Living with Cystic Fibrosis

A Complete Guide for Patients, Caregivers & the Community

Developed by the Americas Health Foundation (AHF) with an unrestricted grant from Vertex.

This guide is for educational and self-learning purposes only and is not a substitute for professional medical advice, diagnosis, or treatment. Always consult qualified healthcare professionals regarding any questions or decisions about cystic fibrosis care.

What This Guide Covers

How to Use This Guide

This guide is designed as a standalone resource; it does not require internet access. Keep it nearby as a reference for CF management, drawn from expert input. Complete the personal templates with your care team and share relevant sections with your school, workplace, and family.

SECTION 1: UNDERSTANDING CYSTIC FIBROSIS

What Is Cystic Fibrosis?

Cystic Fibrosis (CF), also called mucoviscidosis, is a chronic hereditary disease caused by mutations in the CFTR gene. This gene produces a protein that regulates the movement of salt and water in cells. When the protein is defective, mucus up to 30 to 60 times thicker than normal accumulates in the lungs, pancreas, liver, and digestive system, causing infections, inflammation, and progressive organ damage.

Cause: CF is autosomal recessive: both parents must carry a mutated CFTR gene for a child to have CF. If both parents are carriers, each pregnancy has a 25% chance of CF, a 50% chance of being a carrier, and a 25% chance of having neither mutation.

Main Symptoms: Chronic cough with secretions, recurrent pneumonia and bronchitis, difficulty gaining weight, fatty and foul-smelling stools, digestive problems, and characteristically salty skin.

Diagnosis: Newborn screening (heel prick test with IRT), confirmed by sweat chloride test, followed by genetic CFTR sequencing, which is also essential for determining eligibility for modulator therapy.

Treatment: There is currently no cure, but treatment has advanced significantly. It includes CFTR modulators, mucus-thinning medications, antibiotics, pancreatic enzymes, respiratory physiotherapy, and nutritional support.

CF affects the lungs, pancreas, liver, and digestive system, requiring ongoing specialized follow-up from a multidisciplinary care team.

Inheritance: How CF Is Passed Down

Each box below represents the chance for each child when BOTH parents are carriers (not affected, but carry one mutated CFTR gene):

25% CF	**50%** Carrier	**25%** Unaffected

MODULE 1: WELLBEING AND SUPPORT

1.1 Modulators: Access and Impact on Patients’ Quality of Life

CFTR Modulators: What They Are and What Has Changed

CFTR modulators are the most significant advance in CF history. Unlike previous treatments that only managed symptoms, modulators address the underlying protein defect, the root cause of CF. They do not work for everyone: eligibility depends on each patient's specific CFTR mutation, which is why genetic testing is essential.

Evolution of Modulator Therapy

- **Kalydeco (ivacaftor, 2012):** First modulator, applies to approximately 4%–5% patients, targets G551D and related mutations
- **Orkambi (lumacaftor/ivacaftor, 2015):** Broader mutation coverage
- **Trikafta / Kaftrio (elexacaftor/tezacaftor/ivacaftor, 2019):** Covers approximately 90% of eligible mutations,the most impactful advance to date
- **Alyftrek (2024):** Newest option, expands eligible mutation coverage by 94 additional mutations, offering once-daily dosing for eligible patients

[Documented Clinical Impact: Preliminary, unpublished data comparing 50 Latin American children followed in the United States (Texas Children’s Hospital, Houston) with 50 Mexican children treated at *Hospital Infantil de México Federico Gómez* suggest notable differences across three key areas: nutritional status, pulmonary function, and quality of life scores. Children with access to CFTR modulators demonstrated substantially better outcomes across all domains.]

> *“The difference was frankly significant across all three areas: nutrition, pulmonary function, and quality of life questionnaire.”*
> - Dr. José Luis Lezana Fernández, Medical Director, Asociación Mexicana de Fibrosis Quística, Mexico

> *“The first thing they tell us, in less than a week, is that the cough and the secretions decrease in a significant way.”*
> - Dra. Catalina Vázquez, Pediatric Pulmonologist, Colombia

> *“It is a revolution. There is no other way to describe it. We are witnessing a complete shift in the landscape.”*
> - Cristiano Silveira, Public Policy and Advocacy Director, Instituto Unidos pela Vida (IUV), Brazil

Who qualifies: A patient must have at least one eligible CFTR mutation. Genetic sequencing is required to determine eligibility. Ask your CF specialist whether your mutations have been sequenced and whether you may qualify.

Inequality in Access Across Countries

Regulatory approval of medication does not guarantee that patients can access it. Across Latin America, the gap between approval and real-world access is one of the most urgent challenges in CF care.

Argentina: CF law mandates coverage of modulators and all CF medications through health insurers (obras sociales and prepagas). Access has been available since modulators first emerged. If your insurer denies coverage, administrative and judicial remedies are available: contact AAER.

Brazil: Trikafta was included within SUS (unified public health system) in 2023. Access is now broader but not yet universal. Private insurance: judicial action remains an important tool if denied.

Colombia: Modulators are available through EPS health insurers, although of 1,282 CF patients, only 253 are on modulators. If denied, a tutela (constitutional rights protection) has a high success rate. ACFQ can provide legal support templates.

Mexico: Trikafta is COFEPRIS-approved but not in the public medication catalog (Cuadro Básico). Public patients must often use the judicial amparo mechanism. Private insurance patients have broader access. Patient associations can provide legal support and equipment or medications at reduced cost.

