

INTERNATIONAL STUDENT CONGRESS



ABSTRACT
BOOK
2023

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**Cell/ Genetics
Oral Session**

Validation of a bioinformatic procedure with the analysis of TP53 gene mutations

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Introduction: Considering that genetic analysis has a fast-growing significance in the diagnosis of various malignant tumours, bioinformatic applications become more and more inevitable, thus their capability of processing genetic information. TP53 gene mutations are one of the most common genetic changes in colorectal cancer, but also in other types of malignant diseases, representing an excellent subject for bioinformatic analysis.

Aim: The aim of our research is to validate the bioinformatic application we created, by analysing 25 samples of TP53 genes from patients diagnosed with malignant colorectal cancer.

Material and Methods: In our work, we compared the sequences of 25 healthy TP53 genes with the structure of 25 genes sequenced from the sample of patients diagnosed with colorectal cancer, and then we determined the most common mutation hotspots in the examined samples.

Results: Using our program to compare the sequences, we found out that the most frequent mutations were in exon 8, presenting differences in eight samples. These mutations were mostly between the 815th and 860th positions. The second exon, containing the most mutations was exon 4 with five samples presenting alterations mostly between the 250th and 275th positions.

Discussion: Based on our results, these type of comparison-based bioinformatic applications provide a perfect way for analysing genetic information and identifying mutations in the structure of such significant genes, as the TP53. Considering the mutations in both healthy and malignant samples, the position of these can conclude to a more precise analysis, helping the genetic diagnosis of these malignant diseases.