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The structure of state-of-art gene fusion-finder algorithms

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Fusion genes are hybrid genes that combine parts of two or more original genes. They can be generated as a result of chromosomal rearrangements or abnormal transcription, and have been shown to act as drivers of malignant transformation and progression in many human cancers. The biological significance of fusion genes together with their specificity to cancer cells has made them into excellent targets for molecular therapy. Fusion genes are also used as diagnostic and prognostic markers to confirm cancer diagnosis and monitor response to molecular therapies. High-throughput sequencing has enabled the systematic discovery of fusion genes in a wide variety of cancer types. A significant number of bioinformatics algorithms have been developed to detect fusion genes. Detection strategies are quite variegated. Many algorithms have been proposed, and each of them has specific biases at the level of sensitivity or specificity. In this presentation,the author will focus on the limits and strength of the available tools and highlight RNAseq characteristics that significantly affect fusion detection performance, e.g. stranded libraries, minimal coverage threshold, etc.

Biography

R A Calogero joined the The Bioinformatics and Genomics Unit (B & Gu) group in 1999, when he moved from the Naples University Federico II, Italy. The Bioinformatics and Genomics Unit (B &Gu) is an interdisciplinary group devoted to the study of multifactorial diseases by mean of high throughput technologies - ie microarray, Next Generation Sequencing - and bioinformatics. B & Gu is part of the Tumor Immunology Group which has long been engaged in research on tumor immunogenicity. His background in molecular biology and bioinformatics and his consolidated experience in microarray data analysis and database mining have served to switch the laboratory's classical immunological approach to a genome-wide vision.

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