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THE ROLE OF THE NURSING TEAM IN THE CARE OF PATIENTS WITH KABUKI SYNDROME

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A – study design, **B** – data collection, **C** – statistical analysis, **D** – interpretation of data, **E** – manuscript preparation, **F** – literature review, **G** – sourcing of funding

ABSTRACT

Background: Kabuki syndrome is a rare genetic condition characterised by pathological changes within all the systems of the body, but with variable gene expression. All the patients described in the literature so far have specific facial features resembling the masks of actors from the Japanese Kabuki Theatre and mild to moderate mental impairment. Diagnosis is made based by genetic testing for mutations of the KMT2D and KDM6A genes. Therapy is mainly based on symptomatic alleviation of the effects of mutation, rehabilitation and improvement of the quality of patients' life. Then prognosis of patients with Kabuki syndrome is closely related to the severity of symptoms, which is very variable.

Aim of the study: The purpose of the study is to present the nursing problems based on the case report and to present complications caused by the disease entity.

Material and methods: The criterion for including the patient in the study was the legal guardian's (parent's) consent for the child to participate in the study. Qualitative research was conducted using analysis of medical records, interview with the child and the child's legal guardian, direct and indirect observation of the child for psycho-social changes related to the disease and interpretation of the data in the context of the theoretical knowledge and our own observations.

Case report: The report is based on the case of a 16-year-old girl, diagnosed (at the age of fourteen) with rare genetic disorder – Kabuki syndrome. The patient experiences some characteristic symptoms – big, red lips, low-set and sticky-out ears, drooping eyelids and short fingers and toes. One of the first symptoms suggesting Kabuki syndrome were: spitting up during breastfeeding, problems with swallowing, motor clumsiness and epilepsy. The role of the nursing team during hospitalization was to take care because of habitual dislocation of patella, and to provide psychological suport. At present time the girl uses a wheelchair or she moves on her knees

Results: The manifestations of chronic disease contribute to the feeling of excessive stress, regardless of the patient's age. An adequate diet enriched with proteins contributes to the prevention of bedsores among individuals with Kabuki syndrome, immobilised to various degrees. Regular consultations with specialists, such as cardiologist, neurologist, gastroenterologist, orthopaedist, ophthalmologist, psychologist, orthodontist, speech therapist, immunologist, endocrinologist and dietician reduce the risk of complications associated with the disease in the patient with Kabuki syndrome. Early implementation of rehabilitation, focused to increase muscle tension, contributes to maintaining autonomy and self-care in patients with Kabuki syndrome.

KEYWORDS: Kabuki syndrome, child, care, nursing team

Background

Kabuki syndrome is a rare genetic disease diagnosed for the first time in Japan in 1981 by two independent Japanese physicians, Norio Niikawa and Yoshikazu Kuroki [1,2]. To date, only 300 cases of people with Kabuki syndrome have been reported worldwide [2]. In Japan, this disease occurs as often as 1:320,000–86,000 people [3,4]. The condition is caused by a mutation in the KMT2D and KDM6A genes. Niikawa–Kuroki syndrome is a multisystemic disease, characterized by specific facial features, which resemble the faces of Japanese actors of the Kabuki Theatre, namely, large, red, prominent lips, drooping eyelids, arched eyebrows, long eyelid fissures and a short nasal septum [3,5,6].

The clinical picture of the Kabuki syndrome includes bone abnormalities, such as growth retardation, brachydactyly (shortening of fingers), clinodactyly (bending of the fifth finger of the hand) and spine abnormalities [5]. Pathological changes occur in other body systems, but with different intensity. The most commonly diagnosed

and described conditions in Niikawa-Kuroki syndrome are defects in the left side of the heart, abnormalities in the functioning and structure of the genitourinary tract, frequent recurrent infections of the respiratory tract, vision and hearing impairment, dental changes, mobility and speech problems [7]. The Kabuki syndrome is also characterised by a mental impairment of varying degrees and social-emotional retardation. Because of these numerous systemic changes, patients with Kabuki syndrome need frequent consultations, rehabilitation and surgical interventions in many cases [3].

