ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 02 | February - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

DEMOGRAPHY OF CYSTIC FIBROSIS AT A KASHMIRI SPECIALTY HOSPITAL

Paediatrics

Dr Mohammad Imran Malik Registrar, Department of Pediatrics, GMC Srinagar, J&K, India

Prof. (Dr.) Khurshid Ahmad Wani Professor, Department of Pediatrics, GMC Srinagar, J&K, India

Dr Wamiq Farooq Registrar, Department of Pediatrics, GMC Srinagar, J&K, India

Dr Irshad Ahmad Kumar* Medical Officer, Directorate Health Services Kashmir. *Corresponding Author

ABSTRACT

INTRODUCTION Cystic fibrosis (CF) is a recessive genetic disorder caused by a mutation in the epithelial chloride channel—CFTR (CFTR). Our study's primary objective was to examine the demographic profile of cystic fibrosis at our specialised hospital. **MATERIAL AND METHODS** The current research was a hospital-based observational study of individuals suspected of having cystic fibrosis undertaken at the G.B. Pant Children's Hospital in Srinagar between 2019 and 2022. In this research, 200 patients were chosen according to inclusion and exclusion criteria. **RESULTS** Out of 25, thirteen of our patients were male and twelve were female, for a ratio of 58% males to 42% females. 40% of the patients in our research were between 1 and 24 months of age. 60% were offspring of non-consanguineous marriage. Positive patients who were married consanguineously were third-degree relatives. During the course of our investigation, we discovered that the majority of patients were from Kupwara, totaling 25% of the study group. In our study, 72% of patients presented with recurrent respiratory tract infections. **CONCLUSION** In our study we concluded that Cystic fibrosis in Kashmir is more prevalent in men with non-consanguineous marriages and in the Kupwara district.

KEYWORDS

Cystic Fibrosis, Consanguineous marriage, Family history

INTRODUCTION

Cystic fibrosis (CF) is a recessive genetic disorder caused by a mutation in the epithelial chloride channel—CFTR (CFTR). CF is the most common genetic illness, with disease severity ranging from moderate to fatal. The proportion of CF patients within a community varies by ethnicity. CF affects one in every 3000 births in Northern Europe, but only one in every 10,000 newborns in Latin America, according to previous studies. [1,2]

A faulty gene is responsible for the buildup of a thick mucus coating that coats the airways and pancreas. This gene is known as the CFTR gene, and it encodes the Cystic Fibrosis Transmembrane Conductance Regulator protein (CFTR). In epithelial cells, the CFTR is a transmembrane channel that regulates the passage of chloride and bicarbonate ions into and out of the cell. If both CFTR genes are faulty, no functional CFTR protein will be produced, and the patient will develop CF symptoms. [3]

Typical first manifestations include recurrent and chronic lung infections and pancreatic insufficiency with increased sweat chloride levels. [4] Patients are diagnosed with diverse modes of presentation at various ages, from infancy to maturity; however, a small number of youngsters may have unusual presentations, which may influence the delay in diagnosis and prognosis. [5] In individuals with cystic fibrosis, the sweat glands are more impermeable to chloride ions, resulting in a higher concentration of chloride in the sweat reaching the skin's surface. In order to preserve electroneutrality, sweat glands decrease the reabsorption of sodium ions, leading to an increase in the concentration of sodium in sweat. [6] However, normal chloride and salt transport has been found in certain instances with cystic fibrosis, suggesting the involvement of other pathophysiologic mechanisms. In certain instances of cystic fibrosis, bicarbonate secretion across epithelial cells has been observed to be diminished. Bicarbonate, the principal buffer in the body, maintains the alkaline pH in the epithelia, and a decrease in bicarbonate production generates an anti-acidic environment that precipitates mucins and clogs ductal networks. [7] . Our study's primary objective was to examine the demographic profile of cystic fibrosis at our specialised hospital.

MATERIALAND METHODS

The current research was a hospital-based observational study of individuals suspected of having cystic fibrosis undertaken at the G.B. Pant Children's Hospital in Srinagar between 2019 and 2022. The Institute's Ethical Clearance Committee authorised the research (GMC Srinagar). Our research included all individuals aged 1 month to 18 years who were diagnosed or suspected to have cystic fibrosis based on sweat-chloride and CTFR gene mutation testing, or who had a sibling with cystic fibrosis. Patients younger than one month, those with an aberrant karyotype, and those with acute conditions such as anorexia nervosa, hypothyroidism, Addison's disease, and electrolyte imbalance were excluded from the research. A thorough medical history, physical examination, and all essential investigations were performed. Data were gathered and put into Microsoft Excel before being analysed using SPSS V 20.