> *"I wish it could reach everyone, but since it doesn't reach everyone, we cannot forget that group that we still have to keep fighting for."*
>
> - Dra. Claudia Jimenez, Respiratory Therapist, Fundación Colombiana para Fibrosis Quística, Colombia

1.2 Psychosocial Challenges

Psychosocial Challenges in CF

Key sources of psychosocial burden:

- Treatment burden: approximately 3 hours per day, every day of the year
- Uncertainty about the future, often amplified by unguided online searches
- Prolonged hospitalizations (2–3 weeks for IV antibiotics)
- Mandatory social isolation: CF patients cannot have direct contact with each other due to cross-infection risk (minimum 2 meters distance)
- Financial and occupational impact: caregivers frequently reduce or leave employment

School and Work: Advocating for Yourself

For Students and Parents

- Request a meeting with the school principal or health coordinator to explain CF and its daily demands

- Ask the school to provide reasonable accommodations: Permission to take enzymes with meals, access to a clean bathroom, flexibility for medical appointments, exemption from smoky or dusty environments, a recovery plan for extended hospitalizations
- Provide the school with a written medical summary from your CF specialist and keep it updated annually
- Many countries have legal frameworks for disability accommodation in education. Ask your patient association what applies in your country

For Working Adults

- You are not required to disclose your CF diagnosis; you may simply request medical accommodation based on a doctor's note
- Useful accommodation: flexible start hours for morning physiotherapy, work-from-home options during illness, scheduled breaks for medication, proximity to clean bathrooms
- Patient associations often have workplace accommodation letter templates
- Know your rights: Most countries have labor protections for workers with chronic conditions

Remember: CF is part of who you are, it is not all of who you are. A good workplace or school will support you in maintaining your health so you can contribute fully.

1.3 Age-Specific Challenges (Children, Adolescents, and Adults)

CF Across the Life Cycle

The experience of CF changes with age. Understanding the distinct challenges at each stage helps patients, families, and care teams provide better-targeted support.

Children

- Establish CF care as a normal part of daily life from an early age, so treatment is integrated into routine rather than perceived as a burden.
- Schools are key: teachers and staff need basic education about CF to ensure inclusion and proper support
- Parents must balance rigor (ensuring treatment happens every day) with warmth and emotional reassurance
- Give children age-appropriate explanations for why each treatment matters; this dramatically improves cooperation and adherence. For this purpose, keep normalized communication at home, using puppets, books, and videos as tools
- Children with CF can participate in most school activities, sports, and social events

Adolescents

- The tension between independence and treatment compliance is the defining challenge of this stage;
- Sadness, fear of hospitalization, and disruption to school and social life are common.

- Transfer treatment responsibility gradually, not all at once

- Peer relationships are vital; online CF communities reduce isolation while respecting cross-infection rules
- Reproductive health and family planning are topics to raise with the care team during adolescence

Adults

- Navigating relationships, career, and family planning with CF requires specific guidance
- The transition from pediatric to adult care is a critical gap: Many patients "graduate" at 18 without a functioning adult referral network or proper medications available for them
- New adult CF care teams are essential as the population reaching adulthood grows
- Work accommodation is a right: patients are entitled to flexibility for medical appointments and treatment time

> *"What we see is that every year we discharge more young people at 18 and we have nowhere to send them."*
>
> - Dr. José Luis Lezana Fernández, Medical Director, Asociación Mexicana de Fibrosis Quística, Mexico

1.4 Strategies for Managing Anxiety, Depression, and Social Isolation

Patients and caregivers with CF are up to 3 times more likely to experience anxiety or depression than the general population. This is not a personal weakness; it is a direct consequence of living with a chronic, demanding, life-limiting condition.

Adaptive vs. Pathological Anxiety

Not all anxiety is problematic. Reasonable concern about health is adaptive. Anxiety becomes pathological when it is disproportionate to the real situation, focused on worst-case scenarios, or prevents participation in daily activities or treatment. Signs that professional attention is needed: fears disproportionate to the situation, persistent worst-case thinking, inability to complete daily activities or treatment routines.

The Vicious Cycle: Poor mental health makes it harder to maintain treatment routines. Worse adherence leads to worse physical outcomes. Worse physical health worsens mental health. Breaking this cycle requires addressing mental health as central to care and seeking care from a psychologist or psychiatrist as needed.

Practical Strategies for Mental Health

- Regular physical exercise (adapted to CF capacity)
- Hobbies and activities are completely unrelated to illness
- Social connection, including online communities, since in-person CF meetups are prohibited
- Professional psychological support as a standard part of the multidisciplinary team; For patients without modulator access, mental health support is even more essential

- Medication side-effect monitoring (including modulators and corticosteroids), which can trigger or worsen psychiatric symptoms

> *“If a person is depressed or very anxious, that can become an obstacle to their ability to adhere correctly to their treatment.”*
> - Lic. Sabrina Brancatisano, Psychologist, Asociación Argentina de Fibrosis Quística, Argentina

1.5 Comprehensive Treatment

Early Diagnosis

Early diagnosis is the foundation of optimal outcomes. The earlier CF is identified, and treatment begins, the better the long-term results are. The recommended diagnostic pathway includes:

1. **Newborn screening** using the IRT/IRT or IRT/DNA (immunoreactive trypsinogen) protocol, that is, the standard heel prick test
2. Confirmatory **sweat chloride test**
3. **CFTR gene sequencing**, especially for complex or ambiguous cases, and to determine eligibility for modulator therapy

Multidisciplinary Care Team

The **ideal CF care team** includes:

- Pulmonologist (specialist lead)
- Nutritionist
- Physiotherapist or respiratory therapist
- Psychologist
- Social worker
- Nurse
- Endocrinologist (currently absent from most teams, but strongly recommended given the metabolic complications of CF and modulator therapy)

Mental Health Effects of Treatment: What to Watch For

Some medications used in CF, including CFTR modulators and corticosteroids, can affect mental health in ways that patients and families often do not recognize as medication-related. This is one of the most underreported aspects of CF treatment.

