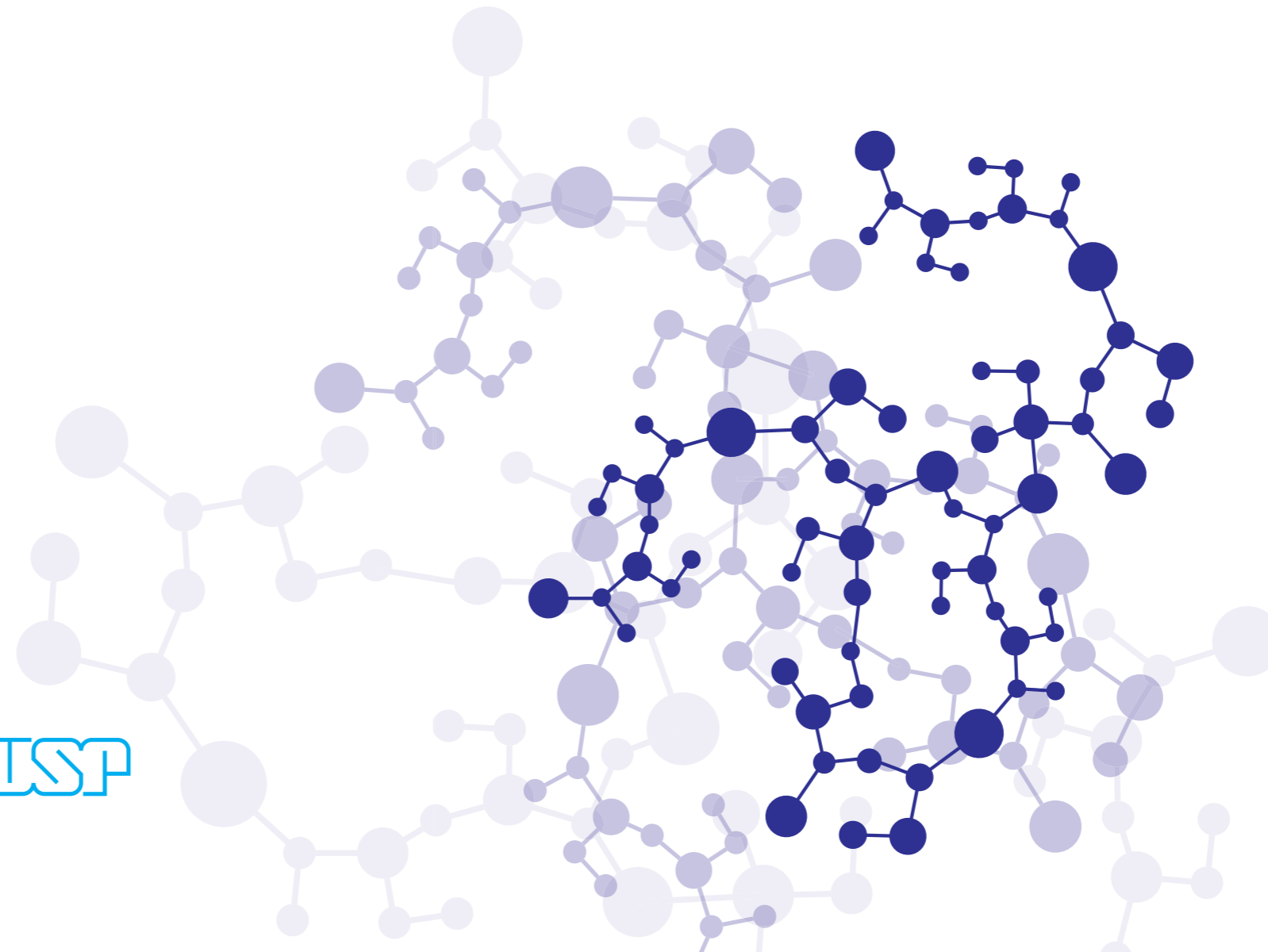


2012

Brazilian
Cystic Fibrosis
Patient Registry



www.gbefc.org.br





BRAZILIAN CYSTIC FIBROSIS PATIENT REGISTRY (REBRAFC) 2012 ANNUAL REPORT

To all persons interested in cystic fibrosis,

In 2013 the Brazilian Group for the Study of Cystic Fibrosis (GBEFC) completed 10 years of existence and ended the year with a record of participation in the Brazilian Cystic Fibrosis Patient Registry (REBRAFC), reaching the mark of 2,669 patients registered in 35 Centers throughout the country (page 38). The initiative aims to improve CF patients care in our country, through the knowledge of the real situation of our country, including diagnostic and treatment practices, and clinical, microbiological and functional outcomes. It is clear that much remains to be done by patients in the country, and that regional differences probably reflect the model of public health care (SUS), which despite proclaiming an universal access to health, has a decentralized approach that results in significant difficulties in access to diagnostic and therapeutic resources in several regions of the country.

The community of health professionals involved in the care of CF patients, through participating in the REBRAFC, manifests itself by exposing the reality of healthcare practice and patients' status, with the hope that this initiative may contribute to changes in government attitudes and result in better health care for individuals with CF.

Institution	City	State	Responsible
Hospital de Clínicas de Porto Alegre - UFRGS	Porto Alegre	RS	Fernando Abreu e Silva
Hospital de Clínicas de Porto Alegre - Adultos	Porto Alegre	RS	Paulo de Tarso Roth Dalcin
Hospital São Lucas - PUCRS	Porto Alegre	RS	Paulo Cauduro Maróstica
Santa Casa de Porto Alegre	Porto Alegre	RS	Gilberto Bueno Fischer
Hospital Santa Isabel	Blumenau	SC	Glaurir Maria Foletto
Hospital Infantil Joana de Gusmão	Florianópolis	SC	Norberto Ludwig Neto
Hospital Infantil Jeser Amarante Faria	Joinville	SC	Tiago Neves Veras e Rafaela C. Benvenuti da Costa
Hospital das Clínicas da UNESP	Botucatu	SP	Giesela Fleischer Ferrari
Hospital das Clínicas da UNICAMP (pediatria)	Campinas	SP	Antonio Fernando Ribeiro
Hospital das Clínicas da USP Ribeirão Preto	Ribeirão Preto	SP	Lidia Alice Gomes M. M. Torres
Hospital de Base Fac Med de SJ Rio Preto	São José do Rio Preto	SP	Katia Izabel de Oliveira
Irmadade da Santa Casa de Misericórdia de São Paulo	São Paulo	SP	Neiva Damaceno
Instituto da Criança do Hospital das Clínicas da FMUSP	São Paulo	SP	Joaquim Carlos Rodrigues
Hospital da UNIFESP	São Paulo	SP	Sonia Mayumi Chiba
Hospital das Clínicas da FMUSP	São Paulo	SP	Rafael Stelmach
Consultorio Fabiola Adde	São Paulo	SP	Fabiola Vilac Adde
Centro de Puericultura - CPAP	São Paulo	SP	Luiz Vicente Ribeiro F. da Silva Filho



Centers that contribute to this report by inserting follow-up data of 2012 (alphabetical order per Brazilian state).

Institution	City	State	Responsible
Hospital Universitário Prof. Alberto Antunes – UFAL	Maceió	AL	Katharina Vidal de Medeiros Moura
Hospital Especializado Otavio Mangabeira	Salvador	BA	Maria Angélica Santana
Hospital Universitário Prof. Edgar Santos	Salvador	BA	Edna Lúcia Santos de Souza
Hospital Infantil Albert Sabin	Fortaleza	CE	Cláudia de Castro e Silva
Hospital da Criança de Brasília José Alencar	Brasília	DF	Luciana de Freitas Velloso Monte
Hospital Infantil Nossa Senhora da Glória	Vitória	ES	Roberta de Cássia Melotti
Hospital Dr Dório Silva	Vitória	ES	Daniele Menezes Torres
Hospital das Clínicas da UFGO	Goiânia	GO	Lusmaia Damaceno Camargo Costa
Hospital Infantil João Paulo II	Belo Horizonte	MG	Alberto Andrade Vergara
Consultorio Francisco Reis	Belo Horizonte	MG	Francisco José Caldeira Reis
Hospital Julia Kubitschek	Belo Horizonte	MG	Marcelo de Fuccio
Hospital Universitário da UFJF	Juiz de Fora	MG	Marta Cristina Duarte
Instituto Materno Infantil de Pernambuco	Recife	PE	Murilo Carlos Amorim de Britto
Hospital Pequeno Príncipe	Curitiba	PR	Paulo Kussek
Hospital das Clínicas da UFPR	Curitiba	PR	Carlos Antônio Riedi
Instituto Fernandes Figueira	Rio de Janeiro	RJ	Laurinda Yoko Shinzato Higa
Hospital Universitário Pedro Ernesto - UERJ	Rio de Janeiro	RJ	Agnaldo J. Lopes
Hospital de Pediatria da Universidade do Rio Grande do Norte	Natal	RN	Vera Maria Dantas

About Cystic Fibrosis and the GBEFC:

Cystic fibrosis (CF) is an autosomal inherited recessive disease with multisystemic involvement (respiratory, gastrointestinal, liver and genitourinary). It is a complex disease still little known in our country, despite the existence of some specialized centers with health professionals dedicated to patient care for many years. Treatment is quite complex and involves high cost medications, but access to care and medications is not uniform in the country.

