Americas Health Foundation

Living with

CYSTIC FIBROSIS

A Self-Learning Program for Patients, Families, and Caregivers

Program Overview

1. **MODULE 1:** Wellbeing & Support
2. **MODULE 2:** Family Support Systems
3. **MODULE 3:** Patient Education and Treatment Adherence

Module 1

Wellbeing and Support

The treatment landscape has transformed, but access remains unequal, and the mental health burden is real

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

1.2 Psychosocial Challenges

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

1.4 Strategies for Managing Anxiety, Depression, and Social Isolation

1.5 Comprehensive Treatment

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Module 1 - Wellbeing and Support

1.1 Modulators

What are CFTR Modulators?

CFTR Modulators address the root defect

In CF, the **CFTR protein is misfolded or absent**; it cannot perform its job of moving salt and water across cell membranes. This causes the thick, sticky mucus that damages the lungs, pancreas, and other organs over time.

CFTR modulators correct or compensate for that protein defect directly, making them the first treatments to change the course of the disease, not just manage its consequences.

The most significant advance in CF history

~90% of people with CF carry mutations that respond to Trikafta (elexacaftor/tezacaftor/ivacaftor)

Alyftrek (2024) offers once-daily dosing, the latest generation.

Across the Region: Approval ≠ Access

Argentina

Mandate by Law

National CF Law requires coverage of ALL CF medications, including modulators, through Obra Social. Still, there is an access lag.

Brazil

Public System (2023)

After years of advocacy and judicial decisions, Trikafta was added to SUS in 2023. Access is expanding slowly, but is not yet universal.

Colombia

Partially Available

Modulators are available through EPS, with ongoing efforts for direct industry negotiation and centralized access.

Mexico

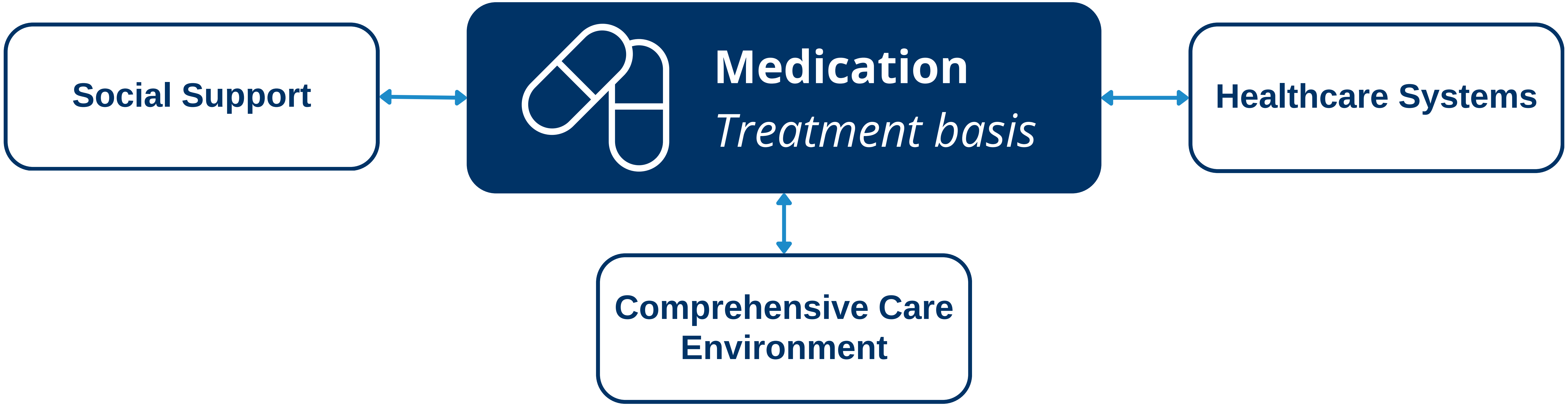
Private Only

Trikafta is approved by COFEPRIS, but not in the public sector drug catalog; only accessible via private insurance.

In all four countries: regulatory approval ≠ patient access. The gap between policy and reality is the defining challenge.

Health as a Multifactorial Concept

Holistic and Integrated Health Approach



Social Support

Medication

Treatment basis

Healthcare Systems

Comprehensive Care Environment

Health is multifactorial: it does not only depend on access to a drug, but on social support, health systems, and comprehensive care.

Clinical Evidence and Comparative Outcomes

50 Latin American children
(Texas Children's Hospital)
Using Modulators

↑ Nutrition
↑ Lung Function
↑ Quality of Life

50 Mexican children
(Hospital Federico Gómez)
Not on Modulators

↓ Nutrition
↓ Lung Function
↓ Quality of Life

> *"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 Modulator Pipeline:
From Symptom Relief to Disease Modification

First Modulator

~4% of mutations
G551D gating

2012
Kalydeco
(Ivacaftor)

2015
Orkambi
(Lumacaftor/
Ivacaftor)

Expand mutations

First for
F508del homozygous
~45% of patients

Transformative Impact

Triple combination
~90% of mutations

2019
Trikafta /
Kaftrio

2024
Alyftrek

Once-daily dosing

Next-gen
Adds 94 additional eligible
mutations beyond Trikafta
Improved efficacy

Barriers for Regulatory Approval

- High Costs
- Access to Information
- Slow uptake as regulators adopt modulators as effectiveness is proven

Regulatory Agencies

Argentina
ANMAT

Brazil
ANVISA

Colombia
INVIMA

Mexico
COFEPRIS

> ***It is a revolution. There is truly 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 · Brazil

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Module 1 - Wellbeing and Support

1.2 Psychosocial Challenges

The Mental Health Reality in CF

3x more likely to develop **anxiety or depression** than the general population.