Reported effects linked to modulators in some patients include:

- Sleep disturbances
- Mental fog or difficulty concentrating
- Worsening of pre-existing anxiety or depression
- In some patients, increased irritability (also observed with corticosteroids)

What to do: These effects do not occur in all patients, and when they do, they can often be managed without stopping the medication. The most important first step is recognizing the *possible* connection with treatment. Patients and families often attribute these symptoms to stress, poor sleep, disease progression, and may not report them. If you or your family member notice changes in mood, sleep, or mental clarity after starting or changing a medication, inform your CF care team as soon as possible. **Do not adjust or stop treatment on your own**. Management decisions (including whether to continue, adjust, or temporarily interrupt a medication and whether to add psychological or psychiatric support) must be made together with your healthcare team.

New Metabolic Realities with Modulator Therapy

CFTR modulators have transformed nutritional reality for many patients. For decades, CF care focused on preventing weight loss and achieving hypercaloric diets. With modulators, a new challenge has emerged: some patients gain weight rapidly and may develop metabolic complications as a result.

This shift **has two practical implications:**

- Dietary plans that were appropriate before modulators may need to be revised toward a more balanced, or in some cases, calorie-restricted diet. This should only be done under clinical supervision.
- Endocrinologists are now an important addition to the CF multidisciplinary team, even though they are currently absent from most centers. If your center does not have an endocrinologist and you are experiencing significant weight changes or metabolic symptoms since starting modulator therapy, ask your team for a referral.

Fragmented vs. Integrated Care: What It Means for Your Family

In many parts of Latin America, integrated CF centers, where all specialists see the patient on the same day, are the exception, not the rule. Many patients receive fragmented care: respiratory follow-up in one place, nutritional guidance in another, and no psychological or social support included.

If your care is fragmented: Contact your patient association to identify the nearest reference center and ask about transfer or shared-care options. Fragmented care is not inevitable. Advocacy for integrated care is part of what patient associations do, and knowing your options is the first step.

MODULE 2: FAMILY SUPPORT SYSTEMS

2.1 Best Practices

Recommendations for High-Quality Care

- Access to medication is essential and non-negotiable
- Broad access to early diagnosis should be guaranteed through neonatal screening
- Multidisciplinary consultations in a single visit on the same day significantly reduce the burden on families and improve continuity of care. Patients should not need to travel to multiple separate appointments
- Each consultation should produce documented goals for the next visit, communicated in accessible, patient-appropriate language
- Follow-up frequency should be individualized: quarterly at minimum, more frequent depending on clinical status
- Guidelines must be clear for clinical care, caregivers, and patients
- Socioeconomic context must be factored into all clinical recommendations. Nutritional guidance and treatment plans must be feasible for each patient’s real circumstances

> *“The patient does not need to keep going to the hospital for different appointments. Everything is done on the same day, at the same time.”*
>
> - Dr. José Luis Lezana Fernández, Medical Director, Asociación Mexicana de Fibrosis Quística, Mexico

A Changing Epidemiology: CF Is No Longer Only a Childhood Disease

Until recently, CF was understood primarily as a condition of childhood and early adulthood, with a median life expectancy that few surpassed. This reality has changed substantially: the combination of early diagnosis through neonatal screening, improved multidisciplinary care, and the arrival of CFTR modulators means that more patients than ever are reaching adulthood, building careers, forming families, and living into middle age. Countries with access to modulators and integrated care are already seeing this shift in their patient populations.

This shift has direct implications for how the health system must organize itself:

- Transition protocols from pediatrics to adult care are urgently needed; many patients currently 'graduate' at 18 without a functioning adult care network
- Adult CF services and specialists need to be developed and trained, as this population did not exist in sufficient numbers before
- Health policies, registries, and financing structures must evolve to serve a lifelong condition, not just a childhood one

Registries and Orphan Disease Recognition as Best Practices

Patients and families should advocate for:

- National CF registries, which enable data-driven decision-making, continuity of care across institutions, and evidence to support access to new treatments. Colombia's mandatory reporting system and Mexico's national registry initiative are examples to follow

- Legal recognition of CF as an orphan disease, as it creates specific frameworks for financing, access, and care organization. Colombia's definition of orphan diseases and its centralized purchasing strategy through PAHO are examples of how this recognition translates into practical benefits for patients

2.2 Support Networks

CF is not experienced by the patient alone; it affects the family as a whole. A support network is key.

Roles in the Support Ecosystem

Role	Responsibilities
Patient	Adhering to treatment, with support proportional to age, capacity
Family and caregivers	Providing daily, direct support across all aspects of care; their own wellbeing should also be treated as a clinical priority
Healthcare team	Delivering clinical care, education, shared goal setting, and specialist referrals
Regional patient associations	Offering direct support to families, local presence, community knowledge, and practical guidance
National patient associations	Leading policy advocacy, working to secure medication access, supporting national registries, and providing legal guidance
General community	Promoting flexibility and inclusion, and helping reduce stigma around invisible disability

Patient Associations by Country

National and regional associations are key allies for accessing medications, navigating legal and administrative systems, and finding peer and psychosocial support. Whenever possible, contact them proactively.

Country	Association	Contact	What They Offer
Argentina	AAER - Asociación Argentina de Enfermos Respiratorios	aaerargentina.org.ar	Medication access support, legal guidance, family resources, national registry
Brazil	ABCF - Associação Brasileira de Assistência à Mucoviscidose	abcf.org.br	SUS access support, judicial action guidance, family resources, advocacy
Colombia	ACFQ - Asociación Colombiana de Fibrosis Quística	acfq.org.co	EPS/tutela support, family resources, care team navigation, legal templates

Mexico	Asociación Mexicana de Fibrosis Quística	Contact via national CF center or IMSS/ISSSTE patient services	Amparo guidance, treatment access advocacy, family support
Other countries	Latin American CF Consortium / National health ministry CF program	Contact through national health ministry or regional hospital	Regional network, referrals, specialist support

Online Communities: Since CF patients must maintain physical distance from one another (minimum 2 meters), online peer groups are a vital tool for reducing isolation. Ask your care team or patient association about trusted online communities for patients and families.

