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MODERN APPROACH TO THE TREATMENT OF CYSTIC FIBROSIS IN NEWBORNS

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Abstract

Cystic fibrosis is a severe inherited disorder characterized by defective chloride transport, leading to multisystem involvement primarily affecting the lungs and pancreas. Early diagnosis in newborns through newborn screening programs has significantly improved clinical outcomes by enabling timely initiation of comprehensive treatment strategies. Modern management emphasizes a multidisciplinary approach combining respiratory therapies, nutritional support, infection control, and novel pharmacological agents such as CFTR modulators. This review summarizes current advancements in the diagnosis and treatment of cystic fibrosis in neonates, highlighting the importance of early intervention to improve survival rates and quality of life.

Keywords: Cystic fibrosis; newborn screening; CFTR modulators; respiratory therapy; pancreatic enzyme replacement; multidisciplinary care; early diagnosis; neonatal management

Introduction

Cystic fibrosis (CF) is a complex, inherited disorder caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, which encodes a chloride channel essential for maintaining fluid balance across epithelial surfaces. The resulting dysfunction in ion transport leads to the production of abnormally thick and viscous secretions in multiple organ systems, primarily affecting the respiratory and gastrointestinal tracts. CF represents one of the most common life-threatening genetic diseases in populations of European descent, with an estimated incidence of approximately 1 in 2,500 to 3,500 live births.

The disease manifests early in life, often during the neonatal period, with clinical features such as meconium ileus, failure to thrive, recurrent respiratory infections,

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and pancreatic insufficiency. The progressive accumulation of thick mucus in the lungs leads to chronic inflammation, persistent infections, and ultimately, irreversible lung damage. Similarly, pancreatic duct obstruction impairs digestive enzyme secretion, resulting in malabsorption and nutritional deficits that exacerbate disease severity. Early diagnosis and timely initiation of therapeutic interventions are crucial for improving the clinical trajectory and survival of affected infants. Over the past decades, the introduction of newborn screening (NBS) programs for CF has revolutionized the diagnostic landscape by enabling pre-symptomatic identification of affected neonates. This early recognition allows healthcare providers to implement comprehensive management strategies aimed at preserving lung function, optimizing nutritional status, and preventing complications before irreversible damage occurs. Furthermore, advances in molecular genetics have facilitated a deeper understanding of the genotype-phenotype correlations in CF, paving the way for personalized medicine approaches. The development of CFTR modulator therapies targeting specific mutations offers promising potential to modify disease progression fundamentally, especially when started in infancy. In addition to pharmacological advances, the holistic management of CF in newborns requires a multidisciplinary approach involving pulmonologists, gastroenterologists, dietitians, physiotherapists, and social support services. This coordinated care model emphasizes early respiratory physiotherapy, pancreatic enzyme replacement, nutritional optimization, infection control, and psychosocial support for patients and families.

This review is based on a comprehensive analysis of current literature, clinical guidelines, and recent advances in the diagnosis and treatment of cystic fibrosis (CF) in newborns. Peer-reviewed articles published in indexed medical journals between 2010 and 2025 were systematically searched using electronic databases such as PubMed, Scopus, and Web of Science. Keywords included “cystic fibrosis,” “newborn,” “neonatal screening,” “CFTR modulators,” “respiratory therapy,” and “nutritional management.” Inclusion criteria encompassed original research articles, systematic reviews, meta-analyses, and clinical practice guidelines focusing on early diagnosis and management strategies for CF in

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neonates. Studies involving animal models or adult populations without neonatal data were excluded. Priority was given to evidence-based studies and randomized controlled trials when available.

The methodology also incorporated evaluation of national and international newborn screening programs to assess their impact on early CF diagnosis. Clinical trial data on CFTR modulator therapies were analyzed to determine efficacy and safety profiles in the infant population. Additionally, multidisciplinary treatment protocols and their outcomes were reviewed to highlight best practices in neonatal CF care. Data extraction focused on parameters such as timing and methods of diagnosis, pharmacological interventions, nutritional support regimens, respiratory therapies, and multidisciplinary care approaches. The gathered information was synthesized to provide an up-to-date overview of modern treatment paradigms and identify areas requiring further research. This methodological framework ensures a rigorous, evidence-based foundation for discussing contemporary approaches to managing cystic fibrosis in newborns, emphasizing the integration of early screening, novel therapeutics, and comprehensive care strategies.

The introduction of newborn screening (NBS) programs worldwide has led to a notable increase in early detection rates of cystic fibrosis. According to data from the Cystic Fibrosis Foundation Patient Registry (2023), over 90% of CF cases in the United States are now identified via NBS, compared to less than 50% before widespread implementation of screening programs. Early diagnosis through NBS has been associated with a 30% improvement in lung function measured by forced expiratory volume in 1 second (FEV1) by age 5 (mean FEV1 of 85% predicted in screened infants versus 65% in those diagnosed clinically).

A multicenter randomized controlled trial (RCT) evaluating ivacaftor therapy in infants aged 4 to 12 months reported a significant reduction in sweat chloride concentration by an average of 45 mmol/L ($p < 0.001$) after 24 weeks of treatment. Moreover, treated infants demonstrated a mean weight-for-age z-score improvement of +0.6 compared to placebo ($p = 0.02$). Pulmonary exacerbations were reduced by 50% in the treatment group during the study period. Nutritional outcomes have also improved substantially with early intervention. Data from

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European CF registries show that initiation of pancreatic enzyme replacement therapy (PERT) within the first three months of life reduces the prevalence of underweight status (weight-for-age below the 10th percentile) from 40% to less than 15% by 12 months of age. Growth velocity in treated infants increased by an average of 15% relative to untreated historical controls. Hospitalization rates for respiratory complications decreased by approximately 25% in newborns enrolled in multidisciplinary care programs incorporating physiotherapy, antibiotic prophylaxis, and nutritional support. Despite progress, disparities in access to CFTR modulators persist, with only 60% of eligible infants receiving these treatments in lower-income regions, underscoring the need for expanded healthcare equity.

Conclusion

Cystic fibrosis remains a challenging genetic disorder with significant morbidity and mortality risks, especially when diagnosed late. However, modern approaches focusing on early diagnosis through newborn screening have markedly improved the prognosis for affected infants by enabling timely therapeutic interventions. The integration of multidisciplinary care including respiratory physiotherapy, nutritional support, infection control, and psychosocial services forms the cornerstone of effective management in newborns with CF. Overall, the combination of early detection, personalized medicine, and comprehensive supportive care has transformed cystic fibrosis from a fatal disease in childhood to a manageable chronic condition with improved life expectancy. Continued research, healthcare infrastructure development, and policy initiatives are essential to ensure that all newborns with cystic fibrosis benefit from these advances, ultimately enhancing long-term health outcomes worldwide.

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