~3 hrs of **treatment required every single day**, including nebulizations, physiotherapy, enzymes, and medications.

2-3 wks of **mandatory hospitalization** for IV antibiotic courses, a major disruption to work, school, and family life.

2 meters is the **minimum distance** required between CF patients, making in-person peer support impossible.

Modulators themselves can trigger or worsen psychiatric symptoms (sleep disturbance, brain fog, mood changes).

Poor mental health must be treated as a clinical priority, not a secondary concern. Unaddressed anxiety and depression directly undermine treatment adherence and outcomes.

Family Emotional Environment

Grief-like stages

Shock

Grief

Acceptance

The **family's reaction to the diagnosis is fundamental to shaping the patient's experience**, especially when the patient is a child.

Chronic illness is experienced not only by the patient but collectively by their family and within their broader environment.

Emotional Needs in Each Stage of Life

Children

- **Consistent family routines** that integrate treatment into daily life
- **Support**, as children begin questioning their condition and why they need treatment as they grow
- **Age-appropriate CF information** that deepens understanding of the condition over time

Adolescents

- **Support** to develop autonomy and identity
- **Help** balancing treatment demands with social life, relationships, and school or university commitments
- **Access** to emotional support to manage sadness and anxiety

Adults

- **Tailored support** to manage relationships, career decisions, and daily treatment demands in adult life
- **Flexible, understanding work environments** that accommodate CF-related care and health needs
- **Opportunities and support** to maintain social connections and activities beyond CF to protect mental health

Early diagnosis is the key to improving acceptance and quality of life.

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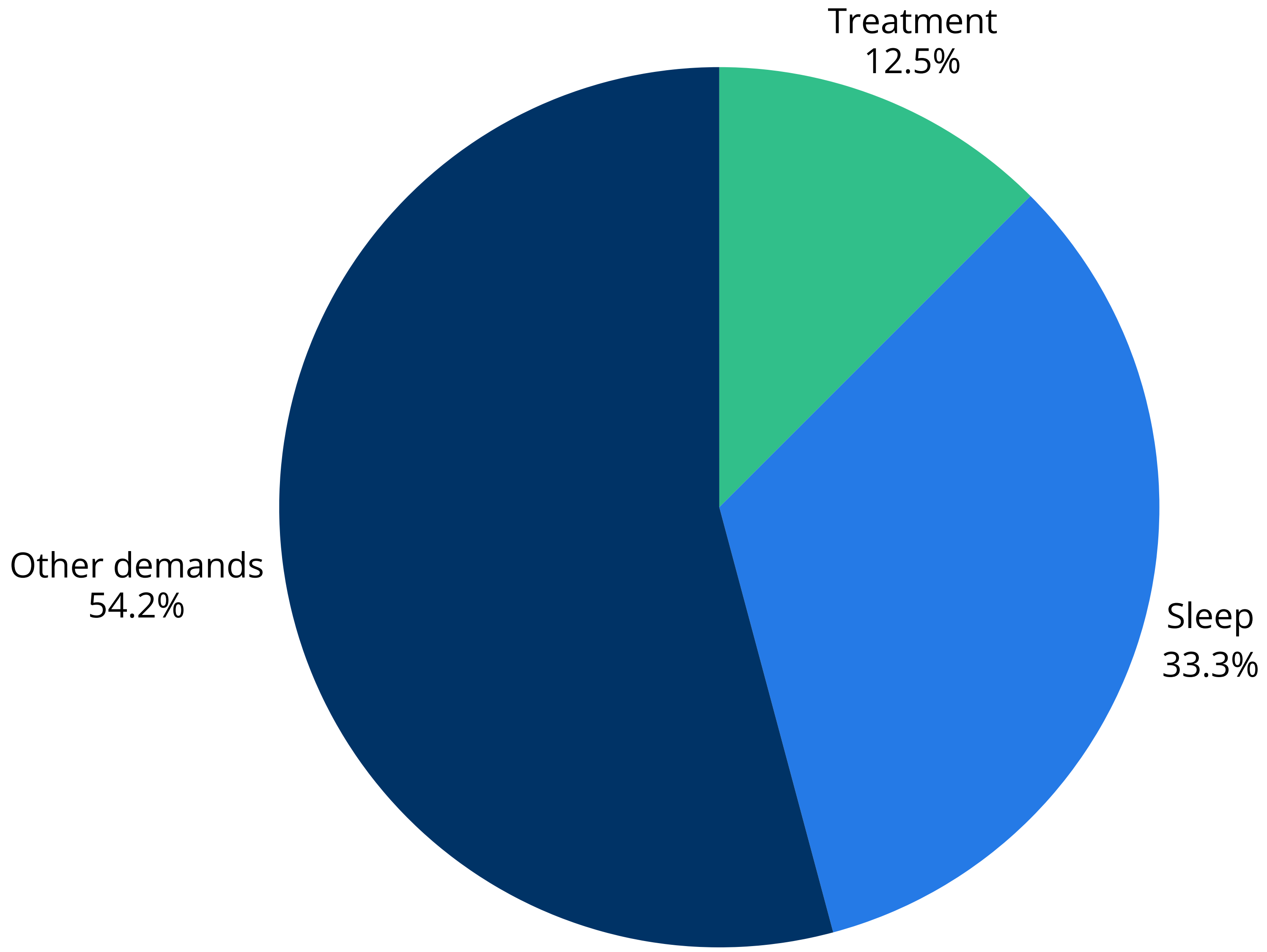
1.3 Age-Specific Challenges

Disease Cycle

From the earliest age, patients and their families should go through:

1. **Acceptance**
2. **Understanding**
3. **Learing how to communicate**

CF Treatment can take up to 3 hours a day



CF is a life-long disease. Building autonomy and individuality beyond the treatment is essential.