> *"No association could have the reach to be everywhere. The presence of small regional organizations is very important."*
> - Cristiano Silveira, Public Policy and Advocacy Director, Instituto Unidos pela Vida (IUV), Brazil

Caring for the Caregiver

Caregivers, most often mothers (around 90% in many CF families), are at high risk of burnout, emotional exhaustion, anxiety, and depression. They frequently neglect their own basic needs due to the demands of daily treatment and coordination of care; c**aregiver wellbeing should be treated as a clinical priority, not an afterthought.**

Signs of Caregiver Burnout

Physical	Emotional	Behavioral
Chronic fatigue not relieved by sleep	Constant anxiety or dread	Withdrawing from friends and family
Frequent illnesses (immune system weakened by stress)	Feeling trapped with no way out	Missing your own medical appointments
Sleep problems despite exhaustion	Resentment (followed by guilt)	Inability to enjoy anything previously enjoyed
Headaches, muscle tension, digestive issues	Emotional numbness or unexpected crying	Irritability or short temper with the patient

Five Daily Self-Care Anchors

1. **Sleep:** Protect at least 6–7 hours, even if you need to ask someone to cover one treatment session
2. **One meal just for yourself:** Eat something you enjoy, prepared or chosen by you, not just convenience
3. **Spend some minutes outside:** Fresh air and daylight are physiological necessities, not luxuries
4. **One person to call:** Maintain at least one relationship outside the caregiving role, even a 5-minute call counts

5. **One activity unrelated to CF**: Do something purely because you want to; whenever possible, make space for hobbies and relaxing activities.

Permission to Rest

Resting is not abandoning your loved one. It is what makes sustainable care possible over the long term. No one has unlimited reserves. Additionally, a caregiver burnout can reduce treatment adherence, increase errors, and strain relationships, all of which directly affect the person with CF. Prioritizing your own rest and asking for help from family, friends, or the care team is a responsible and protective act.

> *"Nobody has inexhaustible batteries. Very often, caregivers neglect their own basic needs."*
>
> \- Lic. Sabrina Brancatisano, Psychologist, Asociación Argentina de Fibrosis Quística, Argentina

If signs of burnout or emotional distress have been present for more than 2 weeks, or if you are having any thoughts of self-harm or wanting to escape, speak with a doctor or mental health professional as soon as possible. Seeking help for yourself is one of the most responsible things a caregiver can do.

MODULE 3: PATIENT EDUCATION AND TREATMENT ADHERENCE

3.1 Equitable Access

Optimizing Your Care When Modulators Are Not Yet Available

A significant number of patients in Latin America cannot access CFTR modulators due to cost, regulatory, or geographic barriers. This does not mean that high-quality care is impossible.
Consistently applying standard CF care, including multidisciplinary follow-up, airway clearance, infection prevention, nutrition optimization, and vaccination, can slow disease progression and improve quality of life, even in the absence of modulators.

Beyond Medications: Social Determinants of Health in CF

Nutrition: Food security

CF patients require up to twice the caloric intake of a person without CF. Meeting this requirement is difficult even in ideal conditions. For families experiencing food insecurity, it can be impossible. Specialists across Latin America report seeing children who are not gaining weight despite receiving the correct enzyme dose because their families cannot afford the protein intake required.

Work and income: Economic stability and employment

Caregivers frequently reduce or leave employment to manage the daily demands of CF care. This creates a reinforcing cycle of economic vulnerability that directly impacts the family's capacity to follow treatment recommendations.

Where you live matters: Geographic location

Patients in rural or peripheral areas face additional barriers to reaching reference centers, attending regular consultations, and accessing equipment.

What to do if social barriers are affecting your treatment: If food insecurity, transportation costs, or employment issues are making it difficult to follow your treatment plan, speak openly with your social worker or care team. These are documented barriers that the healthcare system has a responsibility to help address. Patient associations often have connections to social support programs and can help map local resources.

Understanding Barriers to Access and Adherence

Barrier	What It Looks Like	What Helps
Insufficient health literacy	Patient/family doesn't fully understand why each treatment matters	Ask for a dedicated education session, request written summaries, and ask care team to explain everything in plain language

Treatment fatigue	Feeling exhausted by daily demands after months or years	Adjust routine timing, identify the highest-impact treatments, and allow flexibility on low-risk days with guidance from the care team
Mental health challenges	Depression or anxiety is interfering with the motivation to treat	Address mental health directly (see Module 1); psychological support can help improve adherence
Socioeconomic limitations	Cannot afford recommended foods, transportation, or equipment	Seek referral to a social worker, contact patient associations, and map local resources together with the care team
Lack of social support	Managing everything alone without help	Build shared responsibilities at home, reach out to associations, and join online communities

3.2 Treatment Adherence

Daily CF treatment typically requires approximately 3 hours, including nebulization, respiratory physiotherapy, enzyme intake with every meal, and oral medications. This is a substantial commitment every day, for life.

Core Principles of Adherence

- 100% adherence is not a realistic goal; the objective is the best possible adherence for each individual within their specific circumstances
- Adherence is a system-based responsibility; the healthcare team, family, and institutions all share accountability for making treatment sustainable
- Children who understand why each treatment is necessary adhere better. Build progressive responsibility from an early age
- Treatment must adapt to the patient's life and routines, not the other way around
- CF is a part of a person's life, not the definition of who they are

> *"Adherence is not only the patient's adherence to treatment, but also the adherence of an entire system to this patient."*
>
> - Cristiano Silveira, Public Policy and Advocacy Director, Instituto Unidos pela Vida (IUV), Brazil

3.3 Coping Strategies

Living with CF requires more than following a treatment plan. It also means learning how to navigate the emotional, social, and personal dimensions of a chronic condition. The strategies below can help you build resilience, maintain balance, and live a full life alongside CF.

- **Talk about CF openly.** Avoid secrecy at home, use proper medication names, and explain the reasons behind each treatment step. Familiarity with the condition reduces anxiety for you and those around you.

