

## Determination of F508del Mutation that Cause Cystic Fibrosis by Genosensor Analysis

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### Abstract

Identifying genetic alterations throughout an entire gene is necessary for the confirmation of clinical diagnosis, for the establishment of genotype-phenotype correlation and prenatal diagnosis in many complex inherited diseases. The gene of Cystic Fibrosis Transmembrane Conductance Regulator (CFTR), including characteristics above mentioned, consists of 27 exons. Some of the mutations, such as F508del in exon 10, are associated with severe phenotypes. For this reasons, a powerful mutation-screening tool is needed to scan the exons and intronic splice regions. High Resolution Melting (HRM) analysis provide a rapid and accurate alternative for CFTR analysis. Genosensor is a rapid, closed-tube method that is at least as accurate as other scanning techniques that require heteroduplex separation. It is used to characterize DNA samples according to their dissociation behavior as they are transformed from double stranded DNA to single stranded DNA by increasing temperature. Mutation detection takes place in the same reaction tube together with amplification and it takes only 5 minutes. Different sequences patterns correlate with different mutations in genosensor. In this study, we scanned F508del mutation of 30 cystic fibrosis samples by genosensor analysis. The resonance of the amplified samples were compared with wild-type samples. Heterozygous samples have a different resonance from that of the common homozygous cluster, with low-temperature deviations resulting from heteroduplexes. Three F508del heterozygous samples were detected easily by genosensor analysis. Genosensor analysis provides a convenient way to detect mutations in a multiexon gene like CFTR without performing full gene sequencing.

### Biography

Umut Kökbaş has graduated from Biochemistry Department of Ege University at 2011. He started as researcher assistant at Medical Biochemistry Department of Çukurova University at 2011. He transferred to University of Kyrenia as Associated Professor when PhD degree is ended at 2019. He is subject of interests are Bioelectrochemistry and Genetic Researchers.

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