The Brazilian Cystic Fibrosis Study Group (GBEFC) is a nonprofit organization comprised of healthcare professionals working in the area, created on November 2003. The activities of the GBEFC include research, staff training and aid in the development of CF treatment centers in the country, organizing scientific meetings (four editions of the Brazilian CF Congress), working with the Ministry of Health to define a national protocol of CF care and implementation of Newborn Screening in all Brazilian states.

The GBEFC maintains an Internet site (www.gbefc.org.br) that provides information on cystic fibrosis, scientific publications and resources, and also displays the Patient Registry Reports in Portuguese and English versions for free download.



EXECUTIVE COMMITTEE OF THE BRAZILIAN CYSTIC FIBROSIS REGISTRY:

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- Researcher at the Research and Learning Institute of Hospital Israelita Albert Einstein
- Assistant professor at the Pediatric Pulmonology Unit, Instituto da Criança HCFMUSP
- Vice-President of the Brazilian Cystic Fibrosis Study Group (GBEFC)

Dr. Francisco José Caldeira Reis

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- University of São Paulo (UNIFESP)

Acknowledgements:

This work would not be possible without the support of some Pharmaceutical Companies listed below, who sponsored the initiative with enthusiasm and ethically, even without any perspective neither of a privileged access to data nor of availability of marketing actions.

- Roche Brasil
- Novartis Brasil
- United Medical Ltda.
- Abbott Laboratórios do Brasil
- Moksha8 Brasil

We thank all health professionals involved in the care of cystic fibrosis patients by their cooperation in this initiative, which we believe will result in significant advances in the care of cystic fibrosis patients in our country.



Table 33
Data of adult population.

	Gender		Total
	Male	Female	
Azoospermia or Hypospermia	48 (22.0%)	-	48
Pregnancy	-	9 (3.8%)	9
Oral or injectable contraceptives	-	46 (19.3%)	46
Common law marriage	37 (17.0%)	62 (26.1%)	99 (21.7%)
Employment	79 (36.2%)	72 (30.3%)	151 (33.1%)
Total of patients older than 18 years with follow-up data	218	238	456

1. INTRODUCTION

This report describes data from the Brazilian Cystic Fibrosis Patient Registry in which demographics, diagnosis and treatment data of patients with cystic fibrosis in Brazil are reported. By the time of data extraction for analysis (March 28 2014), a total of 2,669 patients were registered in the database, and 2,510 (94%) of which had some follow-up data in the year 2012.

The number of registries and follow-ups is increasing annually as shown in Figure 1. Additionally, the number of records inserted in this last year (total of 487 new records) was higher than in the previous year (384 cases).

Figure 1
Increase of registries and follow-ups between 2009 and 2012.

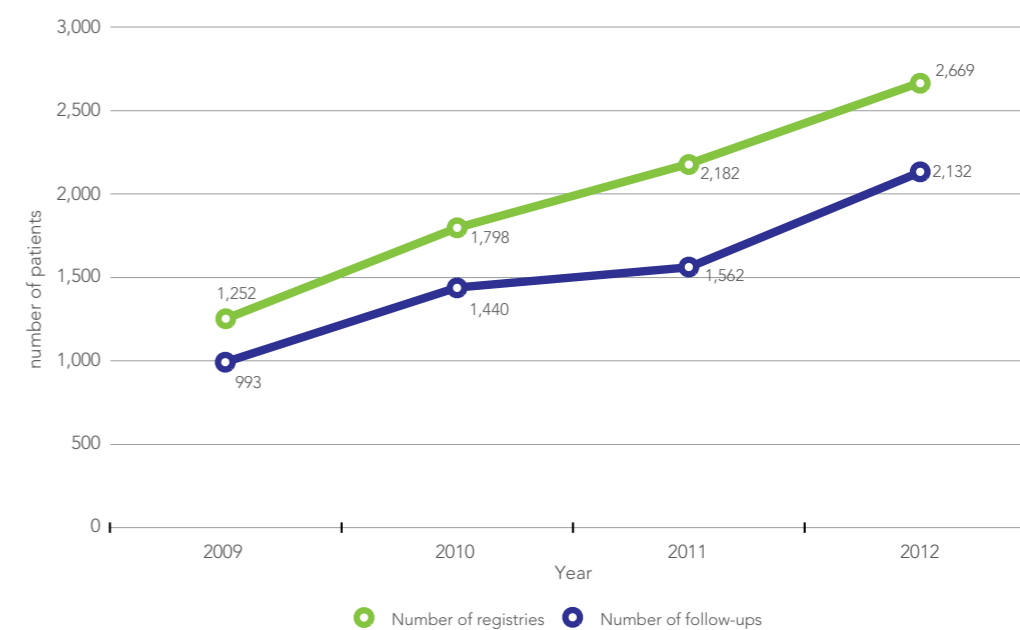


Table 1
Patients' distribution by time of follow-up.

Time of follow-up	N	%	Accumulated %
No follow-up	159	6.0	6.0
1 year	688	25.8	31.7
2 years	578	21.7	53.4
3 years	656	24.6	78.0
4 years	559	20.9	98.9
5 years	29	1.1	100
Total	2,669	100	

n = number of patients.

In the Table 1 it is possible to notice that almost half of the patients (46.6%) have at least three years of follow-up.

In the description of demographic and diagnostic data, all patients were included. Only data from the year 2012 (2,132 patients) were included in the Follow-up data.



2. DEMOGRAPHIC DATA

Table 2
Distribution of patients by Brazilian State of origin (birth place).

Brazilian State of origin	n (%)	Brazilian State of origin	n (%)
São Paulo	829 (31.1%)	Rio Grande do Norte	22 (0.8%)
Bahia	341 (12.8%)	Goiás	18 (0.7%)
Rio Grande do Sul	328 (12.3%)	Mato Grosso	14 (0.5%)
Minas Gerais	254 (9.5%)	Mato Grosso do Sul	9 (0.3%)
Rio de Janeiro	161 (6.0%)	Amazonas	6 (0.2%)
Santa Catarina	152 (5.7%)	Piauí	6 (0.2%)
Paraná	129 (4.8%)	Tocantins	5 (0.2%)
Espirito Santo	95 (3.6%)	Paraíba	4 (0.1%)
Ceará	80 (3.0%)	Sergipe	4 (0.1%)
Pará	56 (2.1%)	Acre	3 (0.1%)
Pernambuco	54 (2.0%)	Rondônia	3 (0.1%)
Não informado	36 (1.3%)	Maranhão	2 (0.1%)
Distrito Federal	31 (1.2%)	Roraima	1 (0%)
Alagoas	26 (1.0%)		

Total 2,669 (100%)

n=number of patients.

Table 31
Intravenous antibiotics: days of treatment by age group.

Days	Age group					Total
	Up to 5 years	> 5 to 10	>10 to 15	>15 to 20	>20 years	
Mean (standard deviation)	18.9 (14.5)	25.2 (24.2)	31.6 (37.1)	27.0 (17.9)	29.3 (26.4)	27.6 (28.4)
Median (p25-p75)	14 (10-21)	17 (14-29,5)	20 (14-35))	21 (14-32)	21 (14-35)	18 (14-30)
Minimum-Maximum	2-66	1-159	9-300	1-90	3-165	1-300
Total of patients	65	72	102	73	97	412

Table 32
Intravenous antibiotics - drugs utilized.

Drugs utilized	n	(%)
Ceftazidime	274	12.9%
Amikacin	244	11.4%
Oxacillin	144	6.8%
Imipenem or Meropenem	111	5.2%
Ciprofloxacin	100	4.7%
Vancomycin	80	3.8%
Trimethoprim-sulfamethoxazole	77	3.6%
Tobramycin	62	2.9%
Others	47	2.2%
Piperaciline/Tazobactam	41	1.9%
Cefepime	40	1.9%
Gentamicin	36	1.7%
Ticarcillin/Piperacillin	12	0.6%
Cefuroxime	8	0.4%
Linezolid	7	0.3%
Colomycin	7	0.3%
Chloramphenicol	3	0.1%
Aztreonam	1	0.04%
Total of patients	2,132	100%

n=number of patients.



Table 30
Intravenous treatments - admissions

Treatment	n (%)
No admission	1,674 (78.5%)
Home care	72 (3.4%)
Hospital admission	366 (17.2%)
Hospital and home care admission	20 (0.9%)
Total of patients	2,132 (100%)

Cycles	n (%)
Mean (standard deviation)	1.75 (1.22)
Median (p25-p75)	1 (1-2)
Minimum-Maximum	1-13
Total of patients	418

Days	n (%)
Mean (standard deviation)	27.55 (28.43)
Median (p25-p75)	18 (14-30)
Minimum-Maximum	1-300
Total of patients	412

Implanted catheter	n (%)
No	2,108 (98.9%)
Yes	24 (1.1%)
Total of patients	2,132(100%)

Table 3
Distribution of patients by Brazilian Region of origin (birth place).

