CYSTIC FIBROSIS REGISTRY OF TURKEY (CFRT) DATA FOR 2022 – SUMMARY REPORT

PREFACE

In this report, we present a summary of the characteristics of our cystic fibrosis (CF) patients followed up in Turkey in 2022.

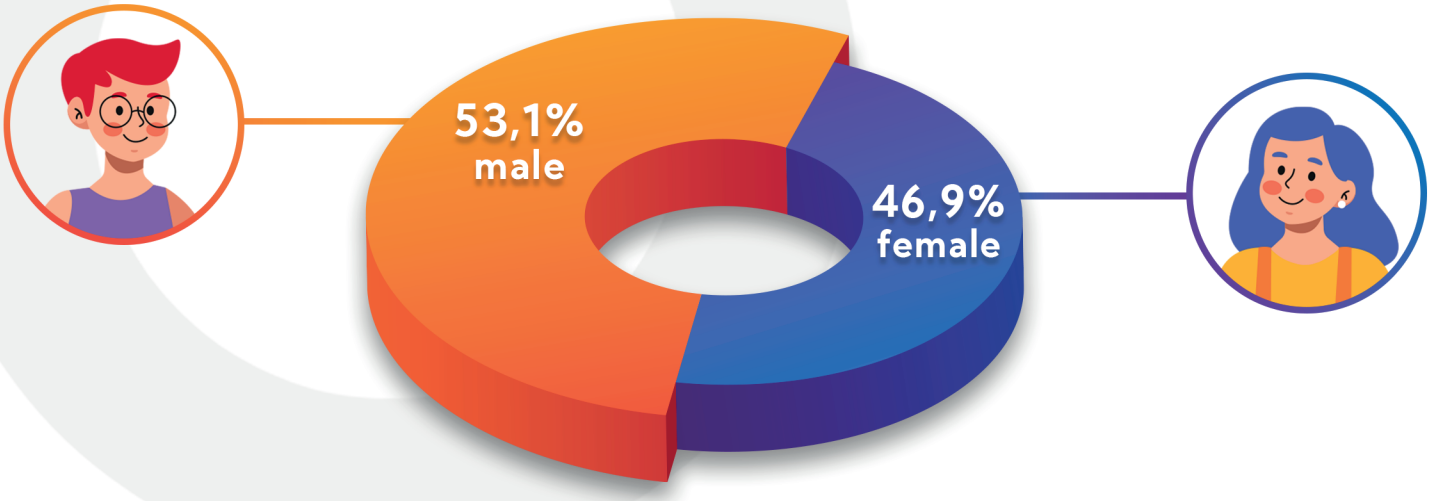
The "Cystic Fibrosis Registry of Turkey" (CFRT) which is established by the "Pediatric Respiratory Diseases and Cystic Fibrosis Association" annually records disease-related information of CF patients followed up in various centers in our country. This information helps to better understand CF disease, to have more information about our patients' disease-related characteristics, quality of life and natural course of the disease, to compare it with our previous data, to see the changing numbers over time and to identify patients who will benefit from special or preventive treatment methods in our country.

You can find more detailed information about the CFRT on our website
https://www.kistikfibrozisturkiye.org/hasta-kayit-sistemi/

Our detailed report for 2022 is available at
https://www.kistikfibrozisturkiye.org/wp-content/uploads/2024/01/2022-UKKS.pdf

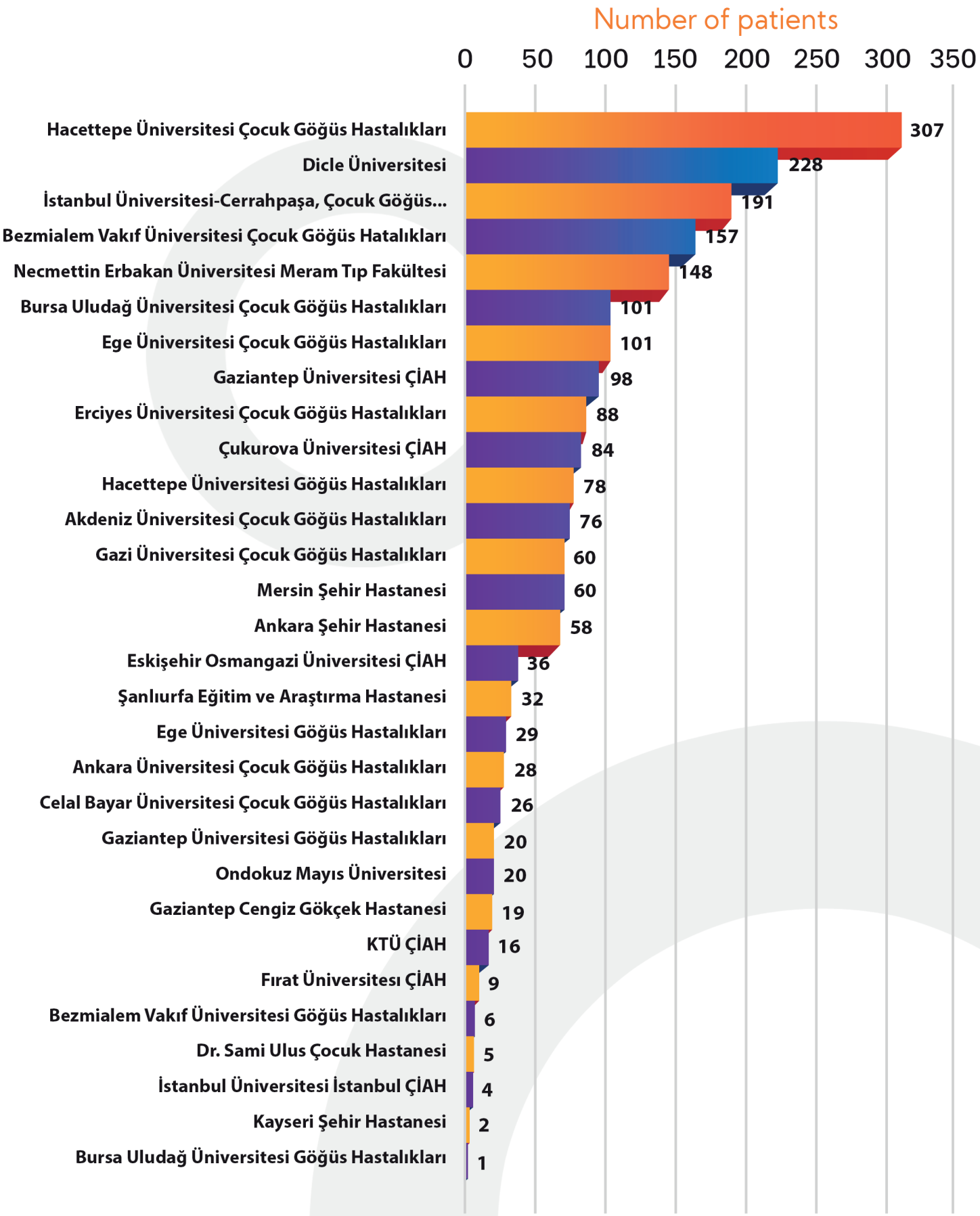
1. NUMBER AND DISTRIBUTION OF PATIENTS

In our country, there are 2088 CF patients from 30 centers registered in the CFRT in 2022.

