



Case Report

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Cystic Fibrosis in India- Need for Study

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Introduction

Cystic fibrosis in earlier days was described as “Cystic fibrosis of Pancreas” in a child. The word cystic fibrosis was coined due to the characteristic cysts and fibrosis found in the pancreas. It was a leading cause of mortality in the people of northern Europe in the 16th century. In the early years of cystic fibrosis, it was known that if the fingers of the child tasted salty after rubbing the forehead, it was going to die soon. The classical description of cystic fibrosis was first reported in a child by Andersen [1], Physician and Pediatric pathologist in 1938. The inheritance of cystic fibrosis was described as an autosomal recessive by Lowe et al. [2]. Farber recognized that the sticky mucus caused many of the symptoms and, hence, labeled it as “Mucoviscidosis” in 1945 [3].

Cystic fibrosis is the most common serious autosomal recessive disorder. It affects Caucasian populations, with a birth prevalence of 1 in 2500 [4]. It is the most common genetic cause of bronchiectasis in the Caucasian population. The disease is characterized by abnormality of exocrine glands which affect lungs, pancreas, reproductive system of males and sweat glands leading to electrolyte abnormalities. These cases can present as dehydration, particularly in summer. It was found that cystic fibrosis patients have excess electrolytes in sweat. This became the basis for the sweat chloride test which measures the salt concentration in the sweat which got established as the diagnostic test [5]. Measurement of sweat chloride became the gold standard test with development of pilocarpine iontophoresis method by Gibson & Cooke [6].

Cystic Fibrosis Trans-membrane Regulator (CFTR) mutations cause defective regulation of the chloride channels which result in drying up of secretions leading to increase in viscosity of mucus and decrease in the mucociliary clearance. These mutations also result in abnormalities of epithelial ion and water transport, which disturb airways, mucociliary clearance and other cellular functions [7].

CFTR mutations are found in about two third of cystic fibrosis cases. There is a loss of a phenylalanine at amino-acid position 508 (F508del). The majority of these mutant alleles are single base-pair (bp) substitutions, micro insertions /deletions, with a 3-bp deletion [8].

There is no complete cure for cystic fibrosis till today and treatment for cystic fibrosis patients is mainly palliative. Hence, it is important to give supportive therapy for these patients. Equally, prevention becomes an important step, which includes genetic counseling and pre-implantation genetic diagnosis (PGD).

There are very few studies on clinical and molecular aspects of cystic fibrosis in India and hence no proper data exists on the number of affected patients in the population. In majority of cases, cystic fibrosis is not diagnosed in the early stage which may lead to complications with significant morbidity and mortality. Cystic fibrosis is not confirmed in majority of suspected cases due to lack of adequate diagnostic facilities like sweat chloride testing and molecular testing. We do not know the prevalent mutations in the Indian population and identification of prevalent mutations is the need of the hour. This will help in developing strategies for genetic counseling and prenatal diagnosis of cystic fibrosis.

Though cystic fibrosis is the most common genetic disorder amongst Caucasians, in India cystic fibrosis was supposed to be extremely rare. In majority of cases, it is not suspected or it is misdiagnosed. In majority of cases, when the children are referred for testing, the disease is seen in advanced stage. The knowledge about the incidence and molecular genetics of cystic fibrosis in the Indian subcontinent is limited. In all probability, it may be under reported or diagnosed in late stages. Recently there have been many reports from Indian researchers which suggest that the disease burden may be more [9-12]. The present project aims to study cystic fibrosis in Maharashtra state with respect to clinical profile and the mutations.

It is important to develop early diagnostic and better treatment facilities for patients of cystic fibrosis to prevent mortality and morbidity. It is critical that a database of cystic fibrosis patients with respect to clinical profile and pattern of mutations in Indian patients be built. An appropriate strategy for patient identification, diagnosis and molecular study is very much required. This study focuses on identifying common symptoms and clinical signs of cystic fibrosis so that physicians will be able to identify the disease in the early stages. The effort is also to educate the parents about cystic fibrosis, so that they can adopt a preventive strategy of cystic fibrosis like prenatal diagnosis and PGD in the subsequent pregnancies. The aim is to create an action plan for genetic counseling and a support group for cystic fibrosis patients. Understanding molecular profile of cystic fibrosis will help us in setting up a panel of common CFTR mutations which can be used in carrier detection and prenatal diagnosis of cystic fibrosis.

In India, there is no protocol for newborn screening programme and no specific strategy for newborn screening particularly with respect to cystic fibrosis. The diagnostic facilities for cystic fibrosis are not uniform in all parts of the country. There are no specialty clinics for cystic fibrosis which can deliver holistic management for these patients. We need to have a well defined management protocol for cystic fibrosis patients with respect to genetic diagnosis and treatment plan. There are very few diagnostic centers all over the world and hence, emphasis should be on increasing physicians' awareness and use of clinical screening tools. At present, high index of clinical suspicion is mandatory for early detection and prompt treatment intervention.

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