Brazilian Region of origin	n (%)
Southeast	1,339 (50.2%)
South	609 (22.8%)
Northeast	517 (19.4%)

Brazilian Region of origin	n (%)
North	96 (3.6%)
Center West	72 (2.7%)
Not informed	36 (1.3%)



Figure 2
Distribution of patients by Brazilian Region of origin (birth place).

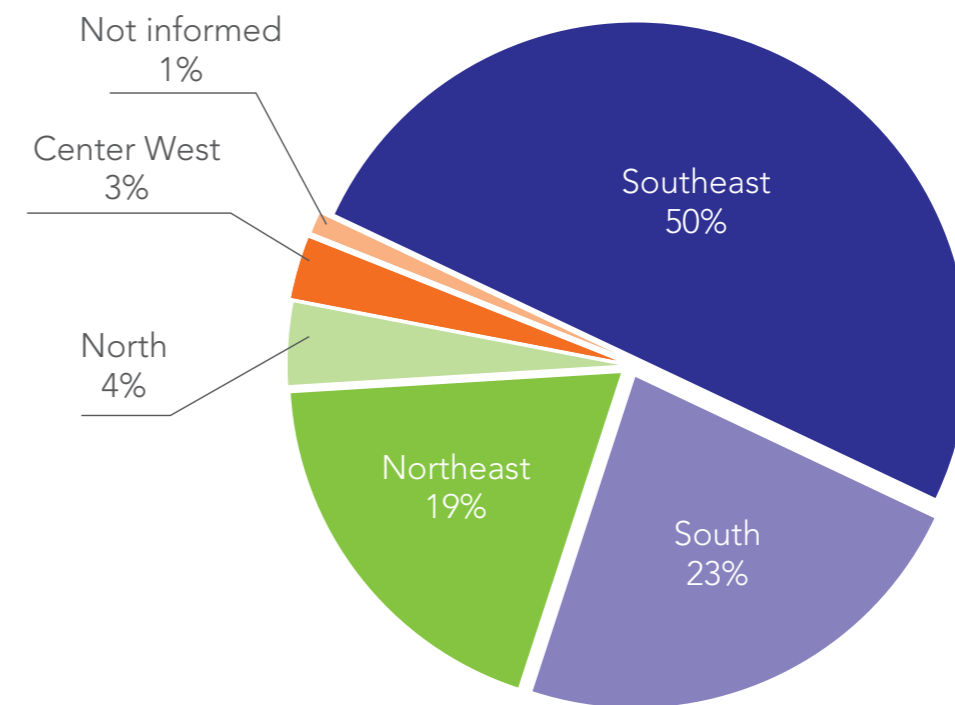




Table 4
Distribution of patients by Brazilian State where they are treated.

Brazilian State of treatment	n (%)	Brazilian State of treatment	n (%)
São Paulo	887 (33.2%)	Pará	55 (2.1%)
Rio Grande do Sul	356 (13.3%)	Distrito Federal	50 (1.9%)
Bahia	348 (13.0%)	Pernambuco	50 (1.9%)
Minas Gerais	241 (9.0%)	Alagoas	26 (1.0%)
Rio de Janeiro	158 (5.9%)	Rio Grande do Norte	23 (0.9%)
Paraná	138 (5.2%)	Goiás	17 (0.6%)
Santa Catarina	134 (5.0%)	Mato Grosso do Sul	2 (0.1%)
Espírito Santo	101 (3.8%)	Paraíba	1 (0.04%)
Ceará	82 (3.1%)		

Total of patients 2,669 (100%)
n=number of patients.



Table 26
Oral medications

	n (%)		n (%)
Pancreatic enzymes	1,706 (80.0%)	Azithromycin	797 (37.4%)
Less than 5.000 U/kg/dia	575 (27.0%)	Ursodeoxycholic acid	439 (20.6%)
5.000 - 10.000 U/kg/dia	976 (45.8%)	Proton pump inhibitors	452 (21.2%)
More than 10.000 U/kg/dia	129 (6.1%)	H2 blockers	137 (6.4%)
Unknown	26 (1.2%)	Corticosteroid	112 (5.3%)
Dietary supplements	1,300 (61.0%)	Ibuprofen (for lung disease)	16 (0.8%)
Oral	1,174 (55.1%)	Ibuprofen or other NSAID* (for arthropathy)	6 (0.3%)
Gastrostomy	32 (1.5%)		
Gastric tubes	9 (0.4%)		
Unknown	85 (4.0%)		

Total of patients 2,132 (100%)

n=number of patients.
* Non-steroidal anti-inflammatory

Table 27
P. aeruginosa eradication treatment

P. aeruginosa eradication treatment	n (%)
Yes	462 (21.7%)
No	986 (46.2%)
Unknown	684 (32.1%)
Total of patients	2,132 (100%)



Table 25
Inhaled medications

Bronchodilators	n (%)
Short acting Beta 2 agonist	730 (34.2%)
Long acting Beta 2 agonist	426 (20.0%)
Anticholinergic	88 (4.1%)

Antibiotics	n (%)
Inhaled Tobramycin solution 300mg	733 (34.4%)
Colomycin	496 (23.3%)
Gentamycin	44 (2.1%)
Other	47 (2.2%)
Injectable Tobramycin solution	12 (0.6%)
Vancomycin	6 (0.3%)
Amikacin	9 (0.4%)

Mucolytics	n (%)
Alfa dornase	1,542 (72.3%)
N Acetylcystein	65 (3.0%)

Saline solutions	n (%)
0.9% saline solution	465 (21.8%)
Hypertonic saline 3%	88 (4.1%)
Hypertonic saline 5%	87 (4.1%)
Hypertonic saline 7%	380 (17.8%)

Total of patients 2,132 (100%)

n=number of patients.

Figure 3
Distribution of patients by Brazilian State where they are treated.

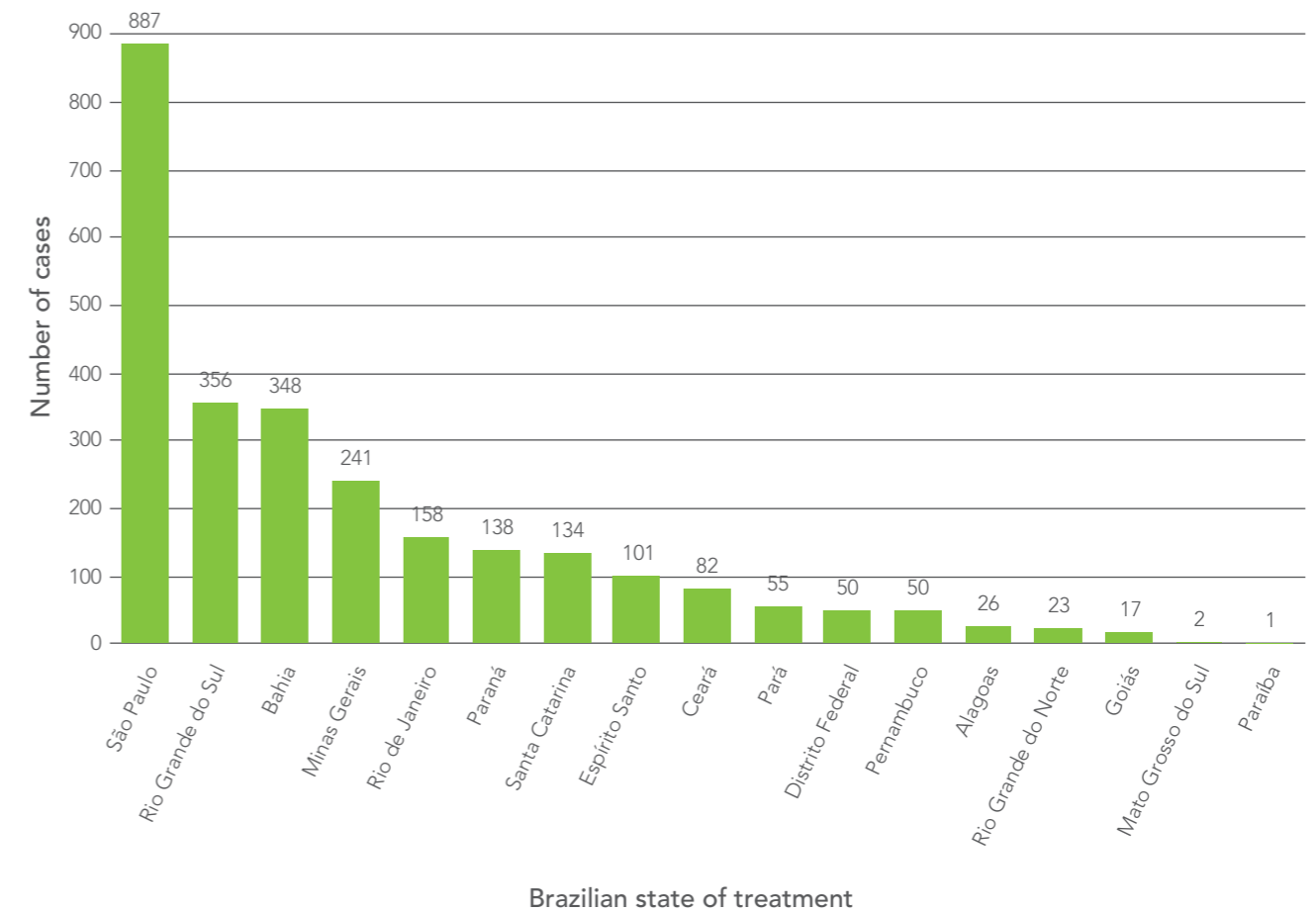




Table 5
Gender and ethnic group of Brazilian patients.

