International Journal of Modern Medicine
VOLUME05 ISSUE01
PUBLISHED DATE: - 31-01-2026
DOI: https://doi.org/10.55640/ijmm-05-01-05

eISSN: 2156-5198 pISSN: 2156-518X

RESEARCH ARTICLE

Analysis Of The Frequency Of Detection And Clinical Manifestations Of Cystic Fibrosis In Children

Shamsiev F.M.
Republican Specialized Scientific and Practical Medical Center for Pediatrics of the Ministry of Health of the Republic of Uzbekistan, Uzbekistan

Uzakova Sh.B.
Republican Specialized Scientific and Practical Medical Center for Pediatrics of the Ministry of Health of the Republic of Uzbekistan, Uzbekistan

Inomov B.N.
Republican Specialized Scientific and Practical Medical Center for Pediatrics of the Ministry of Health of the Republic of Uzbekistan, Uzbekistan

Azizova N.D.
Republican Specialized Scientific and Practical Medical Center for Pediatrics of the Ministry of Health of the Republic of Uzbekistan, Uzbekistan

Abdullaeva M.K.
Republican Specialized Scientific and Practical Medical Center for Pediatrics of the Ministry of Health of the Republic of Uzbekistan, Uzbekistan

Karimova M.Kh.
Republican Specialized Scientific and Practical Medical Center for Pediatrics of the Ministry of Health of the Republic of Uzbekistan, Uzbekistan

Abstract: Objective of the study. To investigate the frequency of detection and the characteristics of clinical manifestations of cystic fibrosis in children. Materials and methods. A total of 106 patients with cystic fibrosis aged from 0 months to 14 years were observed. All patients were hospitalized and treated in the departments of pulmonology, early childhood pathology, and gastroenterology. Results. In children with cystic fibrosis, the main clinical symptoms of the bronchopulmonary system were cough, dyspnea, oral rales, lethargy, and loss of appetite. Upon admission to the hospital, the general condition of most patients was assessed as severe. Conclusion. The high frequency of repeated hospitalizations and early mortality caused by severe respiratory complications indicate the need to improve programs for early detection, long-term follow-up, and multidisciplinary management of children with cystic fibrosis.

Key words: Cystic fibrosis, sweat test, children, cough, meconium ileus.

INTRODUCTION

In recent years, the hereditary disease cystic fibrosis in children with an autosomal recessive type of inheritance, caused by a pathogenic variant of the nucleotide sequence (mutation) of the cystic fibrosis transmembrane conductance regulator (CFTR) gene, has been increasingly

International Journal of Modern Medicine
VOLUME05 ISSUE01
PUBLISHED DATE: - 31-01-2026
DOI: https://doi.org/10.55640/ijmm-05-01-05

eISSN: 2156-5198 pISSN: 2156-518X

RESEARCH ARTICLE

diagnosed worldwide. According to estimates by experts from the World Health Organization (WHO), "... worldwide, 45-50 thousand children with cystic fibrosis are born annually, and the number of heterozygous carriers of the disease amounts to tens of millions..." [3]. "... The first symptoms appear in 65% of children under six months of age, 80% under one year, and 90% under two years..." [5]. To date, the increase in the number of cases, insufficient understanding of pathogenetic mechanisms, features of the clinical course, and the occurrence of severe complications of cystic fibrosis necessitate scientific research on this nosology.

The prevalence of CF varies significantly by country, with the United States being the leader in the number of CF patients, where in 2017 there were 29,887 patients; in the EU at that time, 48,204 patients were registered. The average age of CF patients in the EU is 20.8 years, in the USA — 21.7 years, and mortality from CF — at an average of 30.6 years, in the EU — 31.2 years [1]. The incidence of CF fluctuates depending on racial and geographic affiliation. The pathology is among congenital diseases, but it is worth noting a 40% increase in the development of CF in the last ten years in adults [8].

For a long time, it was believed that the mucous membranes of the respiratory tract were sterile. However, numerous studies refute this view. Of course, the lung biocoenosis cannot be compared in diversity with the intestinal microflora, but the airways are an ecological niche for a large number of microorganisms.

