



#### Genome Rearrangements Joao Meidanis

Campinas, Brazil 2015



#### 1. Summary

- 2. Genome comparison
- 3. Rearrangement events
- 4. Example: mouse vs. human (X-chromosome)
- 5. Rearrangement distance
- 6. Known results
- 7. Current research lines



#### SUMMARY

- One of the big challenges of contemporary Biology is to measure evolution
- Besides point mutations, evolution is known to occur by means of movements of large chunks of DNA (genome rearrangements)
- The advent of entire genomes brings a whole new facet to this issue
- As a first estimate of the amount of evolution between two species, one can use the formula

number of events

unit of time

 Our research focus on efficient ways of computing the number of rearrangement events between two or more genomes



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## GENOME COMPARISON Point mutations

...TATCGATAGACCACTG...

... TATC--TAGACGACTA...



## GENOME COMPARISON Genome rearragements



Movement of large segments within the genome. Above, segment E - F - G flips over



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## **REARRANGEMENT EVENTS**

- Insertion / Deletion
- Reversal
- Transposition
- Fission / Fusion
- Block Interchange
- Others: duplication, genome doubling



## **INSERTION / DELETION** Gene gain / loss between genomes





## **REVERSAL** A segment is reversed between genomes





# TRANSPOSITION

A segment moves to a new position (or: exchange of two adjacent segments)





## FISSION / FUSION Genome breaks in two / Two genomes join





## **BLOCK INTERCHANGE** Exchange of two nonadjacent segments





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## EXAMPLE: HUMAN AND MOUSE X-CHROMOSOME Pavel Pevzner et al. Genome Res. 2003; 13: 37-45



Figure 2. X-chromosome: from local similarities, to synteny blocks, to breakpoint graph, to rearrangement scenario

#### Thanks to:



Cold Spring Harbor Laboratory Press



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## **REARRANGEMENT DISTANCE** Maximum parsimony approach

Given two genomes, and a set of events, the rearrangement distance between the genomes is the length of the shortest series of events that transforms one genome into the other.

In the previous example: Genome 1: mouse X-chromosome Genome 2: human X-chromosome Set of events: reversals only

**Distance: 7 events** 



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## **KNOWN RESULTS**

- Insertion / Deletion distance : efficient algorithm known
- Reversal distance : efficient algorithm known
- Transposition distance : NP-hard; approximative algorithms
- Fission / Fusion distance : efficient algorithm known
- Block Interchange distance : efficient algorithm known



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### **CURRENT RESEARCH LINES**

- Comparions of three or more genomes
- More realistic models with several operations, or basic operations that can form several others, e.g., DCJ or SCJ