Gender	n (%)
Male	1,417 (53.1%)
Female	1,252 (46.9%)
Total of patients	2,669 (100%)

Ethnic Group	n (%)
Caucasian	1,866 (69.9%)
Mulatto	624 (23.4%)
Black	170 (6.4%)
Asian	6 (0.2%)
Indian	3 (0.1%)
Total of patients	2,669 (100%)

Figure 4
Distribution of patients by gender.

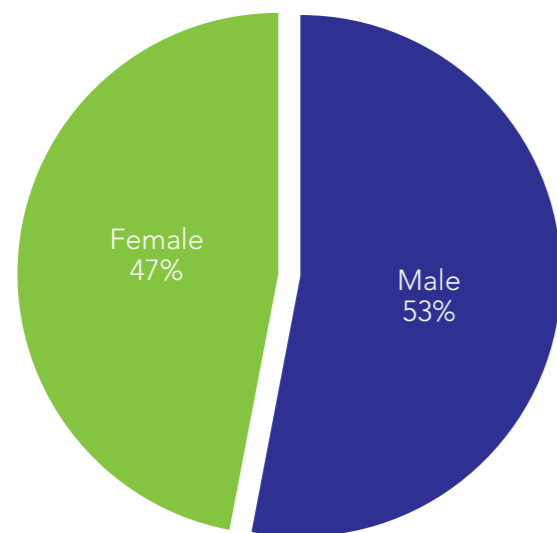


Figure 5
Distribution of patients by ethnic group.

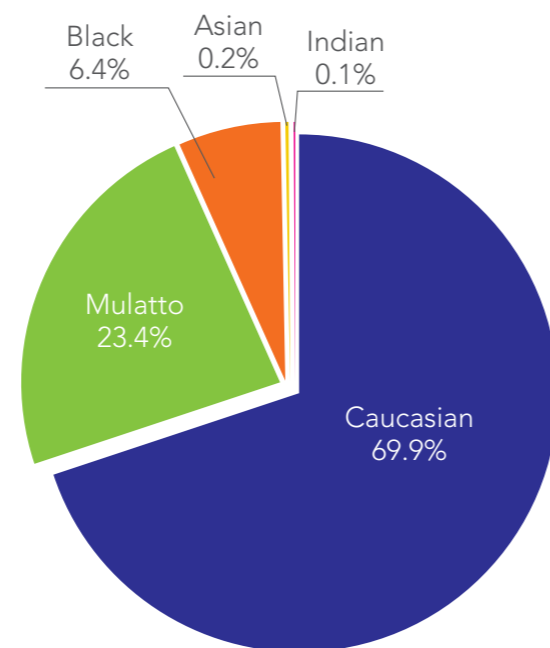


Table 21
Complications in the given year (2012)

Complications in the given year (2012)	n (%)
Asthma	291 (13.6%)
Gastroesophageal reflux	145 (6.8%)
Evidences of hepatic disease	146 (6.8%)
Nasal polyposis	85 (4.0%)
Hemoptysis	71 (3.3%)
Diabetes	79 (3.7%)
Osteopenia / Osteoporosis	62 (2.9%)
Chronic atelectasis	35 (1.6%)
Allergic broncopulmonary aspergillosis	12 (0.6%)
Distal intestinal obstruction	17 (0.8%)
Colelythiasis	16 (1.2%)
Pulmonary hypertension	19 (0.9%)
Cirrosis with portal hypertension	18 (0.8%)
Pneumothorax	3 (0.1%)
Pancreatitis	8 (0.4%)
Hematemesis	2 (0.1%)
Colonic stenosis	1 (0.047%)
Total of patients	2,132 (100%)

n=number of patients.

Table 22
Transplantation.

Transplantation	n (%)
Pulmonary transplantation	
Corpse	18 (0.84%)
Live donor	0
Liver transplantation	1 (0.04%)
Total of patients	2,132 (100%)

Table 23
Oxygen therapy

Oxygenoterapia	n (%)
No	2,046 (96.0%)
Yes	86 (4.0%)
Continuous	46 (2.2%)
Nocturnal	40 (1.9%)
Total of patients	2,132 (100%)

Table 24
Insulin usage

Insulin usage	n (%)
No	2,053 (96.3%)
Yes	79 (3.7%)
Total of patients	2,132 (100%)



Figure 15
Graph showing 95% confidence interval values of Shwachman-Kulczycki score by age group.

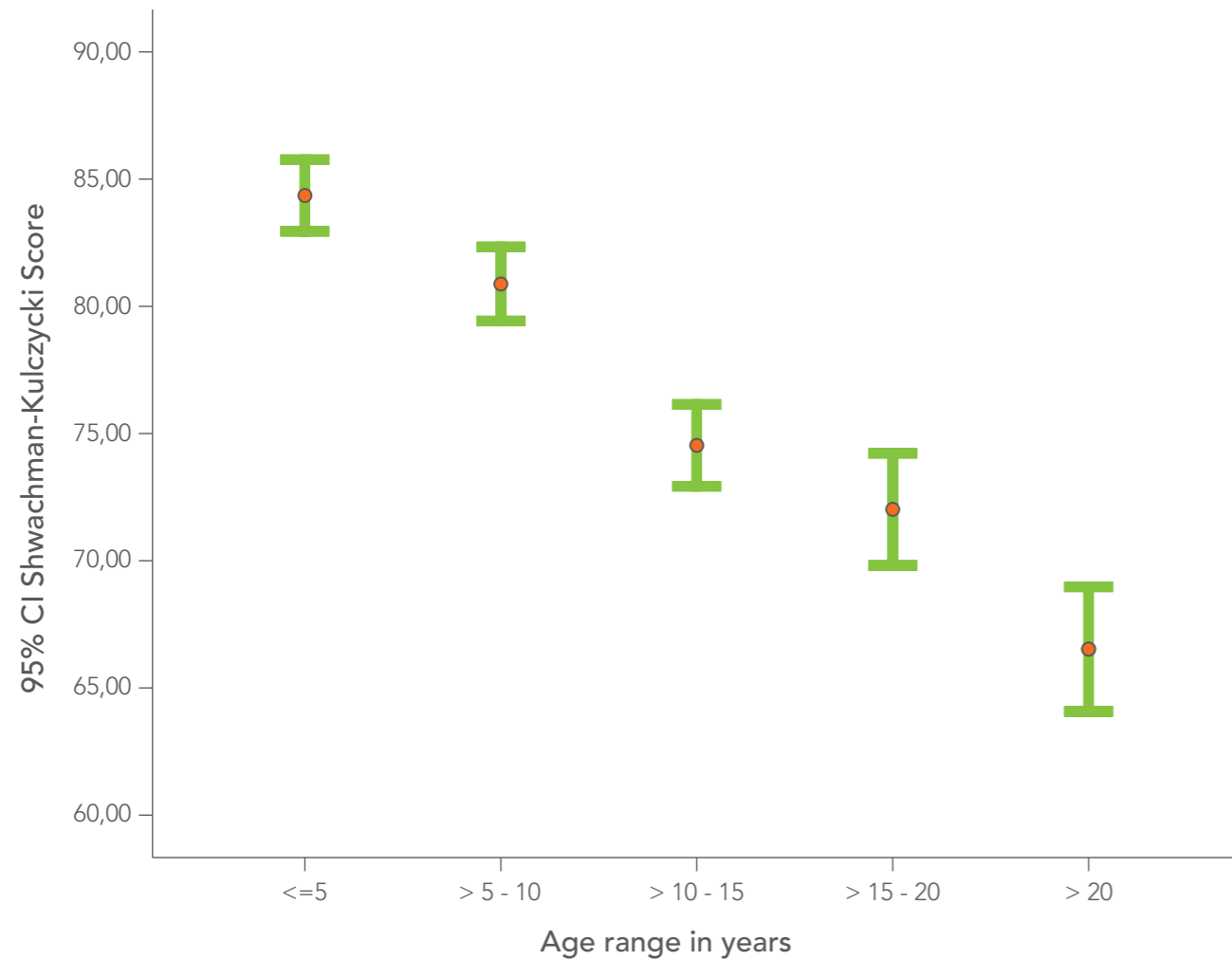


Table 6
Description of current age data (best spirometry or last clinical visit of the year)

Age (years)	
Mean (standard deviation)	13.49 (11.01)
Median (p25-p75)	11.21 (6.16 – 17.16)
Minimum-Maximum	0.08 – 85.24
Total of patients	2,469
No information	200

n=number of patients; p25 = 25th percentile; p75 = 75th percentile

Figure 6
Distribution of patients according to current age (years).