RESULTS

In this research, 200 patients were chosen according to inclusion and exclusion criteria. Eight of the 200 chosen individuals had a previous diagnosis of cystic fibrosis. The remaining 192 individuals had two sweat chloride tests on two different days. 17 out of 192 individuals tested for cystic fibrosis were diagnosed with the disease. All 25 patients were analysed for the 7 most prevalent CFTR mutations (delta508p, 3849+10kb c>t, N1303K, G551D, and G542X).

As shown in Table 1, thirteen of our patients were male and twelve were female, for a ratio of 58% males to 42% females.

Table1: Sex Wise Distribution Of Cystic Fibrosis

Sex	Frequency	Percentage
Male	13	52%
Female	12	48%
Total	25	100

40% of the patients in our research were between 1 and 24 months of age, 24% were between 24 and 60 months of age, 20% were between 60 and 120 months of age, and 16% were older than 120 months. Our study group had a mean age of 59.12 months (Table 2).

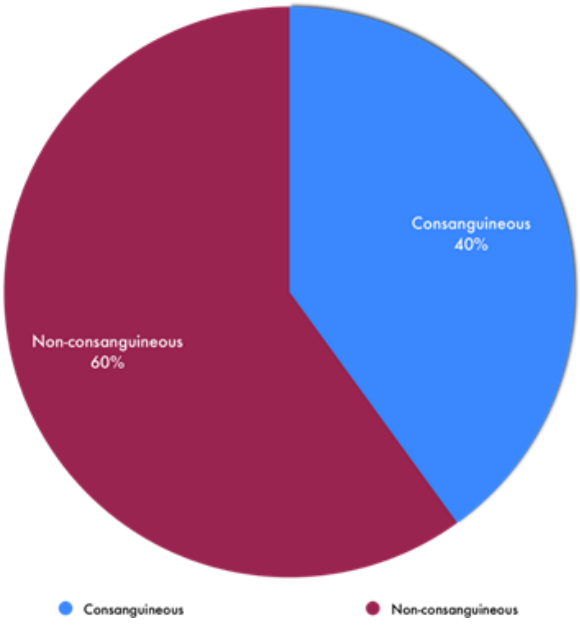
Table2: Age Distribution Of Cystic Fibrosis(n=25)

Age At Presentation	Frequency	Percentage	Mean Age At Diagnosis
1-24 Months	10	40%	16.3 Months±4.94
24 Months to 60 Months	6	24%	35.5 Months±9.1

60 Months to 120 Months	5	20%	90 Months±14.84
>120 months	4	16%	163 Months±21.46
Mean age 59.12			

40% of the individuals in our research were offspring of consanguineous marriage, whereas 60% were offspring of non-consanguineous marriage (Graph 1). Positive patients who were married consanguineously were third-degree relatives.

In addition, a family history of cystic fibrosis was present in 32% of the research participants, whereas it was lacking in 68%, as indicated in Table 3.



Graph 1. : Showing Consanguinity

Table3: Family History In Cystic Fibrosis(n=25)		
Family History Of Cystic Fibrosis	Frequency	Percentage
Present	8	32%
Absent	17	68%

We also gathered information on each patient's geographic location. During the course of our investigation, we discovered that the majority of patients were from Kupwara, totaling 25% of the study group, whereas patients from Shopian and Kulgam made up just 4% of the study group (Table 4).

Table 4: Geographical Distribution Of Cases Of Cystic Fibrosis In Kashmir		
Area	Number Of Cases	Percentage
Kupwara	6	25%
Bandipora	4	16%
Baramulla	4	16%
Srinagar	3	12%
Anantnag	2	8%
Budgam	2	8%
Pulwama	2	8%
Shopian	1	4%
Kulgam	1	4%
Ganderbal	0	0%

In our study, 72% of patients presented with recurrent respiratory tract infections, followed by 52% with failure to thrive, 32% with liver function abnormalities, 28% with chronic diarrhoea, and 28% with anaemia, as shown in Table 5. Greasy stools, clubbing, abdominal distention, abdominal pain, nasal polyps, and rectal prolapse were found infrequently in our patients.

Table 5: Most Common Clinical Presentation		
Clinical feature	Frequency	Percentage
Recurrent Respiratory Tract Infections	18	72%
Failure To Thrive (malnutrition)	13	52%
Liver Function Abnormalities	8	32%
Chronic Diarrhea	7	28%
Pallor (Anemia)	7	28%
Greasy Stools	6	24%
Clubbing	4	16%
Abdominal Distension	4	16%
Abdominal Pain	3	12%
Nasal Polyposis	2	8%
Rectal Prolapse	2	8%

