

Functional Variants Detection by RNAseq

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Abstract : RNAseq represents an attractive methodology for the detection of functional genomic variants. RNAseq results obtained from polyA+ RNA selection protocol (POLYA) and from exonic regions capturing protocol (ACCESS) indicate that ACCESS detects 10% more coding SNV/INDELS with respect to POLYA. ACCESS requires less reads for coding SNV detection with respect to POLYA. However, if the analysis aims at identifying SNV/INDELS also in the 5' and 3' UTRs, POLYA is definitively the preferred method. No particular advantage comes from ACCESS or POLYA in the detection of fusion transcripts.

Keywords : fusion transcripts, INDEL, RNA-seq, WES, SNV

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