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Psychosocial Functioning of the Child with Primary Ciliary Dyskinesia- Case Study Based on the Theory of Marjory Gordon

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Abstract

Primary ciliary dyskinesia is a rare genetic disease. It is an autosomal recessive inherited disease. This rare disease affecting the respiratory system exposes the child to numerous new and, simultaneously, difficult situations that would be a significant challenge in the normal functioning for an adult.

Nursing care over the patient suffering from primary ciliary dyskinesia creates numerous nursing problems for the patient treated both in home conditions as well as hospital ones. Good knowledge of the patient, his or her ailments, attitude towards the disease as well as planned and implemented treatment methods play an important role. It is essential to collect as much information about the child and his or her family as possible to plan care over the patient properly. The theory of Marjory Gordon may be helpful in this issue.

Keywords: Rare disease; Primary ciliary dyskinesia; Nursing care

Introduction

According to the American act entitled 'Orphan Drug Act 2002", in the United States of America rare diseases or rare disorders are defined as those which affect small populations of patients, usually smaller ones than 200,000 people. In Europe a rare disease is the one that occurs in less than 1 person for 2,000 inhabitants [1,2]. There are approximately 7,000 rare diseases where- according to American Office of Rare Diseases Research- genetic diseases constitute on average 80% of them. Most frequently, these are chronic diseases whose features include severe progressing course. They cause pain and suffering, lead to disability and they bear a high mortality rate [1].

As a result of the fact that these are rare diseases, patients who suffer from them encounter numerous problems on their way. In the medical environment there is a lack of sufficient knowledge regarding the signs of the disease with marginal interest in the scientific research relating to the natural course of the disease and planning the therapy. This leads to implementation of the risky or improper forms of treatment in patients, which can result in severe complications in patients.

One of such diseases is the disorder of ciliary motility. Primary ciliary dyskinesia is a rare genetic disease. It is an autosomal recessive inherited disease. The prevalence of primary ciliary dyskinesia is estimated at 1:16,000 for 1:20,000 live births [3]. The cause of acquired ciliary dyskinesia can be bacterial and viral infections, environmental pollution, administered medications [4,5]. It is characterized by abnormal ultrastructure of the cilia and their impaired motility. The main signs include recurrent infections of the respiratory system, chronic otitis, chronic rhinosinusitis, male infertility, in half of the cases there appears situs inversus. Situs transversus, bronchiectasis, sinusitis constitutes the classic triad of symptoms of the primary ciliary dyskinesia of the so-called Kartagener's syndrome [6]. In several percent of the patients, situs ambiguus is diagnosed (change in visceral positioning only above or below the diaphragm). Diagnosis of the primary ciliary dyskinesia is established on the basis of the clinical picture and assessment of the ciliary ultrastructure and/or mutation in the DNAH5 and DNAI1 genes [7,8]. The clinical picture of the disease depends on the age of the child. In the neonatal period the clinical signs of the primary ciliary dyskinesia usually occur in the form of the raised respiratory rate, pneumonia, respiratory distress syndrome of the infants, chronic rhinitis and difficulty in feeding related to this due to impaired nasal patency, dextrocardia, situs inversus and the following defects: complex heart defect, oesophageal atresia, iliary atresia, hydrocephalus [9,10]. In the late neonatal period and in older children bronchial asthma is often diagnosed due to recurrent obstructive bronchitis, chronic cough, raised respiratory rate [6].

This rare disease affecting the respiratory system exposes the child to numerous new and, simultaneously, difficult situations that would be a significant challenge in the normal functioning for an adult. The attitudes of the children suffering from chronic diseases of the respiratory system towards their own disease are ones of the important factors influencing prognosis, they affect the functioning of the sick child and the course of therapy [11,12]. The impact of the chronic disease on the mental health of the child has become the object of interest of many researchers [12,13]. In the child's psyche there appear changes caused by the rare disease. This influences the change in self-esteem, psychophysical fitness, own availability as well as conviction regarding the child's own appearance, sometimes a different one than in healthy peers. Sometimes, there may appear disorders as a result of the lack of self-confidence. The consequences can be defense mechanisms in the form of the escape, withdrawal. The chronic disease in children may also become a source of frustration and deprivation of the needs of the child as well as improper actions of the parents and teachers [11,13].