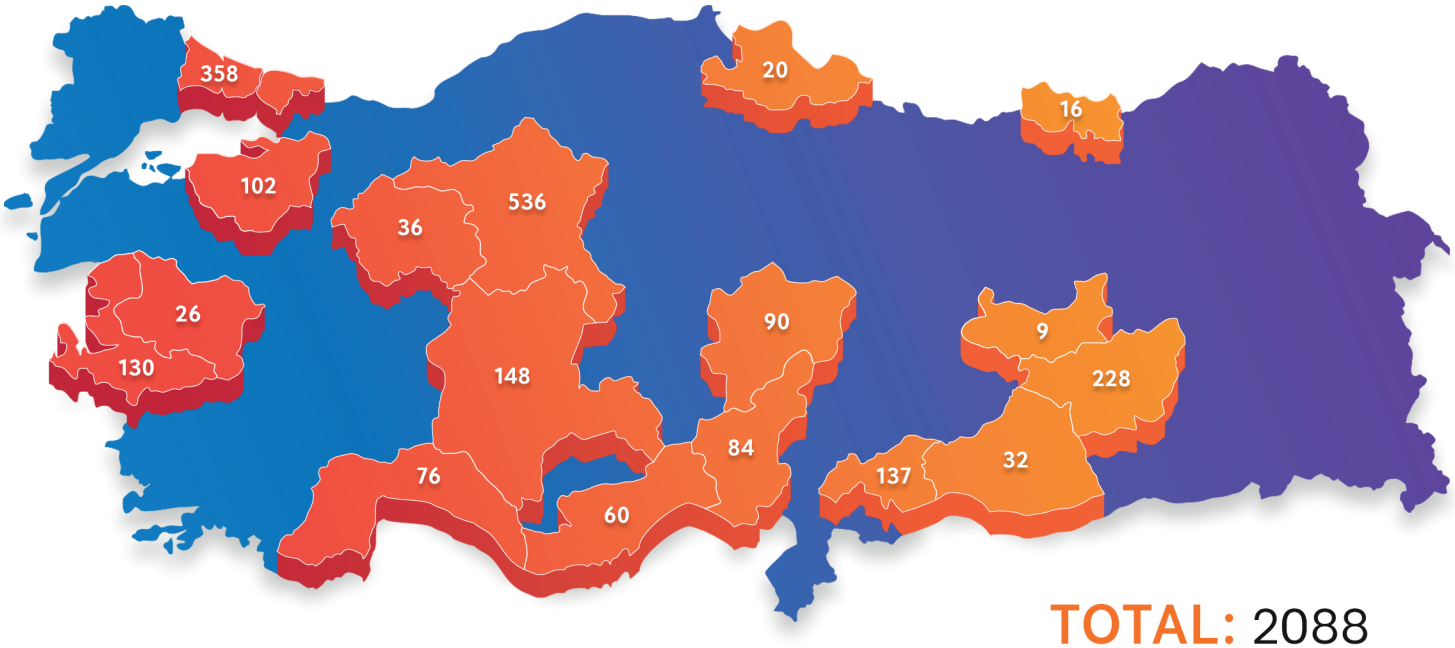


There are 53.1% male and 46.9% female patients registered in the CFRT.

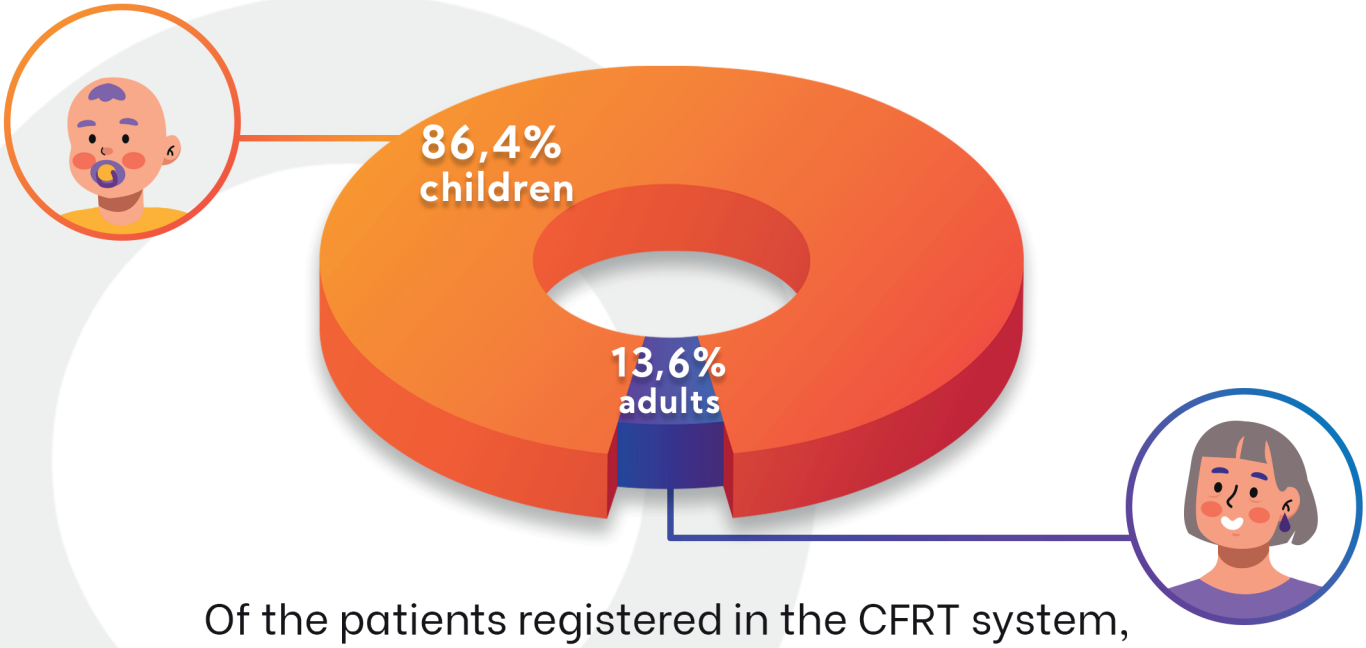
Number of patients registered by centers



