

# Roadmap

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- Chromosome evolution:
  - Inversion
  - Transposition
  - Fission and fusion
- If today's lecture is perplexing, there is a useful film at <http://www.youtube.com/watch?v=ZcnyMMHLkAw>

## One-minute responses

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- Q: What is the difference between subfunctionalization and specialization?
  - Subfunctionalization: the gene has two functions, and the two new copies split them
  - Specialization: the gene has one function, but the two new copies adapt to do it in different tissues, times, or situations

## **One-minute responses**

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- Q: Are blue receptors and rhodopsins related to R/G and part of a larger gene family?
  - Yes. But those duplications are much older and not tandem.
- Q: How about olfactory receptors?
  - Yes, a HUGE superfamily with some human-specific branches (why?)

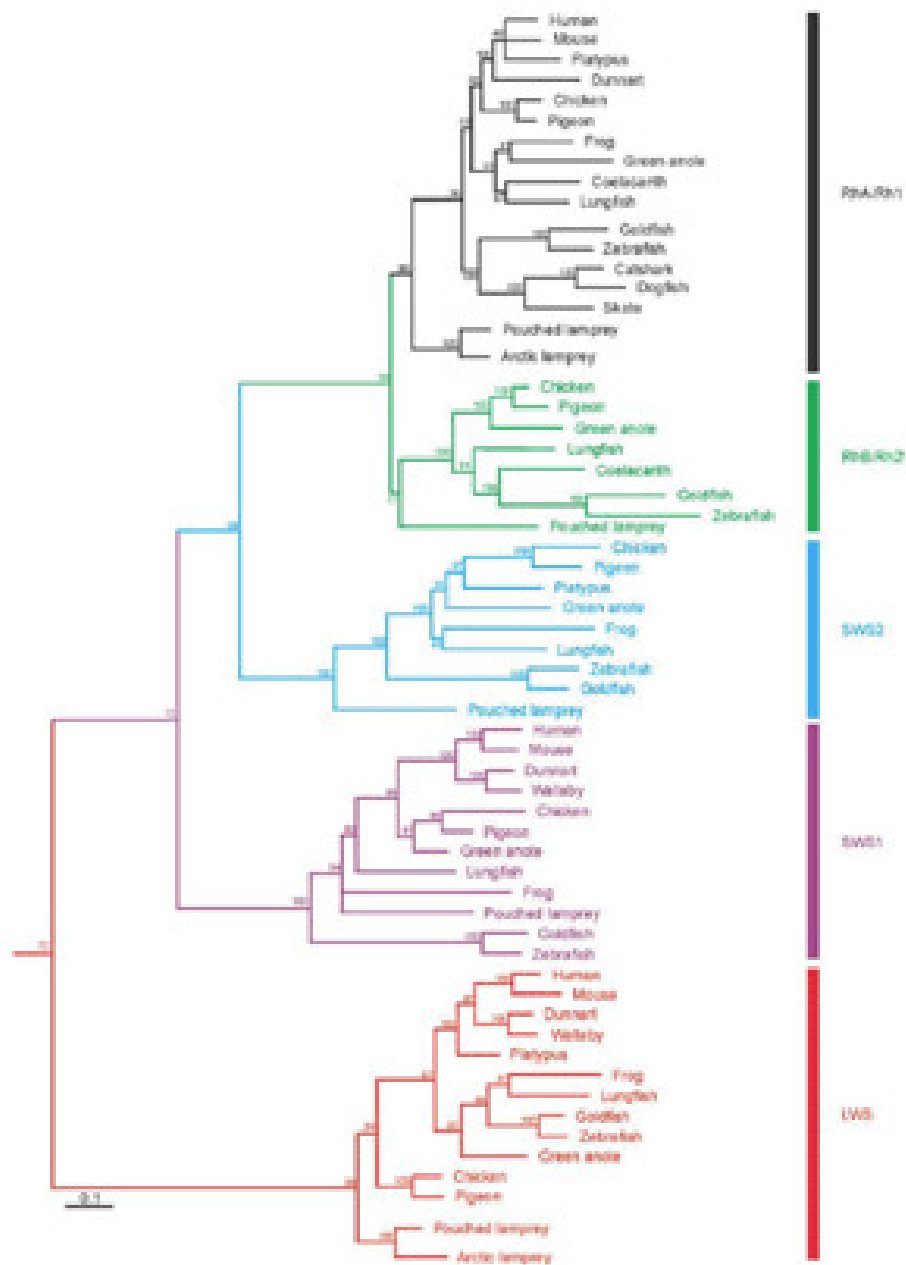