CF Across Life Stages: Different Challenges, Different Recommendations

Children

- **Encourage normalization of treatment** from an early age,
- Support parents in **balancing structure with warmth and a sense of normalcy**
- Prioritize **psychological support for parents** when a diagnosis occurs in the newborn period
- Promote **basic CF education in school** settings
- **Ensure access to nutritional support,** recognizing that feeding challenges can generate significant caregiver anxiety

Adolescents

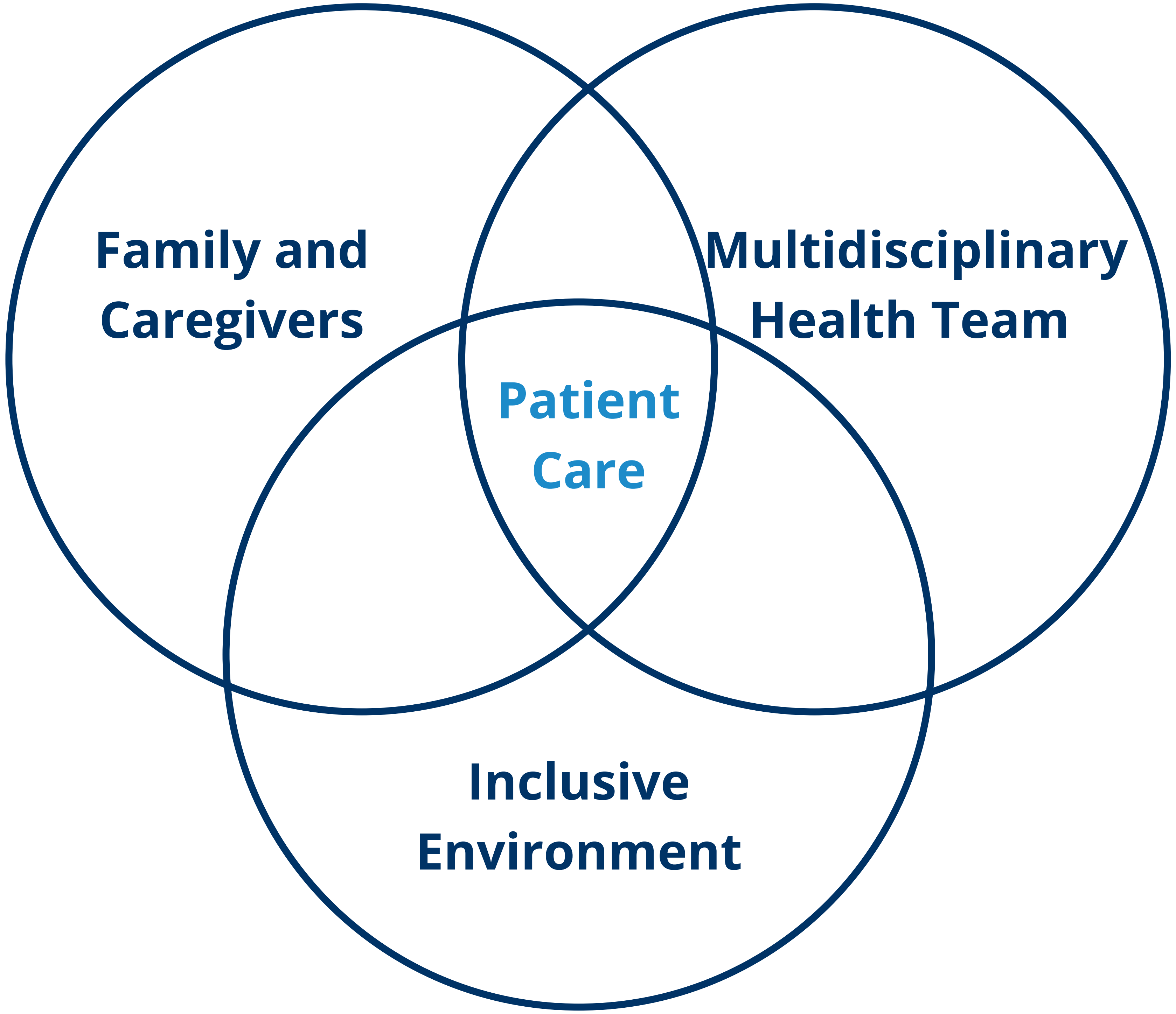
- Recognize the **tension between independence and treatment adherence** as a defining challenge
- Acknowledge that the **treatment burden (~3 hours/day) can impact social life and identity** development
- Support **early and gradual transfer of treatment responsibility** to improve adherence
- **Be aware that sadness, fear of hospitalization, and school disruption** are common and may require additional support

Adults

- Recognize that r**elationships, career development, and family planning require tailored support**
- Address the **transition from pediatric to adult care** as a critical gap, particularly where adult services lack CF-specialized teams
- Acknowledge that **many Latin American countries lack fully established adult CF programs**, requiring system-level support
- Promote **workplace awareness and accommodations** to support ongoing treatment needs

Multidisciplinary care must be provided throughout life.

