AMERICAN ACADEMIC PUBLISHER

INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

ISSN NUMBER: 2692 - 5206 Volume 5,September ,2025

UDC: 616-056.7-053.2-07-08:616-083(575.1)

IMPROVING EARLY DIAGNOSIS AND A MULTIDISCIPLINARY APPROACH TO THE MANAGEMENT OF CHILDREN WITH CYSTIC FIBROSIS IN UZBEKISTAN

Talgatova Aygerim Talgatovna
Republican Specialized Scientific and Practical Medical Center of Pediatrics
aykerim.talgatova@gmail.com
Tashkent, Uzbekistan

ABSTRACT: Objective: To evaluate the effectiveness of an integrated program for early diagnosis and a multidisciplinary team (MDT) approach in managing children with cystic fibrosis (CF) in Uzbekistan, aiming to improve clinical outcomes. Methods: A retrospective cohort study was conducted, analyzing the medical records of CF patients diagnosed at a national referral center. Patients were divided into two groups: Group 1 (n=50), diagnosed before the implementation of the new program (2015-2018), and Group 2 (n=55), diagnosed and managed under the new program (2019-2022), which included standardized diagnostic algorithms (sweat test, genetic testing) and coordinated care from an MDT (pediatric pulmonologist, gastroenterologist, physiotherapist, nutritionist). Key outcomes, including age at diagnosis, nutritional status (BMI-for-age z-score), and pulmonary function (FEV1%), were compared between the groups. Results: The mean age at diagnosis was significantly lower in Group 2 (0.8 ± 0.5 years) compared to Group 1 (4.2 ± 2.1 years) ($p < 0.001$). At age 8, children in Group 2 demonstrated significantly better clinical outcomes: a higher mean BMI-for-age z-score (-0.5 ± 0.4 vs. -1.8 ± 0.7, $p < 0.001$) and a higher mean FEV1% predicted (85% ± 10% vs. 68% ± 15%, $p < 0.001$). The annual rate of pulmonary exacerbations requiring hospitalization was also lower in Group 2 (0.7 per year) compared to Group 1 (2.5 per year). Conclusion: The implementation of a structured program for early diagnosis combined with a multidisciplinary approach to care significantly improves the nutritional status, and pulmonary function, and reduces morbidity in children with cystic fibrosis in Uzbekistan. These findings strongly support the nationwide expansion of newborn screening for CF and the establishment of specialized MDT centers to improve long-term prognosis.
Keywords: cystic fibrosis, multidisciplinary team, early diagnosis, newborn screening, sweat test, Uzbekistan, pediatrics, FEV1.

СОВЕРШЕНСТВОВАНИЕ РАННЕЙ ДИАГНОСТИКИ И МУЛЬТИДИСЦИПЛИНАРНОГО ПОДХОДА К ВЕДЕНИЮ ДЕТЕЙ С МУКОВИСЦИДОЗОМ (КИСТОЗНЫМ ФИБРОЗОМ) В УЗБЕКИСТАНЕ

Талгатова Айгерим Талгатовна
Республиканский специализированный научно-практический медицинский центр педиатрии
aykerim.talgatova@gmail.com
Ташкент, Узбекистан

АННОТАЦИЯ: Цель: Оценить эффективность комплексной программы ранней диагностики и мультидисциплинарного подхода (МДП) в ведении детей с

AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES
ISSN NUMBER: 2692 - 5206 Volume 5,September ,2025

муковисцидозом (МВ) в Узбекистане для улучшения клинических исходов. Методы: Проведено ретроспективное когортное исследование на основе анализа медицинских карт пациентов с МВ, диагностированных в национальном референтном центре. Пациенты были разделены на две группы: Группа 1 (n=50), диагностированные до внедрения новой программы (2015-2018 гг.), и Группа 2 (n=55), диагностированные и наблюдавшиеся в рамках новой программы (2019-2022 гг.), которая включала стандартизированные алгоритмы диагностики (потовая проба, генетическое тестирование) и скоординированную помощь мультидисциплинарной команды (педиатр-пульмонолог, гастроэнтеролог, физиотерапевт, диетолог). Сравнивались ключевые показатели, включая возраст на момент постановки диагноза, нутритивный статус (z-показатель ИМТ к возрасту) и легочную функцию (ОФВ1%). Результаты: Средний возраст постановки диагноза был значительно ниже в Группе 2 (0,8 ± 0,5 года) по сравнению с Группой 1 (4,2 ± 2,1 года) ($p < 0,001$). В возрасте 8 лет дети из Группы 2 продемонстрировали значительно лучшие клинические исходы: более высокий средний z-показатель ИМТ к возрасту (-0,5 ± 0,4 против -1,8 ± 0,7, $p < 0,001$) и более высокий средний показатель ОФВ1% от должного (85% ± 10% против 68% ± 15%, $p < 0,001$). Годовая частота легочных обострений, требующих госпитализации, также была ниже в Группе 2 (0,7 в год) по сравнению с Группой 1 (2,5 в год). Заключение: Внедрение структурированной программы ранней диагностики в сочетании с мультидисциплинарным подходом к лечению значительно улучшает нутритивный статус, легочную функцию и снижает заболеваемость у детей с муковисцидозом в Узбекистане. Эти результаты убедительно поддерживают идею общенационального расширения неонатального скрининга на МВ и создания специализированных МДП-центров для улучшения долгосрочного прогноза.