Figure 22. Phylogeny of vertebrate cone and rod opsins

## **An opsin mystery**

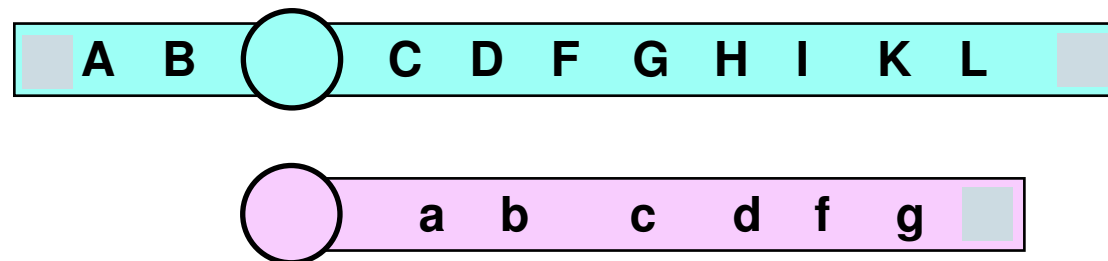
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- Dragonfish live in the deep sea, where most fishes see only blue light
- Dead specimens proved to have members of the red-receptor gene family—why?
- As soon as deep-sea explorers saw a live one, they understood....

# Chromosomes

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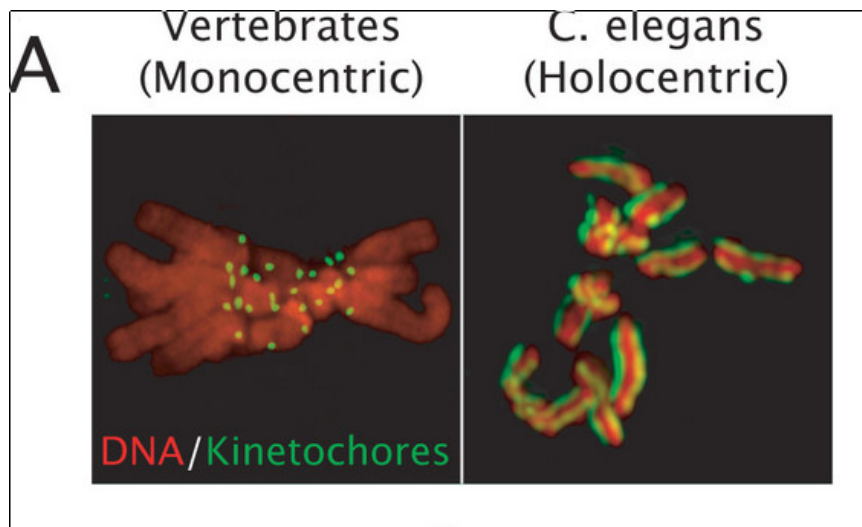
- Bacterial chromosomes are circular
- Most higher organisms have linear chromosomes with a centromere that attaches them to the spindle
- Telomeres at the ends protect the DNA from unravelling



## An exception to the rule

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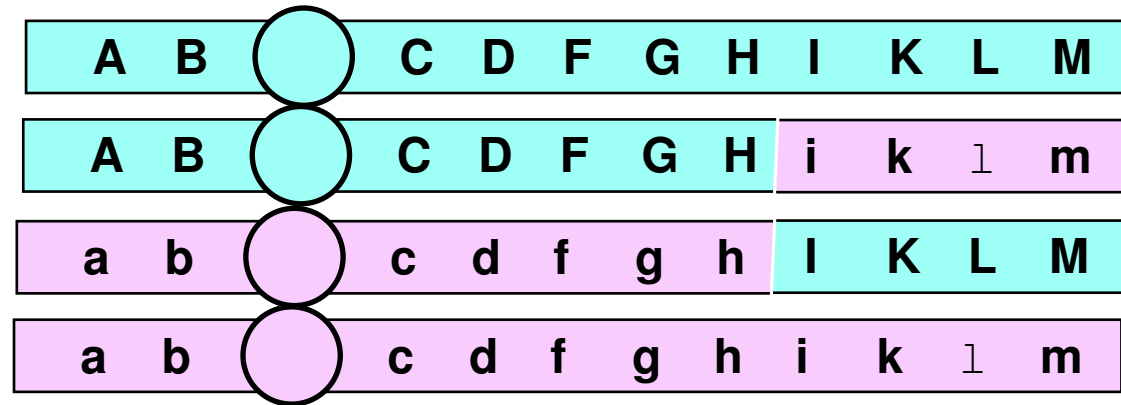
- *C. elegans*, the lab nematode, has holocentric chromosomes
- The normal function of the centromere is diffused across the whole chromosome
- How did this evolve? Why did it evolve?