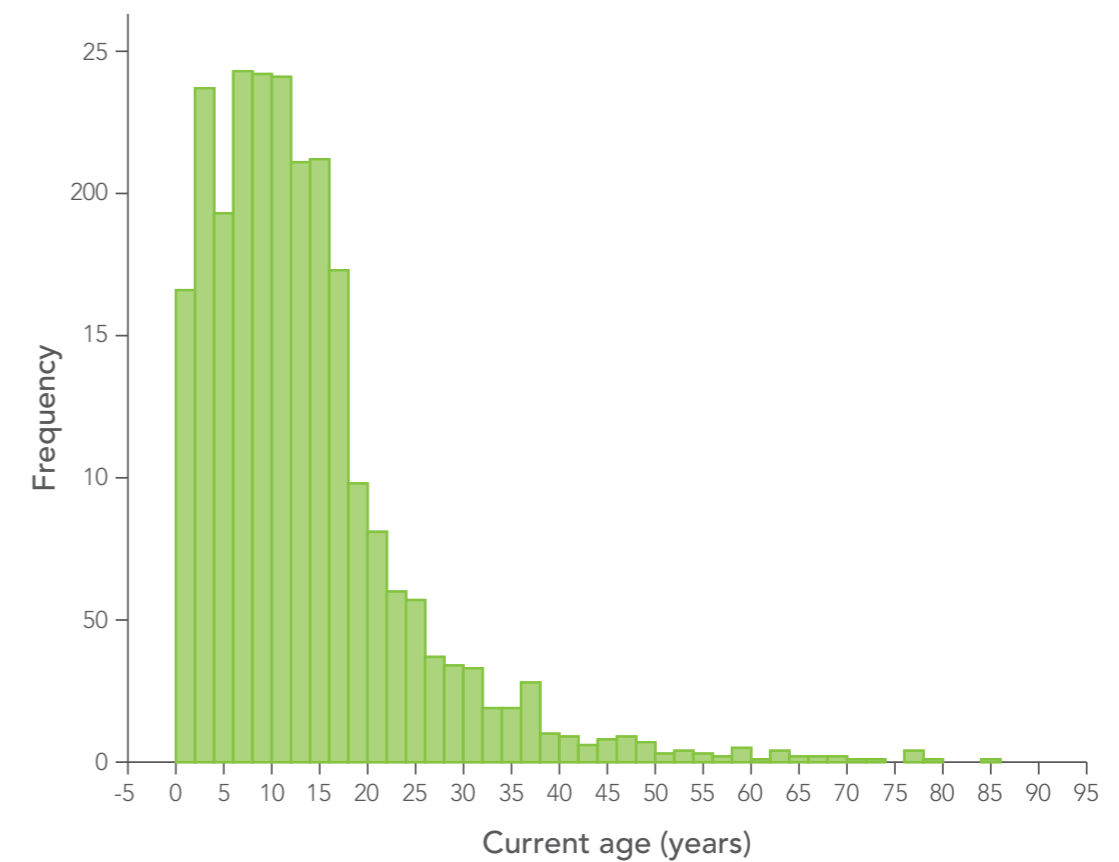




Table 7
Distribution of patients by age group.

Age Group	n (%)
Up to 5 years	503 (20.0%)
> 5 to 10	578 (23.0%)
>10 to 15	562 (22.4%)
>15 to 20	373 (14.9%)
>20 to 25	176 (7.0%)
>25 to 30	93 (3.7%)
>30 to 35	62 (2.5%)
>35 to 40	47 (1.9%)
>40 to 45	22 (0.9%)
>45 to 50	17 (0.7%)
>50 years	36 (1.4%)
Total of patients	2,469 (100%)
Patients without information	200

Age group (adult-pediatric)	n (%)
Younger than 18 years	1918 (77.6%)
18 years or older	551 (22.3%)
Total of patients	2,469 (100%)
Patients without information	200

n=number of patients.



Table 19
Deaths

Death	n (%)
No	2,107 (98.9%)
Yes	25 (1.2%)
Causes of death*	
Respiratory failure – chronic/acute	17
Post-transplant complications	4
Septic shock / sepsis	3
Acute pulmonar exacerbation	3
Dehidration	2
Total of patients	2,132 (100%)
Age at death (years)	
Mean (standard deviation)	20.58 (11.14)
Median (p25-p75)	21.07 (13.15-25.95)
Minimum-Maximum	2.42-54.93

* more than one may be indicated for each case.

Table 20
Shwachman-Kulczycki score.

Total score	n (%)
Mean (standard deviation)	76.79 (17.44)
Median (p25-p75)	80 (65-90)
Minimum-Maximum	10-100
CLASSIFICATION	
Severe (≤ 40)	77 (4.6%)
Moderate (41 a 55)	165 (9.9%)
Median (56 a 70)	356 (21.3%)
Good (71 a 85)	544 (32.6%)
Excellent (86-100)	526 (31.5%)
Total of patients	1,668 (100%)



8. CLINICAL TREATMENT DATA

Figure 14
Distribution of the number of consultations per patient in 2011.

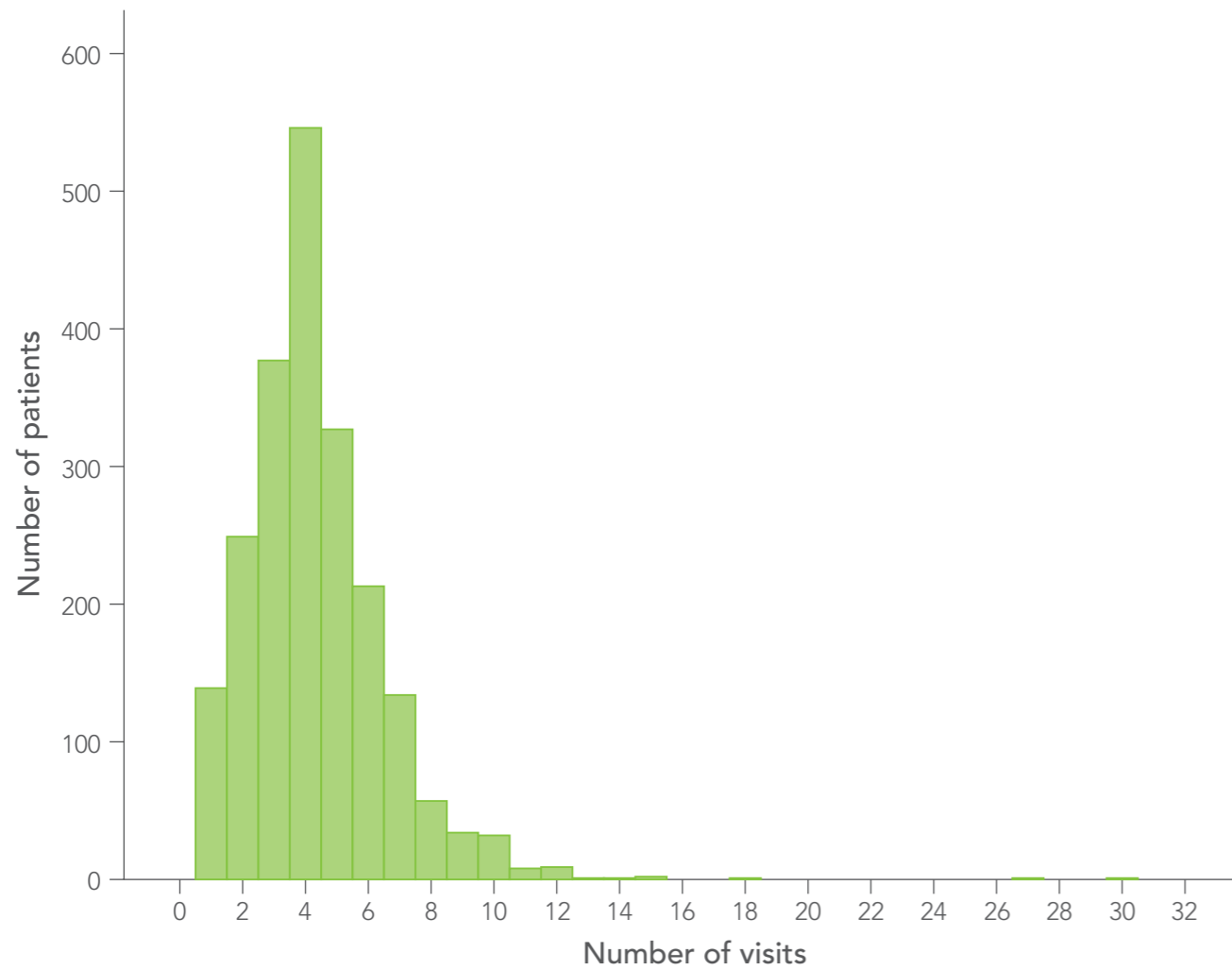


Figure 7
Distribution of patients by age group.