DISCUSSION

From September 2019 to September 2022, an observational research was done at G. B. Pant Hospital, an affiliated tertiary care hospital of Government Medical College, Srinagar. It consisted of 25 patients, 13 of whom were male and 12 of whom were female. With a male-to-female ratio of 1.08:1, which is close to the gender distribution of patients in the research by GR Khatami et al. [8] In our research, the majority of patients were between 1 and 24 months of age. 10 (40%) of the 25 patients were between the ages of 1 and 24 months, 6 (24%) were between 24 and 60 months, 5 (20%) were between 60 and 120 months, and 4 (16%) were older than 120 months. Fateme Ziyaee et al. did a research on cystic fibrosis in children aged 0 to 18 at a university clinic in Iran, where they discovered that the most afflicted age group was between 6 months and 2 years. [9] GR Khatami et al. discovered that the typical age of illness start was between 0 and 12 months, and that more than 90 percent of the patients belonged into this group. [8]

40% of our patients were the result of consanguineous marriages, and 32% had a family history of cystic fibrosis, which is comparable with the findings of GR Khatami et al., who discovered 42% were the result of consanguineous marriages and 27% had a family history of cystic fibrosis. [8]

The majority of our patients were from the district of Kupwara (25%), followed by Bandipora (16%), Baramulla (16%), and Srinagar (12%). Anantnag, Budgam, and Pulwama each provided 8% of the afflicted population, while Shopian and Kulgam each contributed 4%. This information is consistent with the findings of Manzoor A.Raina et al. at the Clinical Biochemistry, Skims Soura, who discovered that Kupwara had the highest incidence of cystic fibrosis, followed by Baramulla and Srinagar in decreasing frequency. [10]

The most prevalent presenting symptoms at our hospital were respiratory tract infections (72%), followed by failure to thrive (52%) and persistent diarrhoea (28%) In decreasing order, further symptoms were LFT abnormalities, pallor, oily stools, and recurrent sinusitis. This is similar to the research undertaken by Fatemeh Faraahmad and colleagues. They discovered that 71% of their patients had recurring respiratory tract infections, and 90% of their patients had failure to thrive. [11]

However, the most prevalent manifestation in their research was failure to thrive, which accounted for 90% of their cases. This is in contrast to our result of 52%, which may be related to the nature of their research cohort, in which almost 95% of the individuals were less than one year old.

Similarly, respiratory tract symptoms and failure to thrive dominated the clinical picture of cystic fibrosis patients in a research undertaken by Masarat Sultana Kawoosa and colleagues.[12]

Conclusion:- In our study we concluded that Cystic fibrosis in Kashmir is more prevalent in men with non-consanguineous marriages and in the Kupwara district.

REFERENCES

1. O'Sullivan, B.P.; Freedman, S.D. Cystic fibrosis. Lancet 2009, 373, 1891–1904. [CrossRef]
2. Ratjen, F.; Bell, S.C.; Rowe, S.M.; Goss, C.H.; Quittner, A.L.; Bush, A. Cystic fibrosis. Nat Rev Dis Primers Nat. Rev. Dis. Primers 2015, 1.
3. Derichs, N., Targeting a genetic defect: cystic fibrosis transmembrane conductance regulator modulators in cystic fibrosis. Eur Respir Rev, 2013. 22(127): p. 58-65.
4. Doull IJ. Recent advances in cystic fibrosis. Arch Dis Child 2001; 85: 62-66.
5. Chernick V, Boat T, Wilmott R, Bush A. Kindig's Disorders of Respiratory Tract in Children. Amsterdam (HOL): Saunders Elsiever; 2006.
6. Hinda Kopelman. Cystic fibrosis: 6. Gastrointestinal and nutritional aspects. Thorax 1991;46:261-267.
7. Jeffery W Wine. Cystic fibrosis: The 'bicarbonate before chloride' hypothesis. Current Biology 2001;11:R463-R466.
8. GR Khatami et al. Characteristics of Patients with Cystic Fibrosis: Experience in a Large Referral Children's Hospital in Tehran, Iran. Middle East Journal of Digestive Diseases. 2010;2(1):20-23.
9. Ziyaee F et al. Clinical manifestation, laboratory findings, and outcome of children with cystic fibrosis over a 10-year period in South Iran. Egyptian Pediatric Association Gazette. 2020; 68(33):1-6
10. Raina MA et al. Assessment of Correlation between Sweat Chloride Levels and Clinical Features of Cystic Fibrosis Patients. Journal of Clinical and Diagnostic Research.

2016;10(12): 1-5.
11. Farahmand et al. Clinical presentation of cystic fibrosis at the time of diagnosis: a multicenter study in a region without newborn screening. The Turkish Journal of Gastroenterology. 2013; 24 (6): 541-545.
12. Kawoosa MS, Bhat MA, Ali SW, Hafiz M and Shastri S. Clinical and Mutation Profile of Children with Cystic Fibrosis in Jammu and Kashmir. Journal of Indian Pediatrics. 2014;15:176-8.