The role of the nurse in the care of a patient with Kabuki syndrome

The concepts of nursing theory form the basis of professional care. Using these concepts and the practical actions of the nursing staff and combining them with their own experience is a measure of highly specialised care. In the case of rare genetic diseases, which include the Kabuki syndrome, there are no universal models of management, requiring the therapeutic team to be innovative in taking therapeutic action. Since the Niikawa-Kuroki syndrome is a newly identified disease involving multiple conditions and disorders within all organs of the body, each patient requires a highly individual care plan..

The number of symptoms and their different intensities necessitates that the therapeutic team constantly acquires new knowledge on the Kabuki syndrome in order to help the patient and his/her family effectively.

Among the state institutions that help and support patients with genetic diseases are foundations and associations such as "Wspólnie" [Together] – Association for Children with Rare Genetic Diseases and Their Families [8], and "Neuron" Foundation [9]. As mentioned by Florek-Łuszczki M. and Lachowski S. in the article "Institutional activities for the disabled people", the State Fund for Rehabilitation of the Disabled (PFRON) [10] also provides great support for the disabled.

A common feature of patients affected by the Kabuki syndrome mutations is facial dysmorphism, which attenuates with age, with the facial features become less striking, comparable to those of healthy people [11]. Different appearance and the feel of isolation may be connected with psychological stress for both the patient and his or her family. Stress among individuals with chronic genetic diseases was addressed in the work by Ziarko M. titled "Struggling with stress in the chronic disease" [12]. The destructive impact of stress on making therapeutic decisions in genetic diseases, a critical event that is the chronic disease and its somatic symptoms do have an effect on the quality of life because they impair the physical fitness [13].

The goal of the nursing team caring of the Kabuki patient is to provide comprehensive consultancy to find support and assistance from state institutions and to propose possible modifications and facilities for self-care and self-help. Due to the possibility of motor problems in patients with Kabuki syndrome, it is important, depending on the severity of the disease, to ensure a sense of safety during locomotion, by providing give physical support. One of the goals of care for a person with a genetic disease is to offer methods for coping with stress that help maintain good quality of life in the face of an incurable chronic disease, financial problems and a fear of losing autonomy [14,15].

It is important to ensure the continuity of nursing care both during hospitalisation and at home. In most cases, the burden of caring for a chronically ill child falls on the family's shoulders. In order to prevent burnout of caregivers of a child with a genetic disease, it is necessary to assist the patient and his or her caregivers in the home environment by providing a long-term care or palliative care [7,16]. The role of the therapeutic team in the care of a patient with Kabuki syndrome is primarily to educate caregivers and patients about the need for regular specialist consultations, shaping correct habits, such as taking an upright posture while sitting, and avoiding crossing legs to minimize the risk of posture defects. Early implementation of rehabilitation procedures (daily exercises at home adjusted by physiotherapists to patient's abilities, corrective exercises, correct body posture during everyday activities) is also of great importance in order to maintain autonomy and prevent contractures [17,18].

Aim of the study

The purpose of the study is to present nursing issues based on a case report, to propose implementation of nursing interventions for a child with Kabuki syndrome, and to present complications caused by the disease.

Material and methods

This is a case report of a 16-year-old girl patient, 157 cm in height and 65 kg in weight. Her BMI of 26.37 indicates that she is overweight. The study was conducted using medical records analysis [19]. The tools used in the analysis of records were a nurse's observation sheet, laboratory testing results, case history and outcomes of examinations and consultations with the psychologist, speech therapist, neurologist, endocrinologist and physiotherapist.

A scheduled interview with the patient was used to gather additional information on the current state of her physical health, mental well-being, as well as on the symptoms as a result of the Kabuki syndrome. An interview with the child and her legal guardian focused on active listening and creating a current, written record.