For patients with cystic fibrosis, monitoring the state of respiratory tract microorganisms is important for preserving lung function. The life expectancy of such patients is closely linked, first and foremost, to the preservation of respiratory function, which is seriously compromised due to impaired mucociliary clearance, leading to the development of severe chronic respiratory tract infections from an early age and the progression of respiratory failure [9].

One of the features of pathogenetic changes in the bronchopulmonary system in cystic fibrosis is the development of chronic colonization. The severity of the condition in this case is mainly associated with the presence of bacterial infection in the bronchial secretion, which requires constant monitoring of microbial composition and the use of antibiotic therapy (ABT) throughout life [7,10,11]. In early life, children's bronchopulmonary systems are colonized by gram-positive cocci, but later, gram-negative non-fermenting bacteria predominate, contributing to the development of severe forms of infection [4, 12].

OBJECTIVE OF THE STUDY

To study the frequency of detection and features of clinical manifestations in children with cystic fibrosis.

METHODS

We examined 106 patients aged from 0 months to 14 years, who were hospitalized in the departments of pulmonology, early childhood pathology, and gastroenterology, in 15 of whom the diagnosis of cystic fibrosis was confirmed by clinical examination. The CF diagnosis was verified based on comprehensive clinical and instrumental studies: based on complaints, a carefully collected history (a case of CF in the family), clinical symptoms, a positive result of neonatal screening for immunoreactive trypsin, by collecting and analyzing sweat, and mandatory radiological confirmation. Analysis of the general condition of the body was also conducted based on consultations with related specialists. The CF diagnosis was confirmed using a sweat test on MACRODUCT USA VESCOR-3700 and NANODUCT-1030 devices.

The nosological diagnosis in the observed children with CF was formed in accordance with the "International Statistical Classification of Diseases and Related Health Problems" Tenth Revision (ICD-X) [2].

RESULTS AND DISCUSSION

The frequency of detection of cystic fibrosis was studied based on a retrospective analysis of medical histories of children treated in the pulmonology departments of the RSNPMCP of the Ministry of Health of the Republic of Uzbekistan and the RMMC of Namangan region for the period 2022-2024.

International Journal of Modern Medicine
VOLUME05 ISSUE01
PUBLISHED DATE: - 31-01-2026
DOI: https://doi.org/10.55640/ijmm-05-01-05

eISSN: 2156-5198 pISSN: 2156-518X

RESEARCH ARTICLE

In 2022-2024, 28,449 children aged 0 to 18 years with bronchopulmonary pathology were hospitalized for treatment, of whom 305 children had a confirmed diagnosis of cystic fibrosis, accounting for 1.1%. Each child who returned for treatment underwent inpatient treatment from 5 to 8 times a year.

By 2024, 23 new patients (6.9%) were registered, and in 2025, 54 (16.4%) new patients were registered, which is 2.4 times more than in 2024 (Fig. 1).

