

Application of Next-Generation Sequencing Technology for CFTR Mutation Screening

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Objective: We report here the results of next-generation sequencing analysis of *CFTR* gene in first ten patients.

Methods: Genomic DNA was isolated from blood samples of ten patients who had been referred to our center for *CFTR* gene mutation screening for different indications. AmpliSeq libraries were produced by using Ion AmpliSeq Library Kit for coding regions (NT_007933.16) of the *CFTR* gene (NG_016465.3). Amplicons were enriched by using Ion PGM Template OT2 200 Kit and sequenced on Ion Torrent Personal Genome Machine by using Ion PGM Sequencing 200 Kit v2. hg19 (Genome Reference Consortium GRCh37) was used as a reference. Torrent Suite Software v4.2, VariantCaller (v4.2-r88446) and Ion Reporter Software 4.2

were used for the analysis. IGV_2.3.8 was used to visualize the sequences. Mutations were confirmed using the Sanger sequencing method.

Results: There was no pathogenic mutation in five out of ten cases. There was one patient each for the following mutations: homozygous c.1521_1523delCTT, heterozygous c.1521_1523delCTT, c.3154T>G (p.Phe1052Val), and heterozygous c.3683A>G (E1228G). There was a heterozygous c.1576C>G (L526V) mutation in a patient directed to us for *CFTR* mutation screening before conception.

Conclusion: The patient that we found to have a homozygous c.1521_1523delCTT mutation has being followed-up for cystic fibrosis. c.1576C>G (L526V) mutation that we found in the other patient was not reported in the literature before. This mutation was tested in Mutation Taster and Polyphen and it was concluded that it may have a pathogenic affect. As a result, we suggest that next-generation sequencing method can be used as a successful method to screen *CFTR* gene mutations.

Key words: CFTR, next generation sequencing analysis, c.1521_1523delCTT, c.3154T>G (p.Phe1052Val), c.3683A>G (E1228G)