

# Cystic fibrosis, are we missing in India?

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## ABSTRACT

**Introduction:** This study was undertaken to evaluate and improve the availability and accessibility of CF services in Maharashtra, Pune, India.

**Material and methods:** Tool used was a questionnaire (Annexure I) which was prevalidated by the departmental faculty with 15 items which included experience regarding CF patients in relation to knowledge about diagnosis and prognosis of CF patients, availability about diagnostic and management services of CF was administered to the pediatricians who participated in the training workshop at Pune Regional centre, Aundh, Pune, Maharashtra, India. (n = 40).

**Results:** 92.5% agreed that Cystic fibrosis (CF), is now a pan ethnic disease. 100% accepted that the factors that are associated with decreased survival in CF patients from developing countries are early age of onset of symptoms, severe malnutrition at the time of diagnosis and frequent episodes of pneumonia. Severe malnutrition, not responding to nutritional therapy, neonatal meconium ileus and recurrent pneumonia, 87.5% 100% and 85% suspect cystic fibrosis respectively. 90% do not have facilities for diagnosis of cystic fibrosis like sweat chloride test and 87.5% felt absence of facilities makes the diagnosis of cystic fibrosis difficult. 90% agreed that the education of pediatricians about the disease, can improve the quality of life and survival in CF. **Conclusion:** Inadequacy of diagnostic services for CF may be reason for missing CF cases in India and we need to sensitize the health professionals for CF diagnosis and develop better diagnostic and patient support services for CF.

## Introduction:

Cystic fibrosis (CF) is an autosomal recessive genetic disorder. CF is thought to be more common in Caucasians. However, recent reports suggest that Indian children do suffer from CF. The estimated prevalence in migrant populations in the UK and US vary from 1 in 10,000 to 1 in 40,000 (1, 2). It is very important to understand the basic genetics and pathophysiology of CF which is pertinent to Indian geneticists and clinicians which will lead to the discovery of more CF patients. Precise incidence of CF in the Indian population is not known. Even if the prevalence of CF in India is 1 in 10,000 births, there may be 3000 children born with cystic fibrosis annually in different parts of India. Therefore, India would hold the largest population of CF patients in the world today (3).

Maharashtra University of Health Sciences (MUHS) is the apex University in the State of Maharashtra. The Department of Genetics, Immunology and Biochemistry of this University endeavors to be the key centre for diagnosing genetic disorders using modern molecular biology and genetic techniques. The study is an effort to find out the knowledge about CF and availability of genetic services for CF in Maharashtra, India before starting full fledged genetics services for CF.

## Aim of the study

This study aims to find out the availability and accessibility of CF genetic services in Maharashtra, Create and support a CF Services Network to share strategies for providing quality, accessible and comprehensive services to CF patients subsequently.

## Material and methods

Tool used was a questionnaire (Annexure I) which was prevalidated by the departmental faculty with 15 items which included their qualification, experience regarding CF patients in relation to knowledge about diagnosis and prognosis of CF patients, availability about diagnostic and management services of CF. The questionnaire was administered to the pediatricians who participated in the training workshop at Pune Regional centre, Aundh, Pune, Maharashtra, India.

The sample size was n = 40

**Results:**

The faculty who participated in the study were pediatricians from medical colleges with experience ranging from 2 years to 25 years. 92.5% agreed that Cystic fibrosis (CF), is now a pan ethnic disease. Almost all pediatricians 100% agreed that the factors that are associated with decreased survival in CF patients from developing countries are early age of onset of symptoms, severe malnutrition at the time of diagnosis and frequent episodes of pneumonia. In cases of severe malnutrition not responding to nutritional therapy 87.5% suspect CF. Similarly in cases of neonatal meconium ileus 100% suspect CF and recurrent pneumonia not responding to nutritional therapy, 85% suspect cystic fibrosis. 90% do not have facilities for diagnosis of cystic fibrosis like sweat chloride test and 87.5% felt absence of facilities for diagnosis (sweat chloride estimation and genetic studies), makes the diagnosis of cystic fibrosis difficult. 85% felt care of children with CF is not a priority for governments in developing countries. 80% wanted indigenous technology in sweat chloride test which may be alternative to diagnosis which is the need of hour. 90% agreed that the education of pediatricians about the disease, early diagnosis can improve the quality of life and survival in CF.

