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Title:	Understanding the Impact of Non-coding Mutations in Colorectal Cancer
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Abstract:	<p>The studies characterizing the genomic landscape of cancers have been majorly focused on the identification of driver mutations within the protein-coding gene regions. However, the non-coding region occupies a significantly larger proportion of the genome, and functional mutations have been reported in the regulatory regions (of non-coding regions) which can affect signaling pathways implicated in cancer. Cis-regulatory elements (CREs) are an enriched subset of the non-coding DNA and can regulate the gene expression of neighboring genes. CREs can be highly tissue-specific and hence, it becomes important to study tissue type-specific gene regulation. In this study, we used capture Hi-C data for 19,023 promoter fragments in the Colorectal cancer cell line (HT-29) from Orlando et al. and integrated it with the whole genome somatic mutation and gene expression data from PCAWG. We used the SMuRF tool to identify significantly mutated ($qvalue < 0.05$) CREs. We identified five genes, ALCAM, PRKCH, TSC22D1, NFIB and FGFR2 with significant differential expression ($p-value < 0.05$ and absolute fold change ≥ 1) in the mutated group (samples having mutations in the CRE interacting with the gene) versus the non-mutated group. Out of these five genes, we focused our analysis on FGFR2 which is a well-known cancer-driver gene, but the impact of non-coding mutations on this gene in colorectal cancer has not been reported before. We identified multiple TFBS and histone modifications in the FGFR2 CRE. We thus report a non-coding CRE interacting with the FGFR2 gene as a potential non-coding cancer driver in colorectal cancer.</p>
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