- **Keep learning about your condition.** Use videos, stories, and other accessible resources, and revisit your understanding at each life stage. Knowing your condition well is a form of self-care.
- **Build your independence gradually.** Take on small treatment responsibilities early and work toward adapting your routine around your lifestyle, not the other way around.
- **You own your diagnosis.** Decide for yourself what to share, with whom, and when. There is no obligation to disclose, and your boundaries deserve full respect.
- **Educating people around you reduces isolation.** Others often fall short of support because they don't understand specific needs related to the condition. Sharing information about CF can open real networks of support at school, work, and beyond.
- **Seek support beyond medical care.** Psychologists, social workers, and interdisciplinary teams are part of your treatment. Don't hesitate to ask for emotional and social support, especially during major transitions.
- **Allow yourself to feel difficult emotions.** Anger, grief, and denial are a natural part of the process. Acknowledging them rather than suppressing them protects both your mental health and your ability to stick to treatment.
- **Build a life with purpose beyond CF**. Invest in projects, relationships, and experiences that motivate you. A meaningful life strengthens resilience and makes treatment more sustainable over time.
- **Plan for the long term**. Treatment advances have significantly improved life expectancy. Permit yourself to set real goals around education, career, relationships, and family.
- **Use early diagnosis as a head start**. Starting treatment early and building knowledge gradually leads to better long-term outcomes and greater autonomy over time.
- **Fit treatment into your life, not life into your treatment.** Work openly with your care team to adapt your routine so that CF management and your personal goals can coexist.
- **You are more than your diagnosis**. CF is part of your story, not the whole of it. Maintaining diverse activities and relationships is essential to your identity, motivation, and quality of life.
- **Protect your mental health as actively as your physical health**. Working, studying, and maintaining social connections are not distractions from treatment. They are a part of it.

> *"I have cystic fibrosis, but I want to have a social life, a work life, a professional life. I don't feel the need to make everything revolve around cystic fibrosis."*
> - Luz Angela Sanchez, CF Patient, Fundación Colombiana para Fibrosis Quística, Colombia

3.4 Nutrition and Supplements

Nutrition: A Clinical Pillar

Nutritional management in CF is among the most demanding and consequential aspects of daily care. **The pancreatic insufficiency present in approximately 85% of CF patients requires continuous, individualized attention.**

- **Caloric needs:** CF patients often require up to 1.5–2x the calories of people without CF due to increased metabolic demand and malabsorption
- **Pancreatic enzyme replacement therapy (PERT)** must be taken with every meal and every snack. Dosing is dynamic and adjusted regularly

- With modulators, some patients **transition from hypercaloric to more balanced diets**, but this requires clinical monitoring
- **Food insecurity** is documented across the region: some patients cannot meet requirements. This does not come from non-compliance; lots of families are unable to afford recommended foods
- **Good hydration is especially important in hot climates**, as patients with CF lose more salt in sweat

> *"If a person needs one thousand calories, a person with CF may need up to 2,000 calories."*
>
> - Dra. Claudia Angarita, Nutritionist, Fundación Colombiana para Fibrosis Quística, Colombia

3.5 Respiratory Therapy

Respiratory Therapy: Essential and Irreplaceable

CFTR modulators dramatically reduce mucus burden but must not replace respiratory physiotherapy and other treatments. Airway clearance techniques remain essential for all eligible patients.

- Clearance respiratory physiotherapy should be done at least 2x a day, up to 3-4x a day during exacerbations
- The recommended nebulization sequence is bronchodilator → hypertonic saline → dornase alfa (DNase) → airway clearance techniques → inhaled antibiotic
- Equipment quality matters: Aluminum spacer chambers deliver over 65% of inhaled medication vs. approximately 30% for standard plastic chambers
- The respiratory therapist's role includes ongoing education, routine adjustment, and emotional support in addition to therapy delivery

> *"Even if they are on modulators, we must explain to them that this does not replace respiratory physiotherapy."*
>
> - Dra. Claudia Jimenez, Respiratory Therapist, Fundación Colombiana para Fibrosis Quística, Colombia

Respiratory Physiotherapy: A Non-Negotiable Foundation

Minimum frequency: at least twice daily (morning and evening), every single day, including when you feel well. For people without access to modulators, airway clearance is one of the most important interventions to protect lung function. However, it is also one of the first to be shortened or skipped during busy periods.

- **Autogenic Drainage (AD):** A self-directed breathing technique moving mucus from small to large airways
- **Active Cycle of Breathing Techniques (ACBT):** Cycles of relaxed breathing, deep breaths, and huff-coughs
- **Oscillating PEP Devices (Flutter, Acapella):** Hand-held devices creating vibration and positive pressure to loosen mucus
- **Postural drainage:** Positioning the body to use gravity to drain different lung segments

Nebulization Sequence: Order Matters

Step	Medication / Action	Purpose
1	Bronchodilator (Salbutamol)	Open and relax airways before treatment
2	Hypertonic Saline (3% or 7%)	Draw water into the airways to thin and loosen mucus
3	Dornase Alfa (DNase) (if prescribed)	Break down DNA in mucus, making it thinner and easier to clear
4	Airway Clearance	Use your chosen technique: AD, ACBT, or oscillating PEP device
5	Inhaled Antibiotic (if prescribed)	Target lung bacteria; given last to reach cleaner airways

3.6 Patient Registry

A patient registry is a systematic database that collects health information about people living with CF over time. Registries are one of the most powerful instruments available to the CF community to improve care, expand access, and influence policy.

Why Registries Matter: Three Levels of Impact

1. For You Personally

When your medical information is stored in a national registry, your records travel with you. If your family relocates to another state, province, or city, your full clinical history, including diagnoses, treatments, lung function results, sputum cultures, and nutritional status, can move electronically to your new care center. You do not start from zero. Continuity of care is preserved without depending on paper folders or the memory of any single doctor.

