Research Paper Medical Science

Cystic Fibrosis needs more attention

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ABSTRACT

Cystic fibrosis (CF) is a rare inherited disorder characterized by abnormality of exocrine glands which can affect lungs, pancreas, reproductive system of males and dehydration particularly in summer. Sweat chloride though considered as the gold standard still not final answer for the diagnosis of CF. Our clinical and laboratory profile of 4 patients showed growth failure, Sweat chloride positive in only two cases., history of rectal prolapse with stool examination fat globules , recurrent lower respiratory tract infection with negative sweat chloride test,bronchiectasis who had sweat chloride positive and fourth case of severe pneumonia, chronic diarrhoea, failure to thrive, respiratory distress, anaemia. was referred for cystic fibrosis work up. Investigations reveal severe dimorphic anemia hypoalbuminemia, elevated LDH and liver enzymes. USG abdomen was normal. Upper Gastrointestinal Endoscopy was suggestive of moderate villous flattening with deficiency of folds, Liver biopsy done showed extensive fatty change with portal and septal fibrosis but sweat chloride test was negative in this case. CT & Chest X ray showing consolidation Lt lower lober. Molecular analysis of this case for DF508 showed homozygous status. Inadequacy of diagnostic services for CF may be reason for missing CF cases and we need to 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

Cystic fibrosis (CF) is a rare inherited disorder characterized by abnormality of exocrine glands which can affect lungs, pancreas, reproductive system of males and dehydration particularly in summer. Sweat chloride is considered as the gold standard for the diagnosis of CF.

Herewith sharing the clinical and laboratory profile of 4 patients referred to us for molecular analysis for CF. All four had growth failure. Sweat chloride was done in all cases which was positive in only two cases. 9 years old male with consanguinity term normal delivary with weight 20kg and height 122cm less than 3rd percentile failure to thrive with history of rectal prolapse with stool examination showing fat globules and sweat chloride positive. Second case of a 10 year old male with history of recurrent lower respiratory tract infection with negative sweat chloride test. Third case was 12 years old with recurrent LRI CT scan showing bronchiectasis who had sweat chloride positive. The fourth was2 years old toddler full term born by caesarean section of 3rd degree consanguineous marriage who was diagnosed with severe pneumonia, chronic diarrhoea, failure to thrive, respiratory distress, anaemia was referred for cystic fibrosis work up. Investigations reveal severe dimorphic anemia hypoalbuminemia, elevated LDH and liver enzymes. USG abdomen was normal. Upper Gastrointestinal Endoscopy was suggestive of moderate villous flattening with deficiency of folds, Liver biopsy done showed extensive fatty change with portal and septal fibrosis, sweat chloride test was negative. CT & Chest X ray showing consolidation Lt lower lober. Molecular analysis for DF508 showed homozygous status.

KEYWORDS cystic fibrosis,DF598 mutation

Table No:1

Age in yrs	Clinical profile	Sweat chloride test	Molecular analysis DF 508
9	Chronic diarrhea	Positive	Negative
10	Recurrent Lower Respiratory Tract	Negative	Negative
12	Recurrent Lower Respiratory Tract Bronchiectasis	Positive	Negative
2 yrs	with severe pneumonia, chronic diarrhoea, failure to thrive, respiratory distress,	Negative	Positive

Table No:2

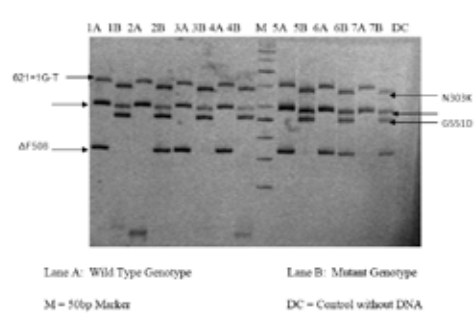

Table No:3

Lane No.	Sample I.D.	Result	Sweat chloride test
1A/1B	CF suspect 1	Wild Type	positive
2A/2B	CF suspect2	Homozygous Mutant	Negative
3A/3B	CF suspect3	Wild Type	Negative
4A/4B	CF suspect4	Wild Type	Positive
5A/5B	Wild Type Control	Wild Type	
6A/6B	Heterozygous Control	Heterozygous	
7A/7B	Homozygous Mutant	Homozygous Mutant	

Fig : 1 : T, A, Y and V were suspected cases of Cystic fibrosis with sweat chloride results

DISCUSSION: 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). . 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).

In western countries, CF services are well developed which resulted in better survival of CF patients. There are few centers which offer diagnostic facilities and clinical care to CF patients in India. 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 (5). 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 a very important point that even though pediatricians do suspect CF clinically but non availability of CF related diagnostic services is the hindrance for diagnosing and hence further management of CF. The feasibility of the sweat chloride test to diagnose CF in early childhood is difficult and delay in diagnosis can lead to severe malnutrition, lung disease, or even death. 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 confirmation of diagnosis and proper management of CF patients in India is difficult due to poor genetic testing services of CF.Considering the heterogeneity of the disease associated phenotypic manifestations, and lack of awareness, it is likely that we are missing many cases in India..

Conclusion: Inadequacy of diagnostic services for CF may be reason for missing CF cases and we need to 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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