Doctors, directly and indirectly, observed the child for psycho-social changes related to the disease. The observations were made at home as well as during rehabilitation activities. Doctors directly observed her for disease-specific, dysmorphic facial changes, unnatural appearance of hands and feet, body structure, and

difficulties that may result from the necessity to use a wheelchair. Indirect observation by the rehabilitation team and the family provided information on the progress, the patient's involvement in the rehabilitation as well as her responses to the disease. All information was recorded and further analysed by comparing it with reports in the published literature. The recorded data were interpreted in the light of theoretical knowledge and our own observations. The study was granted the approval no. 68/2016 by the Bioethical Commission of the State Medical Vocational School. The legal guardians of the child provided informed consent

Specific case report

Health interview and analysis of the patient's medical records

The patient was born by spontaneous labour in the 38th week of pregnancy with a birth weight of 3300 grams, a height of 54 cm, head circumference of 33 cm, and assigned an Apgar score of 10. During pregnancy, the mother suffered from viral infections of the upper respiratory tract. After the delivery, the newborn was diagnosed with general swelling, bruising of the thumb of the left hand and the pit (dimple) in the coccygeal skin.

The girl's psychomotor development was normal as she started sitting at 9 months, walking at around 1.5 years, speaking at 2 years. The patient had motor clumsiness manifested by waddling, running with a huddled posture, and dropping objects from her hands. Her main problem as an infant were frequent, recurrent infections of the upper respiratory tract and the need for hospitalization due to episodic febrile convulsions at the age of 2, 5.5 and 9.

The patient also had epilepsy. The first epileptic seizure occurred when she was 5.5, while the most recent one in 2010 lasted for 4 hours.

During hospitalization in 2010, a neurological consultation was conducted, which showed a disturbing characteristic, namely dysmorphic lesions of the skull, face, hands and feet, and as a result, the patient was referred to for extended neurological, genetic and endocrinological diagnostics. The examination also showed a generally reduced motor efficiency, walking disorders and obesity. As part of the neurological consultation, Romberg's test was performed to assess static balance.. Deep reflexes were poorly symmetrical but expressed.. There was poor coordination of the upper and lower limbs. Strength and muscle tone were normal. In order to confirm the suspicion of epilepsy, a transverse MR imaging of the head was performed with intravenous contrast agents.

The first genetic testing was performed in 2010 in the Medical Genetics Laboratory in Łódź. During the physical examination, a strong, yet obese body structure was noted. The following features attracted the consultant's attention: large, square forehead; large, protruding, low-set ears; flat root of the nose; short, prominent philtrum, large mouth, widely spaced teeth, small chin, and drooping eyelids. The patient's palms were very soft with tapered fingertips. Her feet were wide with shortening of the second to the fifth toes, while the hallux was large. Due to dysmorphic changes in the face, hands and feet, peripheral blood lymphocytes were taken and used for an in vitro cell culture. The test was aimed to determine patient's karyotype. The test resulted in normal female 46, XX karyotype. There was no chromosome abnormalities that could explain the dysmorphia observed. However, a change in the number of copies of genes, that is a duplication below 1 Mb, was detected in the patient's genome, which may be a risk factor for the emergence of diseases with recessive inheritance patterns. On the basis of the above examination, no diagnosis or a suspicion of Kabuki syndrome was made.

The first alarming symptoms that might have indicated a rare genetic disease were noted by an orthopaedist who thereafter referred the patient for genetic consultation. A test for changes in the number of copies of genes by the aCGH method was carried out in 2014 and revealed the most likely clinically insignificant micro-duplications: duplication of 2q31.1 (HOXD9 gene) and duplication of 7q11.23 (WBSCR16 gene). In ongoing diagnostics, the patient was referred to the Genetic Clinic with a referral from her family practitioner. Considering the entire medical history and clinical symptoms, the dysmorphic syndrome called Niikawa-Kuroki syndrome was suspected. On the Makrythanasis scale, 3/10 points were assigned to patient's Kabuki syndrome and molecular testing of the KMT2D gene was ordered. The results of the genetic test confirmed the suspicion of Kabuki syndrome by showing a pathogenic mutation of p.Gln3905.

Since she was 4 years old, patient's main problems have been habitual dislocations of the patellae leading to difficulty in walking and gradually impaired physical fitness. Due to the lack of stability within the knee joint, the patient has had difficulty walking since the age of 9 years. She moved on her knees and in a wheelchair. At the age of 10, the patient underwent MR imaging of her right knee joint in the fibular, anterior and transverse planes. The imaging revealed complete patellar dislocation with features of cartilagino-osseous necrosis in the lateral condyle of the right femur. In addition, dislocation was accompanied by swelling, reduced thickness of patella cartilage, an increased amount of fluid in the recesses of the knee cavity and signs of damage to the patellar retinacula.

