



Genome Rearrangements

João Meidanis

São Paulo, Brazil

December, 2004

AGENDA

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

$$\frac{\text{number of events}}{\text{unit of time}}$$

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

AGENDA

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

GENOME COMPARISON

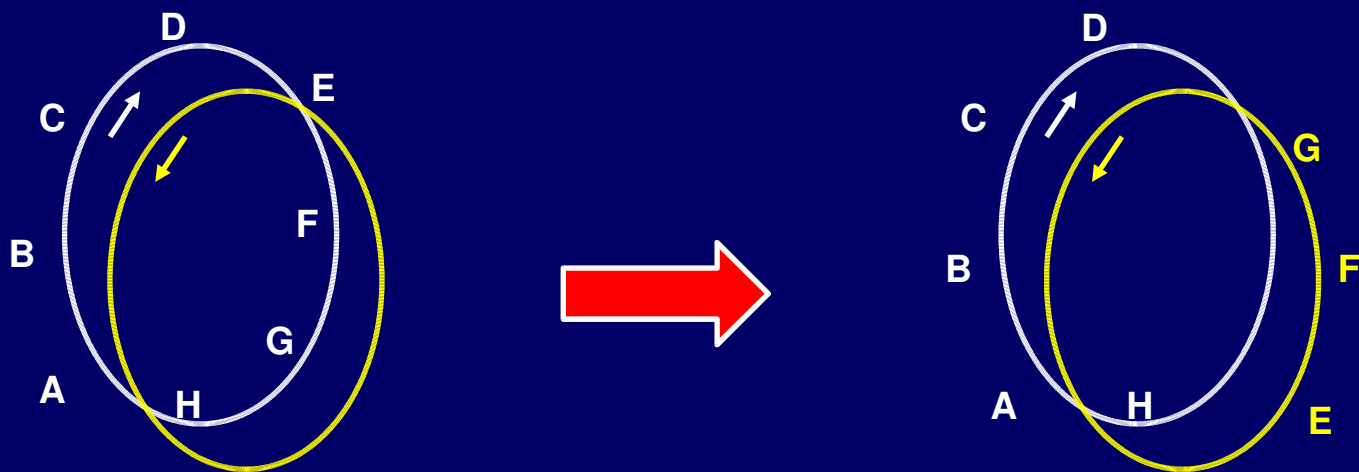
Point mutations

...TATCGATAGACCACTG...

...TATC--TAGACGACTA...

GENOME COMPARISON

Genome rearrangements



Movement of large segments within the genome.