Distribution of registered patients



TOTAL: 2088

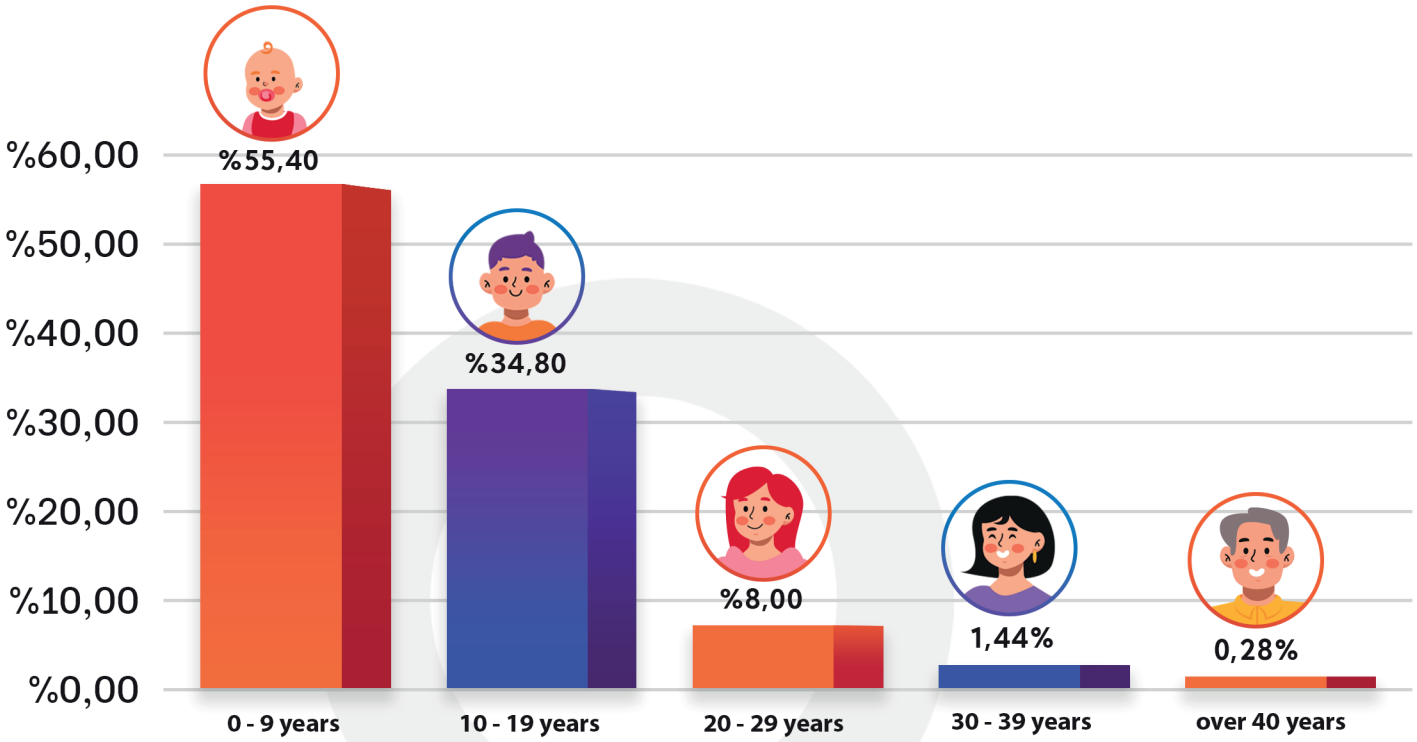


Of the patients registered in the CFRT system, 86.4% were children and 13.6% were adults.

Median diagnostic age for CF patients is

4 months

Age distribution of patients



2. GENETICS

• CF is a genetic disease. Individuals with CF have two CF-causing mutations, one inherited from the mother and the other from the father.

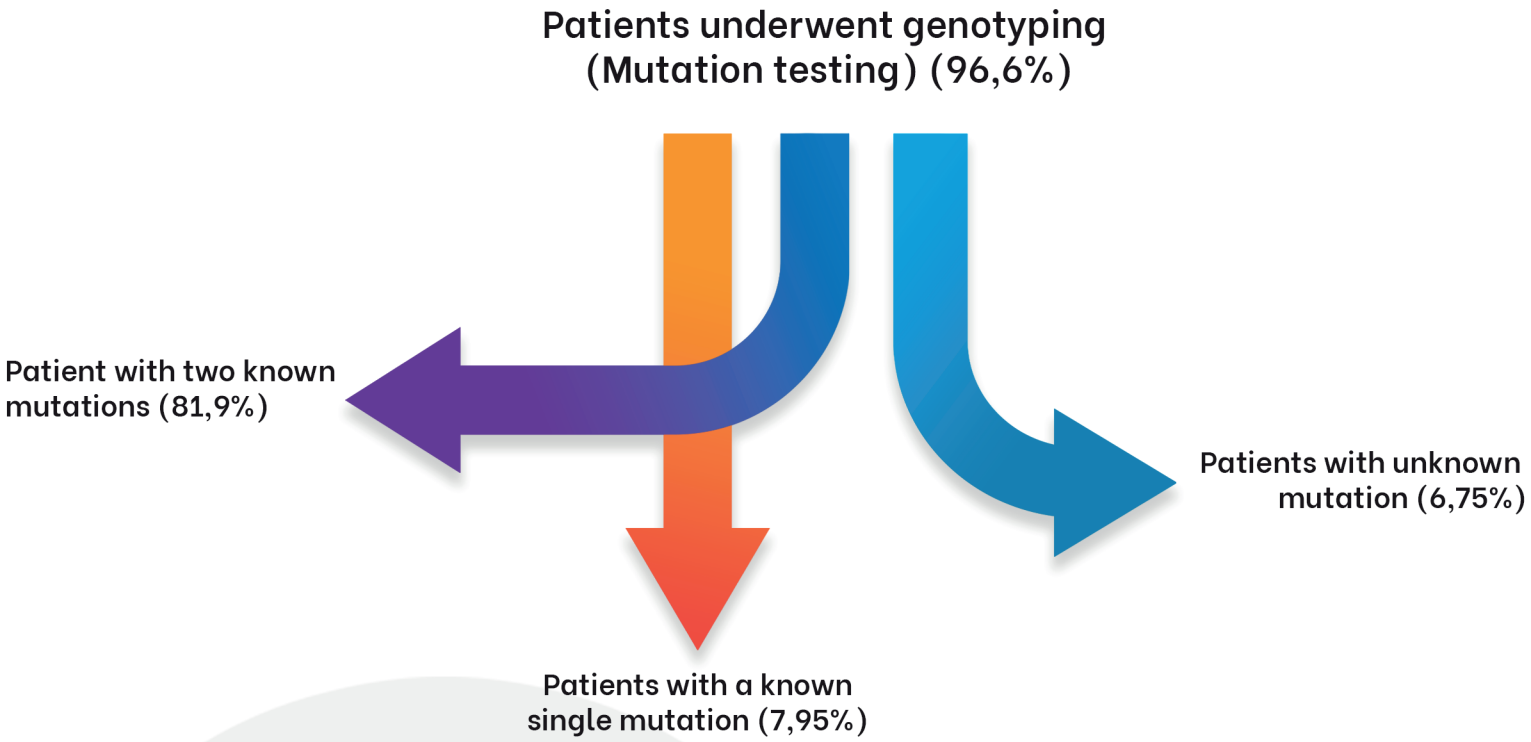
• In our country, F508 deletion (F508del) is the most common mutation in patients who underwent genetic analysis and whose mutation could be detected (22.25%).

