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The prevalence of gene mutation in Cystic Fibrosis (CF) in Children medical center Tehran from 1395-1400

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Keywords: Cystic Fibrosis; Genetic Mutation; CF prevalence; Pancreatic Insufficiency

Abstract

Introduction: Cystic fibrosis (CF) is a multisystem disorder caused by pathogenic mutations of the CFTR gene (CF transmembrane conductance regulator). Pulmonary disease remains the leading cause of morbidity and mortality in patients with CF. It is also responsible for many cases of hyponatremic salt depletion, nasal polyposis, pansinusitis, rectal prolapse, pancreatitis, cholelithiasis, and non-autoimmune insulin-dependent hyperglycemia. CF occurs most frequently in white populations of northern Europe and Australia/New Zealand. CF is inherited as an autosomal recessive trait. The CF gene codes for the CFTR protein, which is 1,480 amino acids. CFTR is expressed largely in epithelial cells of airways, the gastrointestinal tract (including the pancreas and biliary system), the sweat glands, and the genitourinary system. CFTR is a member of the adenosine triphosphate–binding cassette superfamily of proteins. It functions as a chloride channel and has other regulatory functions that are perturbed variably by the different mutations. More than 1,900 CFTR polymorphisms have been described, But Those with clinical manifestations may be grouped into 6 main classes based upon how they impact upon protein structure and function.Mutation class I-III are generally considered to be severe mutations in that they lead to a complete or nearly complete absence of CFTR function, whereas class IV-VI mutations are associated with some residual functional protein. The most prevalent mutation of CFTR is the deletion of a single phenylalanine residue at amino acid 508 (F508del)

Method: This is a Retrospective cross-sectional observational study that will study patients that have been admitted in Children Medical Center from 1395-1400. We will assess all of the patient's files and extract useful correlated data. We will evaluate all data including, patient's admission code. Statistically we will analyze the data by using SPSS software.

Introduction

Cystic fibrosis is an autosomal-recessive, monogenetic disorder caused by CFTR gene mutation which occurs on the long arm of chromosome 7. This gene transport ions across the epithelial cell surface. Primary function of CFTR is that it's an apical anion channel of chloride and bicarbonate [1]. Most common allele mutation is on F508del which is responsible of approximately 66% of all cases Worldwide it's a three-nucleotide deletion at 508th codon which cause deletion of phenylalanine residue and defects on intracellular processing of the CFTR protein [2].

The CFTR genetic mutation can't be predicted all the time because a lot of mutations have been identified and been classified into different classes by respect of their different abnormal functions. Initially the classification was proposed by Tsui which was then refined by Welsh and Smith they have classified 5 classes but in the new era of treatment of CF we can see there have been 6 classes identified and here they are summarized with CFTR defects and type of mutations with examples:

- Class I represents with mutation type of nonsense or frameshift with no functional CFTR protein i.e., Gly5421x, Arg553x
- Class II represents with mutation of missense aminoacid deletion with CFTR trafficking Defect i.e., Phe508del, Arg560Thr

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• Class III represents with mutation of missense aminoacid change with Defective channel regulation i.e., Gly551Asp, Gly178Arg
• Class IV represents with mutation of missense aminoacid change with decreased channel conductance i.e., Arg 117His, Arg 334Trp
• Class V represents with mutation of Splicing defect missense with reduced synthesis of CFTR i.e., 3849+10kbC=T, 5T
• Class VI represent with mutation of missense aminoacid change with decreased CFTR stability i.e., 4326delTC, Gln1412X [3, 4].

