Abstract

Cystic Fibrosis (CF) is a well-known life threatening inherited disease caused by mutations in the Cystic Fibrosis Trans Membrane Conductance Regulator gene (CFTR gene). Those mutations produce a defect in the encoded CFTR protein that functions mainly as a chloride channel and as regulator of other channels. CF patients are characterized by progressive lung disease, pancreatic dysfunction, elevated sweat electrolytes and male infertility. This study included a total of 73 patients (43 Males / 30 Females) (60 unrelated) residing in Palestine (West Bank and Gaza) who were clinically diagnosed with cystic fibrosis. The aim of the study was to determine the types and rates of mutations present in the CFTR gene and to develop allele specific mutation analysis test for the most common mutations among Palestinians. This test will make molecular testing for CF in Palestine possible and will have a direct impact on a better treatment of these patients. Whole blood was collected and DNA from these samples was extracted by automated methods. For each patient, PCR amplifications were performed for the coding region of the CF gene. Consequently, sequencing by Next Generation Sequencing was performed which enabled us to identify the CF mutations present in this cohort. After validating these mutations by different methods like Sanger sequencing, we defined a set of 18 mutations present in the Palestinian population. In a next step, we designed an easy and fast allele specific diagnostic test for 8 of the identified mutations that represent more than 80% of the CF mutations found in this population.