Above, segment E – F – G flips over

AGENDA

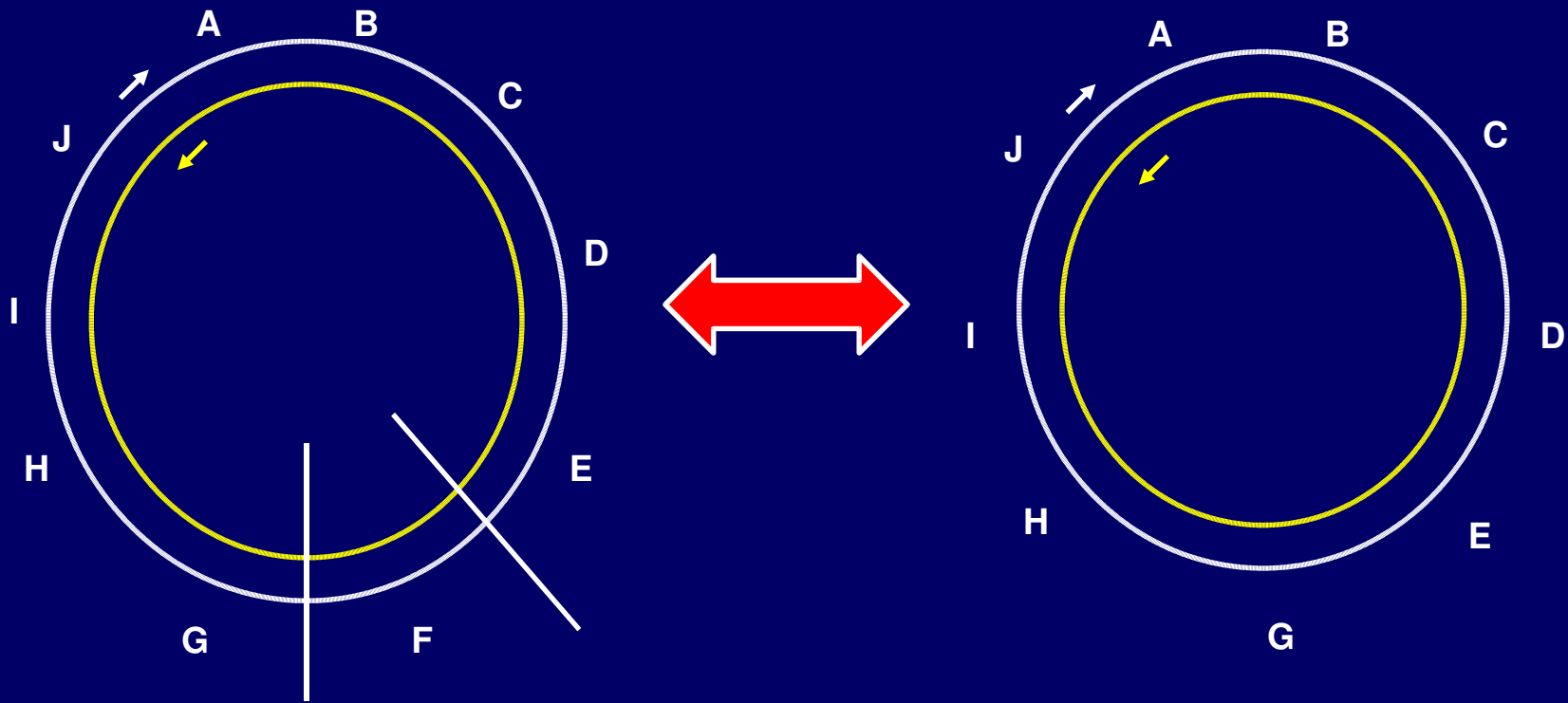
1. Summary
2. Genome comparison
3. Rearrangement events
4. Example: mouse vs. human (X-chromosome)
5. Rearrangement distance
6. Known results
7. Current research lines

