## OTR.28 - Targeted Next Generation Sequencing for *CFTR* gene analysis in a Brazilian cohort of cystic fibrosis patients

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**Introduction:**

Cystic Fibrosis (CF) is an autosomal recessive disease, recognized as one of the most prevalent in Caucasoid populations. CF occurs due to mutations in the *CFTR* gene, located on chromosome 7q-31.2. This gene encodes the *CFTR* protein, a transmembrane channel responsible for the flow of chlorine, sodium and water. Patients with CF have mutations that affect normal function or cause the absence of this protein. To date, more than 2000 sequence variations have been described in the *CFTR* gene. The characterization of these mutations leads to the diagnosis and a possibility of a specific treatment for each patient from personalized therapies.

**Objective:**

The aim of this study was to perform the complete screening of the *CFTR* gene through next generation sequencing to increase the knowledge about the mutations that circulate in the population of Rio de Janeiro and to define a panel for the diagnosis of the disease.

**Methodology:**

The screening of mutations was performed throughout the entire length of the *CFTR* gene, including coding and no coding regions. We have used the Illumina Next Generation Sequencing (NGS) platform. For library preparation and sample enrichment we used the TruSeq® method with a custom panel of 990 amplicons designed by Illumina DesignStudio. Ninety-three individuals were divided into two groups according to the results previously observed in a routine screening of 30 known *CFTR* variants. Fifty patients had one mutated allele previously identified and 43 patients had no mutation observed. All 93 patients were than analysed by NGS.





**Results:**

A total of 93 patients were analyzed, and 38 mutations were identified, one of which was never described. Forty-seven patients had their diagnosis completed, one of whom was part of the group with no allele defined before the NGS. To date, forty-nine different pathogenic genetic variations define the panel of mutations of our laboratory.

**Methodology:**

Through the study of the *CFTR* gene by the NGS, we were able to increase the knowledge of the mutations that circulate in the population of Rio de Janeiro. In addition, the NGS also enabled the increase of the panel of mutations investigated by our laboratory, expanding the detection rate to 69% in this specific population.

**Keywords: Cystic Fibrosis; CFTR; NGS**