Le réseau régional d'ingénieurs en bioinformatique de Lille et le PPF bioinformatique vous convient à une conférence
Mercredi 22 Mai 2012, à 14h, Amphithéâtre de l'Institut de Biologie de Lille, 1, rue du Pr Calmette, LILLE.

SVDetect : a tool to identify genomic structural variations from paired-end and mate-pair sequencing data

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The detection of structural variants in the human genome plays an important role in the understanding of many genetic diseases, including cancer.
Recently, with the arrival of new high-throughput sequencing technologies, along with the application of paired-end sequencing strategies, our current power to detect structural variants has significantly improved. New computational methods are needed that can reliably identify such genomic rearrangements. I will present a new method for structural variant identification and prediction of variation type from paired-end mapping data provided by current short read aligners, and implemented into a bioinformatic tool called SVDetect. The central idea is to use paired-end reads where were anomalously mapped onto a reference genome sequence to detect signatures and call the underlying variants. Clusters of such paired-ends sharing a similar genomic location improve the confidence in the detection of variants. We use then all the characteristics of paired-ends (strand orientation, order and clone insert size) as parameters of decision to identify a particular signature and to call a specific structural variant type. By this way, we are able to discriminate various types of structural variants such as insertions/deletions, small or large duplications, inversions, or balanced/unbalanced inter-chromosomal translocations. SVDetect is compatible with any NGS technology handly paired-end/mate-pair sequencing, and directly create output files for convenient graphical view of predicted structural variants.

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