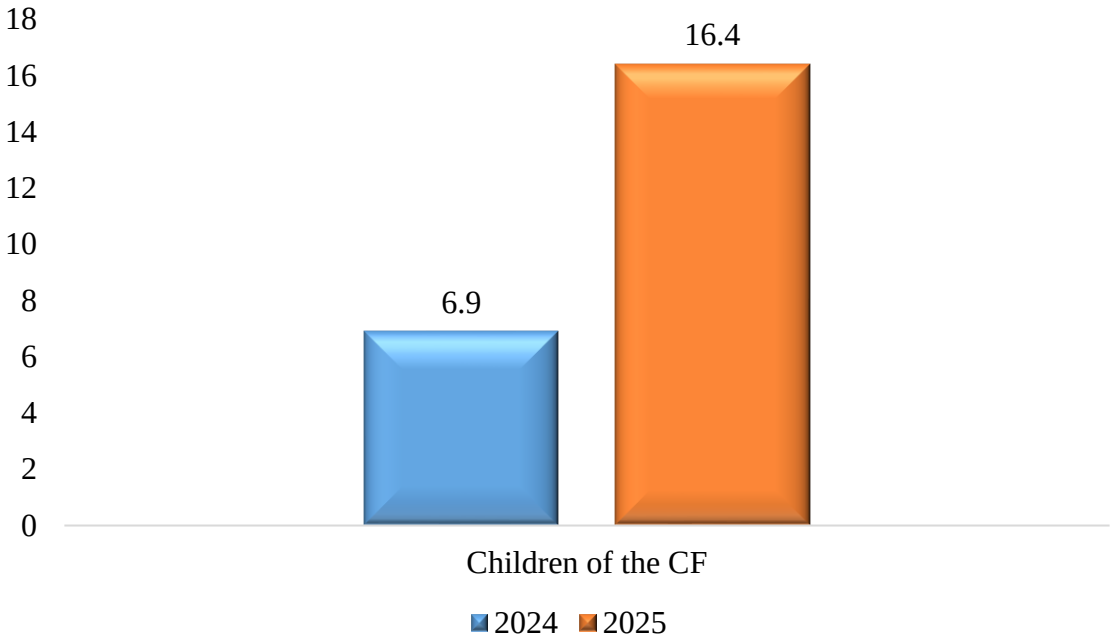

Figure 1. Newly identified children diagnosed with cystic fibrosis (%)

In 2024, 22 patients died, and in 2025, 20 patients died. The cause of death was the presence of multiple pulmonary bullae, respiratory failure developed against the background of microbial-inflammatory damage to the respiratory system. The age at death was 1 and 2 years, respectively. Of all patients, 9 (2.7%) children with CF receive treatment outside the Republic of Uzbekistan. In the first year of life, the CF diagnosis was first established in 32 children (9.7%), in 13 (3.9%) children at the time of diagnosis they were 2 years old, at 3 years old in 3 children (0.9%), at 7 years old in one child, and at 14 years old the diagnosis was made in 2 (0.6%) children (Fig. 2).

The next stage of our present research is the results of observations and examination from a prospective analysis of 106 children with cystic fibrosis of a mixed nature.

International Journal of Modern Medicine
VOLUME05 ISSUE01
PUBLISHED DATE: - 31-01-2026
DOI: https://doi.org/10.55640/ijmm-05-01-05