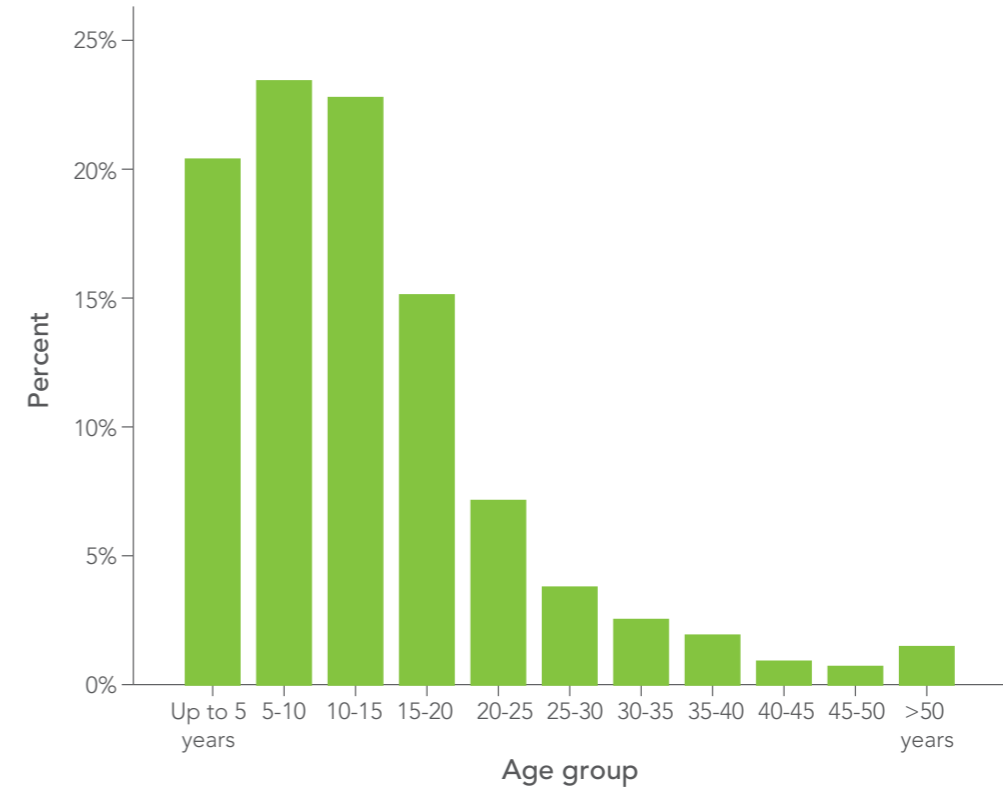
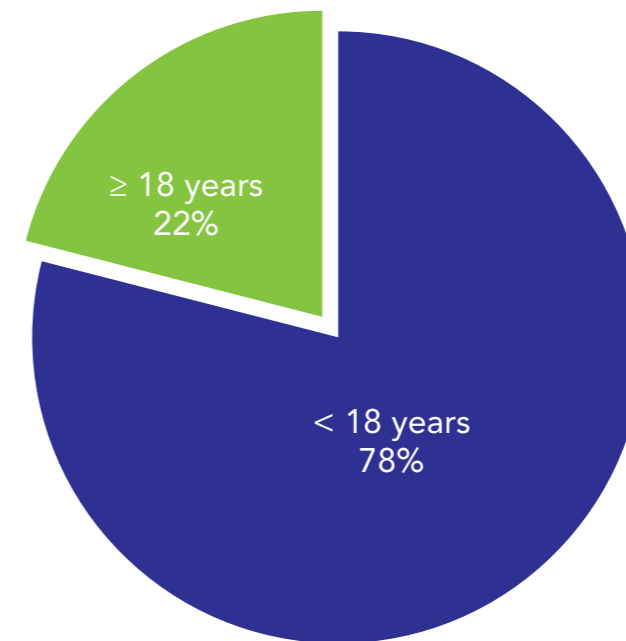


Figure 8
Distribution of patients by age group category (pediatric / adult).





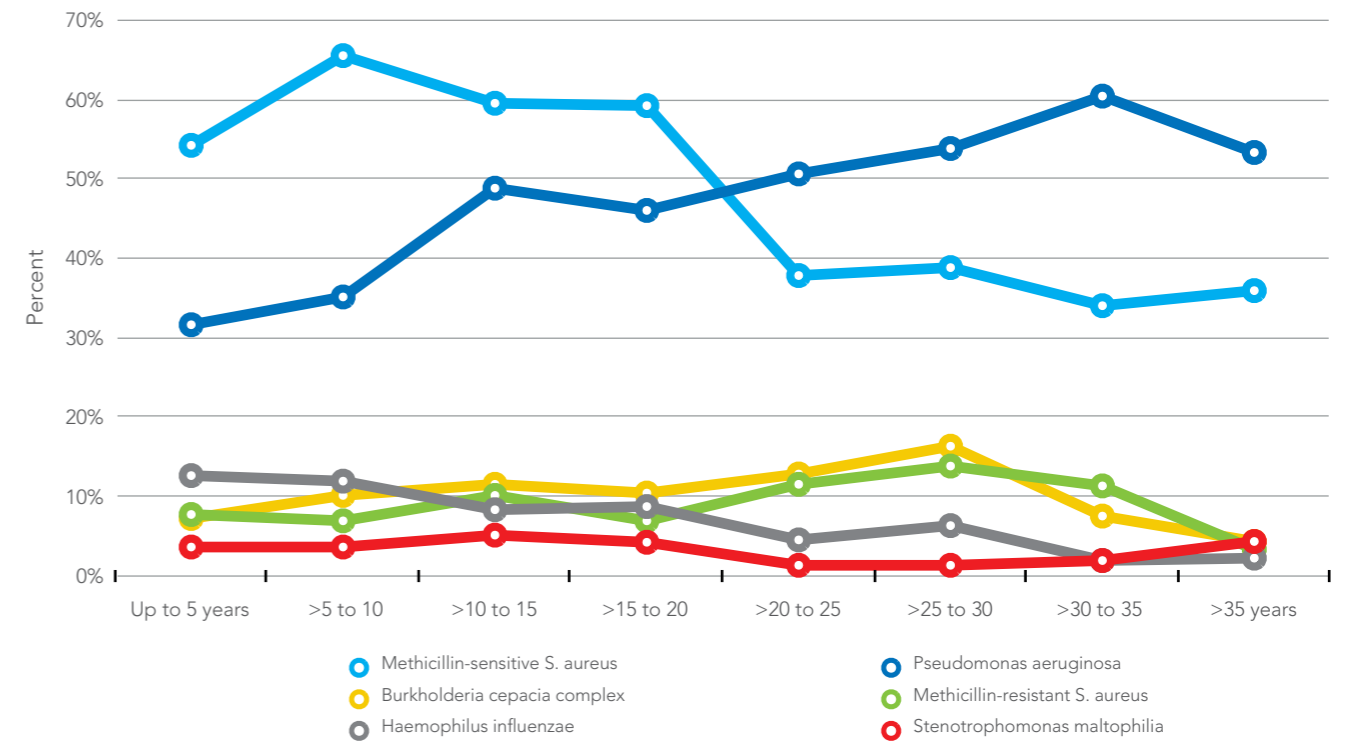
3. DATA AT DIAGNOSIS

Table 8
Age of patients at diagnosis.

Age (years)	
Mean (standard deviation)	5.71 (9.85)
Median (p25-p75)	1.53 (0.25 – 7.34)
Minimum - Maximum	0 – 82,95
Total of patients	2,663
Patients without information*	6

n=number of patients; p25 = 25th percentile; p75 = 75th percentile
* Incorrect birth dates.

Figure 13
Prevalence of identified pathogens by age group.





7. MICROBIOLOGY DATA

Microbiology data describe positive results for the respiratory pathogen at least once in the given follow-up year; since there is not a standardization of microbiology processing of respiratory tract samples of cystic fibrosis patients in our country, these data have to be cautiously interpreted.

Table 18

Description of microorganisms identified.