CFTR is mainly in Pancreatic ductal epithelium, and this ductal system produce large amount of alkaline fluid which is made up of sodium chloride and bicarbonate around 1-2L/day. We got 2 forms of pancreas in CF the first one is pancreatic insufficiency where the exocrine part is severely damaged and other one is pancreatic sufficiency where person have enough pancreatic function to maintain health without Pancreatic enzyme replacement therapy (PERT). There are multiple organs effected but the exocrine pancreas is most reliable phenotypic barometer of CFTR function. Pancreatic sufficient patients are diagnosed at older age. Pancreatitis is uncommon in CF because it needs the pancreatic acinar tissue to be damaged to become symptomatic which is present in Pancreatic sufficient CF but as we know around 85-90% of CF going to cause Pancreatic Insufficiency [5, 6]. The earliest manifestations of CF are Gastrointestinal and Nutritional Disorders. Around 15% comes with meconium Ileus. Pancreatic disorders cause lack of enzymatic activity which leads to fats and proteins malabsorption and then causing Failure to thrive. Additional manifestations can be Digital Clubbing, Chronic sinusitis and nasal polyps. In newborn period respiratory manifestations are less common but older infants can present with persistent coughing, recurrent wheezing, tachypnea and frequent lung infections. For respiratory clearance we have an essential Mucociliary system. Which is consist of 2 hydrogels: A mucus layer and a periciliary layer. Effective mucociliary transport depends upon on proper hydration of it. Mucociliary clearance action can be abnormal because of ciliary motion or by change is the composition of mucus that make it less responsive to ciliary propulsion. In a Healthy person there is a normal preserved airway surface hydration. A high level of osmotic pressure in periciliary layers ensures that the provides an appropriate lubrication for ciliary activity. And a well hydrated mucus layer makes it transports more rapidly from the distal of the airways towards trachea. So, in CF patients one of the causes of lungs problem is abnormal concentration of Mucus level which primarily reflects on abnormality of airway epithelial ion-water transport. Because of the defect in chloride and bicarbonate anions which are secreted by CFTR it leads to hyperabsorption of fluid susceptibility in Lung epithelium. This epithelium abnormality leads to deplete in the airway surface of fluid which further leading towards hyper concentrated mucus causing impaired mucus transport and mucus adhesion. Airflow obstruction and recurrent bacterial infections are common features of CF lung disease.

Hyper concentrated respiratory secretions lead to chronic airway obstruction, colonization by bacteria, acute and chronic infection. In early years of life, a greater diversity of Bacteria i.e., Staphylococcus Aureus, Hemophilus Influenza are common but as the lung disease progresses typical CF pathogens i.e., Pseudomonas aeruginosa, Burkholderia cepacia, Achromobacter xylosoxidans colonize the airway. A 3-decade study in CF foundation shows us that the prevalence of P.aeruginosa has decrease continuously but prevalence of multi-drug resistant remained constant which can be due to widespread implementation of therapy to eradicate P.aeruginosa, which is most common bacterial pathogens of adults.

Inhaled antibiotics towards eradicating the P.aeruginosa have a success rate of around 80% [7-9]. Chronic Intralumenal bacterial infection produces a highly proinflammatory milieu in the airway lumen. Which clears both bacterial products and as well as host response products I.e., macrophages and epithelial cells [10]. Inflammatory response in CF lung disease early in life and becomes persistent and ultimate results ends up in bronchiectasis. Interrupting the early cascade of the disease process with drug i.e., CFTR potentiators and correctors, mucolytics and antibiotics may have subsequent beneficial effects on inflammation. For the long-term CF treatment Ibuprofen is only anti-inflammatory which is recommended [11].

Since the CFTR genotype is not a useful predictor of the severity of lung disease in the individual patient, it should not be used as an indicator of prognosis [12].

Rationale

Aim of our study is to find out the prevalence of gene mutation in Cystic Fibrosis (CF) in Children medical center Tehran from 1395-1400.

Objectives

Main objective

To construct a cross-sectional case series study and determine the prevalence of genetic mutation in Cystic Fibrosis (CF) in Children medical center Tehran from 1395-1400.

Specific Objectives

lTo determine the common genetic mutation of cystic fibrosis in CF patients in Children's Medical Center, Tehran.
lTo determine the common genetic mutation of cystic fibrosis in CF patients in Children's Medical Center, Tehran.

Goals

To determine the genetic mutations in Children's Medical Center, Tehran, from 1395-1400.To determine the prevalence of F508del in CF patients.As the prevalence of CF is increasing in Iran we want to know about the different genetic mutations and the best outcomes we can make for each patient.