Supporting families and caregivers in staying healthy and emotionally balanced is key to sustained caregiving



Family and Caregivers

Multidisciplinary Health Team

Patient Care

Inclusive Environment

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1.4 Strategies for Managing Mental Health Issues

“

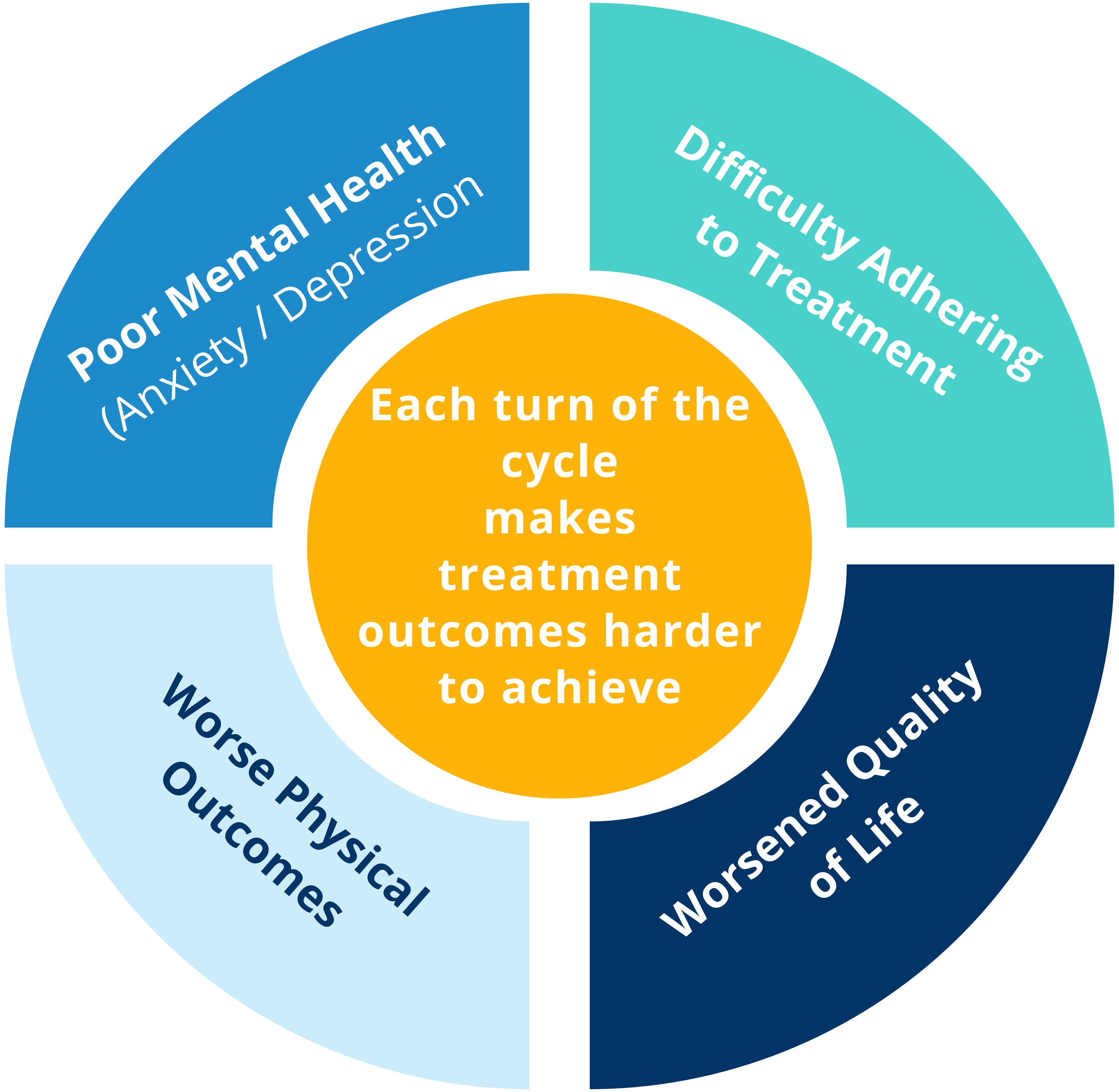
Both people with CF and their caregivers are three times more likely to develop anxiety or depression compared to the population that does not have a chronic condition.

Lic. Sabrina Brancatisano

Psychologist · Asociación Argentina de Fibrosis Quística. · Argentina

The Vicious Cycle: Mental Health and CF Outcomes

Breaking this cycle requires **addressing mental health as a clinical priority** alongside physical treatment.



Poor Mental Health
(Anxiety / Depression)

Difficulty Adhering to Treatment

Worsened Quality of Life

Worse Physical Outcomes

Each turn of the cycle makes treatment outcomes harder to achieve

Coping with Mental Challenges

ADAPTATIVE ≠ PATHOLOGICAL

Helpful Anxiety Harmful Anxiety

Promoting strategies to manage worry, strengthen alignment with personal values, and reinforce identity beyond the disease can help lessen the impact of mental health challenges on treatment engagement and outcomes

Strategies for Supporting Mental Health in CF

Physical Exercise

Regular exercise adapted to CF capacity improves mood, lung function, and quality of life.

Online Community

Virtual peer connection bridges the mandatory physical isolation rule (minimum 2 meters between CF patients).

Medication
(When needed)

Anxiety and depression are clinical conditions. Pharmacological treatment is appropriate when indicated.

Meaningful Hobbies

Activities completely unrelated to illness preserve identity beyond the patient role.

Psychologic Support

A psychologist on the CF care team is a necessary standard team member.