No more than a year after surgery of the right patella, an MRI of the left knee joint was performed. A correct amount of fluid was found in the left knee joint cavity, however, the examination revealed a complete dislocation of the left patella that was located next to the lateral condyle of the femur. The case had features of severe dysplasia of the right patellofemoral joint and

a non-developed intercondylar line of the femur. The cartilage of the lateral condyle of the femur and tibia was thinner, which is a feature of chondromalacia. In 2012, the patient underwent further surgery for the habitual left patella dislocation, which also took place without complications.

In 2014, the patient underwent another MRI of the lumbosacral region because of the flaccid paraparesis of lower limbs. The examination showed physiological lumbar spinal lordosis, moderate right-convex scoliosis and normal morphology of lumbar vertebrae. No clear pathological changes were found in the soft tissues around the spine. Other than minor disorders of statics, the MR image of the L-S section of the spine was normal. The next hospitalization was due to unspecified paralytic syndrome, central nervous system disorders, cryptogenic encephalopathy, epilepsy, generalised muscle hypotonia, hypothyroidism, psychomotor retardation and upper respiratory tract infection. The programme of neurological speech therapy included exercises of the articulatory organ, introduction of correct speech patterns for "s, z, c, dz" sounds in sentences, words, expressions, strengthening of introduced speech patterns, getting used to systematic exercises, and motivating for effort.

Based on psychological and pedagogical examination, it was found that the development of the patient was slightly below normal in relation to her age. The patient also became demotivated quickly when facing difficulties and rehabilitation exercises and easily avoided attempts to achieve goals. In addition, doctors found normally developed verbal functions, wide general knowledge and concepts, the ability to express her thoughts and feelings, the willingness to make verbal contact. The biggest difficult was arithmetic reasoning, acquiring knowledge and skills in mathematics, even in elementary addition and subtraction of numbers up to 10. As regards visuo-motor coordination, her learning ability was at a level lower than expected, which may contribute to poor motor ability and activity, controlled by visual perception, resulting in reduced lower manual dexterity. Based on the observation of patient's social functioning in the area of emotions and social behaviour, it was found that she correctly made emotional contact and had a positive relationship with people around her. The "Describe yourself" test also indicated a tendency to experience negative emotions, sadness, anxiety, agitation, unrest, tension and fears, which the patient seemed to be suppressing, seeming to not want to be considered better than she was in reality. The therapeutic programme included psychological assistance and supportive conversations addressing her strong anxiety and fears of standing on her own two feet. The patient also participated in rehabilitation holidays due to generalised muscle hypotonia, motor clumsiness and poor motor coordination. Additional difficulties were flexural contractures of the group of ischiotibial muscles of the right and left leg due to a long-term immobilisation.

Description of the patient's current health situation

Muscle hypotonia, contractures and elastic subcutaneous tissue are some of her prevailing problems. In the dysmorphological examination, we observed features of facial dysmorphy distinctive for Kabuki syndrome: thick hair; wide root of the nose; slanting, upward-facing eyelid fissures; thinned eyebrows on both sides; long eyelashes; prominent, relatively large ears; hypertelorism; high cheekbones; short column; wide end tip of nose; wide, prominent mouth; corners of lips facing downwards; widely spaced teeth and hypodontia.

In the upper extremities, the 5th bone is shortened, so the hands and feet are relatively small. Although most patients with Kabuki syndrome have mild to moderate intellectual disabilities, the patient's intellectual level is normal for her age.

The patient's body feels doughy, which gives the impression of being overhydrated. The girl has a difficulty in daily routines such as combing and dressing, with which she need help. All activities requiring precise manual skills, such as tying shoelaces, putting on shoes, using cutlery, combing, were problematic for the patient and she often dropped objects. The patient requires regular rehabilitation and multi-profile support of her development. She is now a 16-year-old patient, who, from the age of 9, moves on her knees or in a wheelchair.

