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THE IMPORTANCE AND ROLE OF NURSES IN THE THERAPY OF CHILDREN WITH CYSTIC FIBROSIS

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Abstract

The aim of this paper is to present guidelines and algorithms for the care of children with cystic fibrosis to adopt new knowledge and apply it in practice to ease the lives of those affected. Cystic fibrosis, or mucoviscidosis, is a hereditary autosomal recessive disease that affects one in 2,500 to 3,500 newborns among Caucasians. Timely diagnosis and initiation of treatment are crucial. Treatment is daily and lifelong, encompassing pharmacological, nutritional, and physical measures. Physical therapy is especially important, including lung drainage exercises performed by another person or devices like vibrating vests. The nurse should encourage the child to drink age-appropriate amounts of fluids to facilitate easier expectoration. Physical therapy of the chest and a drainage position are recommended one hour before and two hours after meals. Deep breathing exercises can be performed through play, with the child being instructed. The nurse should also educate parents on conducting physical therapy and the proper nutrition of the child, as the child will require adequate care and treatment throughout their life.

Key words: *Cystic fibrosis, health care, physical therapy, diagnosis, parent education.*

Introduction

Cystic fibrosis (CF), also known as mucoviscidosis, is one of the most common genetic diseases among Caucasians, affecting between 2,500 and 3,500 newborns. The incidence of cystic fibrosis varies globally: in some parts of the UK, one in 380 newborns is affected, while in the Asian regions of the Himalayas, that figure is 1 in 90,000 (Berham, 2014). In Europe, the prevalence ranges from 1 in 3,000 live births, and the frequency of heterozygotes varies between 1 in 31 and 1 in 35. In Serbia, 318 cases have been registered, with an estimated additional 100 individuals undiagnosed or being treated under a different diagnosis [1].

Cystic fibrosis is the most common autosomal recessive inherited multisystem disease that shortens life expectancy. It is characterized by the secretion of thick and viscous mucus from all exocrine glands that produce mucus, tears, sweat, saliva, and digestive

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juices. While normal secretions are thin and slippery, in affected individuals, the defective gene causes their thick and sticky consistency. Instead of facilitating flow, these secretions can obstruct airways and ducts, particularly in the pancreas and lungs. Most children with this disease suffer from chronic lung disease, pancreatic insufficiency, and elevated chloride concentrations in sweat, which forms the well-known triad for cystic fibrosis [2, 3].

The gene responsible for this disease is located on the seventh chromosome and regulates the flow of chloride through cell membranes. This gene is called CFTR (cystic fibrosis transmembrane regulator). To date, more than 1,900 mutations have been identified, with the delta F508 mutation present in 70% of patients. The defective gene is inherited in a recessive manner, meaning that a child must inherit one copy of the gene from both parents for the disease to manifest. If a child inherits only one copy, they do not develop cystic fibrosis but may become a carrier and pass the defective gene to their offspring.

Cystic fibrosis - hereditary disease and its effects on health

Cystic fibrosis is a hereditary disease that is transmitted in an autosomal recessive manner. When both parents are carriers of the defective gene, the probability that their child will have cystic fibrosis is 25%, with a 50% chance that the child will be a carrier of the gene but healthy, and a 25% chance that the child will neither be a carrier nor affected [4-7]. The primary disorder in cystic fibrosis is the inability to retain chloride ions in the bronchi, leading to the dehydration of secretions. The secretion becomes thick and difficult to clear, which can cause obstruction of the bronchioles, creating favorable conditions for the development of infections. Similar changes occur in the pancreas, bile ducts, and vas deferens, leading to obstruction and fibrosis of these organs. The pancreas plays a crucial role in food digestion by producing enzymes that aid in the breakdown of food. In individuals with cystic fibrosis, the pancreas is often obstructed, preventing the secretion of these enzymes. The result is poor absorption of nutrients, which manifests as specific stools that may contain oil droplets and have an unpleasant odor. Symptoms of cystic fibrosis can vary at different ages. The first symptom is often meconium ileus, while later symptoms may include jaundice, poor digestion, and accelerated sweating, marked by distinctly salty sweat. In the first months of life, children with cystic fibrosis often experience digestive issues, abdominal pain, and frequent, bulky stools. As the child grows, symptoms worsen, especially in the respiratory system. Thick mucus accumulates in the lungs, making breathing difficult and leading to chronic infections. Bacteria, such as Staphylococcus aureus, are often found in the sputum. Children tire more quickly and struggle to endure physical exertion.