REARRANGEMENT EVENTS

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

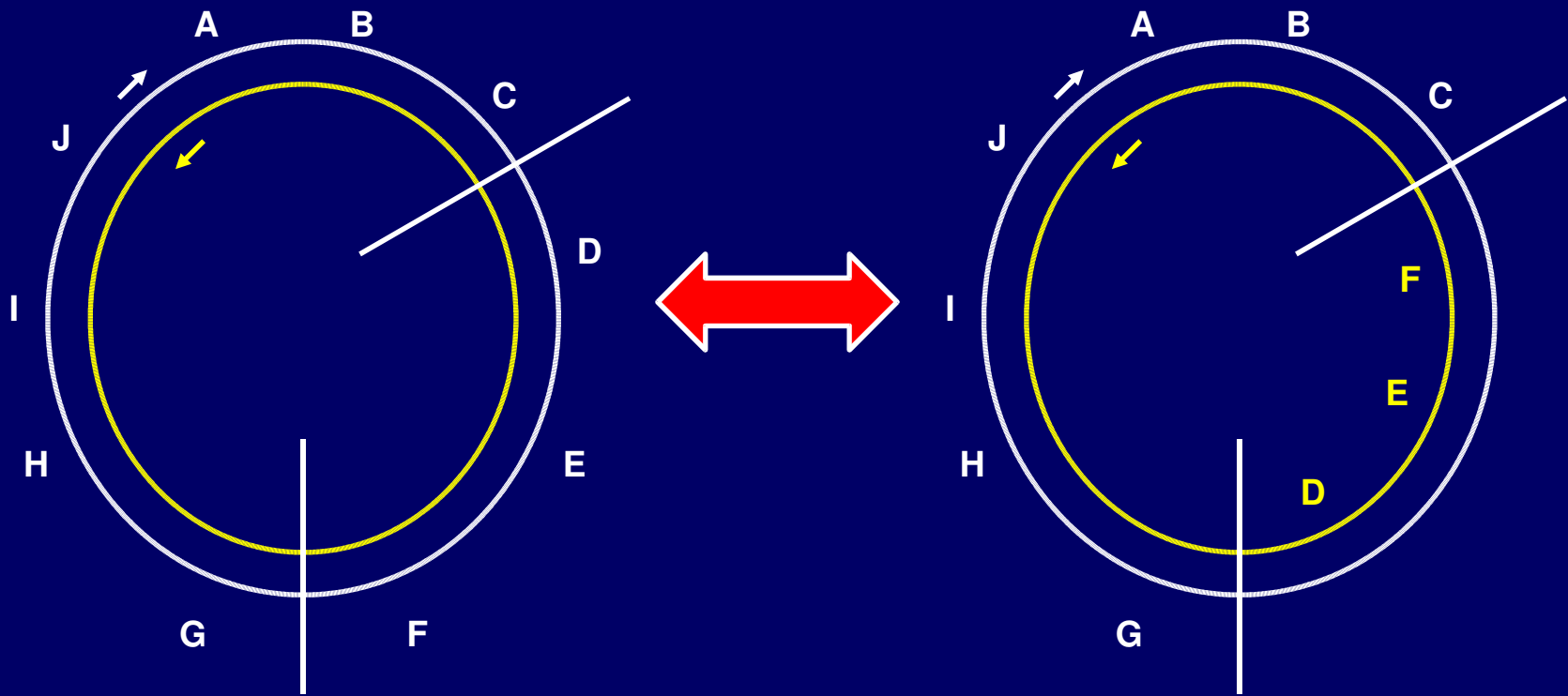
INSERTION / DELETION

Gene gain / loss between genomes



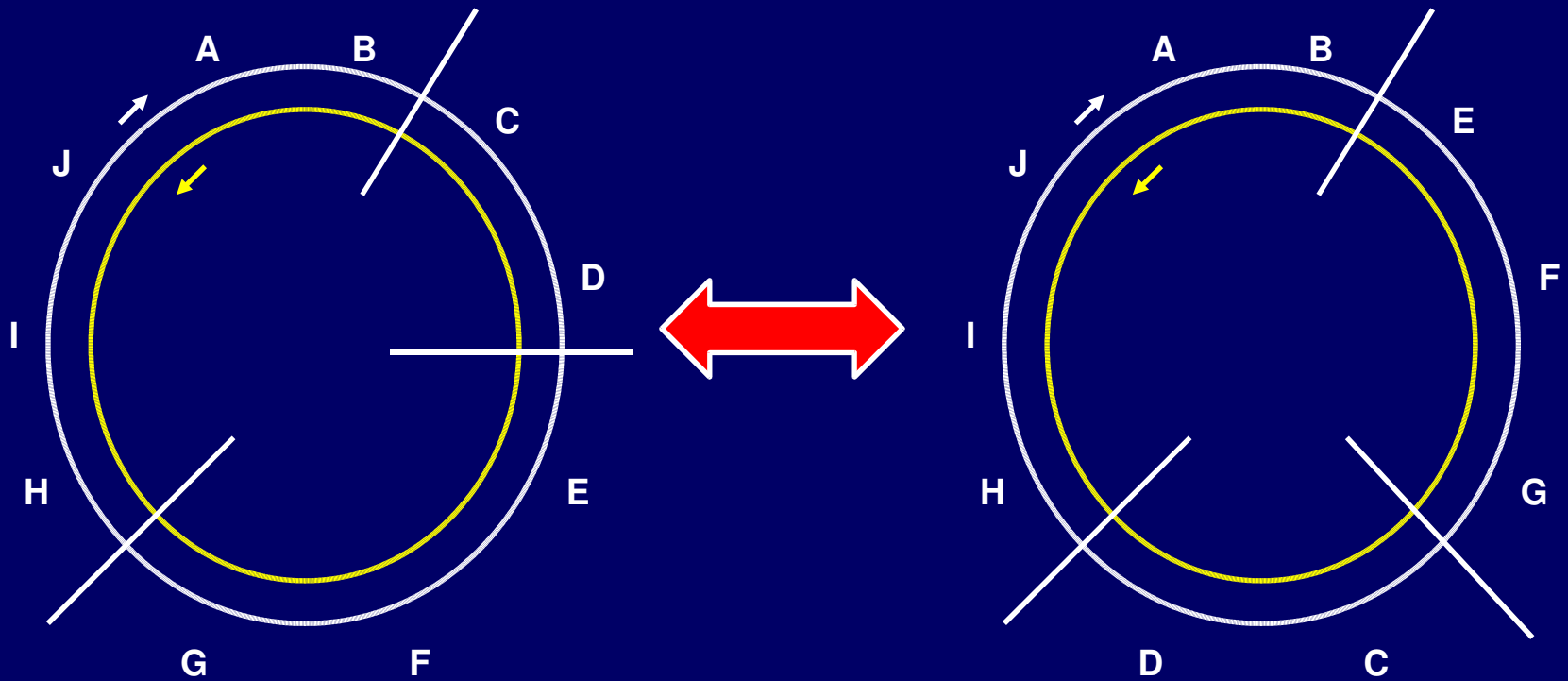
REVERSAL

A segment is reversed between genomes



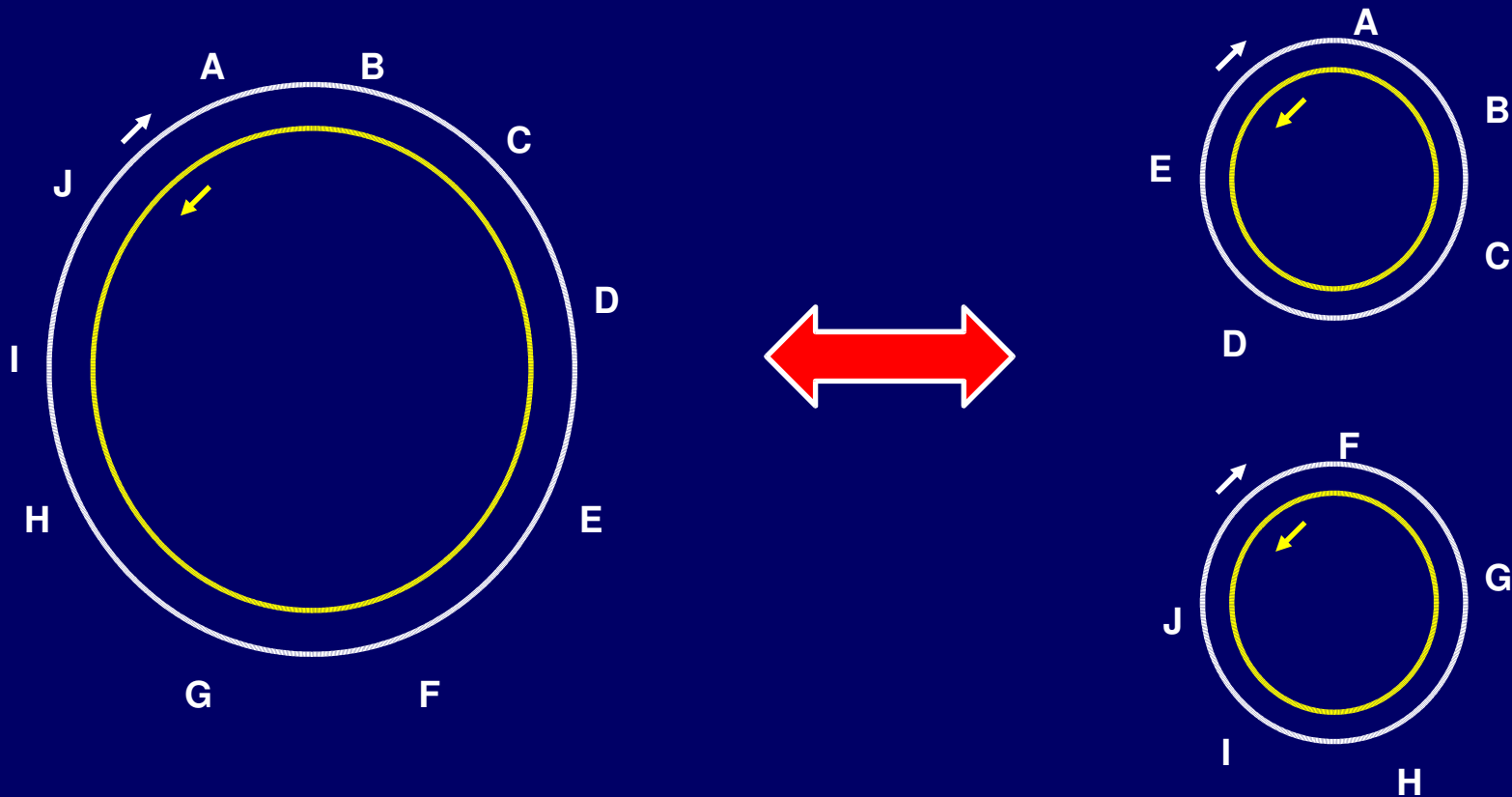
TRANSPOSITION

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



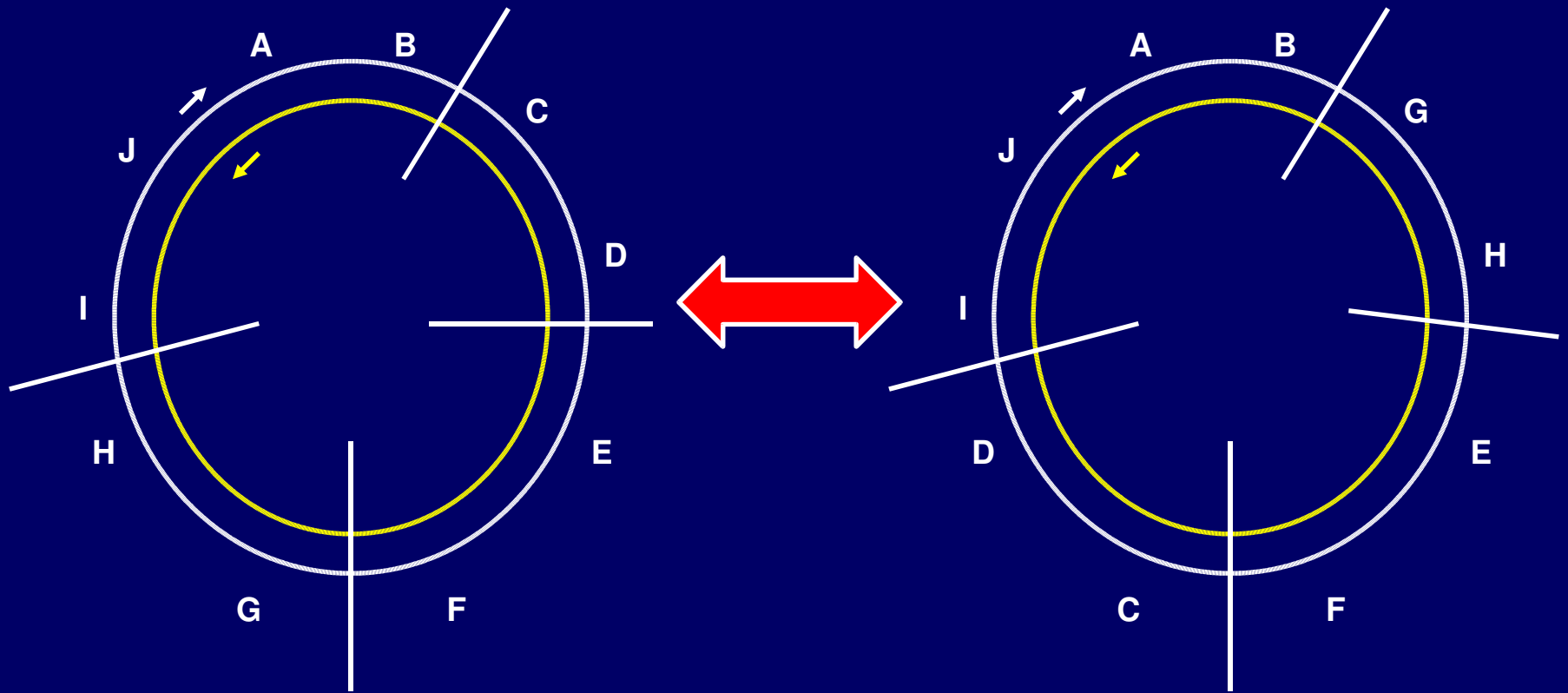