**DISCUSSION:**

This study highlights the need to improve genetics services in India. Many advances in genetics, from mapping the human genome through to the Human Genome Project continue to have significant impact on public health policy and service delivery (4). The changing scenario in genetics suggests that every physician will need to use genetic knowledge for patients presenting with specific symptoms and genetic testing will be increasingly important to aid in prescribing and clinical management of patients with genetic disorders (5). Genetic service experts feel that integration of genetics in health care will improve health by reducing morbidity and mortality in genetic disorders. It is important to provide education and training to health care professionals to enhance awareness, build competencies which will help them in proper management of CF patients. The pattern of genetic education was studied in different parts of world. UK genetic services are among the most highly developed in Europe, having evolved from academic departments into regional centers serving populations of 26 million (6). With a very large population and high birth rate, and due to consanguineous marriages, there is a high prevalence of genetic disorders in India. Due to inadequate diagnostic, management and rehabilitation facilities, the burden of these disorders is greater than in Western countries (7). Indian scenario of genetic education is also not different. Our survey results about genetics education show that at undergraduate level (64.65%) and at post graduate level (69.31%) hours of teaching in genetics is inadequate (less than 5) while nursing schools in India do have 15 hours of teaching in genetics (unpublished data). Medical Genetics course represents 54 hours of the fourth semester of Medicine and it is also taught in Dentistry, Nursing, and Health Technology (8). Guttmacher et al. (2007) summarized disconnect between basic sciences and clinical experiences during training. Failure to integrate genetics across the curriculum and inadequate representation of genetics on certifying exams are other important factors leading to dearth of genetics professionals (9). Surveys of health professionals demonstrate a lack of basic knowledge and confidence to deal with genetics-related issues that arise in the clinical setting. Confirmation for those findings about the genetics knowledge and competence of health professionals also comes from survey data from 5,915 consumers of genetics services (Harvey et al. 2007a) data, collected by the Genetic Alliance and the National Coalition for Health Professional Education in Genetics (NCHPEG), show that 32% of respondents rated the genetics knowledge of their providers as poor, across all disciplines, and 78% reported that they received no genetics-education materials from the provider designated as most important to the family (10). The results of our survey also are giving similar results. A strategic plan developed by the Genomics Directorate in consultation with the Western Australian Genetics Council Allocate resources stressed on need improving genetics knowledge of health professionals and the community. In western countries in, CF services are well developed which resulted in better survival of CF patients. CF services in India are in developing state, There are few centres which offer diagnostic facilities and clinical care to CF patients. It is important to develop early diagnostic and better treatment facilities for patients of CF to prevent mortality and morbidity. It is very much the need of the hour to have database of CF and recognize pattern of mutational analysis of CF for Indian patients. This is also stressed by Kabra et al. Appropriate strategies for patient identification, diagnosis and molecular study need to be devised which will help in planning strategies for prenatal diagnosis (11). Our study brings out very important point pediatricians do suspect CF but non availability of CF related diagnostic services is the hindrance for further management of CF. The availability of the sweat test, cystic fibrosis (CF) has been difficult to diagnose in early childhood, and delays can lead to severe malnutrition, lung disease, or even death. screened group had a higher proportion of patients with pancreatic insufficiency, their growth indices were significantly better than those of the control group during the 13-year follow-up evaluation and, therefore, this randomized clinical trial of early CF diagnosis must be interpreted as unequivocally positive (12). This study highlights need for establishing good services for diagnosis of CF. It also brings out that pediatricians do suspect CF in practice but further confirming diagnosis and proper management of CF patients in India is difficult due to poor genetic services of CF.

**Conclusion:** Inadequacy of diagnostic services for CF may be reason for missing CF cases and we need to sensitize the health professionals for CF diagnosis and develop better diagnostic and patient support services for CF. There is a need for development of CF diagnostic services so that more and more CF cases will be diagnosed. Early diagnosis will improve patient management and reduce mortality.

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Annexure : 1

Cystic fibrosis, are we missing in India?

Dear participant,

This questionnaire is an effort to find out problem of Cystic fibrosis in our society. We would be thankful to you if you can give us the feedback from your clinical practice about this disease.

Name:

Designation with Qualification:

Years experience:

Address:

Phone No:

Signature:

1. Cystic fibrosis (CF), earlier believed to be non existent in non Caucasians, is now a pan ethnic disease, having being reported from various regions of the world over last one decade.  
a. Strongly agree   b. Agree   c. neutral   d. disagree   e. strongly disagree
2. Care of children with CF is not a priority for governments in developing countries  
a. Strongly agree   b. Agree   c. neutral   d. disagree   e. strongly disagree
3. Cystic fibrosis is rare in India  
a. Strongly agree   b. Agree   c. neutral   d. disagree   e. strongly disagree
4. Factors that are associated with decreased survival in CF patients from developing countries are age of onset of symptoms <2 months, severe malnutrition at the time of diagnosis and frequent episodes of pneumonia .  
a. Strongly agree   b. Agree   c. neutral   d. disagree   e. strongly disagree
5. Absence of facilities for diagnosis (sweat chloride estimation and genetic studies), makes the diagnosis of cystic fibrosis difficult  
a. Strongly agree   b. Agree   c. neutral   d. disagree   e. strongly disagree
6. Education of pediatricians about the disease, early diagnosis can improve the quality of life and survival in CF  
a. Strongly agree   b. Agree   c. neutral   d. disagree   e. strongly disagree

7. Using indigenous technology in sweat chloride test may be alternative to diagnosis and is the need of hour
  - a. Strongly agree
  - b. Agree
  - c. neutral
  - d. disagree
  - e. strongly disagree
8. In cases of severe malnutrition not responding to nutritional therapy, I do suspect cystic fibrosis.
  - a. Strongly agree
  - b. Agree
  - c. neutral
  - d. disagree
  - e. strongly disagree
9. Aggressive physiotherapy with nutritional management and judicious use of antibiotics can improve the quality of life and survival in CF
  - a. Strongly agree
  - b. Agree
  - c. neutral
  - d. disagree
  - e. strongly disagree
10. I do come across cystic fibrosis patients in my practice
  - a. Strongly agree
  - b. Agree
  - c. neutral
  - d. disagree
  - e. strongly disagree
11. Many of the above mentioned hurdles can successfully be solved by establishing CF services.
  - a. Strongly agree
  - b. Agree
  - c. neutral
  - d. disagree
  - e. strongly disagree
12. In cases of recurrent pneumonia I do suspect cystic fibrosis.
  - a. Strongly agree
  - b. Agree
  - c. neutral
  - d. disagree
  - e. strongly disagree
13. I do have facilities for diagnosis of cystic fibrosis like sweat chloride test
  - a. Yes
  - b. No
14. Cystic fibrosis is never considered in my practice
  - a. Yes
  - b. No
15. In cases of neonatal meconium ileus, suspect cystic fibrosis
  - a. Yes
  - b. No