Discussion

The Kabuki syndrome is a disease entity diagnosed very rarely in Poland, as evidenced by the limited number publications in the Polish literature [20]. Therefore, it is necessary to publish new case reports in order to increase awareness and knowledge of the disease among healthcare professionals. As the first diagnoses of Kabuki syndrome were made more than 40 years ago, it can be concluded that patients with Niikawa-Kuroki syndrome can live to an old age [21,22]. For this reason, early diagnosis of coexisting diseases and intervening with surgery and rehabilitation is a priority for improving the patient's quality of life and extending it. This is related to the necessity for frequent hospitalizations, which are associated with increased anxiety and stress, both of which the medical staff should alleviate, providing patients with peace of mind and a sense of security. It is also necessary to carry out a population study based on a precise estimation of the prevalence of Kabuki syndrome in Poland. Due to multi-system abnormalities in patients with Kabuki syndrome, cooperation of specialists from various fields of medicine is required [23,24].

In the presented case of a 16-year-old patient who was diagnosed as having Kabuki syndrome at the age of 14, the prevailing problem is weakening of muscle tone, which was also observed and reported by Cheon, Ko and team in the paper on the clinical and molecu-

lar features of Kabuki syndrome [4]. According to the authors, muscle tone becomes weaker mainly in the neonatal period among 51–98% of patients. Moreover, the article highlighted the relationship between the type of mutation and phenotype. It has been shown that in patients with KMT2D mutation, hypotonia is more frequent than in individuals with KDM6A mutation. With the introduction of systematic rehabilitation procedures, satisfactory results were obtained, allowing patients to perform activities without the assistance of other people [4].

The article by Haller, Kruk presents a case report of a patient with epilepsy, which manifested itself at age of 13 years [21]. Based on record analysis, the authors pointed out that epilepsy is one of the dominant symptoms in Kabuki syndrome and occurs at a frequency of 10–80% in all of the 300 cases reported so far [18,22]. The research showed that epilepsy seizures were manifested only in children aged from 12 months to 12 years, which was also observed in this case report.

The problem of anxiety and stress caused by the presence of multiple conditions and the patient's different appearance was also addressed by Badenci and Cengiz [26]. The authors of the article presented the case of a 5-year-old patient, paying particular attention to her and her family's mental state. The patient had significant separation anxiety and fear of death and invasive medical procedures. In order to alleviate the excessive fear of the care and medical activities, a method was used, where patient enacted brief scenes with dolls and toys imitating medical tools, having the opportunity to play the role of a doctor or nurse. The measures had a positive effect. Additionally, the patient received psychotherapy. Thanks to the role playing technique, the psychologist could learn about the emotions and fears that the patient was experiencing. Due to the age at which this patient was diagnosed as having Kabuki syndrome, the above methods were not applied. However, psychological assistance was implemented for both the patient and the closest family members.

According to the published data, obesity occurs in children increasingly frequently. In some cases, increased body weight is caused as diabetes or hypothyroidism. Lack of physical activity can also have a direct impact on weight. In the article by Skowrońska and Fichna, the case of a 16-year-old boy patient with obesity, type II diabetes and metabolic syndrome was reported. The nursing care included taking sample materials for laboratory tests, observation of the patient, checking the blood glucose level. A low-fat diabetic diet and pharmacotherapy were also implemented. The patient also consulted with dietician and diabetologist. He was also educated on changing his lifestyle and nutrition habits [27,28]. Preventive measures proposed by Skowronska, Fichna et al. were also applied in the case report of a girl patient with Kabuki Syndrome, who was diagnosed as obese. As in the case reported in this paper, the following issues have been highlighted – the need to perform regular laboratory check-up tests and to observe the patient for symptoms of weakness, fatigue, dryness of skin layers, mood worsening as these may indicate a decrease in thyroid hormone levels [24–27].

Conclusions

1. Early implementation of rehabilitation, focused on increasing muscle tone, contributes to maintaining autonomy and self-care in patients with Kabuki syndrome.
2. The manifestation of a chronic disease contributes to feelings of excessive stress, regardless of the patient's age.
3. An adequate diet enriched with proteins contributes to the prevention of bedsores among individuals with Kabuki syndrome, who may be immobilised to varying degrees.
4. Regular appointments with consultants, such as cardiologist, neurologist, gastroenterologist, orthopaedist, ophthalmologist, psychologist, orthodontist, speech therapist, immunologist, endocrinologist and dietician reduce the risk of complications associated with the disease in patients with Kabuki syndrome.

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