# Chromosomes

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- Through crossing-over chromosome strands can mix and recombine



- If gene order differs on the homologs this process gets into trouble



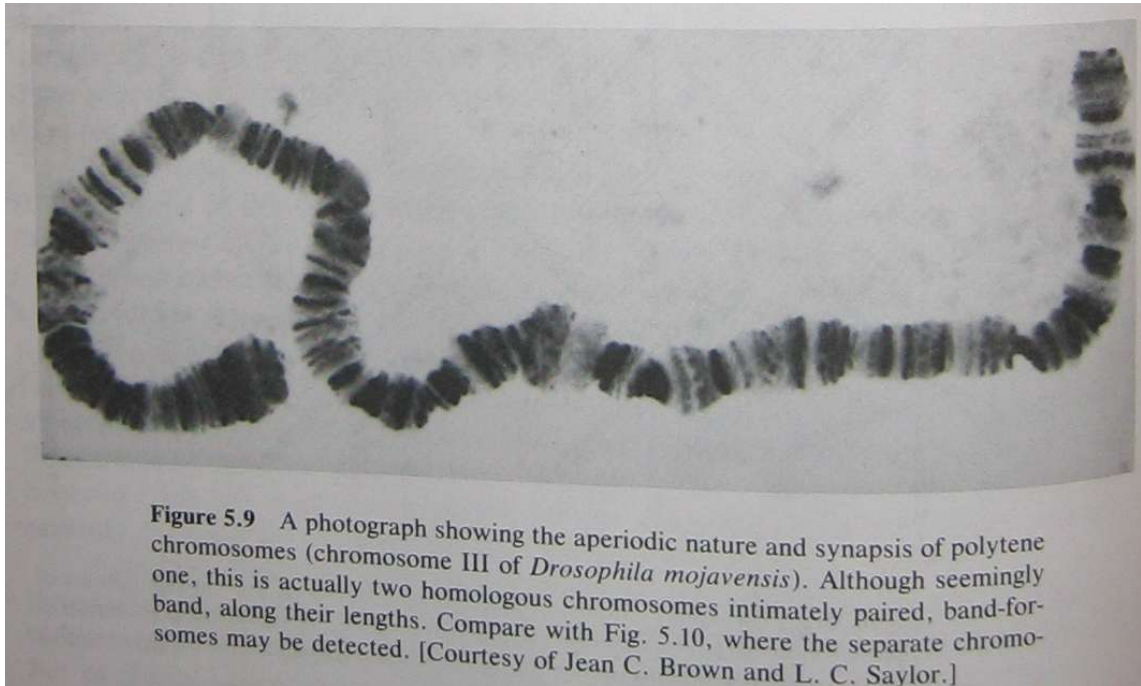
## Chromosome rearrangements

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- Inversion (changing the direction of part of the chromosome)
- Translocation (exchanging material between different chromosomes)
- Transposition (moving material to another location on the same chromosome)

# Polytene chromosomes

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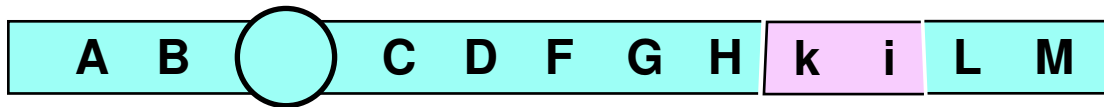


- In flies and mosquitoes, “giant chromosomes” in salivary glands
- Chromosomes pair and then reduplicate themselves up to 1024 copies

