

**Evaluation of somatic point mutations associated to splicing mechanisms in colorectal cancer**

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Colorectal cancer (CRC) is a multifactorial disease and the third more common cancer worldwide. Cancer development involves numerous abnormal regulations, such as gene expression, mRNA processing and somatic point mutations. To improve the comprehension of these factors, we evaluated the correlation of differential splicing with differentially expressed genes (DEG), differentially exon usage (DEU) and somatic point mutations through interatomic network analysis. For this purpose, the RNA-seq dataset (GSE50760) of 12 paired tumor and adjacent normal tissue samples was employed. After reads alignment on the reference genome, the data was submitted to DEG analysis supported with gene and transcripts quantification and DEU analysis assisted with exonic quantification. Additionally, samples were analyzed for somatic point mutations detection. The results were integrated in an interatomic network for the proceeding analysis. Furthermore, the variants effects prediction was estimated to understand their impact in the splicing mechanisms of CRC. According to the obtained results, it was possible to elaborate an interatomic network composed by DEG, which was assigned to gene ontology, hub-bottlenecks and clustering analysis. These analyzes allowed the detection of DEG that are highly representative for CRC development and maintenance. The DEG was mainly involved in processes such as cell cycle regulation, extracellular matrix organization and immune response. The data also permitted the identification of DEG with specific somatic point mutations in essential regions for splicing mechanism. Hence, the methods employed in this work promoted a better and innovative understand of cancer development.

**Biography**

Joice de Faria Poloni is currently a PhD candidate in the Cellular and Molecular Biology Post-graduation program of the Federal University of Rio Grande do Sul, Brazil. Her area of expertise is in the bioinformatics field with emphasis on systems biology, systems chemo-biology and transcriptomic analysis to study tumoral processes and developmental mechanisms. She also works with somatic point mutations and alternative splicing isoforms in different tissues. In this sense, her scientific production involves book chapters, research and review articles in these fields.

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