Research Questions

- What is the common genetic mutation of cystic fibrosis patients?
- What are the common genetic mutations of cystic fibrosis patients in Children's Medical Center, Tehran?
- What are the common genetic mutations of cystic fibrosis patients in Children's Medical Center, Tehran, sine 1395-1400?
- What is the prevalence of the F508del gene in Children's Medical Center, Tehran?
- Which genetic mutation is the most prevalent in Iran?

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Review of Literature

In 2019 Al-Abadconducted a retrospective study on 777 sera samples for patients clinically suspected to have cystic fibrosis over a six year period 1/1/2013-1/10/2018. The patient's age ranged between 1year and 33 years, of which 59.2% (460) were male and 40.8% (317) female. Blood samples were analyzed at Princess Iman Centre for Research and Laboratory Sciences at King Hussein Medical Centre. The samples were tested for 34 mutations of CFTR gene using CF Strip Assay VIENNALAB Diagnostics GmbH, Austria by polymerase chain reaction (PCR).

A total of 777 patient samples were analyzed for cystic gene mutations. Twelve (12) mutations were identified. In 49 patients (6.3%) were heterozygous genotype mutant and 28 (3.6%) were homozygous. The most frequent mutation F508del was found in 32/77 (41.5%). 20 (25.9%) of them were heterozygous genotype mutants and 12 (15.6%) were homozygous genotype mutants. The second frequent mutation was N1303K with frequency rate 15.6% (12/77), 9 (11.7%) of them were heterozygous and 3 (3.9%) were homozygous. Regarding frequency of cystic fibrosis gene mutation depending on sex, 55.8% (43/77) of mutations were found in male, whereas 44.2% (34/77) in female. (Al-Abadi et al. 2019)

In 2022 Shen reported 158 different variants including 23 novel observations were identified after sequencing. The majority of CFTR variants (82.3%) in Chinese have been observed only once or twice. 43.7% of the variants were only identified in patients of Chinese origin. The c.2909G>A(p.Gly970Asp), c.1766+5G>T and c.1657C>T(p.Arg553X) were the most frequent variants among Chinese patients, with allele frequency of 12.1%, 5.4% and 3.6%, respectively. The first two variants both showed significant Chinese ethnic tendency, while the latter one most likely came from Europeans for historical reasons. They also demonstrated significant differences in geographical distribution. c.1521_1523delCTT(p.F508del) was rarely observed in patients of pure Chinese origin, with an allele frequency of 1.8%. Two de novo variants (c.960dupA[p.Ser321IlefsX43] and c.2491-2A>G) and two deep-intronic variants (c.3718–2477C>T and c.3874-4522A>G) were identified, which were also quite rare among Chinese. (Shen et al. 2022)

In 2022 Qi Ni researched the 53 manually curated P/LP variants in CFTR gene, we excluded individuals identified or suspected with CF and their parents in our cohorts and estimated the Chinese CF prevalence is approximately 1/128,434. Only 21 (39.6%) of the 53 variants were included in Caucasian specific CF screening panels, resulting in significantly under-estimation of CF prevalence in our children cohort (1/143,171 vs. 1/1,387,395, $P = 5e{-}24$) and parent's cohort (1/110,127 vs. 1/872,437, $P = 7e{-}10$). The allele frequencies of six pathogenic variants (G970D, D979A, M469V, G622D, L88X, 1898+5G->T) were significantly higher in our cohorts compared with gnomAD-NFE population (all P-value < 0.1). Haplotype analysis showed more haplotype diversity in Chinese compared to Caucasians. In addition, G970D and F508del were founder mutations of Chinese and Caucasians with two SNPs (rs213950-rs1042077) identified as related genotypes in the exon region.(Ni et al. 2022)

In 2016 Stewart and Pepper reported with a total of 79 variants were detected in the included reports, of which 39 have been shown empirically to cause CF, 10 are of varying clinical consequence,61.,62. 4 have no associated evidence of pathogenicity, 4 are synonymous, 5 are novel, and 21 are unique to Africa (4 of the 21 were novel). The most frequently detected alleles were ΔF508 (992 chromosomes), 3120+1G>A (83 chromosomes), G542X (58 chromosomes), N1303K (51 chromosomes), W1282X (48 chromosomes), E1104X (41 chromosomes), 711+1G>T (36 chromosomes), 3272-26A>G (17 chromosomes), and 394delTT (15 chromosomes). It should also be noted that for 224 chromosomes, the methods used did not result in the resolution of the patient's molecular CF status. (Stewart and Pepper 2016)

Methodology

Research design and method

The study is based on retrospective cross-sectional case series study.