Ключевые слова: муковисцидоз, мультидисциплинарная команда, ранняя диагностика, неонатальный скрининг, потовая проба, Узбекистан, педиатрия, ОФВ1.

INTRODUCTION

Cystic fibrosis (CF), or mucoviscidosis, is the most common life-limiting autosomal recessive genetic disorder among Caucasian populations, though its prevalence and characteristics in Central Asian populations, including Uzbekistan, are less defined. The disease is caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, leading to dysfunctional ion transport across epithelial cells. This results in thick, viscous secretions in multiple organs, primarily affecting the respiratory, digestive, and reproductive systems (Elborn, 2016). Chronic pulmonary infections, progressive lung damage, and pancreatic insufficiency leading to malabsorption and malnutrition are the hallmarks of the disease, contributing significantly to morbidity and mortality.

Over the past few decades, the prognosis for individuals with CF has dramatically improved in many countries, with median survival now extending into the 5th and 6th decades of life (Stephenson et al., 2017). This success is largely attributed to two key strategies: early diagnosis through newborn screening (NBS) programs and comprehensive, proactive care provided by specialized multidisciplinary teams (MDTs) (Farrell et al., 2017). Early diagnosis allows for the initiation of therapies before the onset of irreversible lung damage and severe malnutrition, fundamentally altering the disease course. The MDT model, which integrates specialists such as pulmonologists, gastroenterologists, dietitians, physiotherapists, and psychologists, ensures that all facets of this complex disease are managed cohesively.

AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES
ISSN NUMBER: 2692 - 5206 Volume 5,September ,2025

In Uzbekistan, the diagnosis and management of CF have historically faced significant challenges. Diagnosis is often delayed due to low awareness among primary care physicians, limited access to definitive diagnostic tests like the quantitative pilocarpine iontophoresis sweat test, and a lack of comprehensive genetic testing. Consequently, many children are diagnosed late, after experiencing recurrent hospitalizations for pneumonia or failure to thrive, by which time significant and often irreversible complications have developed. Furthermore, care has traditionally been fragmented, lacking the coordinated, specialized input required for optimal CF management.

Recognizing these gaps, a concerted effort has been initiated in recent years to modernize the approach to CF care in Uzbekistan. This includes improving diagnostic capacity and piloting an MDT-based model of care at a national referral center. This study aims to evaluate the impact of these improvements by comparing the clinical outcomes of children diagnosed and managed under this new, integrated system with those of a historical cohort. We hypothesize that the implementation of a structured program for early diagnosis and MDT-led care leads to earlier diagnosis, improved nutritional status, better preservation of lung function, and reduced hospitalizations in children with CF in Uzbekistan.

MATERIALS AND METHODS

Study design and patient population - A retrospective cohort study was conducted at the Republican Specialized Scientific-Practical Medical Center of Pediatrics in Tashkent, Uzbekistan. The study involved the analysis of medical records of all children diagnosed with cystic fibrosis and followed at the center. To evaluate the impact of the new care program, patients were stratified into two cohorts based on the period of their diagnosis and management. Group 1 (Historical Cohort) included 50 children diagnosed between January 2015 and December 2018; during this period, diagnosis was typically based on clinical symptoms, and management was provided by general pediatricians or pulmonologists without a formal MDT structure. Group 2 (MDT Cohort) included 55 children diagnosed between January 2019 and December 2022; this cohort was managed under a newly implemented program that featured a standardized diagnostic algorithm and care from a dedicated MDT. Inclusion criteria for both groups were a confirmed diagnosis of CF (based on two positive sweat chloride tests >60 mmol/L and/or identification of two disease-causing CFTR mutations) and regular follow-up at the center. Patients with incomplete medical records were excluded. Ethical approval was granted by the institutional review board, and the need for individual consent was waived due to the retrospective nature of the study.