FISSION / FUSION

Genome breaks in two / Two genomes join



BLOCK INTERCHANGE

Exchange of two nonadjacent segments



AGENDA

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

EXAMPLE: HUMAN AND MOUSE X-CHROMOSOME

Pavel Pevzner et al. *Genome Res.* 2003; 13: 37-45

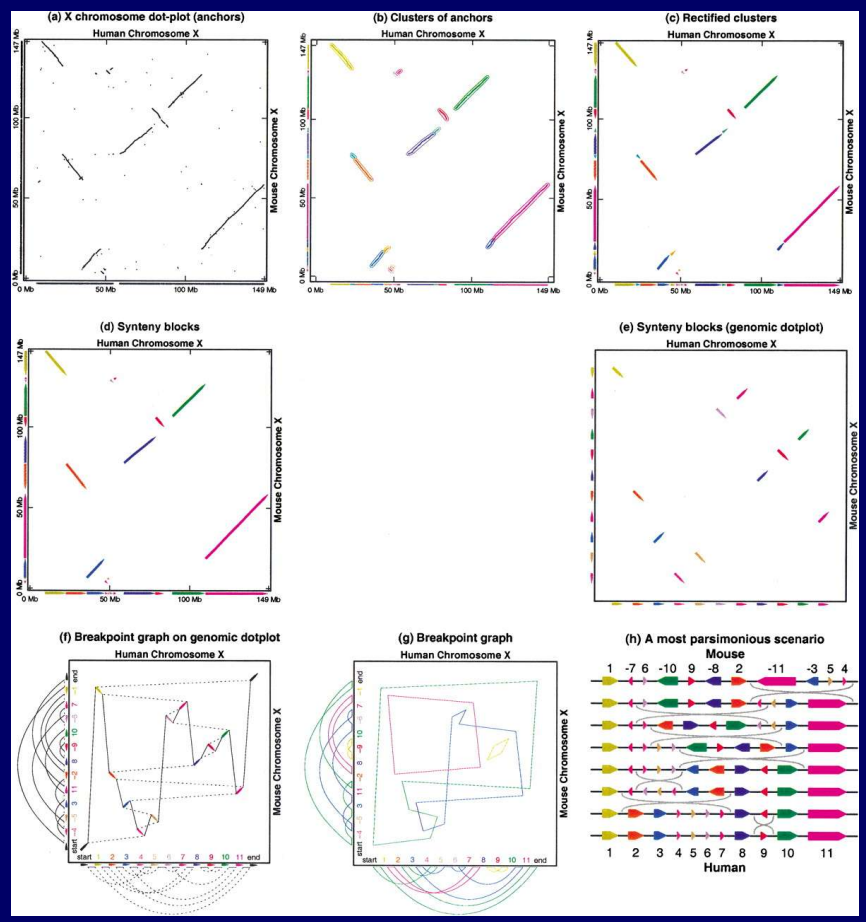


Figure 2. X-chromosome: from local similarities, to synteny blocks, to breakpoint graph, to rearrangement scenario

Thanks to:



AGENDA

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

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

AGENDA

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

KNOWN RESULTS

- **Insertion / Deletion distance: efficient algorithm known**
- **Reversal distance : efficient algorithm known**
- **Transposition distance: no efficient algorithm known; approximative algorithms**
- **Fission / Fusion distance : efficient algorithm known**
- **Block Interchange distance : efficient algorithm known**

AGENDA

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

CURRENT RESEARCH LINES

- **Solve transposition distance problem**
- **Comparisons of three or more genomes**
- **Sets with more than one operation, e.g., reversal and transposition distance**