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EXTENDED ABSTRACTS

Spectrum of CFTR Mutations in the Algerian Population: Molecular and Computational Analysis

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ABSTRACT

Little has been reported on the occurrence of CF in Algerian population. so as to contribute to the few existing data we undertook this study. The aim was in first instance to detect genetics alteration within the CFTR gene of 21 CF Algerian patients by sequencing. 14 different mutations were detected one among them has never been described. Among these mutations the c.680T>G (L227R) which seems to be specific to the Algerian population, it had been in silico studied to work out its impact at a molecular level. this is often the primary study that combined a molecular and computational analysis. These findings will assist in guidance , diagnostic procedure and future screening of CF in Algeria. The nature and frequency of the main CFTR mutations within the North African population remain unclear, although alittle number of CFTR mutation detection studies are wiped out Algeria and Tunisia, showing largely European mutations like F508del, G542X and N1303K, albeit at different frequencies, which presumably emerged via population admixture with Caucasians. Some unique mutations were identified in these populations. this is often the primary study that has a genetic and clinical evaluation of CF patients living in Algeria. so as to supply an efficient diagnostic service and to form accurate risk estimates, we decided to spot the CFTR mutations in 81 Algerian patients. We administered D-HPLC, chemical-clamp denaturing gradient gel electrophoresis, multiplex amplification analysis of the CFTR gene and automatic direct DNA sequencing. We identified 15 different mutations which account for 58.5% of the CF chromosomes. We used a quantitative PCR technique (quantitative multiplex PCR short fragment fluorescence analysis) to screen for deletion/duplication within the 27 exons of the gene. Taking advantage of the homogeneity of the sample, we report clinical features of homozygous CF patients. As CFTR mutations are detected in males with infertility, 46 unrelated Algerian individuals with obstructive azoospermia were also investigated. Cystic fibrosis (CF) may be a severe life threatening genetic disorder commonest among Caucasians with an incidence starting from 1 in 2500 to 1 in 3600 [1]. CF is inherited in an autosomal recessive way and therefore the CF transmembrane conductance regulatory gene (CFTR), located on chromosome 7q31.2[2], has been identified because the responsible gene encoding a transmembrane protein that functions as a chloride channel and a regulator of other channels across the somatic cell membrane. The defective protein impairs water movement across epithelia resulting in formation of viscous mucus that obstructs the airways of the lungs and ducts of the pancreas. CF is characterized by progressive lung disease, pancreatic dysfunction, elevated sweat electrolytes, and male infertility [3]. So far, quite 1900 different CFTR mutations havebeen reported [4]. Although most mutations are rare, the three-base-pair deletion p.(Phe508del) is commonest within the Caucasian population affecting about 70% of the patients whereas within the Jewish population the p.(Trp1282∗) is that the most prevalent with a frequency of 60% [5], clearly indicating that the occurrence of mutations is very population specific. for several ethnic or geographic populations, the mutation spectrum has been determined [6–15]. Recently, CF has been diagnosed within the Middle East starting from 1 in 2,500 to 1 in 16,000 with different mutation frequencies consistent with the ethnic origin of populations [16]. However, reliable information about the frequency of CF among the Palestinians remains lacking and therefore the spectrum and nature of mutations haven't been documented yet hampering molecular diagnostics. an honest insight into the character and frequency of the mutations during a specific population may be a prerequisite to line up adequate and cost-effective molecular diagnostics. The aim of this study was to work out the CF mutation spectrum among the Palestinian patient population. Samples from 60 unrelated CF patients residing within the West Bank and Gaza were collected and their respective CF mutations were determined. Consequently, the mutation spectrum was compared with other ethnic groups residing within the Arabic population.

Keywords: CFTR gene; Mutation; Algerian population; Analysis in Silico

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