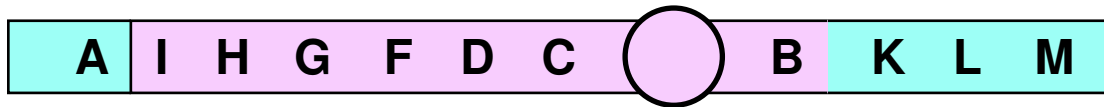
# Inversions

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Paracentric inversion (does not include centromere)



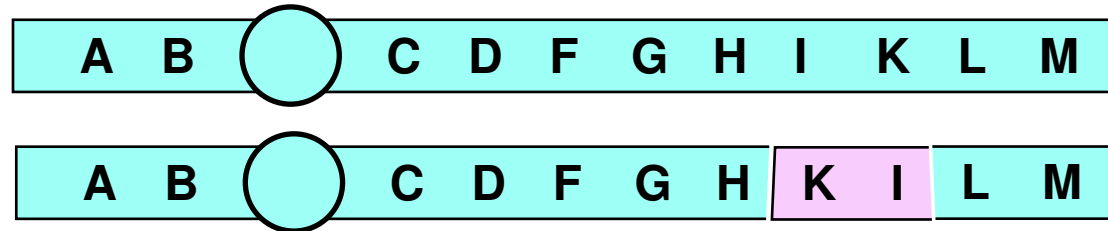
Pericentric inversion (includes centromere)



I prefer not to use these words as they sound too similar to me!

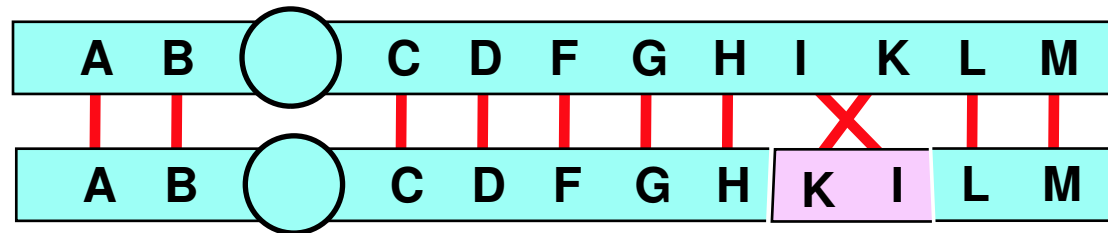
## Paracentric inversion heterozygote

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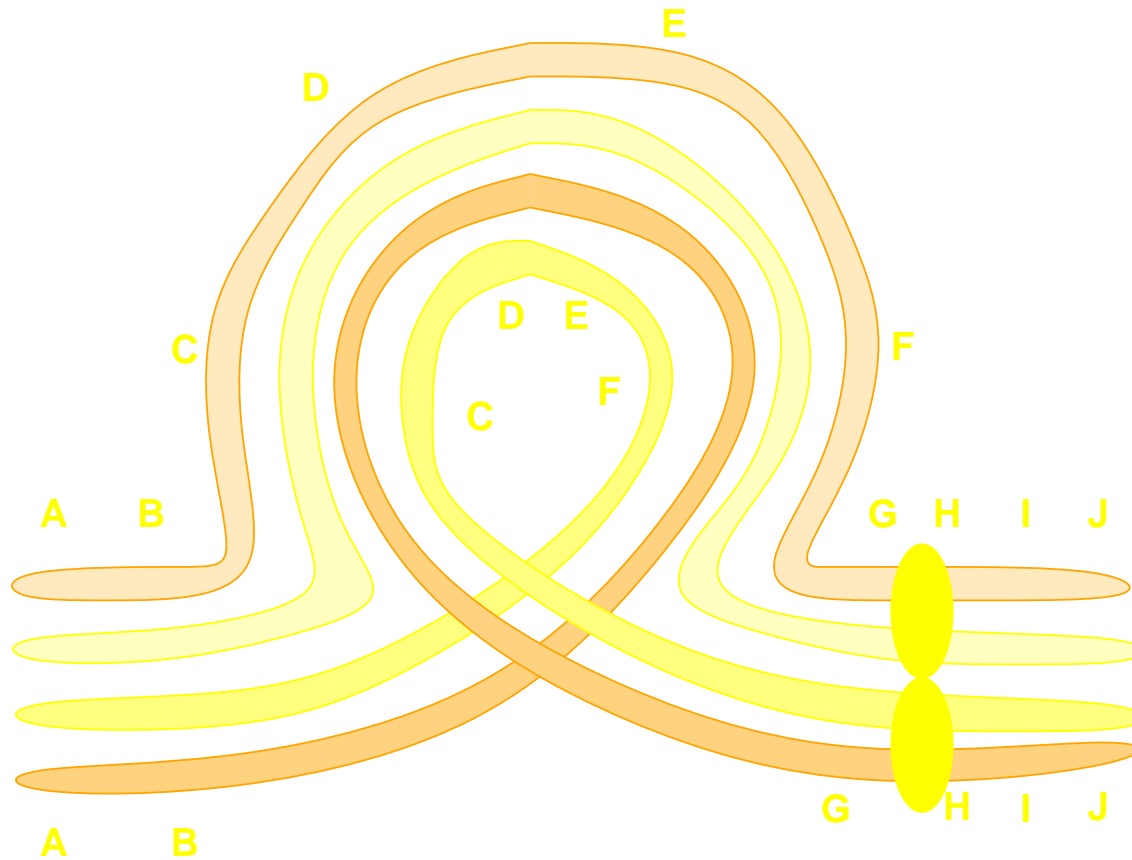