Target Population

Patients referred to Children Medical Center, Tehran from 1395 to 1400.

Sampling

Sampling Method

To construct a cross-sectional case series study and determine the prevalence of genetic mutation in Cystic Fibrosis (CF) in Children's Medical Center, Tehran, from 1395-1400.

Inclusion Criteria

This study includes all patients with diagnosed Cystic Fibrosis since 1395.

Exclusion Criteria

This Study excludes patients admitted at Children's Medical Center, Tehran, before year 1395 and after 1400. Study also excludes patients, who are admitted in hospital but their medical records and files are no more accessible.

Method

The study is a retrospective cross-sectional study. The participants of the study will be composed of patients who visited the Children's Medical Center Tehran with cystic fibrosis with positive mutation. This study excludes patients, who had any underlying clinical disease or comorbid diseases will be excluded from study. Data will be analyzed with SPSS v26 and P value 0.05.

Predictors

Children Age, Children gender, Genetic mutations, Classes of CF, Different outcomes, Microbiological effect on CF, CF systemic effect, Different Clinical manifestations, Diagnosis, Medical Treatment, Prognosis.

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Outcome Variables

Children Age, Children gender, Genetic mutations, Classes of CF, Different outcomes, Microbiological effect on CF, CF systemic effect, Different Clinical manifestations, Diagnosis, Medical Treatment, Prognosis.
Tools used in this study
Measures/instruments
Patients records and files.

Procedures

We study patient files which are accessible and we find out our aimed data from files, patients laboratory results, interventions and patients' complications such as a death or survival. Our study criteria are the main implementation for this research.

Intervention
No intervention is applied in this research.

Sample Size

To construct a cross-sectional case series study and determine the prevalence of genetic mutation in Cystic Fibrosis (CF) in Children medical center Tehran from 1395-1400.

Statistical analysis
By using the latest SPSS software we will analyze this data.

Data Analysis
Statistical analysis will be performed using the Statistical Package for the Social Sciences (SPSS) software version 26 (SPSS Inc., Chicago, USA). Categorical variables will compare using Fisher's exact or χ 2 tests, and continuous variables will compare using student's t-test, ANOVA, and Chi-Square. Univariate analyses and multivariate analyses will be performed. Data will present as mean ± standard deviation (SD) for continuous variables and P value <0.05 will be considered statistically significant.

Ethical Considerations

- Informed consent is essential for safeguarding the patients' rights.
- Full Respect for anonymity and confidentiality must be present.
- Respect for privacy is prudent.
- Research will be designed, reviewed and undertaken to ensure that recognized standards of integrity are met, and quality and transparency are assured.
- Avoid fabrication by the creation of false data or other aspects of research.
- The independence of research should be clear, and any conflicts of interest or partiality should be explicit.

Safety Considerations

- No living being will be harmed in this research.
- No resources will go to waste.

Limitations

- The possibility of the information not being correctly mentioned in the documentsLimitations of the study is that patients may be unwilling or reluctant to participate in the study.
- The possibility that the information provided by the attendant could be biased.
- The information stored in the documents could be incomplete
- All the patients might not be accessible.

