mother, who in the past has limited the dimension of her professional career and, as a pedagogue, took up her son's individual education. Kansagra et al. [15] and Gergont et al. [18] also mention change in behaviour, shouting and stimulation of AHC patients before the attack. In the discussed cases, emotional problems and unjustified symptoms of aggression were also noted. This is not always an omen of an impending attack. It is necessary to separate the symptoms of the disorder from the temperament and character of a person or dysfunctions in this regard (as in the case No. II – hysteric-type neurotic reactions). Libura [3] writes about the social consequences associated with the disease. These include the following: the need of one parent to resign, limit his/her professional work, financial deterioration, resignation from leave, disagreements in the family, the need to use the help of a psychologist, no substitution in caring for the sick, limiting contacts with friends and relatives. She also draws attention to the lack of specialised short-term care, giving parents the opportunity to rest, the deficit in the daily life support system (personal assistants) and the lack of appropriate care and treatment facilities that could be home to patients unable to live independently or needing care after the death of their parents. In both case No. I and II, one of the parents gave up or limited their professional work, which in turn reduced the financial liquidity of the family. Both patients use the help of a psychologist, they have limited mobility not only when it comes to holiday trips, but also about everyday functioning. Going outside the apartment is practically impossible without a wheelchair. This is due to the unpredictability of the next attack. They depend on the support and help of the closest people with whom they often come into conflict. Their guardians, who have to rely on themselves, often tired of long-term care, also see deficiencies in the social assistance system and are worried about the future of their children after their death.

In the care of patients with alternating hemiplegia of childhood, Dorothea Orem's self-care model seems to be the most appropriate. As Glińska et al. [41] write, this is one of the most commonly chosen care theories. It was used by, among others Nieckula et al. in his work on patients with venous ulcers of lower limbs [4]. Orem's theory assumes that every person is capable of self-care, i.e. learned activity in relation to themselves and the environment, undertaken to maintain life, health and complete well-being. Obviously, there are situations when a person is unable to take care of his/her own and its deficit appears. In such an event, I suggest using one of three care systems: fully compensatory, partly compensatory or supportive and educative [39]. The role of the nurse in the implementation of the process of nursing patients with alternating hemiplegia of childhood is to

establish a diagnosis and choose the appropriate care system at a given time. Two cases presented in the study are not enough to determine which nursing theory would be the best and what are the main nursing issues of patients with AHC. Research should be continued in the future to determine nursing priorities not only for patients with AHC, but also for other rare disorders.

Conclusions

1. Patients who suffer from rare diseases have restricted access to diagnosis, cure, rehabilitation and social assistance. They often meet with lack of understanding on the part of society and medical staff resulting from scarce knowledge about this issue.
2. The level of knowledge about rare disorders should be increased not only in society (by providing information on this subject in: leaflets, press, advertising spots, websites), but above all among medical staff by including a block about rare disorders in the training programmes of new staff or training programmes, courses of working staff.
3. Alternating hemiplegia of childhood is an unpredictable disorder and its pathomechanism as well as therapeutic options are still being studied.
4. Nursing care for a patient with alternating hemiplegia disorder depends on the patient's current condition, age, social and family support, and place of residence.
5. The role of the nurse in the implementation of the process of nursing patients with alternating hemiplegia of childhood consists in establishing the diagnosis and choosing an appropriate Dorothea Orem's care system in a given situation.

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Streszczenie

Wstęp: Choroby rzadkie to schorzenia występujące u 1 na 2000 osób, przeważnie o podłożu genetycznym. Do tej grupy należą także jednostki chorobowe powstałe na skutek defektów metabolicznych, immunologicznych, infekcji czy rzadkie nowotwory. Rozwój nauki, a szczególnie genetyki spowodował odkrycie nowych mutacji genowych. Jedną z nich jest mutacja w obrębie genu ATP1A3 odpowiedzialna za m.in. naprzemienną hemiplegię dziecięcą. Naprzemienna hemiplegia dziecięca to choroba o bardzo zmiennym i nieprzewidywalnym przebiegu, której objawy pojawiają się we wczesnym dzieciństwie. Napady mogą występować pod postacią hemiplegicznych czy tetraplegicznych i niekiedy towarzyszą im zaburzenia połykania i oddychania.
Celem pracy było porównanie dwóch różnych przypadków pacjentów cierpiących na chorobę rzadką, jaką jest naprzemienna hemiplegia dziecięca, wyłonienie problemów pielęgnacyjnych w sferze bio-psycho-społeczno-duchowej oraz określenie modelu pielęgnowania na przykładzie teorii Dorothey Orem w opiece nad tymi chorymi.
Materiał i metody: W pracy posłużono się metodą porównawczą opisując dwa przypadki pacjentów. Zastosowano techniki wywiadu, obserwacji, pomiaru i analizy dokumentacji. Kwestionariusz wywiadu zawierający 13 pytań oraz przeprowadzona w warunkach codziennych obserwacja pozwoliły na ustalenie problemów pielęgnacyjnych pacjentów z naprzemienną hemiplegią dziecięcą. Badania przeprowadzono od marca 2018 do maja 2018 r. porównując ze sobą dwa przypadki: 10-letniej dziewczynki i 34-letniego mężczyzny.
Wyniki i wnioski: Pacjenci cierpiący na choroby rzadkie mają ograniczony dostęp do diagnostyki, leczenia, rehabilitacji, świadczeń socjalnych oraz często spotykają się z niezrozumieniem ze strony społeczeństwa i personelu medycznego wynikającego ze znikomej wiedzy na ten temat. Poprzez porównanie ze sobą dwóch przypadków wykazano, że problemy pielęgnacyjne tej grupy pacjentów są niezwykle skomplikowane i zmienne w zależności od wieku, rodzaju napadu, stopnia ciężkości, czasu trwania choroby oraz wsparcia rodziny i społeczeństwa. Najbardziej odpowiedni wydaje się być system pielęgnowania według Dorothey Orem.

Słowa kluczowe: naprzemienna hemiplegia dziecięca, choroby rzadkie, opieka pielęgniarska