Cystic fibrosis is a serious disease that requires continuous treatment and monitoring. Therapies include inhaled medications, digestive enzymes, and infection prevention measures, and proper treatment can significantly improve the quality of life for those affected [8, 9].

Symptoms, diagnosis, and treatment guidelines

Cystic fibrosis is a chronic hereditary disease characterized by reduced oxygen supply to tissues, which can lead to the development of characteristic physical signs, such as clubbing fingers (curved nails resembling a glass dome). Other symptoms include nasal polyps that make breathing difficult and frequent sinus infections. Patients often suffer from chronic diarrhea, with varying degrees of malnutrition, although in treated patients, nutritional status is usually better. In some cases, there may be partial or complete intestinal obstruction due to thick stools, similar to meconium ileus in newborns, as well as the distal intestinal obstruction syndrome, a complication characteristic of cystic fibrosis that may resemble acute appendicitis [9, 10].

Individuals with milder mutations of the gene may experience recurrent pancreatitis with less pronounced disease symptoms. Additionally, this period of life often brings the first signs of liver disease without a clear cause. Reduced nutrient absorption and vitamin deficiencies lead to decreased bone mineral density, which may be a precursor to later osteoporosis. During adolescence, diabetes related to cystic fibrosis frequently occurs as a distinct form of the disease. As pulmonary disease progresses, lung function declines, resulting in difficult and rapid breathing. Coughing becomes intense and productive, with the expectoration of thick yellow-green mucus, and blood traces may often be present in the sputum (hemoptysis). Chronic respiratory infections are common, most often caused by the bacterium Pseudomonas aeruginosa, which forms a biofilm and complicates antibiotic treatment.

In adolescent patients, chronic malnutrition is often associated with a more severe form of lung disease. In addition to diabetes, cystic fibrosis can lead to the progression of liver disease towards cirrhosis, with complications such as splenomegaly, portal hypertension, and bleeding from esophageal varices. Children often experience growth delays and enter puberty later than their peers. [11, 12].

If there is a positive family history of cystic fibrosis, parents are advised to undergo prenatal diagnostics through amniocentesis or chorionic villus sampling. The aim is to detect the presence of the cystic fibrosis gene and enable early treatment. Diagnosis is based on symptoms, family history data, or positive neonatal screening.

Neonatal screening is the most reliable method for diagnosing cystic fibrosis, conducted by taking a blood sample from the newborn's heel. After the onset of symptoms, the following tests are most commonly used:

- Sweat test
- Genetic testing
- Pulmonary function tests
- Sputum culture.

The standardized sweat test using pilocarpine iontophoresis measures the chloride concentration in sweat. A normal concentration is below 40 mmol/L, while children with cystic fibrosis may have concentrations up to five times higher.

Cystic fibrosis is a chronic disease that requires lifelong therapy. The main treatment goals are to maintain adequate nutrition, prevent or treat pulmonary complications,

and preserve physical activity. Treatment includes the use of antibiotics, mucolytic agents, bronchodilators, pancreatic enzymes, and vitamin supplements. Physiotherapy and inhalation therapies are essential to alleviate respiratory issues and clear mucus from the lungs [13-15].

Physical exercises, especially drainage techniques for the lungs, as well as the use of devices such as vibrating vests, significantly improve the quality of life for patients. In the terminal phase of the disease, lung transplantation may be the only option.

Cystic fibrosis is a complex multisystem disease that requires continuous monitoring and a multidisciplinary approach to treatment. Timely diagnosis and proper therapy management can significantly improve the quality of life and extend the lifespan of affected children and young people. Care and treatment algorithms should be clearly defined and tailored to the individual needs of each patient [15-17].