The objective of the work is to present the health problems of the patient with diagnosed primary ciliary dyskinesia. The research method aiming at the best presentation of the specific patient case has been applied in the thesis. The information regarding the physical, mental, emotional and social condition has been collected. The following has been used to collect the information: analysis of the medical documentation, consultation with the physician responsible for treatment of the patient, nursing history, observation of the patient [14,15].

The studies were conducted during the period from September 2015

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to May 2016. Collecting the medical history from the guardians and observation of the patient were conducted in the form of unconstrained conversation in the patient's house and during the appointment in the doctor's office. Legal guardians gave informed consent for the analysis of medical records, observation and describe the case of a child.

Community and Family Interview

The patient aged 10 lives in the village near Mielec, Poland with his parents, a two-year-old brother and grandparents. The patient has his own room. Currently, he studies in the third grade of the primary school. Children at school know that he is sick, however, they do not understand his disease and difficulties in learning due to this cause. Teachers who are informed about the health condition of the boy are supposed to adjust the educational requirements to the individual needs and psychophysical possibilities of the student. The boy can count on his parents' assistance in daily life and at school.

Description of the Disease Process

The patient was born on 27 April 2006 from the first pregnancy, the first childbirth. He was born by caesarean section due to threatening fetal asphyxia with pulse disorders. Body weight: 3700 grams. The Apgar scale in the first minute - 6 points, in the third minute - 7 points. During the next minutes the child's condition did not improve, he required oxygen therapy due to increasing respiratory insufficiency manifested with raised groaning breathing with diaphragmatic and intercostal recession. Due to worsening health condition of the child it was decided to transfer the patient to the Department of Neonatology with Isolation Subunit and Intensive Neonatal Care in the John Paul II City Hospital in Rzeszów where he stayed in May 2006 with the final diagnosis of pneumonia and congenital laryngeal stridor. Further treatment was continued from May 2006 to June 2006 in the Clinical Department of Paediatrics and Pulmonology of the same hospital. During the period of summer 2006 without significant exacerbations in the respiratory system. The only symptoms that parents were concerned with were snuffles, snoring breathing with periodical expectoration of the secretion. At the age of 7 months there was the first inflammatory episode in the bronchi that required intensive treatment with antibiotics and mucolytics. Since November 2006 the child has been under control of the paediatric pulmonological outpatient clinic in Rzeszów. In the chronic treatment the child received Budesonid (Pulmicort) during nebulization for the period of 12 months. Taking inhalation medications, such as: Fluticasone (Flixotide) 50 μg through Aeroscopic inhalation chamber was continued and oral anti-allergic Cetirizine (Zyrtec) was continued. During the period of exacerbations proceeding with dyspnea and very intensive cough systemic steroids, such as: Prednisone (Encorton) 2 mg for a kilo of body weight were applied. The mucolytics, such as: Ambroxol (Mucosolvan) and bronchodilators, such as: Fecosterol (Berodual) were administered through nebulization. The first diagnostics for allergy was conducted at pulmonological outpatient clinic in November 2009- the result of the test was negative. Despite the negative results of the tests, the anti-allergic medication Desloratadine (Aerius) and the anitleucotriene medication Montelukast (Singulair) 4 mg, paroxysmal cough, particularly after the night and difficulties in expectorating the secretion were still observed. Due to persistent snoring breathing, difficulties in breathing through the nose, recurrent otitis the child was consulted by a laryngologist who diagnosed hypertrophy of pharyngeal tonsil, hearing disorders and qualified the child for adenoidectomy and incision of the tympanic membranes with placing the drains. After the procedure of adenoidectomy, the child was feverish, cough intensified, and dyspnea appeared. The child was referred to the Poviat Hospital in Mielec to the Paediatric Department where he stayed with diagnosed pneumonia confirmed radiologically and pharyngitis in March 2010. The treatment included Netilmicin (Netromycine), Cefuroxime (Zinacef) administered intravenously and further continuation of Cefuroxime (Zinnat). During the period from June 2010 to September 2011 the patient was under permanent pulmonological and laryngological control in Rzeszów treated for non-allergic bronchial asthma. The worsening health condition of the child was the reason for concern of both parents and a family doctor who referred the child to the pulmonological outpatient clinic in Mielec for further consultation and follow-up. During the first appointment at the end of September 2011, in the physical examination the physician diagnosed pectus excavatum in the patient. On auscultation above the lung fields numerous rales with discrete lengthening of breath. Over the precordial area the presence of quiet systolic murmur, this sign was earlier consulted by a cardiological who excluded the heart defect. Control allergic tests were performed in the patient and the results were negative as in 2009. Spirometric tests confirmed disturbed flows in the lower respiratory tracts. Despite the permanent laryngological treatment, the chronic rhinitis with retained purulent secretion and increasing hearing disorders were observed in the patient. The signs of grinding of teeth, stomachache, hoarseness, a lack of appetite, which allowed for initial diagnosis of the gastroesophageal reflux. Omeprazole (Helicid) 20 mg in the morning on empty stomach and anti-reflux diet were applied obtaining the gradual regression of the signs from the alimentary tract. Despite the permanent administration of the inhalation steroid medications - Budesonide (Miflonide) 2 mg × 200 mg, the antileucotriene medication. Montelukast (Drimon) in the dose of 5 mg per day and periodical administration of beta-mimetics-Formoterol (Foradil) 2 × 1 it was not possible to obtain longer periods of remission of the bronchial lesions. The next episode of pneumonia occurred in August 2012 with extensive radiological changes. Due to the difficulties in expectorating retained thick secretion the 3% solution of the sodium chloride using nebulization alternately with Ambroxol (Mucosolvan) was applied obtaining gradual improvement of patency of the respiratory tracts.