Mindfullness Practices

Present-moment anchoring is useful for catastrophic, future-focused thinking common in CF.

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Module 1 - Wellbeing and Support

1.5 Comprehensive Treatment

Comprehensive CF Care: What It Looks Like in Practice

The Gold Standard

- Single-day multidisciplinary consultation, where the patient sees the full team in one visit
- Quarterly follow-up at minimum, with more frequent visits for unstable patients
- Each visit results in documented, patient-appropriate goals
- Socioeconomic context is integrated into all planning
- Mental health is monitored at every visit

> *“The patient does not need to keep going to the hospital for separate appointments. Everything is done on the same day.”*
>
> – Dr. José Luis Lezana Fernández, AMFQ, Mexico

The multidisciplinary team:

- Pneumologist (Specialist Lead)
- Nutritionist
- Physiotherapist / Respiratory Therapist
- Psychologist
- Social Worker
- Nurse
- Endocrinologist - currently missing from most teams

Module 2

Family Support Systems

CF is not experienced by the patient alone; it is lived by the entire family and their support ecosystem

2.1 Best Practices

2.2 Support Networks

Module 2 - Family Support Systems

2.1 Best Practices

> ***I wish it could reach everyone, but since it doesn't reach everyone, we cannot forget that group. We have to keep fighting until modulators can reach them and be accessible to every patient.***

Dra. Claudia Jiménez

Respiratory Therapist · Fundación Colombiana para Fibrosis Quística. · Colombia

Early Diagnosis:
The Pathway That Changes Everything

Newborn Screening

IRT/IRT or IRT/DNA protocol (the heel prick test). Standard practice where universal screening exists

Sweat Chloride Test

Confirmatory test measuring chloride in sweat. A sweat chloride ≥60 mmol/L is diagnostic for CF

GFTR Gene Sequencing

Essential for complex cases and to determine which modulator therapy the patient is eligible for

Why it matters: Early diagnosis means families can adapt, build routines, and begin treatment before clinical deterioration. Where newborn screening is not universal, patients often present at age 5–7, losing critical early treatment years

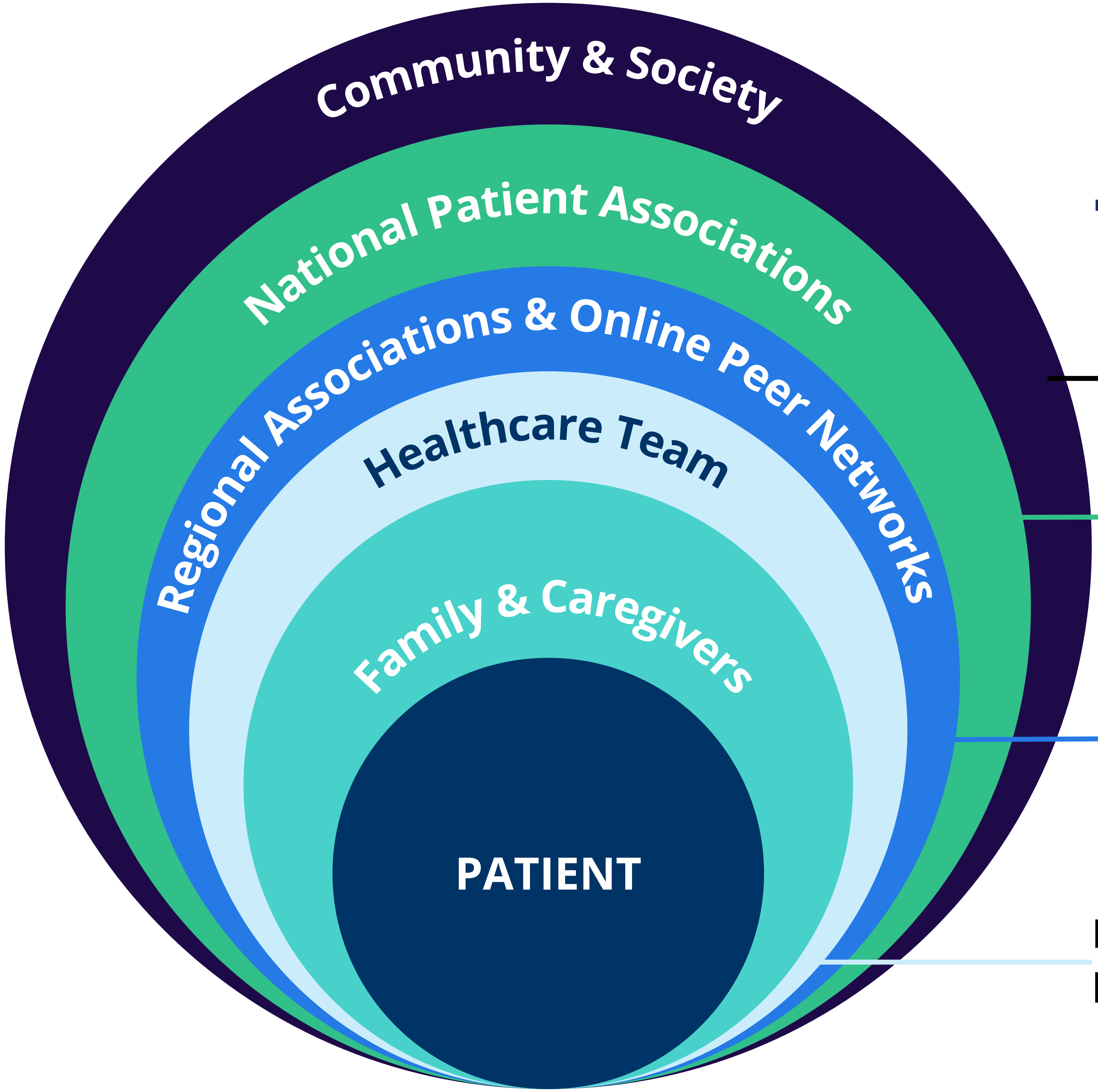
Disciplines within Comprehensive CF Care

