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Analyzing cancer genomics using watson for genomics with structural variants

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Statement of the Problem: As sequencing cost declines, more cancer patients have their tumor samples sequenced to seek for optimum treatments. However, analyzing genomic data requires considerable expertise and efforts. Besides, reports should be generated in clinically relevant time frame, without errors, with objectivity, and in comprehensive manner. It is difficult to meet those criteria in ever increasing flood of new publications, clinical trial information on investigational drugs, and more high-resolution data. As an example of high resolution data, structural variant data is available. However, so far, fusion proteins such as BCR-ABL, EML4-ALK receives attentions; other types of structural variants such as simple disrupts, exon skippings, intron retentions, and internal tandem duplications are underestimated.

Methodology & Theoretical Orientation: Watson for Genomics (WfG) takes variants, copy number alterations, and gene expressions as inputs to generate a report in automated manner. WfG performs molecular profile to identify driver mutations and drug response biomarkers followed by drug analysis including pathway analysis. Structural variants module is being developed to accommodate structural variant data.

Findings: WfG generates reports with high recall rates in drug recommendation compared with human experts using whole genome samples from collaborators such as New York Genome Center and British Columbia Cancer Agency. Structural variants such as EGFR vIII, MET exon skipping event, and tumor suppressor gene disruptions are successfully captured along with de novo events. WfG provides cancer communities with up to date knowledge to benefit their patients.

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