Observing the worsening tolerance of physical effort of the child as well as more and more frequent infectious exacerbations, the physician stated that the whole of the multi-year clinical image did not suggest the typical course of asthma. Still persistent recurrent otitis, permanent rhinitis caused by thick secretion, chest deformation, signs of reflux suggested primary diagnosis of ciliary dyskinesia. In order to verify this diagnosis, the child was referred to the Institute of Tuberculosis and Lung Diseases of the Clinic of Pulmonology and Cystic Fibrosis in Rabka in November 2012. The first diagnostic hospitalization in Rabka in February 2013. On admission above the lungs diffuse wheezes, dry rales, coarse rales- bilaterally. During hospitalization moist cough with expectoration of mucous purulent secretion was observed. Based on the characteristic medical history, endoscopic and microscopic image (a lack of ciliary motility) and low concentration of the nitric oxide in the air exhaled through the nose equaled 55.9 ppb (the result confirming diagnosis of primary ciliary dyskinesia) and concentration of the nitric oxide through the mouth 2.7 ppb (the norm 25 ppb), diagnosis of the primary ciliary dyskinesia was very possible. In the remaining examinations the following abnormalities were detected: impaired patency of - particularly- peripheral bronchi in the spirometric test (bronchial reversibility test- the negative result) and the increase in the numerous *H. influenzae* colonies in the induced sputum culture and bronchial content, with neutrophilia. The treatment included the inhalation and drainage cycle with the mucolytic agent obtaining clinical and spirometric improvement. Treatment with Amoxicillin preparation

and clavulanic acid (Augmentin) and afterwards implementation of the chronic application of Azithromycin in the anti-inflammatory dose supported with the probiotic- *Lactobacillus rhamnosus* (Dicoflor) were recommended. During hospitalization at the Clinic in Rabka in December 2013 on the basis of the examination of the ciliary ultrastructure in the electronic microscope the final diagnosis of the primary ciliary dyskinesia was confirmed. The patient was discharged home with the following recommendations: aerosol therapy using - in the first place- Salbutamol (Ventolin) administered twice a day through the inhalation chamber. Afterwards, nebulization from hypetronic salt solution starting with 3% concentration to obtain 7% concentration (with worse tolerance of higher concentrations- return to the previous concentration) and then Dornasum alfa (Pulmozyme)- used once a day in the dose of 1 ampoule, it is important to remember not to mix it with other substances. Physiotherapy of the respiratory tract twice a day for 20 minutes, however, not earlier than 2 hours following taking Pulmozyme. Perform the procedures of the huffy exhalation technique, breathing through the Acapella. Regular physical activity, particularly in the form of playing with parents and peers was recommended to the patient. The treatment of nasal and sinus changes requires rinsing the nose with warm 0.9% solution of sodium chloride administered through Sinus Rinse from twice to four times a day in the amount of 200-500 ml. Periodical laryngological control with audiometric assessment is advisable. The child requires permanent anti-inflammatory treatment with the antibiotic- Azithromycin every two days in the dose of 125 mg. Furthermore, Flixotide twice in the two doses of 50 μg each through the inhalation chamber. In the event of exacerbation of the bronchial changes and expectoration it is required to increase physiotherapy up to four times a day. Administering the inhalation with Berodual up to two or three times per day as well as mucolytic drugs. In the event of fever and suspicion of bacterial superinfection, empirical administration of Amoxicillin in the daily dose of 90 mg for a kilo of body weight is suggested. Regular control of the course of the disease is also recommended. Permanent pulmonological and laryngological care are advisable. Taking X-ray image is ordered at least once a year. Regular spirometric tests at each appointment at the pulmonological outpatient clinic. Microbiological sputum tests once for three months with appearance of the pathogens that are hazardous for the patient (e.g. *Pseudomonas aeruginosa*) and urgent undertaking the eradication process of the infection are recommended. Control appointment twice a year at the Institute in Rabka are required for control tests and possible verification of the treatment.

