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Clinical profile and molecular diagnosis of Cystic Fibrosis

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This is a study conducted with aim to understand clinical profile and molecular diagnosis of suspected cystic fibrosis CF) in Maharashtra, India over a period of three years from January 2012 to December 2014 in the Genetic laboratory of Maharashtra University of Health Sciences (MUHS), Regional Centre Pune. All (n=113) were tested for DF508 mutation at our laboratory followed by 5 mutational analysis at Hinduja Laboratory Mumbai(n=25). 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 stage. We included children with signs and symptoms of cystic fibrosis like failure to thrive, chronic diarrhea, recurrent pneumonia, newborn with meconium ileus, adults with infertility and excluded HIV positive, Tuberculosis, and malignancy patients. The most common presentation of suspected cystic fibrosis was malnutrition followed by recurrent respiratory infection. In our study group, the age ranged from newborn 1 day to 39 years. Our study also includes29 cases (25.66%) who were less than 1 year which suggests good awareness of cystic fibrosis in physicians even in the absence of a neonatal screening protocol in Maharashtra. These infants were referred based on clinical suspicion. We found malnutrition in 87.75% in 0-18 years age group and 90.47% malnutrition in less than 1 year. We had 28.5% Pneumonia cases and 6.3% cases with bronchiectasis. We found DF 508 mutation in only 4 patients. We found the frequency of DF508 as 6.34% in suspected cystic fibrosis cases in Maharashtra. This is lower than the other reports from India, Kabra et al 2003 (n=120) 19%, Ashavaid et al 2012(n=96) 53% and Mir et al 33.3%. This may be due to inclusion of all suspected cystic fibrosis cases in our study without considering the results of sweat chloride test. Sweat chloride test was done in only 22.2% cases. Sweat chloride test was positive in 35.7% of the suspected cystic fibrosis cases. One of the important cases, 2 years old male child presenting with negative sweat chloride test with recurrent Pneumonia/ Chronic Diarrhea/ Anemia was positive for DF508.This brings out very important fact that any child with multisystemic involvement with malnutrition in spite of sweat chloride test results should be subjected for detection of CFTR mutations. History of consanguity was present in 15.9% and in 7.9% suspected cystic fibrosis cases gave family history of death of siblings.DF508 mutation against gold standard sweat chloride test we found sensitivity 20% and specificity 85.7%. Our study also found DF 508 positive status in 8% of male infertility cases.

Conclusion: We found the most common presentation of suspected cystic fibrosis was malnutrition followed by recurrent respiratory infection and DF508 is not the common mutation in Maharashtra. Probably our mutation profile is different, more detailed study is warranted. The possibility of missing cystic fibrosis cases due to lack of genetic testing services like sweat chloride testing cannot be ruled out.

Speaker Biography

Pramila Menon has completed her PhD at the age of 54 years from Maharashtra University of Health Sciences, Nashik. She is the Sate Consultant in Infant Young Child and Adjunct Professor at Dr D Y Patil Medical College Pune. She worked as Associate Professor at Maharashtra University of Health Sciences (MUHS), Regional Centre Pune. She has over 20 publications.

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