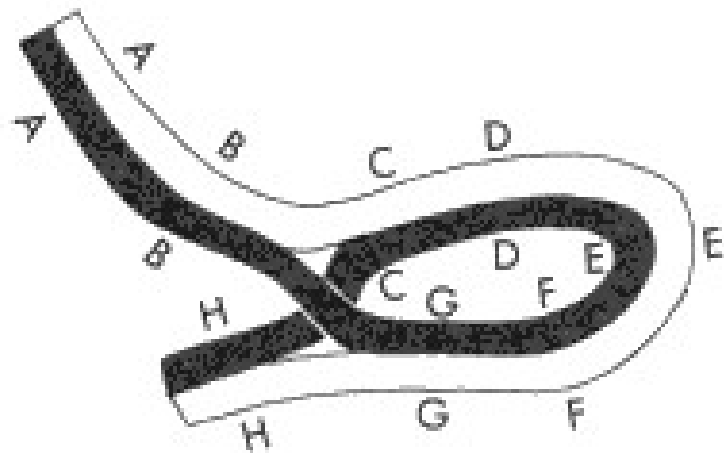
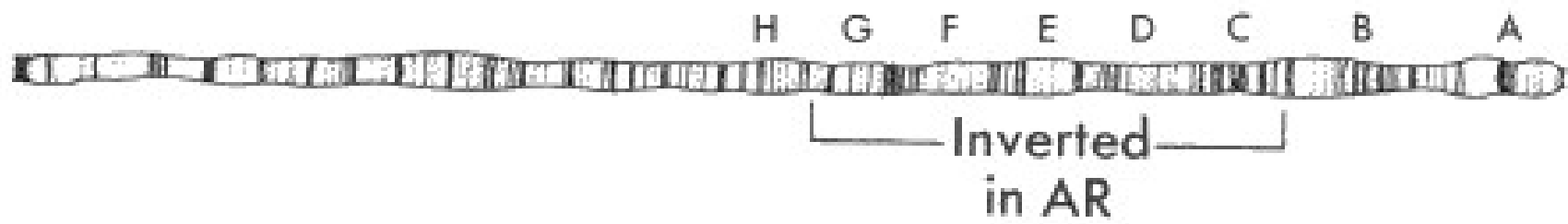
## Paracentric inversion heterozygote