Result And Analysis

Mutation					
		Frequency	Percent	Valid Percent	Cumulative
Valid	F508 del	31	13.7	13.7	13.7
	F508 del	55	24.3	24.3	38.1
	R334w homo	18	8	8	46
	R334W hetro	13	5.8	5.8	51.8
	2183 AA>G	9	4	4	55.8
	2184 AA>G	3	1.3	1.3	57.1
	2789+5 G>A	4	1.8	1.8	58.8
	G542x Hetro	4	1.8	1.8	60.6
	G542X homo	9	4	4	64.6
	N1303K hetro	10	4.4	4.4	69
	N1303K homo	6	2.7	2.7	71.7
	2789+5 G>A	6	2.7	2.7	74.3
	F508del &	4	1.8	1.8	76.1
	R1162x homo	5	2.2	2.2	78.3
	F508 delt &	4	1.8	1.8	80.1

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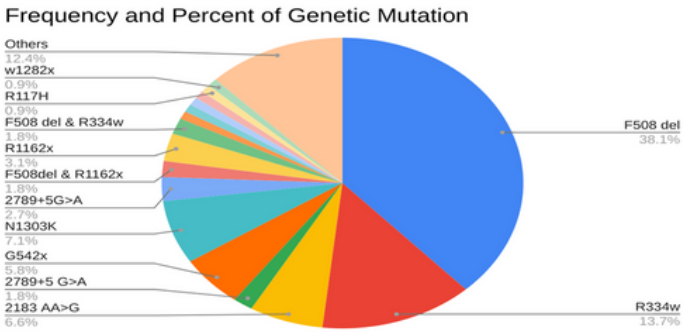
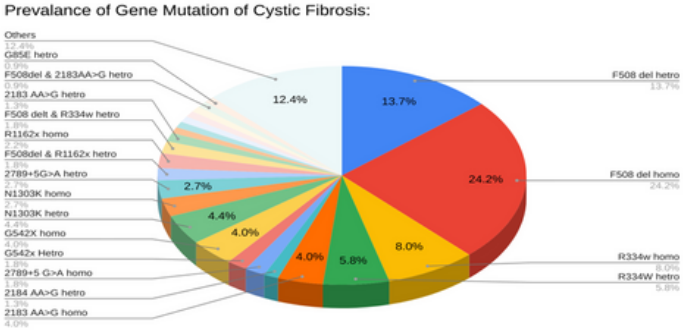
2183 AA>G	3	1.3	1.3	81.4
3120G>A homo	2	0.9	0.9	82.3
3849+10kb C>T	2	0.9	0.9	83.2
F508del & 2183AA>	2	0.9	0.9	84.1
G85E hetro	2	0.9	0.9	85
R1162x hetro	2	0.9	0.9	85.8
R117H hetro	2	0.9	0.9	86.7
w1282x homo	2	0.9	0.9	87.6
Others	28	12.4	12.4	100
Total	226	100	100	

Table 1: The prevalence of different type of mutation





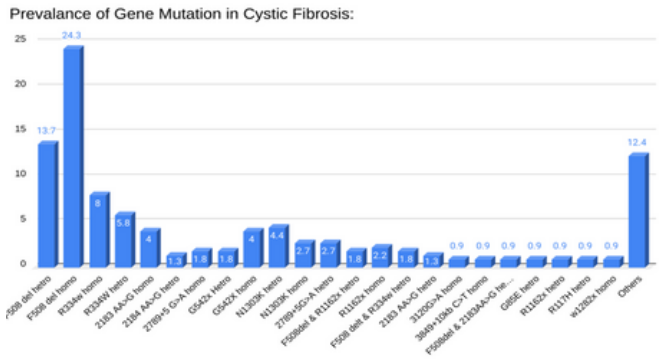


Figure 1: Pie chart for different type of mutation

Sex					
		Frequency	Percent	Valid Percent	Cumulative
Valid	Female	111	49.1	49.1	49.1
	Male	115	50.9	50.9	100
	Total	226	100	100	

Table 2: The frequency of gender

Descriptive Statistics					
	N	Minimum	Maximum	Mean	Std. Deviati
Age	226	0.08	45	9.5981	9.19347
Valid N (listwis	226				

Table 3: The mean age of all patients included in the study

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Age group					
		Frequency	Percent	Valid Percent	Cumulative Percent
Valid		49	21.7	21.7	21.7
	1-5 years old	40	17.7	17.7	39.4
	6-10 years old	53	23.5	23.5	62.8
	11-15 years old	37	16.4	16.4	79.2
	16-20 years old	17	7.5	7.5	86.7
	21-25 years old	15	6.6	6.6	93.4
	>=26 years old	15	6.6	6.6	100
	Total	226	100	100	