- Multidisciplinary Teams
- Socioeconomic Considerations
- Comprehensive Clinical Guidelines
- Health System Strengthening and Policy Strategies
- Proper CF Education
- Data Systems and Orphan Disease Recognition

Module 2 - Family Support Systems

2.2 Support Networks

The CF Support Ecosystem: Layers That Work Together



Increases awareness and **inclusion**

Advocate for **policy change**, build patient registries, and lead medication access campaigns.

Provide **family support** close to home, coordinate access to equipment, and connect local care networks & **Enable peer connection** for patients who cannot travel or meet in person due to infection risk.

Deliver clinical care and bridge patients to all other layers through referrals and coordination.

Understanding the Caregiver's Reality

9 in 10 caregivers are mothers

Often single parents, they carry the full weight of CF management with little shared support or relief.

Many reduce or give up their jobs

The demands of daily CF care frequently force caregivers out of employment, with lasting consequences for family income that largely go unrecorded.

Exhausted caregivers put patients at risk

When caregivers are depleted, treatment consistency suffers, making caregiver wellbeing a clinical concern, not just a personal one.

Patient autonomy eases the load

As the person with CF gradually takes ownership of their treatment, the caregiving burden naturally lightens, benefiting both sides of the relationship.

> *"Nobody has an inexhaustible battery. Very often, caregivers neglect their own basic needs."*
>
> *– Lic. Sabrina Brancatisano · Psychologist, Asociación Argentina de Fibrosis Quística*

Module 3

Patient Education and Treatment Adherence

Concrete tools for daily management, treatment navigation, and system advocacy for patients and families.

3.1 Equitable Access
3.2 Treatment Adherence
3.3 Coping Strategies
3.4 Nutrition and Supplements
3.5 Respiratory Therapy
3.6 Patient Registry

Module 3 - Patient Education and Treatment Adherence

3.1 Equitable Access

Access Equity:
Regulatory Approval ≠ Patient Access

APPROVED ≠ ACCESSIBLE

SYSTEM-LEVEL BARRIERS:

1. Cost and insurance barriers
2. Not included in the public formulary
3. Geographic distribution challenges
4. Food insecurity undermining nutrition targets

What patients still lack access to:

1. Modulators
2. Baseline medications
3. Nebulized antibiotics
4. Enzymes
5. Specialized equipment

Even when therapies are approved, patients face multiple barriers that limit real-world access.
Fighting for access is part of CF care

Legal and Policy Tools for Medication Access

Argentina

Coverage Mandate

- National CF Law requires coverage of all CF medications, including modulators
- Enforcement often still depends on legal action to compel compliance

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Brazil

Judicial Orders + SUS

- TInitial Trikafta access was achieved through court orders, followed by SUS inclusion in 2023
- Some patients still rely on judicial mechanisms to obtain newer therapies

Colombia

Tutela Constitucional

- Urgent constitutional actions (tutelas) can enforce health rights through EPS operators
- Patient associations provide template letters and support for filing

Mexico

Amparo Judicial

- Constitutional amparo actions can challenge government inaction on medication access
- Amparo has been used successfully but typically requires legal representation