Materials and methods

The research employed an indirect descriptive method (analysis of medical documentation). The guidelines and algorithms for health care were presented based on a review of literature and the knowledge of experts in the fields of pulmonology, pediatrics, and nursing. As research instruments, protocols and medical histories of patients hospitalized in the department for the examination and treatment of lung diseases at the Institute for Mother and Child Health Care Dr. Vukan Čupić in Belgrade were used. The study period covered from January 1, 2019, to April 1, 2024. The population included in the research was pediatric patients aged up to 20 years.

During the specified period at the Department for the Examination and Treatment of Lung Diseases at the Pediatric Clinic of the Institute for Mother and Child Health Care Dr. Vukan Čupić, the number of newly diagnosed children with cystic fibrosis by year was:

Table 1. Number of Newly Diagnosed Children with Cystic Fibrosis by Year

Year	Number of Affected Children
2019	11
2020	14
2021	20
2022	17
2023	25
2024	3
Total	90

Based on the data, we can conclude that cystic fibrosis is on a slight increase.

Table 2. Number of Affected Children by Age for the Period from 2019 to 2024

Age (by years of age)	Number	(%)
0-1	36	70
1-5	28	16
5-10	15	8
10-20	8	4
>20	3	2
Total	90	100

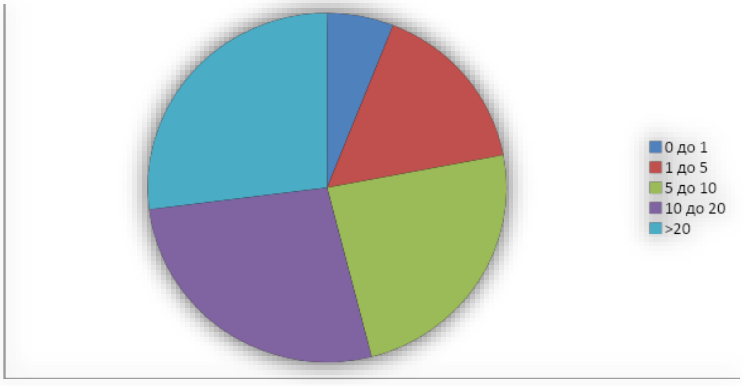

Figure 2. Representation of the Number of Affected Children by Age for the Period from 2019 to 2024

Cystic fibrosis is a hereditary disease, and it was identified in 70% of children aged up to one year. It was then found in 16% of children aged up to five years, while the number drastically declines as the children get older.

Table 3. Number of Affected Children by Gender for the Period from 2019 to 2024

Child's Gender	Number of Affected Children	%
Male	49	56
Female	41	44
Total	90	100

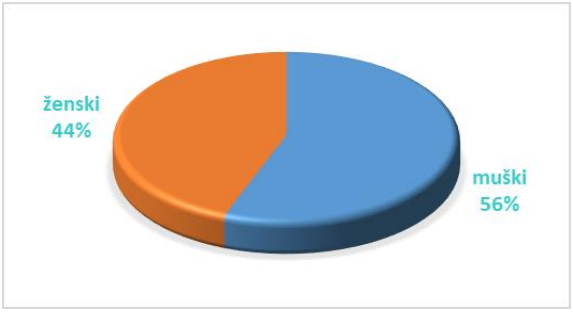

Figure 3. Representation of the Number of Affected Children by Gender for the Period from 2019 to 2024.

Cystic fibrosis is a disease that affects both boys and girls; however, there is a slight predominance of males, accounting for 56% of the total number of affected individuals, compared to females at 44%.