Table 4: The frequency of different age group

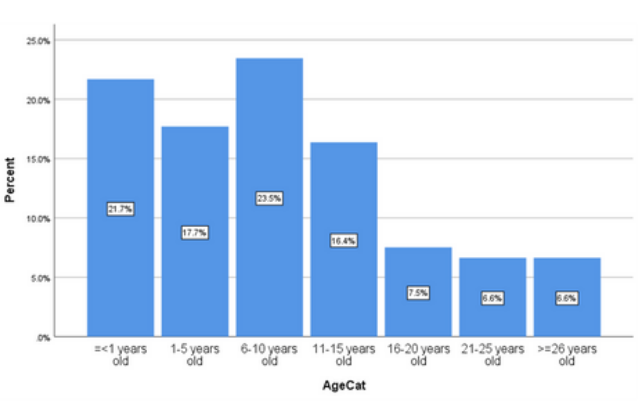

Figure 2: Bar chart for the frequency of different age group

Mutation * Sex Cross tabulation					
			Sex		Total
			Female	Male	
Mutation	F508 del hetro	Count	20	11	31
		% within Sex	18.00%	9.60%	13.70%
	F508 del homo	Count	23	32	55
		% within Sex	20.70%	27.80%	24.30%
	R334w homo	Count	9	9	18
		% within Sex	8.10%	7.80%	8.00%
	R334W hetro	Count	6	7	13
		% within Sex	5.40%	6.10%	5.80%
	2183 AA>G homo	Count	7	2	9
		% within Sex	6.30%	1.70%	4.00%
	2184 AA>G hetro	Count	2	1	3
		% within Sex	1.80%	0.90%	1.30%
	2789+5 G>A homo	Count	2	2	4
		% within Sex	1.80%	1.70%	1.80%
	G542x Hetro	Count	1	3	4
		% within Sex	0.90%	2.60%	1.80%
	G542X homo	Count	5	4	9
		% within Sex	4.50%	3.50%	4.00%
	N1303K hetro	Count	6	4	10
		% within Sex	5.40%	3.50%	4.40%
	N1303K homo	Count	3	3	6

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F508del & R1162...	Count	2	2	4
	% with	1.80%	1.70%	1.80%
R1162x homo	Count	5	0	5
	% with	4.50%	0.00%	2.20%
F508 del & R334...	Count	1	3	4
	% with	0.90%	2.60%	1.80%
2183 AA>G het...	Count	0	3	3
	% with	0.00%	2.60%	1.30%
3120G>A homo	Count	0	2	2
	% with	0.00%	1.70%	0.90%
3849+10kbC>T h...	Count	0	2	2
	% with	0.00%	1.70%	0.90%
F508del & 2183A...	Count	0	2	2
	% with	0.00%	1.70%	0.90%
G85E hetro	Count	1	1	2
	% with	0.90%	0.90%	0.90%
R1162x hetro	Count	0	2	2
	% with	0.00%	1.70%	0.90%
R117H hetro	Count	1	1	2
	% with	0.90%	0.90%	0.90%
w1282x homo	Count	2	0	2
	% with	1.80%	0.00%	0.90%
Others	Count	12	16	28
	% within Sex	10.80%	13.90%	12.40%
Total	Count	111	115	226
	% within Sex	100.00%	100.00%	100.00

Table 5: The frequency of different type of mutation in male and female

Discussion

Al-Abadi et al. in 2019, revealed facts regarding the frequency of cystic fibrosis gene mutation depending on sex, 55.8% (43/77) of mutations were found in male, whereas 44.2% (34/77) in female. 3272-26A and R334W mutations were found only in female patients, with 1.3% frequency rate for each. Male were more frequently had F508 del mutation (21/43-48.8%) in comparison with female, whose more frequently had F508del mutation (11/34-32.4%) N1303K (7/34-20.5%) and G85E (5/34-14.7%). Our findings suggest that cystic fibrosis in Jordan is not a rare disease, and found that the most frequent CFTR gene mutation was F508del. (Al-Abadi et al. 2019) While during our study 111 (49.1%) female and 115 (50.9%) male patients were reported. Secondly, in our study, 27.8% males are F508 del Homo have genes mutation. In female gene F508 del homo is reported 20.7%. So this result strongly affirm our study.