• F508 deletion is the most common CF-causing mutation in Europe.

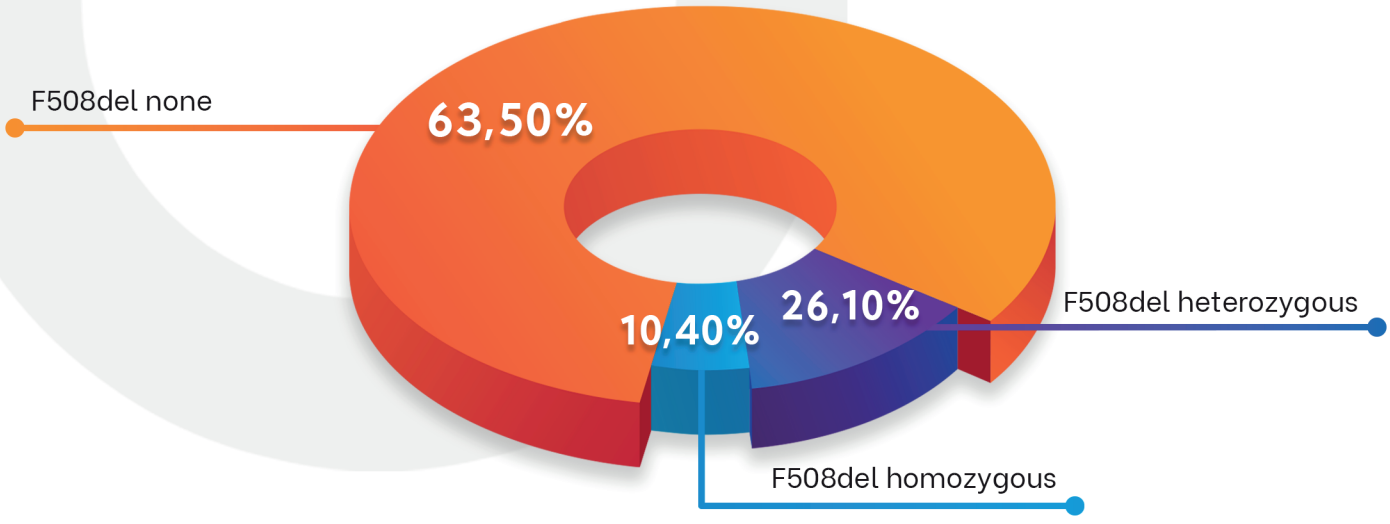
• Homozygous: This means that both mutations in the patient are identical.

• Heterozygous: This means that the mutations in the patient are different from each other.

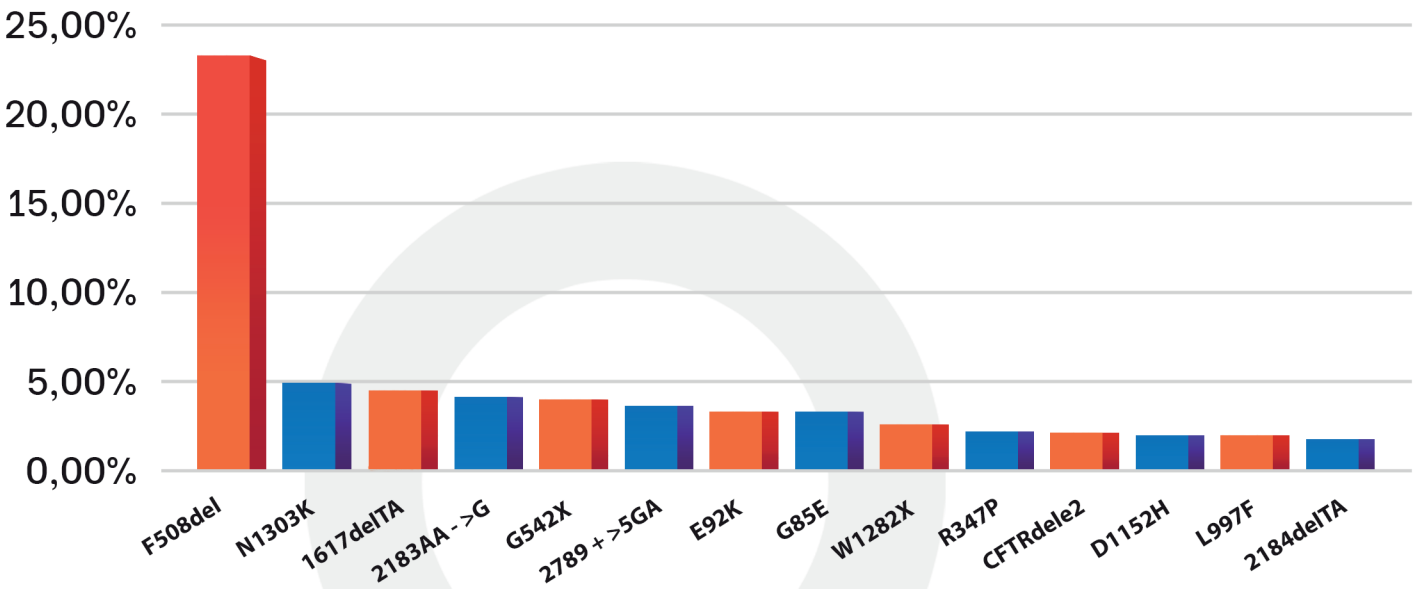
Genotyping results of patients



Distribution of mutations



Most common mutations in genotyping



3. PULMONARY FUNCTIONS

• Pulmonary involvement in CF is the most important factor determining the clinical severity of the disease and the duration and quality of life; therefore, close monitoring of pulmonary functions is important.