Microorganisms identified	n	%
Methicillin-sensitive <i>Staphylococcus aureus</i>	1,182	55.4%
<i>Pseudomonas aeruginosa</i>	1,071	50.2%
Non-mucoid <i>Pseudomonas aeruginosa</i>	590	27.7%
Mucoid <i>Pseudomonas aeruginosa</i>	481	22.6%
<i>Burkholderia cepacia</i> complex	213	10.0%
Methicillin-resistant <i>Staphylococcus aureus</i>	179	8.4%
<i>Haemophilus influenzae</i>	198	9.3%
<i>Stenotrophomonas maltophilia</i>	79	3.7%
<i>Klebsiella pneumoniae</i>	61	2.9%
<i>Serratia</i> sp.	59	2.8%
<i>Achromobacter</i> sp.	52	2.4%
<i>Candida</i> sp.	60	2.8%
<i>Aspergillus fumigatus</i>	28	1.3%
<i>Escherichia coli</i>	27	1.3%
Other <i>Pseudomonas</i>	37	1.7%
Non tuberculous <i>Mycobacteria</i>	5	0.2%
<i>Mycobacterium tuberculosis</i>	5	0.2%
Total de pacientes	2,132	100%

Table 9

Conditions for diagnosis.

Conditions for diagnosis	n (%)
Respiratory symptoms	1,701 (63.7%)
Deficit of growth / malnutrition	1,077 (40.4%)
Steatorrhea or Malabsorption	999 (37.4%)
Neonatal screening (IRT)	603 (22.6%)
Familial history	224 (8.4%)
Clinical or surgical meconium ileus	212 (7.9%)
Sinus disease	179 (6.7%)
Metabolic disturbance	147 (5.5%)
Edema / anemia	93 (3.5%)
Unknown condition	71 (2.7%)
Rectal prolapse	28 (1.0%)
Prolonged jaundice	25 (0.9%)
Infertility	10 (0.4%)
Other	140 (5.2%)
Total of patients	2,669 (100%)

n=number of patients. * List of conditions written by the professional (only few were described); more than a condition per case was allowed.



Table 10
Sweat chloride testing results.

	Chloride (mEq/l)	Sweat mass (mg)	Conductivity (mmol/l)
Mean (standard deviation)	88.20 (26.59)	144.53 (75.93)	104.2 (19.1)
Median (p25-p75)	88.50 (68.7-105)	132.50 (100-180.5)	105.0 (96-115)
Minimum-Maximum	0.38-249.50	0.08-470	33-180
Total of patients	2,252	1,696	263

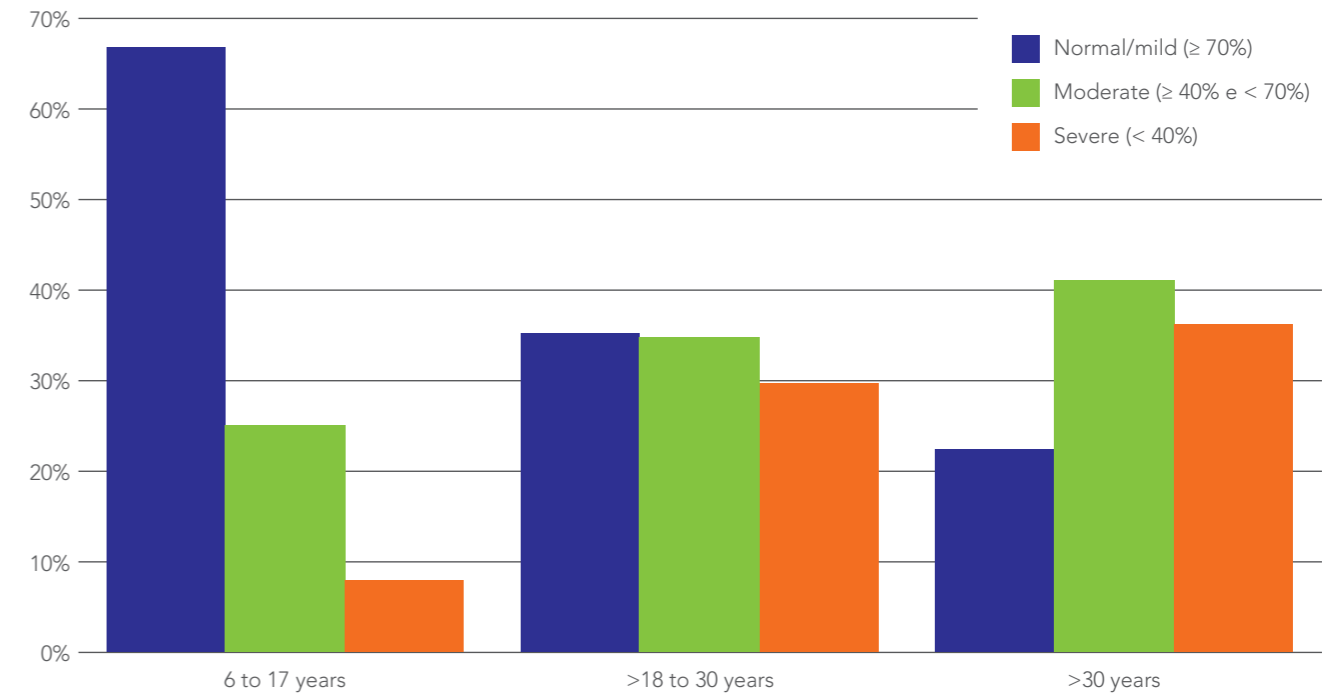
n=number of patients; p25 = 25th percentile; p75 = 75th percentile.

Table 11
Other tests reported for diagnosis.

	n (%)
Measure of nasal potential difference	99 (3.7%)
Rectal biopsy	70 (2.6%)
Total of patients	2,669 (100%)

n=number of patients.

Figure 12
Lung function data (FEV1 percent predicted category) by age group.





6. PULMONARY FUNCTION DATA

The values of FVC, FEV1 and FVC/FEV1 recorded by the participants were those obtained in the best lung function test of the reported year. The predicted values of pulmonary function were obtained from the publication of Stanojevic S et al: Spirometry Centile Charts for Young Caucasian Children: The Asthma UK Collaborative Initiative. American Journal of Respiratory and Critical Care Medicine 2009, 180(6); 547-552.

Table 17
Pulmonary function data (1,073 patients - 50.3%).

Parameter	Mean (standard deviation)	Median (p25; p75)	Minimum; Maximum
Forced Vital Capacity (FVC)			
Percent predicted	83.6 (23.87)	85.81(67.51; 100.13)	20.41; 154.11
Z score	-1.53 (2.11)	-1.33 (-2.94; -0.15)	-7.37; 8.15
Forced Expiratory Volume in the first second (FEV1)			
Percent predicted	74.68 (26.84)	75.9 (54.31; 95.15)	16.46; 148.88
Z score	-2.15 (2.19)	-2.11 (-3.82; -0.6)	-6.74; 9.11
VEF1/CVF			
Value	0.76 (0.13)	0.78 (0.68-0.87)	0.33-1.00

p25 = 25th percentile; p75 = 75th percentile

Table 12
Diagnosis by neonatal screening - Dosage of immunoreactive trypsinogen (IRT).

Dosage of immunoreactive trypsinogen (IRT). (ng/ml)	1st dosage	2nd dosage	Mean of 2 dosages
Mean (standard deviation)	218.0 (128.3)	209.6 (133.4)	218.1 (126.2)
Median (p25-p75)	186 (127-266)	177 (119-251)	183.2 (129.8-269)
Minimum-Maximum	37.5-1255	14-1049	37.5-1152
Total of patients	557	427	562

Cut-off limits for IRT values	1st dosage n (%)	2nd dosage n (%)	Mean of 2 dosages n (%)
< 70 ng/ml	13 (2.3%)	21 (4.9%)	18 (3.2%)
≥ 70 ng/ml	544 (97.7%)	406 (95.1%)	544 (96.8%)
< 110 ng/ml	74 (13.3%)	84 (19.7%)	83 (14.8%)
≥ 110 ng/ml	483 (86.7%)	343 (80.3%)	479 (85.2%)
Total of patients	449	337	454