Discussion

Assessment of the biological and mental condition of the patient

Assessment of the biological condition of the patient: in the physical examination the cardiovascular system without changes, quite murmur over the precordial area (without the influence on the haemodynamics of the circulatory system), the respiratory system: small stench from the oral cavity, cough- occasional, moist, intensifying with retained secretion in the bronchi in the periods of infectious exacerbations, hoarseness - periodically, dyspnea- occasional with significant physical effort; the digestive system- without changes, teeth: mild caries, periodical heartburn, occasional hiccup; the urogenital system without changes; the osteo-articulo-muscular system: chest deformation-pigeon chest; the senses: hearing: periodically the presence of hearing disorders requiring ventilation drainage of the tympanic cavities, the smell: weakened by periodical retention of the secretion in the nose; the skin: without changes.

General mental condition of the patient is at the level of average intelligence. He uses an average scope of vocabulary and notions. He correctly discerns and defines cause and effect relationships, combines the perceived elements into logical whole. He possesses quite significant general knowledge regarding social and natural environment. The level of mathematical knowledge and skills is good. He performs basic accounting operations in memory. Average speed of visual and motor learning. The ability of direct auditory attention, phonological and visual memory as well as visual perception within the scope of differentiating significant elements from less important ones. The above-mentioned difficulties may affect memorizing longer and more complex commands given through the auditory system and influence correctness of writing by ear and from memory. The patient displays a higher level of anxiety, periodically increasing difficulties in attention concentration. The cause of these dysfunctions may be health problems causing higher insecurity in the situation of increasing somatic symptoms, which consequently results in significant emotional lability, tendency for impulsive behavior and frequent outbursts of anger.

Nursing problems

- The 1st nursing diagnosis: Increased insecurity caused by health problems reflected in behavioural disorders, difficulty in attention concentration and anxiety of the child.
- The objective of care: Decrease insecurity, anxiety of the child.
- The 2nd nursing diagnosis: Dyspnea caused by secretion retention in the respiratory tracts reflected in limited ability to perform significant physical effort.
- The objective of care: Decrease dyspnea and improve gaseous exchange by reducing secretion retention in the respiratory tracts.
- The 3rd nursing diagnosis: Impaired reception of aural stimuli caused by retained secretion in the auditory canals reflected in a lack of correct handwriting by ear.
- The objective of care: Reduce secretion retention in the auditory canals.
- The 4th nursing diagnosis: Impaired reception of olfactory stimuli caused by secretion retention in the nasal cavity reflected in difficulties in distinguishing flavours and aromas.
- The objective of care: Improve comfort of breathing through nose.
- The 5th nursing diagnosis: Apathy, low mood, unwillingness for interpersonal contacts caused by frequent hospitalization and limited contacts with children.
- The objective of care: Improve well-being of the child, encourage to contact peers.
- The 6th nursing diagnosis: Fear of the family caused by chronic disease of the child.
- The objective of care: Reduce fear.
- The 7th nursing diagnosis: Possibility of recurrence of the inflammation of the respiratory tracts caused by secretion retention.
- The objective of care: Prevent new inflammations of the respiratory tracts.
- The 8th nursing diagnosis: Possibility of learning difficulties due to frequent absenteeism at school caused by disease and hospitalization.
- Cel pielęgnowania: Zorganizowanie kontynuacji nauki w domu bądź w szpitalu.

• The objective of care: Organize continuation of learning at home or in hospital.