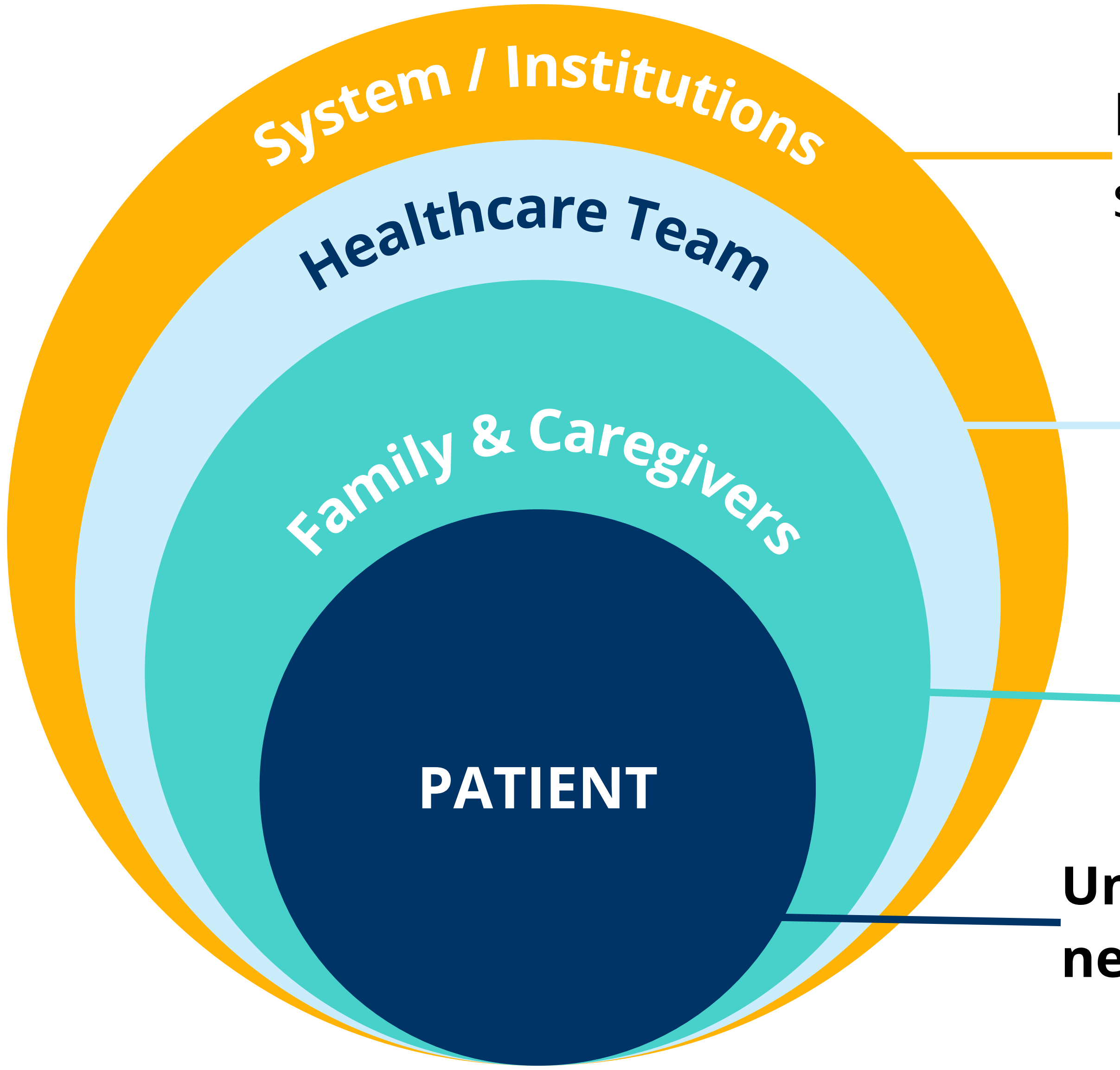
Your patient association can provide guidance, templates, and legal contacts specific to your country.

Module 3 - Patient Education and Treatment Adherence

3.2 Treatment Adherence

Adherence to Treatment:
A Shared Responsibility

The goal is not perfect adherence, but the best possible adherence for each individual



Ensure medications, equipment, and support are **accessible and affordable**

Remove clinical barriers, adjust protocols, and address the root causes of adherence gaps

Build enabling routines without adding burden or shame around adherence gaps

Understands WHY each treatment is necessary and adapts routines to real life

Be aware of the **invisible social gaps**: Financial burden is a real condition in CF patients' families across the region.

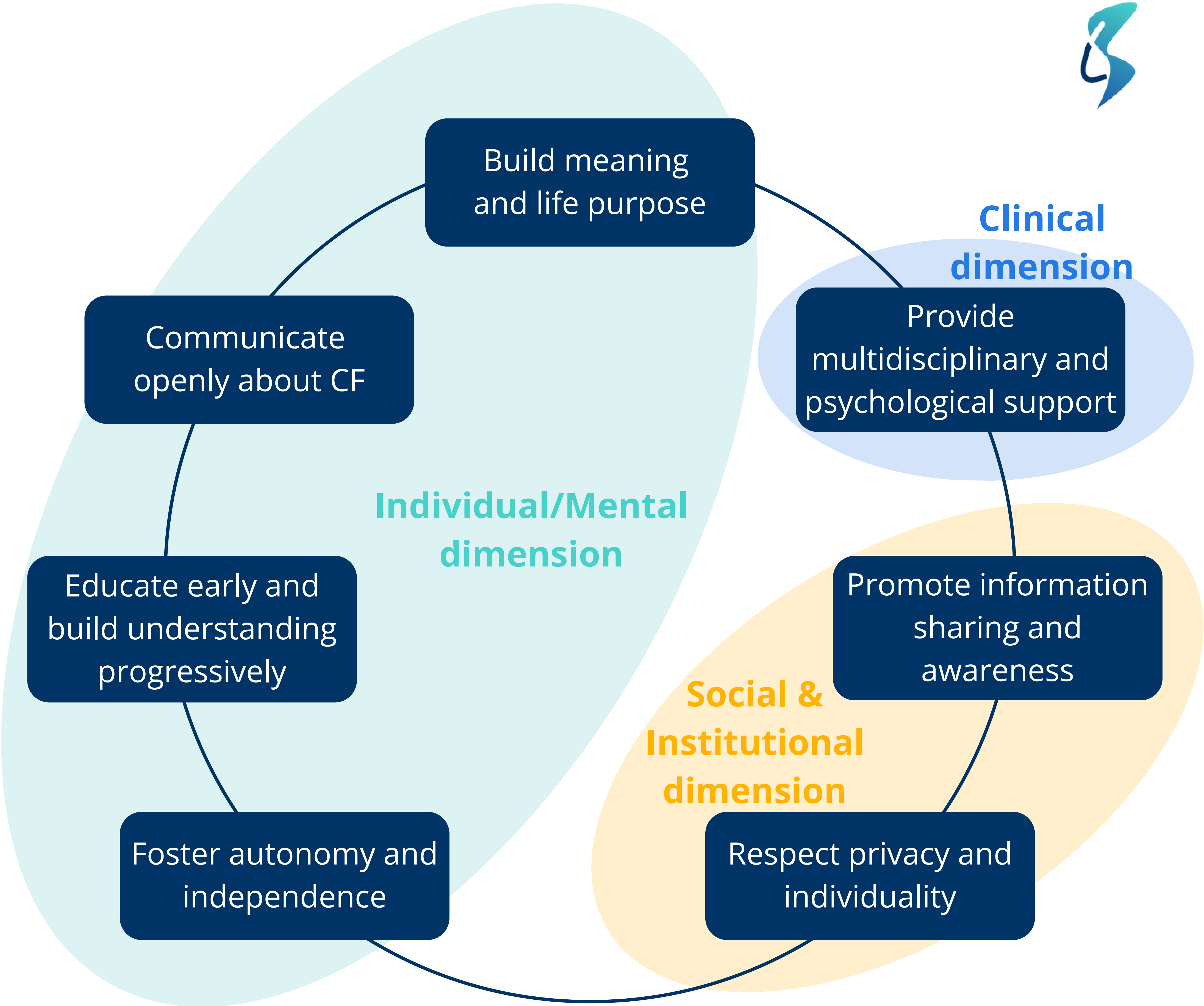
Understanding **WHY adherence gaps occur** is more productive than addressing gaps alone.

