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CFTR Mutations in India: Need to do More!

GENETICIST'S PERSPECTIVE

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Cystic fibrosis is an autosomal recessive multisystem disease with significant morbidity and mortality in all parts of the world. Contrary to the popular belief, early genetic studies on cystic fibrosis confirmed that the condition does occur in India and the most prevalent delta F508 mutation occurred frequently, in 19-56% of cases [1-3]. The largest series on mutation analysis in Indian patients found that delta F508 mutation accounted for 31.1% mutations [4]. It was also noted that Indians may have a different spectrum of mutations as several new mutations were observed [4-6]. The carrier frequency of 0.42% appears to be an underestimate considering a 10 times higher incidence in Caucasians and the disease is not studied well in Indian population [7]. It also needs to be emphasized that it is not easy to diagnose and manage this condition in our country [8].

Kawoosa and colleagues [9] have screened a group of patients with clinical symptoms of cystic fibrosis from Jammu and Kashmir. Then they performed testing for two specific mutations known to cause cystic fibrosis in selected group of 15 patients with the diagnosis of cystic fibrosis. It is important to note that the authors have chosen to screen only for two mutations. Though this is often used as a strategy in a resource-scarce setting, given the allelic heterogeneity, it would have been better to screen the entire gene. Labeling cases with only one mutation as cystic fibrosis transmembrane conductance regulator related metabolic syndrome is also not appropriate as the entire gene was not sequenced in them. They could identify only 16 alleles in 15 patients leaving nearly half the alleles unidentified. Hence it would be premature to draw conclusions on the commonality of the mutations considering the small study group and mutation detection strategy. Nevertheless this study highlights the frequency of the condition and two selected mutations in the studied population and the need for effective strategies for mutation detection that can go a long way to offer genetic counseling services for these families.

Recently mutation-specific treatment has been tried in patients with at least one G551D mutation that was based on prior knowledge of the effect of the mutation on CFTR channel and its repair by the drug [10,11]. Hence it is important that such studies are expanded to provide comprehensive mutation testing facilities for this condition in our country as we get ready to offer newborn screening for this condition.

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