• Pulmonary function tests can be performed in patients over 6 years of age and are evaluated by measuring FEV1.

The severity of lung disease is determined according to FEV1 percentage values:

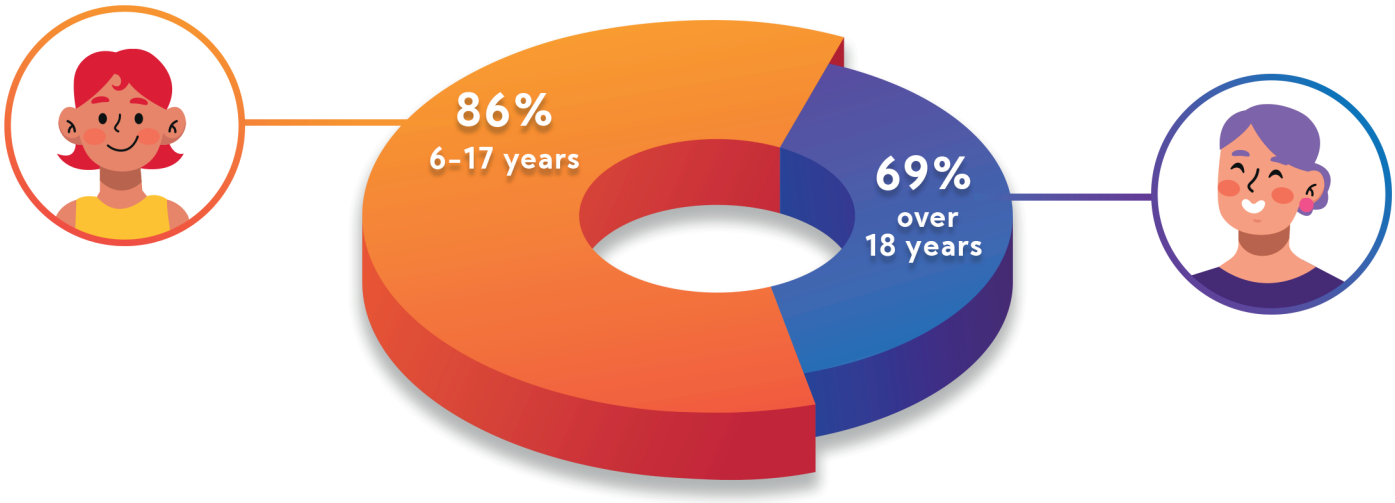
• Normal: 80% and above

• Mild disease: 60%-79%

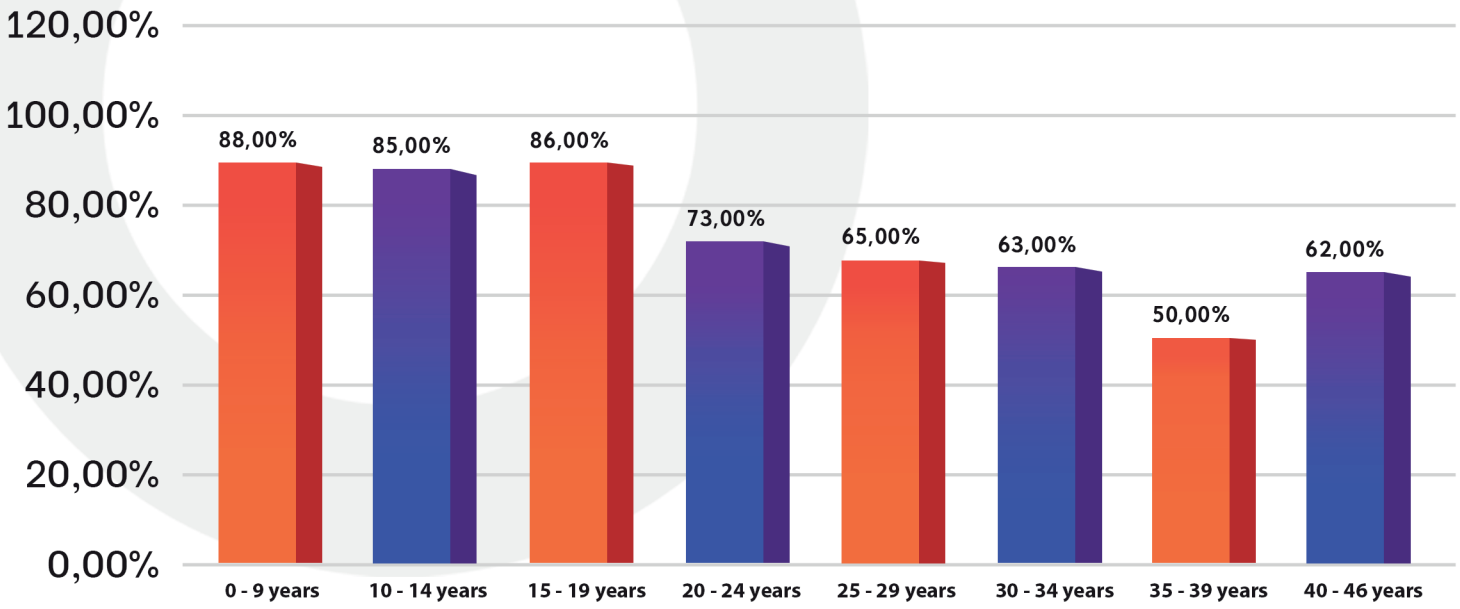
• Moderate disease: 40%-59%

• Severe disease: 39% and below

Median FEV1 percentages of patients



Median FEV1 percentages of patients based on age distribution



4. NUTRITION

• Digestive problems occur in 85% of CF patients as a result of the inability of pancreatic secretions (enzymes) to be released due to the disease or to flow into the intestines due to obstructions in the ducts.

• Since CF patients cannot digest carbohydrates, proteins and fats, they have abundant, loose-fatty, light-colored, foul-smelling and numerous stools, abdominal distension and gas, and if left untreated, individuals cannot gain enough weight and there is retardation in growth and development.

• Good nutrition in CF is important to maintain pulmonary functions.

• Body mass index, height and body weight z scores are used to monitor nutrition. Low values of these values indicate that the nutritional status of the individual is inadequate.

• Z-score is a score that indicates how much a value differs from healthy people of similar age and gender. Since standard values vary for children in different age groups, z scores are used to assess the growth. When this score is below zero, it means that it is low compared to people of similar age and gender.