Our Mutation result has been reported as the following in our 226, in male F508 del hetro 11 (9.6%), F508 del homo 32 (27.8%), R334w homo 9 (7.8%), R334w hetro 7 (6.1%), 2183 AA>G homo 2 (1.7%), 2184 AA>G hetro 1 (0.9%), 2789+5 G>A homo 2 (1.7%), G542x hetro 3 (2.6%), G542x homo 4 (3.5%), N1303K hetro 4 (3.5%), N1303K homo 3 (2.6%), 2789+5G>A hetro 3 (2.6%), F508 del & R1162x hetro 2 (1.7%). R1162x homo 0, F508 del & R334w hetro 3 (2.6%), 2183 AA>G hetro 3 (2.6%), 3120 G >A homo 2 (1.7%), 3849+10KB C>T homo 2 (1.7%), F508 del & 2183AA>G hetro 2 (1.7%), G85E hetro 1 (0.9%), R1162x hetro 2 (1.7%), R117H hetro 1 (0.9%), w1282x homo 0 and others 16 (13.9%) mutation has been reported. In female F508 del hetro 20 (18%), F508 del homo 23 (20.7%), R334w homo 9 (8.1%), R334w hetro 6 (5.4%), 2183 AA>G homo 7 (6.3%), 2184 AA>G hetro 2 (1.8%), 2789+5 G>A homo 2 (1.8%), G542x hetro 1 (0.9%), G542x homo 5 (4.5%), N1303K hetro 6 (5.4%), N1303K homo 3 (2.7%), 2789+5G>A hetro 3 (2.7%), F508 del & R1162x hetro 2 (1.8%). R1162x homo 5 (4.5%), F508 del & R334w hetro 1 (0.9%), 2183

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AA>G hetro 0, 3120 G >A homo 0, 3849+10KB C>T homo 0, F508 del & 2183AA>G hetro 0, G85E hetro 1 (0.9%), R1162x hetro 0, R117H hetro 1 (0.9%), w1282x homo 2 (1.8%) and others 12 (10.8%) mutation has been reported. Hence, in overall in both gender F508 del is more frequent. F508 del males are more vulnerable to homo mutation and females were reported more in hetro mutation. Hence, this result confirmed our data's report.We divided the age in 7 groups. The first group =<1-year-old 49 (21.7%), second group 1 to 5 years 40 (17.7%), third group 6 to 10 years old 53 (23.5%), fourth group 11 to 15 years old 37 (16.4%), fifth group 16 to 20 years old 17 (7.5%), sixth group 21 to 25 years old 15 (6.6%) and in seventh group >= 26 years old are 15 (6.6%). This is the valid percentage of the age group. More patients set their position in group third 6 to 10 years old are 23.5%. The group with the least number seventh, 6.6.% in age =>26 years.

Result

In our study, 111 (49.1%) female and 115 (50.9%) male patients were reported. Secondly, in our study, 27.8% males are F508 del Homo have genes mutation. In female gene F508 del homo is reported 20.7%. Overall in both gender F508 del is more frequent. F508 del males are more vulnerable to homo mutation and females were reported more in hetro mutation. Our Mutation result has been reported as the following in our 226, in male F508 del hetro 11 (9.6%), F508 del homo 32 (27.8%), R334w homo 9 (7.8%) and in female F508 del hetro 20 (18%), F508 del homo 23 (20.7%), R334w homo 9 (8.1%), R334w hetro 6 (5.4%), 2183 AA>G homo 7 (6.3%) reported frequent. More patients set their position in group third 6 to 10 years old are 23.5%. The group with the least number seventh, 6.6.% in age =>26 years.

Conclusion

The most common gene mutation in our study is reported F508 del in both genders. The gender frequency also noted with a major difference in different mutations. Male and female are reported more frequent in homo and hetro respectively.

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