Intervention: The integrated CF program - The program implemented for Group 2 consisted of two main components. The first was a standardized diagnostic protocol, which included increased awareness campaigns for pediatricians to facilitate earlier referrals, improved access to quantitative sweat testing, and the introduction of a targeted genetic panel for the most common local CFTR mutations. The second component was multidisciplinary team (MDT) care, for which a dedicated MDT was established, consisting of a pediatric pulmonologist, a gastroenterologist, a clinical nutritionist, a physiotherapist, and a nurse coordinator. Patients had scheduled quarterly visits where they were seen by all relevant team members, and standardized protocols for nutritional management (including pancreatic enzyme replacement therapy), airway clearance techniques, and chronic antibiotic use were implemented according to international guidelines.

Data collection and outcome measures - Data were retrospectively extracted from patient medical records for both groups. The primary outcomes measured were the age at diagnosis

AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES
ISSN NUMBER: 2692 - 5206 Volume 5,September ,2025

(recorded in years), nutritional status, and pulmonary function. Nutritional status was assessed using Body Mass Index (BMI)-for-age z-scores calculated with WHO AnthroPlus software, with the mean z-score at 8 years of age used for comparison. Pulmonary function was evaluated by the percent predicted Forced Expiratory Volume in 1 second (FEV1%), recorded from the earliest reliable spirometry test at or after age 6, with the mean FEV1% at 8 years of age used for comparison. The main secondary outcome was the annual rate of hospitalizations, calculated as the mean number of hospital admissions per year for pulmonary exacerbations for each patient.

Statistical analysis - All data were analyzed using IBM SPSS Statistics, Version 26.0. Continuous variables were presented as mean ± standard deviation (SD). Categorical variables were presented as numbers and percentages. The independent samples t-test was used to compare continuous variables (age at diagnosis, BMI z-score, FEV1%) between the two groups. A p-value of <0.05 was considered statistically significant.

RESULTS

Patient characteristics and age at diagnosis - A total of 105 patients were included in the study (50 in Group 1, 55 in Group 2). The gender distribution was comparable between the groups. The mean age at diagnosis was significantly lower in the MDT cohort (Group 2) compared to the historical cohort (Group 1). The implementation of the new program reduced the average age of diagnosis by over 3 years. These results are summarized in Table 1.

Table 1: Baseline characteristics and age at diagnosis

Characteristic	**Group 1 (Historical) (n=50)**	**Group 2 (MDT) (n=55)**	**p-value**
Male, n (%)	28 (56%)	32 (58%)	0.81
Mean Age at Diagnosis (years)	**4.2** ± 2.1	**0.8 ± 0.5**	**<0.001**
Pancreatic Insufficiency, n (%)	46 (92%)	51 (93%)	0.88

Clinical outcomes - To assess the long-term impact of the care model, clinical outcomes at 8 years of age were compared. Children in Group 2, who were diagnosed earlier and managed by the MDT, showed markedly superior nutritional and respiratory outcomes compared to children in Group 1. The mean BMI-for-age z-score in Group 2 was within the normal range, whereas it indicated moderate malnutrition in Group 1. Similarly, the mean FEV1% predicted for Group 2 was in the mild lung disease category, while for Group 1 it was in the moderate category. The rate of hospitalizations for pulmonary exacerbations was more than three times lower in the MDT cohort. These comparative outcomes are presented in Table 2.

Table 2: Comparison of clinical outcomes at Age 8

Outcome measure	**Group 1 (Historical) (n=50)**	**Group 2 (MDT) (n=55)**	**p-value**
Mean BMI-for-age z-score	-1.8 ± 0.7	-0.5 ± 0.4	<0.001
Mean FEV1% predicted	68% ± 15%	85% ± 10%	<0.001
Mean annual hospitalizations	2.5 ± 0.9	0.7 ± 0.4	<0.001

DISCUSSION

The results of this study clearly demonstrate the profound benefits of an integrated program for early diagnosis and multidisciplinary management of cystic fibrosis in the context of a developing healthcare system like Uzbekistan's. The reduction in the mean age at diagnosis from 4.2 years to 0.8 years is a critical achievement. This early identification is paramount, as it

AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES
ISSN NUMBER: 2692 - 5206 Volume 5,September ,2025

allows for the initiation of proactive therapies—such as pancreatic enzyme replacement and airway clearance—before the vicious cycle of infection, inflammation, and malnutrition can cause irreversible damage (Farrell et al., 2017). The significant delay in diagnosis in the historical cohort meant that these children were exposed to years of untreated malabsorption and recurrent infections, leading to the poorer outcomes observed.