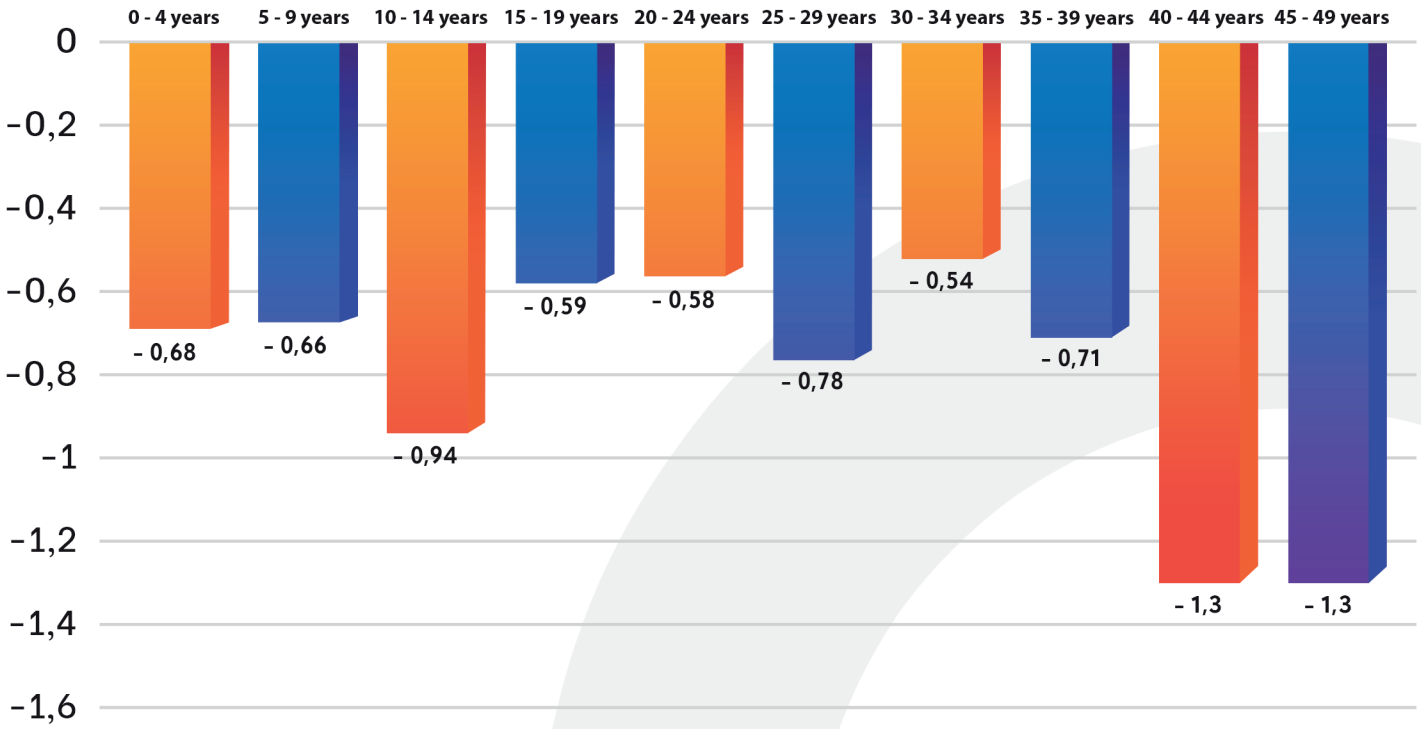
• The low height and body weight z-scores of our patients are an important indicators which show that their growth and nutritional status are not adequate.

a. Weight


Body weight median Z-score distribution
0,6
0,4
0,2
0
-0,2
-0,4
-0,6
-0,8
-1
-1,2
0 - 4 years
5 - 9 years
10 - 14 years
15 - 19 years
20 - 24 years
25 - 29 years
30 - 34 years
35 - 39 years
40 - 44 years
45 - 49 years
- 1,04
- 0,73
- 0,78
- 0,81
- 0,84
- 0,67
- 0,48
- 0,25
0,43
- 0,33

b. Height

Height median Z-score distribution



c. Body Mass Index (BMI):

• BMI is calculated by dividing body weight by the square of height.

BMI = Body weight (kg) / Square of height (m^2)

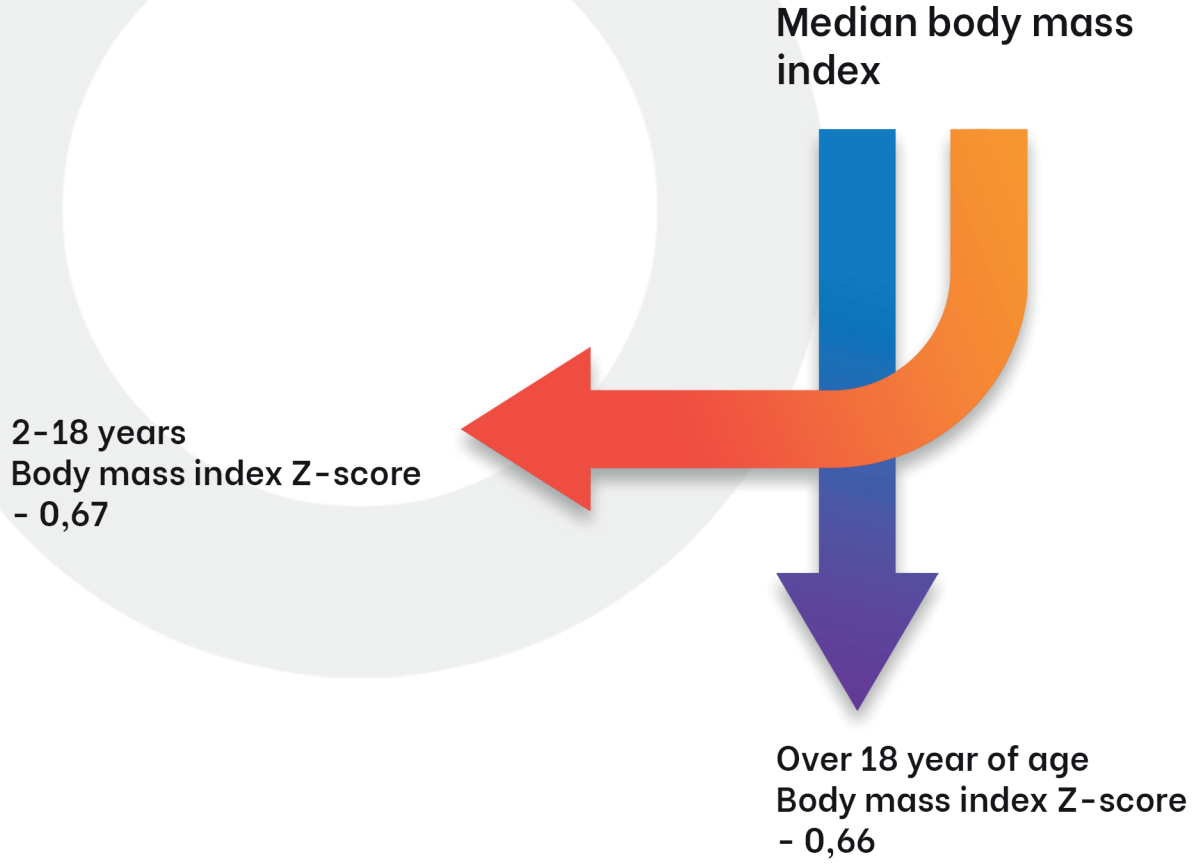
• **BMI assessment:**

2-18 years of age: z-scores are more commonly used.