n=number of patients; p25 = 25th percentile; p75 = 75th percentile

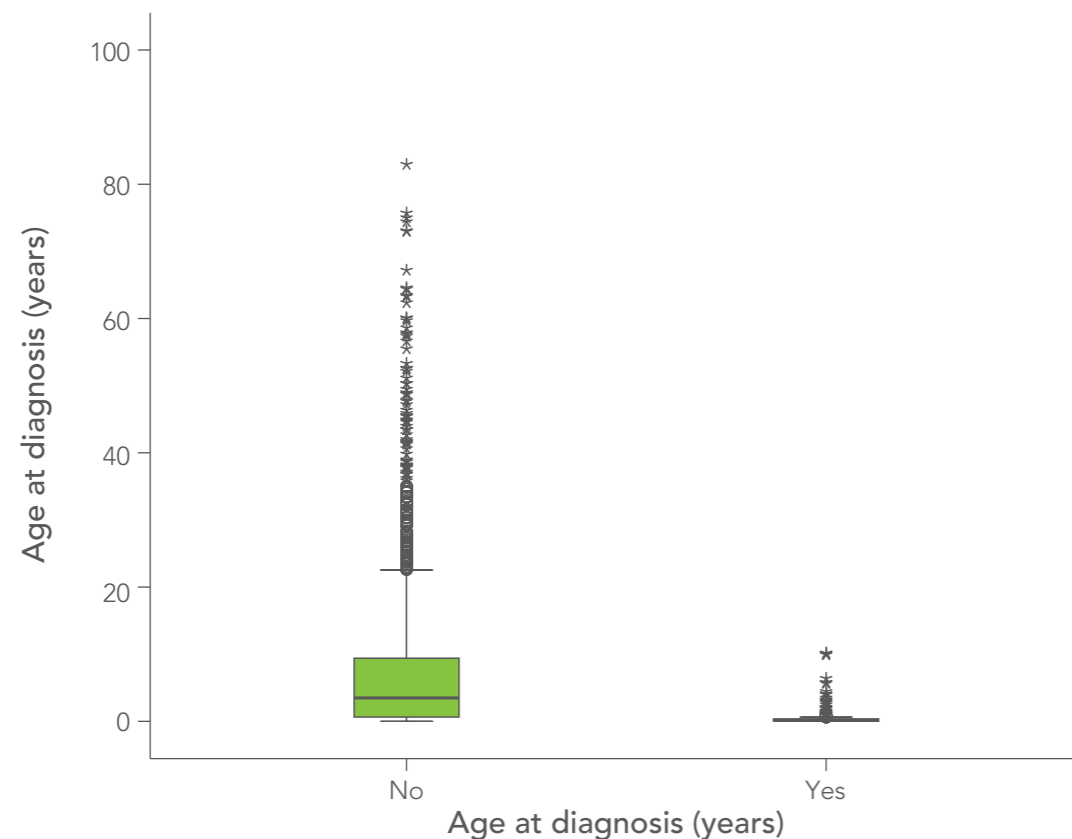


Table 13
Age in years at the diagnosis: with or without neonatal screening.

Age (years)	Neonatal screening		
	No	Yes	Total
Mean (standard deviation)	7.26 (10.69)	0.40 (1.05)	5.71 (9.85)
Median (p25-p75)	3.48 (0.64-9.40)	0.14 (0.09-0.30)	1.53 (0.25 – 7.34)
Minimum-Maximum	0-82.95	0-10.18	0 – 82.95
Total of patients	2,061	602	2,663
Patients without information	5	1	6

p25 = 25th percentile; p75 = 75th percentile

Figure 9
Box-plot graph showing distribution of age at the diagnosis in patients diagnosed with or without neonatal screening test.



5. ANTHROPOMETRIC DATA

Table 16
Anthropometric data

WEIGHT (kg)	NCHS Percentile	Score Z
Mean (standard deviation)	33.84 (30.08)	-0.68 (1.27)
Median (p25-p75)	26.00 (7.00-56.00)	-0.64 (-1.51- 0.15)
Minimum-Maximum	0-100	-3.96; 3.96
Total of patients	1,634	1,634

HEIGHT (cm)	NCHS Percentile	Z Score
Mean (standard deviation)	33.70 (28.76)	-0.63 (1.15)
Median (p25-p75)	27.00 (8.00-54.00)	-0.62 (-1.39-0.11)
Minimum-Maximum	0-100	-3.96; 3.45
Total of patients	1,608	1,608

BMI (kg/m2)	Measure	NCHS Percentile
Mean (standard deviation)	17.76 (3.85)	44.28 (33.54)
Median (p25-p75)	16.91 (15.12-19.60)	40.00 (14.00-75.00)
Minimum-Maximum	3.15-38.01	0-100
Total of patients	2,063	1,267

p25 = 25th percentile; p75 = 75th percentile

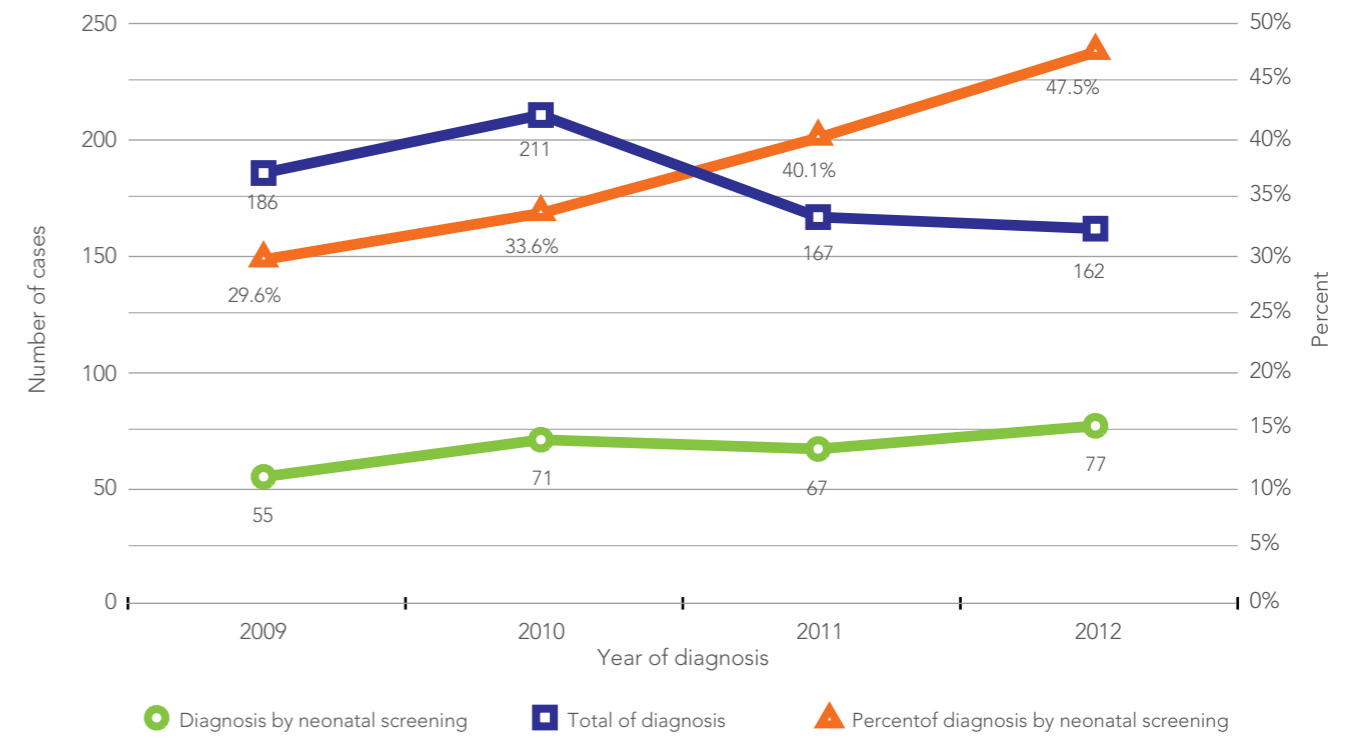


Table 15
Frequency of identified mutations (1,134 patients, 2,268 alleles).

Mutation	n	%
DF508	1,080	47.6%
G542X	94	4.1%
R1162X	18	0.8%
R334W	16	0.7%
G85E	16	0.7%
3120+1G>A	15	0.7%
W1282X	14	0.6%
N1303K	12	0.5%
G551D	6	0.3%
3849+10kbC>T	5	0.2%
R553X	4	0.2%
1078 delT	4	0.2%
S549R	3	0.1%
2183AA>G	3	0.1%
711-1G>T	3	0.1%
1717-1G>A	3	0.1%
W1089X	2	0.1%
D1152H	2	0.1%
Other / Non identified	1,013	44.7%
Total of alleles (1,134 patients)	2,268	100%



Figure 10
New diagnosis of cystic fibrosis and the contribution of neonatal screening.





4. GENETICS DATA

Genetics data contained in this report should be cautiously interpreted, since the approach for CF genetic testing in Brazil is highly heterogeneous. Some Centers only perform detection of DeltaF508 mutation, while others search panels of 2, 4 or up to 30 or more mutations. There are also Centers reporting genetic polymorphisms in the CFTR gene, that were not included in this report because they may not be used as diagnostic criteria without the presence of other mutations.

Table 14
Status of patients regarding genetic testing for CF.

Genetic testing	n (%)
No	1,535 (57.5%)
Yes	1,134 (42.5%)
Total of patients	2,669 (100%)

Quantity of mutations identified per patient	n (%)
None	249 (22.0%)
One	393 (34.7%)
Two or more	492 (43.4%)
Total of patients with genetic testing reported	1,134 (100%)

Genotype - description	n (%)
DF508/DF508	301 (26.5%)
DF508/Other	145 (12.8%)
DF508/Non-identified	333 (29.4%)
Other/Other	46 (4.1%)
Other/Non-identified	60 (5.3%)
Non-identified/Non-identified	249 (22.0%)
Total of patients with genetic testing reported	1,134 (100%)

Figure 11
Distribution of patients by genetic study results (n=1,134).