eISSN: 2156-5198 pISSN: 2156-518X

RESEARCH ARTICLE

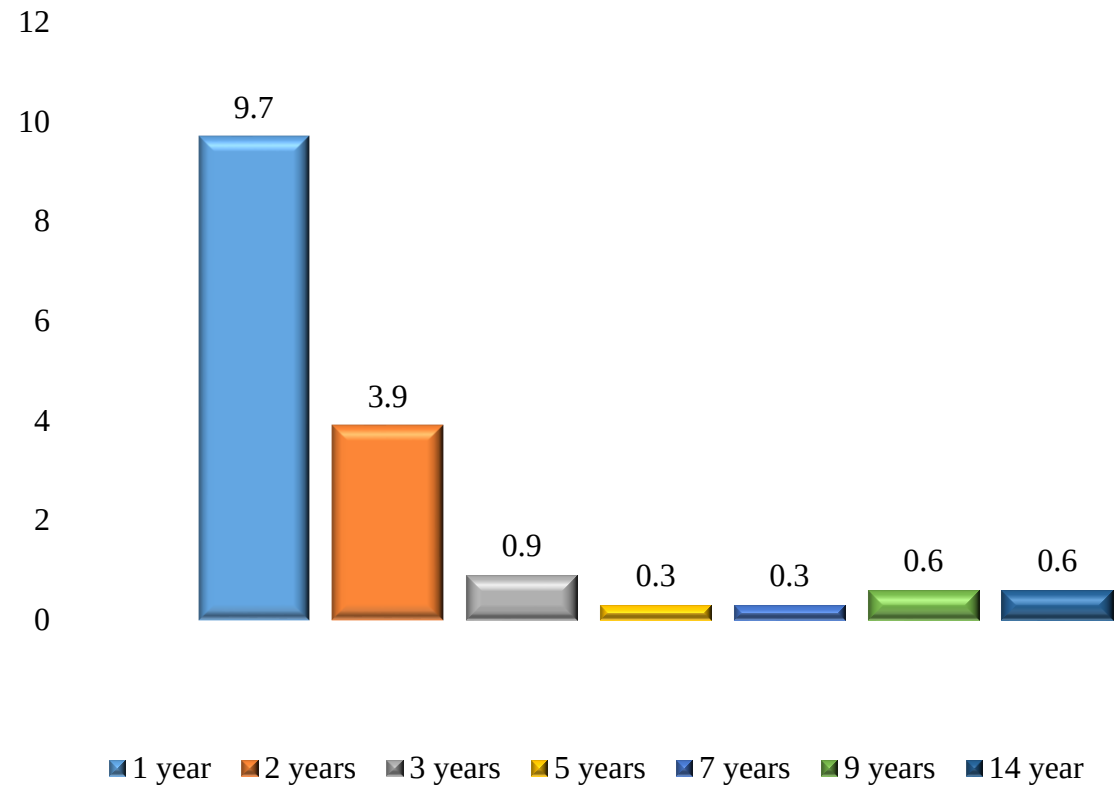

Figure 2. Age of children at the time of diagnosis establishment (%)

The age of children with CF ranged from 1 month to 14 years. When interviewing mothers of children suffering from CF, it was revealed that the course of pregnancy and childbirth did not differ from average population indicators. To verify the CF diagnosis according to the classification used by WHO, the International Cystic Fibrosis Association, and the European Cystic Fibrosis Association, all patients underwent a sweat conductivity test.

Sweat test results were collected from the pulmonology and gastroenterology departments. When studying chlorides in sweat in children with CF, the results were positive in 91 (85.8%) patients; the lowest positive result was 80 mmol/L and the highest result was 149.0 mmol/L. The average sweat chloride content during the sweat test in patients with cystic fibrosis was --- 115.23 ± 1.25 mmol/L. For borderline results, a genetic test of the CFTR gene was additionally used to verify the CF diagnosis.

Children with CF were characterized by disturbances in nutritional status, especially body weight. The height of the patients was at the lower limit of the age norm. In addition, a decrease in the proportionality index was noted in 81 (76.8%) patients, trophic skin changes, signs of pronounced asthenoneurotic disorders with a predominance of asthenia symptoms (increased fatigue, weakness) — in 71 (66.7%), manifestations of intoxication — in 69 (65.2%) children.

From the data presented, among 106 children, standard deviations corresponding to body weight deficiency were established in 94 (88.2%). In 56.5% of children (53/94), the range of standard deviations corresponded to (-) 1 SD — (-) 2 SD, i.e., this group of children had body weight deficiency, and in 43.5% of children (41/94), the range of standard deviations corresponded to (-) 2 SD — (-) 3 SD, which corresponded to pronounced deficiency.

In the neonatal period, clinical manifestations of CF were:

International Journal of Modern Medicine
VOLUME05 ISSUE01
PUBLISHED DATE: - 31-01-2026
DOI: https://doi.org/10.55640/ijmm-05-01-05

eISSN: 2156-5198 pISSN: 2156-518X

RESEARCH ARTICLE

meconium ileus — in 5 (4.9%) children, prolonged jaundice — in 19 (18.0%), low birth weight — in 5 (4.9%) children. Paroxysmal cough with difficult-to-expectorate purulent sputum, chronic sinusitis, obstructive bronchitis, pneumonia, atelectasis, manifestations of respiratory failure, pancreatic insufficiency, enlargement of the liver and spleen, and other clinical symptoms manifested in later periods of the patient's life. It was established that signs of pneumofibrosis — in 50 (47.5%), chronic obstructive bronchitis — in 106 (100%) and as a consequence of the chronic bronchopulmonary process various chest deformities — in 9 (8.1%), cough from rare dry to persistent with difficult-to-expectorate sputum — in 106 (100%) children. Meconium ileus was experienced by 2 (1.67%) children; in other cases, parents were concerned about frequent fatty stools and poor weight gain. The respiratory syndrome manifested later, at three months, and was characterized by a constant cough. The CF diagnosis in children was established in the vast majority of cases in the first year of life based on positive sweat test results and a typical clinical picture of the disease.