Module 3 - Patient Education and Treatment Adherence

3.3 Coping Strategies

Coping with CF: A Multidimensional Response

Remember: You are not your disease.



Build meaning and life purpose

Provide multidisciplinary and psychological support

Promote information sharing and awareness

Respect privacy and individuality

Foster autonomy and independence

Educate early and build understanding progressively

Communicate openly about CF

Individual/Mental dimension

Clinical dimension

Social & Institutional dimension

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Module 3 - Patient Education and Treatment Adherence

3.4 Nutrition and Supplements

Nutrition in CF: Demanding, Consequential, and Often Underresourced

CF patients have 2X caloric needs vs. the general population due to malabsorption and metabolic demand

- **Pancreatic Enzyme Therapy (PERT):** 85% of CF patients have pancreatic insufficiency. Enzymes are required with every meal and snack, with dosing adjusted regularly
- **The Modulator Transition:** Patients starting modulators often shift from hypercaloric to more normal intake. This must be clinically monitored to prevent overcorrection
- **Fat-Soluble Vitamins:** Vitamins A, D, E, and K require monitoring. Vitamin D monitoring is important, but sometimes neglected in routine follow-up
- **Food Insecurity:** A documented and underacknowledged challenge. Nutritional plans must be economically realistic; if not, they are not useful

> *“If children around the age of four reach normal nutritional status, by the age of 18, they will have better respiratory function."*
>
> – Dra. Claudia Angarita, Nutritionist, Fundación Colombiana para Fibrosis Quística

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Module 3 - Patient Education and Treatment Adherence

3.5 Respiratory Therapy

> ***Even if they [patients] are on modulators, we must explain to them that this does not replace respiratory physiotherapy.***

Dra. Claudia Jiménez

Respiratory Therapist · Fundación Colombiana para Fibrosis Quística. · Colombia

The Nebulization Sequence: Order Matters

Bronchodilator
(e.g., Salbutamol)

Opens airways FIRST and ensures subsequent medications reach deeper lung tissue

Hypertonic Saline
(3,5% to 7%, depending on the treatment)

Hydrates and loosens thick mucus, making it easier to mobilize

Dornase Alfa
(DNase / Pulmozyme)

Breaks down DNA in mucus, dramatically reducing its viscosity

Airway Clearance
(ACBT / PEP / autogenic drainage)

An active technique to physically mobilize and expectorate loosened mucus

Inhaled Antibiotic
(If prescribed)

Administered **last;** it penetrates airways that have been cleared and opened

IMPORTANT: CFTR modulators reduce mucus burden but **do NOT replace airway clearance** or other treatment aspects. **Physiotherapy remains essential** for all eligible patients

Module 3 - Patient Education and Treatment Adherence

3.6 Patient Registry

Patient Registries: A Frontline Advocacy Tool

Why Patient Registries Matter

- Registries **generate the data** needed to advocate for medication inclusion, funding, and policy change
- Without registry data, health ministries can claim a condition is too rare for public coverage; **registry data breaks that argument**
- They **track patient populations**, outcomes, and treatment patterns over time
- **Enrollment is an act of advocacy;** every registered patient strengthens the collective case
- Patients can **ask whether their data is included** and how it is being used

In the Region

Argentina

AAER maintains a national patient directory and registry as part of advocacy work

Brazil

GBEFC (Grupo Brasileiro de Estudos em Fibrose Cística) is a specialist network with a national data source

Colombia

Colombia's National Registry of Rare Diseases mandates CF reporting nationwide. Of the 1,282 CF patients currently registered, only 253 have access to modulators

Mexico

AMFQ coordinates data collection; registry expansion linked to public access advocacy

WRAP-UP

Key Messages

The Five Key Messages

TREATMENT

The landscape has changed and continues to change

CFTR modulators have transformed CF. Fighting for access is part of care

MENTAL HEALTH

Caring for the mind is part of the treatment

Anxiety and depression are up to 3× more common. Unaddressed mental health directly undermines outcomes

FAMILY

The patient isn't the only one who needs care

Chronic illness is never lived alone. Caregivers carry an immense and underrecognized load that directly affects patient outcomes

ADHERENCE

Treatment must fit into life, not the other way around

Perfect adherence is not achievable. The goal is the best possible adherence for each individual's real life

ACCESS

Approval is not access, and this must change

In most of Latin America, modulators are approved but out of reach. Registries, legal tools, and advocacy are what close that gap