2. For Clinical Decision-Making

Registry data allows your medical team and health authorities to track trends across the entire CF population: how nutritional status is evolving, whether respiratory function is improving, and how life expectancy is changing over time. These patterns are invisible without systematic data and they directly guide how care is delivered.

3. For Policy and Access to New Treatments

This is where registries have had their most decisive impact in Latin America. When health ministries consider approving a new medication, such as a CFTR modulator, they need to know how many patients are eligible and how many will need the treatment. Countries with robust registries can answer these questions with precision. Countries without them cannot.

The State of Registries in Latin America

Countries are at very different stages:

Country	**Status**
Colombia	Mandatory national registry (Registro Nacional de Enfermedades Huérfanas). CF is reportable like any communicable disease. As of the most recent data: 1,282 registered patients, of whom only 253 were on modulators, a figure that directly informed advocacy efforts
Mexico	Hospital-level registries exist (some running for over 10 years). A national registry project was submitted to the Ministry of Health through the Coordination of National Health Institutes; it is awaiting full approval and rollout across all CF centers
Brazil / Argentina	There are registries and tracking systems, with ongoing efforts to strengthen national coverage and longitudinal data collection
Other countries	All countries are encouraged to build or strengthen CF registries in collaboration with specialist groups, even if resources are limited.

What You Can Do

Action for Patients and Families: Ask your CF specialist whether your data is included in a national or institutional CF registry. Taking part is both a right and an act of solidarity with every person who will benefit from the data you contribute. If your country does not yet have a national registry, your patient association is the best organization to contact and support in building one.

OTHER RECOMMENDATIONS

Vaccinations: Your Shield Against Dangerous Infections

- **Annual influenza (flu) vaccine:** Essential protection - flu is particularly dangerous for CF lungs
- **Pneumococcal vaccine:** Protects against a major bacterial lung pathogen
- **COVID-19 vaccine:** Critical for patients with reduced lung function
- **Pertussis (whooping cough) booster:** Ask your CF team
- **Discuss all vaccine timing with your CF team:** Some live vaccines require precautions

What to Avoid

- Tobacco smoke, including secondhand smoke. Even brief exposure is harmful to CF airways
- Air pollution, dust, mold, and poorly ventilated spaces
- Anyone with an active respiratory infection (cold, flu, COVID)
- Crowded, enclosed spaces during peak infection seasons
- Swimming in natural bodies of water without medical guidance (risk of environmental bacteria)

Regular Monitoring: Early Detection Changes Outcomes

- **CF specialist visits:** Every 3 months if stable, more often if symptomatic
- **Pulmonary function test (spirometry):** At each visit, annually at a minimum
- **Sputum culture:** To identify bacterial colonization and guide antibiotic choices
- **Weight and nutritional assessment:** Every CF visit
- **Annual chest imaging** (X-ray or CT)

Early and Complete Treatment of Infections

Complete ALL antibiotic courses, even when you start feeling better. Stopping early allows bacteria to survive and develop resistance. In CF, unfinished antibiotic courses are a direct risk to future treatment effectiveness.

Mental Health When Access Is Limited:

If you cannot access modulator therapy yet, the emotional and psychological weight of CF is often even heavier. Mental health support, whether through a psychologist, support group, family, or a trusted community, should be treated as an essential part of care, not an optional extra. Investing in this support is a cornerstone of sustainable self-care and long-term treatment adherence.

GETTING WHAT YOU NEED: A COUNTRY GUIDE

The legal and administrative pathways to access CF care differ by country. This section outlines the steps most likely to be effective in each country. Document all requests and denials in writing. Your patient association is your strongest ally.

Argentina

The Argentine CF Law (and Law 23.661) requires all health insurers (Obras Sociales and Prepagas) to cover all CF medications, including CFTR modulators.

Steps:

1. Get a prescription from your CF specialist
2. Submit the prescription to your Obra Social or Prepaga in writing, requesting coverage. Keep a copy of everything submitted
3. If denied: contact AAER immediately,- they have legal advisors and know the most effective pathways in each province
4. If your insurer continues to deny coverage: an amparo or administrative complaint before the Superintendencia de Servicios de Salud are viable options

AAER (Asociación Argentina de Enfermos Respiratorios): aaerargentina.org.ar | Also: RENAC (Registro Nacional de Fibrosis Quística)

Brazil

Trikafta was included in the SUS (Sistema Único de Saúde) in 2023 through CONITEC's decision. This means public health patients now have a pathway to access.

Steps:

1. Obtain a confirmed diagnosis at an accredited CF center (Centro de Referência em Fibrose Cística)
2. Request the medication through your state's Secretaria de Saúde.- Bring the PCDT (Protocolo Clínico e Diretrizes Terapêuticas) documentation
3. If access is delayed: ABCF can advise on your state's specific process
4. Private insurance patients denied access: An Ação Judicial (judicial action) remains highly effective.- ABCF can connect you with legal support

ABCF (Associação Brasileira de Assistência à Mucoviscidose): abcf.org.br | GBEFC (Grupo Brasileiro de Estudos em Fibrose Cística): specialist network

Colombia

CFTR modulators are available through the Colombian health system via EPS (Entidades Promotoras de Salud) authorization.

Steps:

1. Obtain a prescription from a CF specialist
2. Submit an authorization request to your EPS. Include all clinical documentation supporting medical necessity
3. If the EPS denies coverage or does not respond within the legal timeframe: file a tutela (acción de tutela).- This constitutional rights mechanism has a very high success rate for CF medications in Colombia
4. ACFQ can provide tutela templates and legal guidance

ACFQ (Asociación Colombiana de Fibrosis Quística): acfq.org.co | INVIMA: Regulatoria body for medication approvals in Colombia

Mexico

Trikafta is approved by COFEPRIS but has not been included in the Cuadro Básico de Medicamentos (public health catalog). This means public-sector patients must pursue special access routes.