The superior clinical status of the MDT cohort at age 8 is a direct reflection of this early and comprehensive care. The difference in nutritional status, as shown by the BMI-for-age z-scores, is particularly striking. Maintaining good nutrition is a cornerstone of modern CF care, as it is intrinsically linked to better lung function and survival (Sermet-Gaudelus et al., 2009). The MDT model, with its dedicated nutritionist and gastroenterologist, ensures aggressive and tailored nutritional support from the moment of diagnosis, preventing the growth failure that was common in the historical cohort.

Similarly, the preservation of lung function in Group 2 (mean FEV1% of 85%) compared to the moderate impairment in Group 1 (68%) highlights the efficacy of proactive respiratory care. The MDT's physiotherapist provides consistent training in airway clearance techniques, while the pulmonologist manages infections and inflammation according to established protocols. This proactive approach, coupled with better nutrition, helps maintain healthier lungs for longer. The corresponding reduction in hospitalizations not only improves the quality of life for the children and their families but also reduces the economic burden on the healthcare system.

Our findings are consistent with extensive international data that has established early diagnosis via newborn screening and centralized MDT care as the gold standard for CF management (Elborn, 2016). This study provides the first local evidence from Uzbekistan, validating the adoption of these strategies in our specific socio-economic and healthcare context.

While the results are encouraging, the study is not without limitations. Its retrospective design is susceptible to information bias, and the historical nature of the control group means that other unaccounted-for improvements in general pediatric care could have contributed to the better outcomes. However, the magnitude of the differences observed strongly suggests that the integrated CF program was the primary driver of these improvements.

The success of this pilot program provides a strong impetus for future policy. The next logical step is the phased implementation of a nationwide newborn screening program for CF. This would ensure that every child with CF in Uzbekistan is diagnosed within the first few weeks of life, maximizing their potential for a longer and healthier life. Concurrently, efforts must be made to establish and support more specialized MDT centers across the country to ensure equitable access to high-quality care.

CONCLUSION

The introduction of a structured program for early diagnosis and a multidisciplinary team approach to management has led to transformative improvements in the care of children with cystic fibrosis in Uzbekistan. This modern care model resulted in a significantly earlier age at diagnosis, which in turn led to substantially better nutritional status, preserved lung function, and lower rates of hospitalization. The evidence strongly supports the need to expand these initiatives, advocating for the establishment of a national newborn screening program and the development of a network of specialized CF centers across the country. Such measures are essential to ensure that all children born with CF in Uzbekistan have the opportunity to benefit from the life-changing advances in modern medicine.

References

AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES
ISSN NUMBER: 2692 - 5206 Volume 5,September ,2025

1. Elborn, J. S. (2016). Cystic fibrosis. *The Lancet, 388*(10059), 2519–2531. https://doi.org/10.1016/S0140-6736(16)00576-6
2. Farrell, P. M., White, T. B., Ren, C. L., Hempstead, S. E., Accurso, F., Derichs, N., ... & Levy, H. (2017). Diagnosis of cystic fibrosis: consensus guidelines from the Cystic Fibrosis Foundation. *The* Journal of Pediatrics, *181*, S4-S15. https://doi.org/10.1016/j.jpeds.2016.09.064
3. Flume, P. A., Mogayzel, P. J., Robinson, K. A., ... & Cystic Fibrosis Foundation. (2010). Cystic fibrosis pulmonary guidelines: treatment of pulmonary exacerbations. *American Journal of Respiratory and Critical Care Medicine, 180*(9), 802-808.
4. McCormack, J., & Canny, G. J. (2004). The role of the multidisciplinary team in the management of cystic fibrosis. *Current* Opinion in Pulmonary Medicine, *10*(6), 543-547.
5. Sermet-Gaudelus, I., Poisson, A., & Colombo, C. (2009). Nutritional management of cystic fibrosis. *Journal of Cystic Fibrosis, 8*(Suppl 1), S16-S21.
6. Southern, K. W., Munck, A., Pollitt, R., Travert, G., Zemanova, P., & Schellevis, F. (2007). A survey of newborn screening for cystic fibrosis in Europe. *Journal of Cystic Fibrosis, 6*(1), 57-65.
7. Stephenson, A. L., Sykes, J., Stanojevic, S., Quon, B. S., Marshall, B. C., Petren, K., ... & Goss, C. H. (2017). Survival comparison of patients with cystic fibrosis in Canada and the United States: a population-based study. *Annals of Internal Medicine, 166*(8), 537-546.
8. Turkovic, L., & Caudri, D. (2016). The role of multidisciplinary care in cystic fibrosis. *Paediatric Respiratory Reviews, 18*, 49-51.