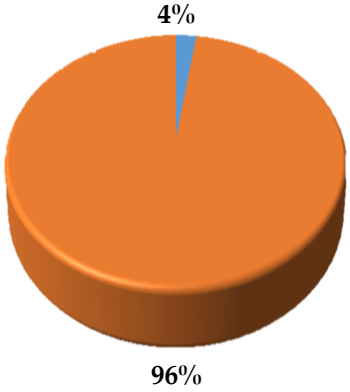

Figure 4. Representation of the Number of Fatal Outcomes for the Period from 2019 to 2024

Discussion

During the research period, a total of 90 new cases of cystic fibrosis were diagnosed and treated within the Service for the Examination and Treatment of Lung Diseases at the Pediatric Clinic of the Institute for Mother and Child Health Care "Dr. Vukan Čupić" in Belgrade. These data suggest a slight increase in the incidence of this multisystem genetic disease in the observed population. Given the hereditary nature of cystic fibrosis, the highest rate of diagnosis was recorded in children up to one year of age (70%), while 16% of cases were noted in children up to five years. With increasing age, the frequency of diagnosis significantly declines, which may be a result of early detection through neonatal screening or recognizable symptoms in early childhood [18-20]. Regarding gender distribution, although the disease equally affects both sexes, the research results show a slight predominance among boys (56%)

compared to girls (44%). These data align with previous studies indicating possible differences in clinical expression and prognosis of the disease based on gender.
During the analyzed period (2019–2024), two fatalities were recorded among children with cystic fibrosis, indicating the severity of the clinical picture and potential complications, despite advancements in diagnostic and therapeutic approaches. These results emphasize the need for continuous monitoring, improvement of therapeutic protocols, and providing adequate support to patients and their families. [20-22].

Conclusion

Nurses are an integral part of the multidisciplinary team in the treatment of children with cystic fibrosis, and their role is crucial in achieving the success of therapeutic protocols and improving the long-term prognosis of the disease. Effective treatment of this genetic disease depends on the synergistic collaboration of doctors, nurses, parents, social workers, psychologists, physical therapists, and other healthcare professionals.
The primary goals of nursing care for patients with cystic fibrosis include:

- Maintaining pulmonary function through the control of pulmonary infections and enhancing the elimination of bronchial secretions.
- Optimizing nutritional status considering malabsorption and increased energy needs of the patients.
- Improving the quality of life through individualized care and support.
- Encouraging normal growth and development, including physical, emotional, and social aspects.
- Gradually empowering the patient to manage their disease independently, thereby enhancing long-term prognosis and confidence in coping with chronic illness.

Key aspects of nursing care for affected children include:

- Early Diagnosis: Timely detection of cystic fibrosis allows for the early initiation of therapy and reduces the risk of developing serious complications. Neonatal screening is becoming a standard protocol in many countries, increasing the chances for early intervention.
- Multidisciplinary Approach: Holistic care for children with cystic fibrosis requires coordinated work among specialists (pulmonologists, gastroenterologists, nutritionists), nurses, and therapists to achieve optimal healthcare.
- Treatment of Pulmonary Complications: Effective management of pulmonary complications, including regular monitoring, the use of antibiotics, respiratory physiotherapy, and inhalation therapies, is a fundamental pillar of treatment.
- Nutrition: Children with cystic fibrosis face significant dietary challenges due to reduced nutrient absorption. An individualized nutritional approach and the use of pancreatic enzyme supplements are crucial for improving nutritional status.
- Physical Activity: Programs of physical activity and respiratory therapy positively impact the maintenance of lung function and the prevention of complications. Physical activity must be tailored to the abilities and condition of each patient.

- Psychological Support: Cystic fibrosis can have a significant psychological impact on children and their families. Continuous psychological support is necessary to ensure the emotional well-being of patients and to reduce the psychosocial stress associated with the disease.

Recommendations for Improving Healthcare:

- Parent Education: Raising awareness about prevention and early recognition of symptoms of cystic fibrosis.
- Genetic Counseling: Counseling parents before pregnancy to assess genetic predispositions for cystic fibrosis.
- Improving Diagnostics: Ensuring access to early diagnostic procedures and prompt initiation of therapy.
- Proper Nutrition: Enhancing nutritional status through education on the specific needs of children with cystic fibrosis.
- Physical Therapy: Consistent application of physical therapy to facilitate secretion clearance and maintain lung function.
- Continuous Education for Healthcare Workers: Additional training for nurses in specific healthcare needs for patients with cystic fibrosis.

Implementing these recommendations aims not only to improve therapeutic outcomes but also to enhance the long-term quality of life for patients with cystic fibrosis through a comprehensive and multidisciplinary approach to treatment.

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