Steps:

1. Obtain a prescription from a CF specialist at IMSS, ISSSTE, or private sector
2. Request access through your institution's 'gastos catastróficos' or 'medicamentos fuera de cuadro básico' mechanisms
3. If denied: file an amparo judicial.- This is the primary legal tool that Mexican CF patients have used successfully to compel IMSS/ISSSTE/Seguro Popular to cover CF medications
4. Contact CF patient associations for legal support: they may have lawyers or legal templates available

Asociación Mexicana de Fibrosis Quística -- contact through major CF centers or CONAMED for patient rights guidance

General Advocacy Tips (All Countries)

- **Document everything in writing:** All requests, all denials, all responses. Keep copies
- **Patient associations have navigated these processes before:** Contact them before a crisis, not only during one
- **A medical letter from your CF specialist** carries significant legal weight in any country
- **Keep complete copies of all your medical records** at home; they are your property
- If you are in a rural or remote area, ask your patient association about **telehealth options** and how other patients in your region have navigated access

EMERGENCY REFERENCE GUIDE

GO TO THE HOSPITAL IMMEDIATELY

- **Severe difficulty breathing: child cannot finish a sentence, nostrils flaring, visible use of neck or chest muscles to breathe**
- **Lips, fingernails, or skin turning bluish or grayish (cyanosis)**
- **Coughing up blood, any amount**
- **Severe chest pain**
- **Fever above 38.5°C (101.3°F) that does not come down with paracetamol or ibuprofen**
- **The person is confused, unusually difficult to wake up, or not responding normally**
- **No improvement after 48–72 hours of worsening symptoms despite full treatment compliance**

CALL YOUR CF DOCTOR TODAY (Not Emergency, But Urgent)

- New cough, or existing cough becoming more frequent or producing more mucus than usual
- New fever lasting more than 2 days
- Unexplained weight loss over 2 or more weeks
- New joint pain or swelling
- Changes in stool: more frequent, oilier, or more foul-smelling than usual
- Increased fatigue beyond the usual level
- New wheezing or shortness of breath with everyday activities
- Any new symptoms you cannot explain

My Emergency Contact Information

CF Center Name	
CF Center Address	
CF Center Phone	
My CF Doctor Name	
My Doctor Direct/On-Call Phone	
Nearest Hospital with CF Experience	
Hospital Emergency Phone	
My Health Insurer / Insurance Number	
Important: My CF Mutations	

MY PERSONAL CF TOOLS

My CF Care Team

Specialist	Name	Phone	Next Appointment

My Daily Treatment Schedule

Time of Day	Treatment / Medication	Duration	Notes
Morning (wake-up)			
With breakfast			
Mid-morning			
With lunch			
Afternoon			
With dinner			
Evening (before bed)			
With snacks			

Questions to Ask at My Next Appointment

For My Pulmonologist

- What is my FEV1 (lung function) today, and how does it compare to my last visit?
- Am I a candidate for CFTR modulator therapy? If not, when should we reassess?
- What bacteria are present in my sputum culture, and what does this mean for my treatment?
- Is my current antibiotic plan still appropriate?
- When should I return? What symptoms should prompt me to call before then?

For My Nutritionist

- Is my weight appropriate for my height and age?
- Is my current enzyme dose correct? Am I taking it correctly (with every meal and snack)?
- Do I need any vitamins or mineral supplements?
- What specific foods should I prioritize this month?
- Are there any nutritional signs of diabetes or liver involvement I should watch for?

For My Physiotherapist / Respiratory Therapist

- Am I performing my airway clearance techniques correctly?
- Should I adjust my nebulization sequence or the timing of treatments?
- What equipment should I have at home, and how do I maintain it properly?
- Are there new techniques I should learn based on my current lung function?

For My Psychologist

- What can I do when I feel overwhelmed by the daily treatment demands?
- How can I talk to my family, teachers, or employer about what I need?
- How do I manage treatment fatigue without reducing the most critical treatments?
- Are there online support groups or resources you recommend?

My Monthly Symptom Log

Bring this log to every appointment. Scale: 1 = Very poor, 3 = Average, 5 = Very good.

Week	Weight (kg)	Cough (1–5)	Energy (1–5)	Stools Normal?	Missed Treatments?	Notes
Week 1						
Week 2						
Week 3						
Week 4						

KEY MESSAGES

1. Treatment: The Landscape Has Changed, and Continues to Change

CFTR modulators have transformed CF from a predominantly childhood disease into a condition increasingly compatible with a long, active life. Early diagnosis combined with appropriate, sustained treatment now offers outcomes unimaginable 20 years ago. If you do not yet have access to modulators, evidence-based care without them remains powerful - and advocacy to close the access gap continues.

2. Mental Health: Caring for The Mind Is Part of Treatment

Anxiety and depression are up to three times more common in CF patients and their caregivers than in the general population. Unaddressed mental health challenges directly undermine treatment adherence and physical outcomes. Psychological support is part of complete CF care.

3. Family: No One Cares Alone, and Caregivers Need Care Too

CF is not experienced by the patient in isolation. The entire family is affected. Building a real support network (and attending to the needs of caregivers) is as essential as any medication in the long-term management of this disease.

4. Adherence: Treatment Must Fit into Life, not the Other Way Around

Perfect adherence is not achievable. The goal is the best possible adherence, adapted to each person's real routine and circumstances. Patients who understand why they are being treated, and who are supported by their team and family, adhere better and live better.

5. Access: Approval is not Access, and this Must Change

Regulatory approval of a medication does not mean patients can access it. National registries, public health policies, patient associations, and sustained advocacy are concrete tools for transforming approval into real-world access. Know your rights and use them.