CONCLUSIONS

According to retrospective analysis data for 2022–2024, cystic fibrosis was detected in 1.1% of children hospitalized with bronchopulmonary pathology, which indicates the continuing relevance of this hereditary pathology in the structure of childhood morbidity.

An increase in the number of newly identified cases of cystic fibrosis was noted: in 2025, the number of new patients increased by 2.4 times compared to 2024, which is associated with improved diagnosis of cystic fibrosis in the Republic.

The high frequency of repeated hospitalizations and early mortality, caused by severe complications from the respiratory system, indicate the need to improve programs for early detection, long-term follow-up, and multidisciplinary management of children with cystic fibrosis.

REFERENCES

1. Voronkova A.Yu., Amelina E.L., Kashirskaya N.Yu., Kondratyeva E.I., Krasovsky S.A., Starinova M.A., Kapranov N.I. /Register of patients with cystic fibrosis in the Russian Federation. - M.: ID "MEDPRAKTIKA-M", 2019.-68p.
2. Geppe N.A., Rozinova N.N., Volkov I.K., Mizernitsky Yu.L. Working classification of the main clinical forms of bronchopulmonary diseases in children//Trudny patsient. -2009. -Volume 7. -No.1-2. -P.35-40.
3. Kachiyan M.M. Clinical and genetic characteristics of children and adolescents with cystic fibrosis in the Krasnodar region and assessment of the role of staphylococcal infection in cystic fibrosis 2021.-140p.
4. Kondratyeva E.I., Krasovsky S.A., Voronkova A.Yu., Amelina E.L., Chernyak A.V., Kashirskaya N.Yu. Register of patients with cystic fibrosis in the Russian Federation. //M.: "Medpraktika-M", 2017. P. 70.
5. Simonova O.I. et al., Cystic Fibrosis: a guide for doctors Moscow: Pediatr, 2018. - 84 p.
6. Skudarnov E.V., Dorokhov N.A., Malyuga O.M., Seroklinov V.M. "Social and genetic aspects of community-acquired pneumonia in children"//Pediatricheskiy vestnik Yuzhnogo Urala. --2017.--No.1.-- P.67-70.
7. Shaginyan I.A., Avetisyan L.R., Chernukha M.Yu., Siyanova E.A., Burmistrov E.M., Voronkova A.Yu. et al. Epidemiological significance of molecular variability of the genome of Pseudomonas aeruginosa isolates causing chronic lung infection in patients with cystic fibrosis. //Klinicheskaya mikrobiologiya i antimikrobnaya khimioterapiya. 2019;21(4):340--351.
8. Yashin S.S., Yunusova Yu.R., Isakova N.V., Serdobolskaya Yu.V. CYSTIC FIBROSIS (MUCOVISCIDOSIS) // Sovremennye problemy nauki i obrazovaniya. -- 2022. -- No. 5. P. 236-247.
9. Caverly L.J., LiPuma J.J. /Cystic fibrosis respiratory microbiota: unraveling complexity to inform clinical practice. // Expert Review of Respiratory Medicine. 2018;12 (10): 857-865.

International Journal of Modern Medicine
VOLUME05 ISSUE01
PUBLISHED DATE: - 31-01-2026
DOI: https://doi.org/10.55640/ijmm-05-01-05

eISSN: 2156-5198 pISSN: 2156-518X

RESEARCH ARTICLE

10. Middleton P.G., Taylor-Cousar J.L. Development of Elexacaftor -- Tezacaftor -- Ivacaftor: Highly Effective CFTR Modulation for the Majority of People with Cystic Fibrosis//Expert Rev Respir Med. 2021;15(6):723--735.

11. Paterson S.L., Barry P.J., Horsley A.R. Tezacaftor and Ivacaftor for the Treatment of Cystic Fibrosis//Expert Rev Respir Med. 2020;14(1):15--30.

12. 12. Uzakova, Sh.B., Inomov, B.N., & Abdullaeva, M.K. (2024). Analysis of clinical manifestations and laboratory parameters of cystic fibrosis in children. Bulletin of the Tashkent Medical Academy, 1, 149-151.