Over 18 years of age: The formula-calculated value of BMI is used:

Low: 18.5 and below
Normal: 18,5-24,9
High: 25 and above

• The low BMI z-score of our patients between the ages of 2 and 18 years and the low BMI values of our patients aged 18 years and older are important indicators which show that the growth and nutritional status of our patients are not adequate.



MICROBIOLOGY

• Mucoid secretions in the lungs of CF patients cause airway obstruction resulting impaired lung ventilation. Some microorganisms easily settle in this environment and frequent lung infections occur from an early age.

• The most common microorganisms that settle in the lungs of individuals with CF are *Staphylococcus aureus* and *Haemophilus influenzae* , at younger ages and *Pseudomonas aeruginosa* at older ages. Pseudomonas in particular causes chronic (long-term) infection of the lungs and is the most important cause of ongoing lung damage.

• Infection control is an important issue as infection transmission is known among individuals with CF.

• *Burkholderia cepacia* complex, *Stenotrophomonas maltophilia*, *Achromobacter xylosoxidans*, Aspergillus species, non-tuberculous mycobacteria and respiratory viruses can also be seen in individuals with CF.

Bacteria	Infection frequency
 Chronic Methicillin sensitive *Staphylococcus aureus*	17,39%
 Chronic *Pseudomonas aeruginosa*	17,24%
 Chronic Methicillin resistant *Staphylococcus aureus* (MRSA) infection	5,08%
 *Haemophilus influenzae*	1,25%
 *Stenotrophomonas maltophilia*	0,29%
 *Achromobacter* infection	0,24%
 Chronic *Burkholderia cepacia complex*	0,38%
 Non-tuberculous mycobacteria infection	1,24%

Hospitalization:

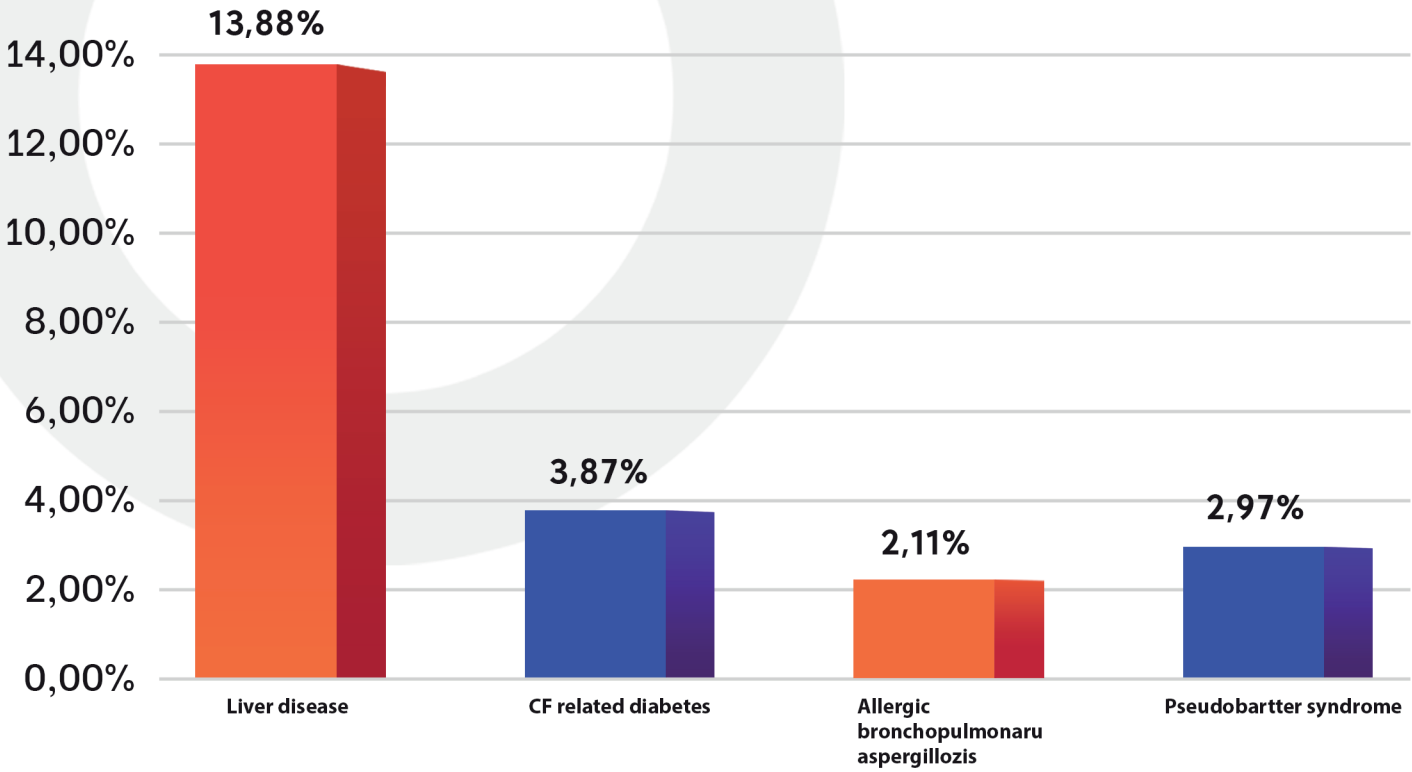
20,44%

Patients hospitalized (due to CF) to receive intravenous antibiotics for at least 1 day: 20.44%

Complications:

• The lungs, pancreas, intestines, liver, sinuses and reproductive organs are affected to varying degrees in CF.