EXPANDED GLOSSARY

Reference Guide for Patients, Families, and Caregivers

Term	Definition
CF / FQ / FC	Abbreviations for Cystic Fibrosis. In English: CF. In Spanish: FQ (Fibrosis Quística). In Portuguese: FC (Fibrose Cística).
CFTR	Cystic Fibrosis Transmembrane Conductance Regulator - the protein regulated by the gene affected in CF. When CFTR does not function correctly, cells cannot properly move salt and water, causing thick mucus buildup.
CFTR Modulator	A medication that corrects the function of the defective CFTR protein at the molecular level, addressing the root cause of CF rather than just the symptoms.
Corrector	A type of modulator that helps the CFTR protein fold into the correct three-dimensional shape so it can reach the cell surface. Examples: elexacaftor, tezacaftor, lumacaftor.
Potentiator	A type of modulator that helps a correctly shaped CFTR protein stay open and function properly. Example: ivacaftor (Kalydeco).
Trikafta / Kaftrio	Brand names for the triple combination modulator (elexacaftor/tezacaftor/ivacaftor). It covers approximately 90% of CF patients with eligible mutations. Widely considered the most transformative advance in CF history.
Alyftrek	The newest CFTR modulator (2024), offering once-daily dosing for eligible patients. Eligibility depends on specific CFTR mutations.
Exacerbation	An acute worsening of respiratory symptoms, typically requiring hospitalization and a course of intravenous antibiotics. Usually manifests as increased cough, more secretions, and reduced lung function.
FEV1	Forced Expiratory Volume in 1 second. The key measure of lung function in CF - how much air you can forcefully exhale in one second. Expressed as a percentage of expected value for your age, height, and sex. Decline over time is a key marker of disease progression.
FVC	Forced Vital Capacity - the total amount of air you can exhale forcefully. Used together with FEV1 to assess lung function.
Spirometry	The lung function test that measures FEV1 and FVC. Performed by blowing as hard and fast as possible into a device. Should be performed at every CF clinic visit.
PERT	Pancreatic Enzyme Replacement Therapy. Capsules or tablets (e.g., Creon) taken with every meal and snack to replace the digestive enzymes the pancreas cannot produce. Dosing is adjusted based on fat content of meals.
Dornase Alfa (DNase, Pulmozyme)	An inhaled medication that breaks down DNA strands in CF mucus, making it thinner and easier to clear from the airways. Taken by nebulization.

Hypertonic Saline	A saltwater solution (typically 3% or 7%) inhaled via nebulizer. It draws water into the airway surface, thinning mucus and improving ciliary clearance. Often taken before airway clearance techniques.
Bronchodilator (Salbutamol / Albuterol)	Inhaled medication that relaxes and opens the airways. Always taken BEFORE other nebulized medications. Important for patients with airway sensitivity.
Nebulizer	A device that converts liquid medication into a fine mist for inhalation into the lungs. Available as jet nebulizers, ultrasonic, and mesh nebulizers. Regular cleaning is essential.
Airway Clearance Technique	Any technique or device used to loosen and remove mucus from the airways. Includes autogenic drainage, ACBT, oscillating PEP devices, and postural drainage.
Autogenic Drainage (AD)	A self-directed breathing technique that uses different lung volumes (low, mid, and high) to progressively move mucus from small to large airways, where it can be cleared with gentle coughs.
Active Cycle of Breathing (ACBT)	A structured breathing technique involving three phases: breathing control (relaxed breathing), thoracic expansion exercises (deep breaths), and forced expiration technique (huff-coughs).
Oscillating PEP (Flutter, Acapella)	Handheld devices that create oscillating positive expiratory pressure during exhalation, producing vibrations in the airways that loosen and mobilize mucus.
Pseudomonas aeruginosa	A common bacterium found in CF airways, particularly in older patients. Its presence indicates a higher level of infection burden. Managed with inhaled and systemic antibiotics.
Burkholderia cepacia complex	A group of bacteria particularly dangerous in CF - associated with rapid lung decline and high mortality. The primary reason is that CF patients must not have physical contact with each other.
Sweat Test (Chloride Test)	The primary diagnostic test for CF. Measures the concentration of chloride in sweat. Values above 60 mmol/L in the presence of symptoms are diagnostic for CF.
IRT (Immunoreactive Trypsinogen)	A pancreatic enzyme measured in the newborn heel prick blood test. Elevated IRT triggers further CF testing. This is how most CF patients are now diagnosed in countries with newborn screening programs.
Pancreatic Insufficiency	The inability of the pancreas to produce enough digestive enzymes, present in approximately 85% of CF patients. Results in malabsorption of fats and fat-soluble vitamins (A, D, E, K).
Malabsorption	Failure to properly absorb nutrients from food. In CF, this is primarily caused by pancreatic insufficiency. Signs include weight loss, fatty stools, and deficiencies in fat-soluble vitamins.
Obra Social (Argentina)	Employer-based health insurer in Argentina, funded by mandatory payroll contributions. Legally required to cover all CF medications including modulators under the CF national law.
SUS - Sistema Único de Saúde (Brazil)	Brazil's unified public health system. Since 2023, Trikafta has included its formulary for eligible CF patients.

EPS - Entidad Promotora de Salud (Colombia)	Health insurer in Colombia's contributory and subsidized health systems. Required to authorize CF medications; denial can be challenged via tutela.
IMSS / ISSSTE (Mexico)	Mexico's largest public health institutions. IMSS covers formal private-sector workers; ISSSTE covers public-sector workers. Both have faced legal challenges to extend modulator access.
Amparo Judicial (Mexico)	A constitutional legal mechanism in Mexico allowing individuals to challenge actions by government entities that violate their fundamental rights. Widely used by CF patients to compel public health institutions to cover modulator therapy.
Tutela (Colombia)	An urgent constitutional legal action in Colombia to protect fundamental rights (including the right to health). It has been highly effective for CF patients denied coverage by their EPS.
Multidisciplinary CF Team	The internationally recommended care model for CF, which includes pulmonologist, nutritionist, physiotherapist/respiratory therapist, psychologist, social worker, nurse, and where possible, an endocrinologist.
Mucoviscidosis	The alternative clinical name for cystic fibrosis, still used in some countries. Referring to the abnormally viscous (thick) mucus that is the hallmark of the disease.

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