Conclusion

1. In the case of primary ciliary dyskinesia quick diagnosis of the disease and early implementation of the treatment as well as undertaken nursing and physiotherapeutic activities play an important role. The children must be under permanent care of many specialists.

2. Parents of the boy possess necessary knowledge regarding disease and care over their son. Family members have accepted the disease of the son and they support him in daily life, however, the disease significantly influences psychosocial functioning of the family.

Summary

Nursing care over the patient suffering from primary ciliary dyskinesia creates numerous nursing problems for the patient treated both in home conditions as well as hospital ones. Good knowledge of the patient, his or her ailments, attitude towards the disease as well as planned and implemented treatment methods play an important role. It is essential to collect as much information about the child and his or her family as possible to plan care over the patient properly. The theory of Marjory Gordon may be helpful in this issue.

Adaptation to hospital environment and making the patient aware that the course of the disease may be progressing is important during hospitalization of the patient.

Primary ciliary dyskinesia does not only influence quality of life of the patient but his or her whole family. It is very difficult for the child and his or her parents who must cope with a sick child properly to accept the disease. This creates the possibility of solving the problems of both physical nature as well as psychological and social one. The role of the nurse as a member of the therapeutic team taking direct part in preparing the child and family for self-care and self-observation in the home conditions is of key importance here. Advice, support and effective assistance in this rare disease contributes to successful therapy and allows the child and his or her family to function in the society. Nowadays, education of the patient is considered to be an important factor of therapy and care playing a considerable role in controlling chronic diseases. Education constitutes an integral part of nursing care over the patient.

Primary ciliary dyskinesia is a disease that- due to its non-specific course- takes different forms in many people. Because of difficulties in its diagnosing, children do not often attend school, which results in their individual course of learning. From an early age sick child are taught new obligations, responsibility and taking care of their health. They are surrounded with orderliness and regularity of performed actions influencing their health and well-being. They often stay in hospitals not only because of the disease but for control appointments as well. From the very beginning the patients are doomed to physiotherapeutic procedures that help to remove the secretion and improve efficiency of the respiratory system. Due to frequency of procedures during the day the children must adjust their day to procedures. Small children do not realize what is happening and often treat this as a form of playing, however, with age it is more difficult to come to terms with the fact that they must devote time for mandatory physiotherapeutic procedures that are time-consuming and have to be performed regularly. The significant role of the nurse and parents themselves is to teach the child that taking medications is a routine and not an unwanted obligation. Frequent infections, recurrent infections of the respiratory tracts, otitis contribute to frequent hospitalizations as well as result in stress and anxiety related to hospital. Due to the fact that this must be multi-specialist treatment it involves frequent control check-ups in hospitals, even when there was not exacerbation of the disease, which results in additional disorders in the sphere of contacts with peers. Consequently, children feel lonely and forgotten by the society. The chronic disease has an impact on the mentality of the child irrespective of his or her age. The main psychological problem is a lack of acceptance of disease and unwillingness for treatment, a lack of understanding why the child is ill and why this occurred to him or her. The chronic disease does not only involve physical suffering but also mental one related to separation from family and friends. The disease may impose numerous prohibitions, limitations, which may obstruct achievement of life plans. Exacerbations of the disease result in worse well-being of the child, his or her functioning causing sometimes depressive states. The role of the nurse is to make the patient aware and motivate the patient to fight against the disease and take care of his or her health. Relationship with the family that should be as close as possible in the infancy plays an important role. It can be stated that the child who was hospitalized in the infancy period for a long time has difficulties in establishing long-lasting emotional bonds and a limited capacity for empathy. Most frequently, they make friends at the departments where they something in common rather than with healthy children who they can differ from taking into account physical aspect (appearance, physical fitness, body posture) and they are often called names. Awareness of the disease itself may influence and lead to depression disorders and even complete withdrawal from treatment.

Younger children have a high self-esteem, which is reflected in their assessment of abilities and skills. Unfortunately, with age awareness of the child and knowledge that he or she stands out from the group are increasing and, consequently, his or her self-esteem is decreasing. The disease influences mentality of the child not only through emotional bonds with his or her family but through contacts with peers as well. This shapes self-esteem and self-image. Understanding and care of the surrounding has a significant impact whether negative effect will increase or will be a factor encouraging willingness to cope with the ailment and development of the child's personality.

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