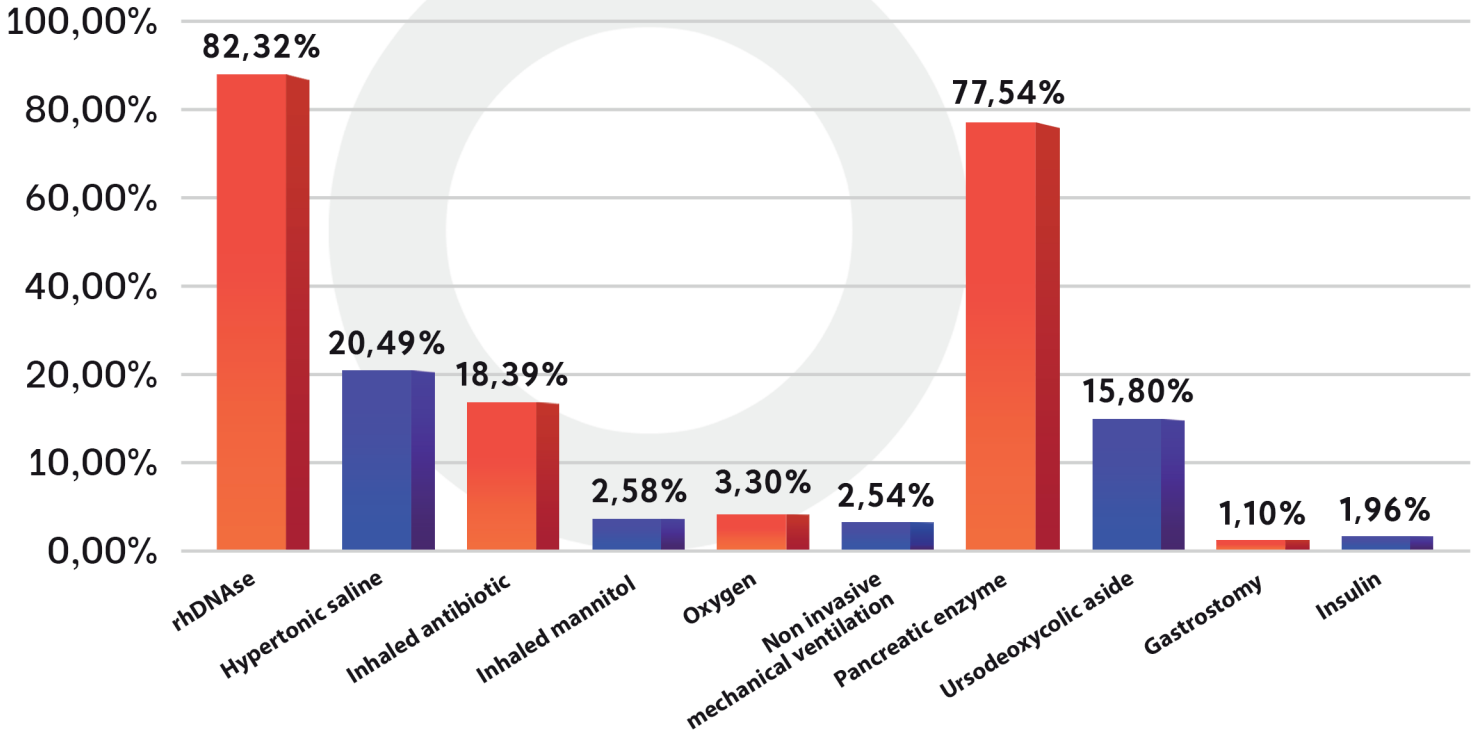
• Complications of CF can include liver disease, diabetes, bone mineralization disorders (osteoporosis) and Pseudobartter syndrome (salt loss).



TREATMENT

• Treatments in CF are aimed to decrease or improve symptoms and prevent complications related with disease.

Treatment distribution of patients



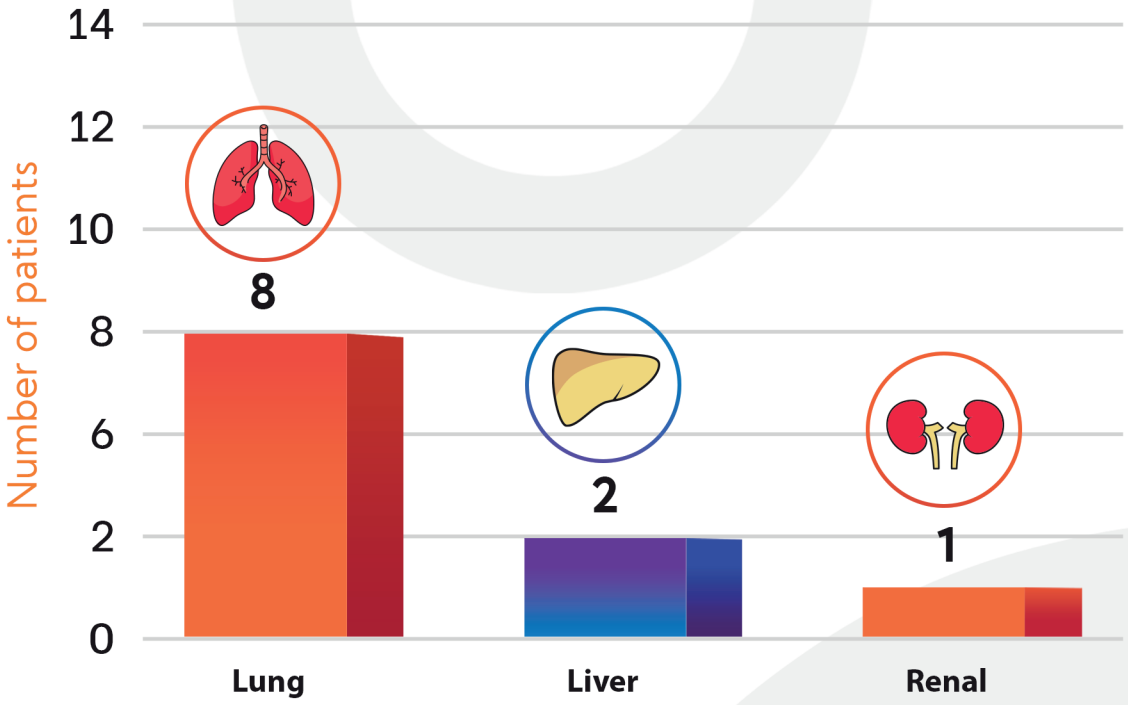
Number of patients using modulator therapy

Modulator treatment	Number of patients
Ivacaftor	10
Lumacaftor/Ivacaftor	10
Tezacaftor/Ivacaftor	8
Elexacaftor/ Tezacaftor/ Ivacaftor	124

Number of people with CF living in 2022 with transplanted organs

- Lung transplantation is recommended in individuals with CF who have progressive respiratory failure and end-stage lung disease.

- Liver transplantation is performed in patients who develop cirrhosis in CF.



TOTAL: 11