



Workshop on Modeling of Genetic Regulatory and Metabolic Networks

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Abstract

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Precise detection of rearrangement breakpoints in mammalian chromosomes

Genomes undergo large scale evolutionary events called rearrangements.

In order to better understand the evolutionary mechanisms involved, it is necessary to precisely identify the regions on the genome where the rearrangements took place in the course of evolution. By comparing the order and orientation of orthologous markers on several genomes of related species, one can identify regions of conserved synteny, also called synteny blocks, and in between those, the breakpoints regions.

We present a new method to precisely localise such regions on a genome by comparison with another. We are given a set of synteny blocks, and the aim is to further extend the synteny blocks and thus refine the breakpoint regions. The method focuses on each breakpoint region bordered by two synteny blocks in the genome of reference. The breakpoint sequence is aligned against the flanking sequences of the orthologous blocks on the second genome. Based on the resulting alignments, the sequence is then partitioned, using a segmentation algorithm, in order to identify the narrowed breakpoint. The method was applied to identify breakpoints regions on the human genome compared with the mouse genome. We obtained 168 breakpoints that are less than 50 Kb in size, out of a total of 355 breakpoints. By comparing them with some publicly available ones, we show that we achieve a better resolution. Finally, we show that breakpoint sequences can be distinguished from the sequences around in terms of segmental duplications, similarity with related species, and transposable elements.