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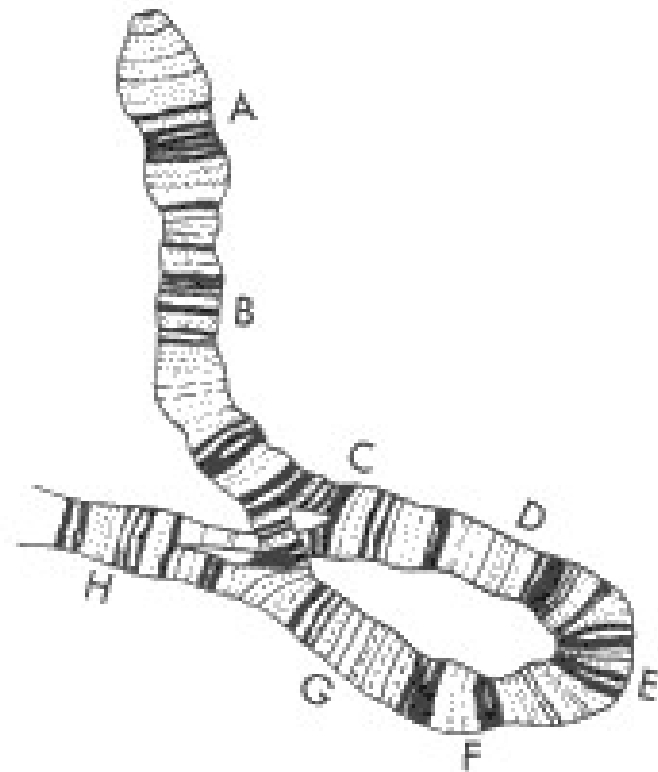


# Pairing in a paracentric inversion heterozygote

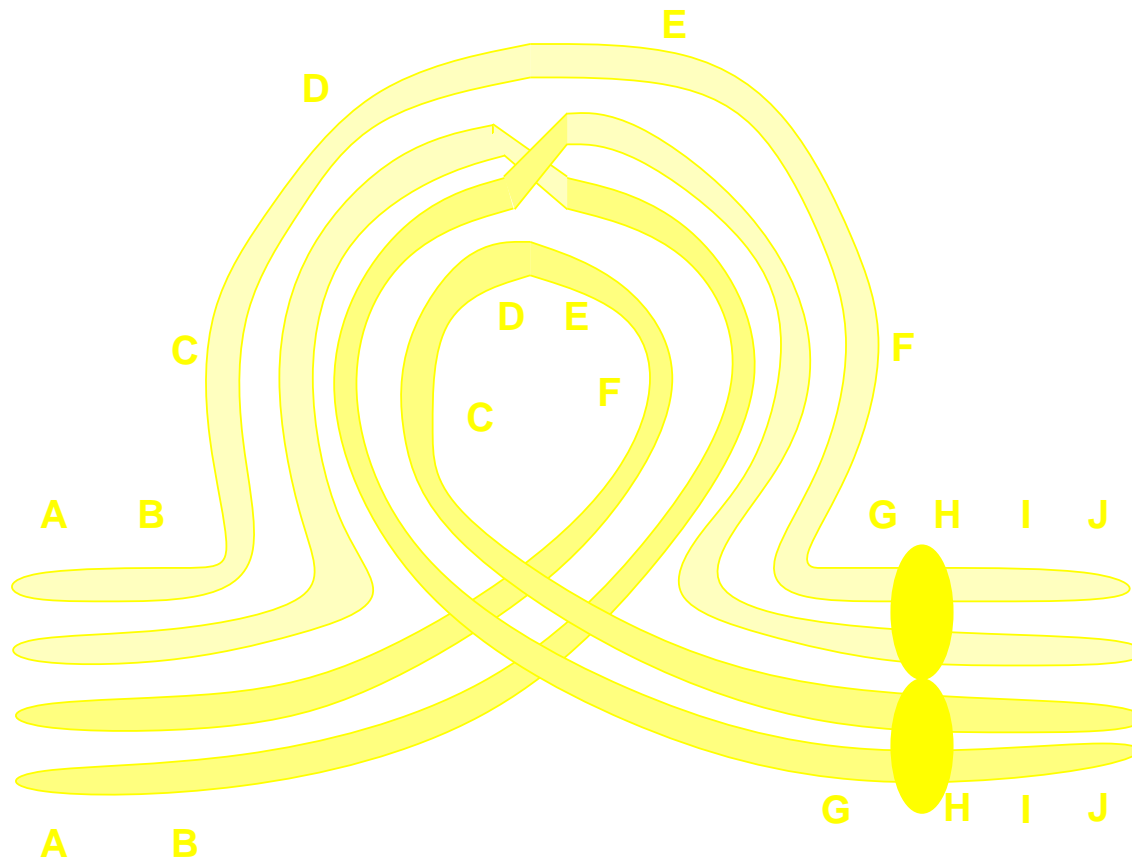




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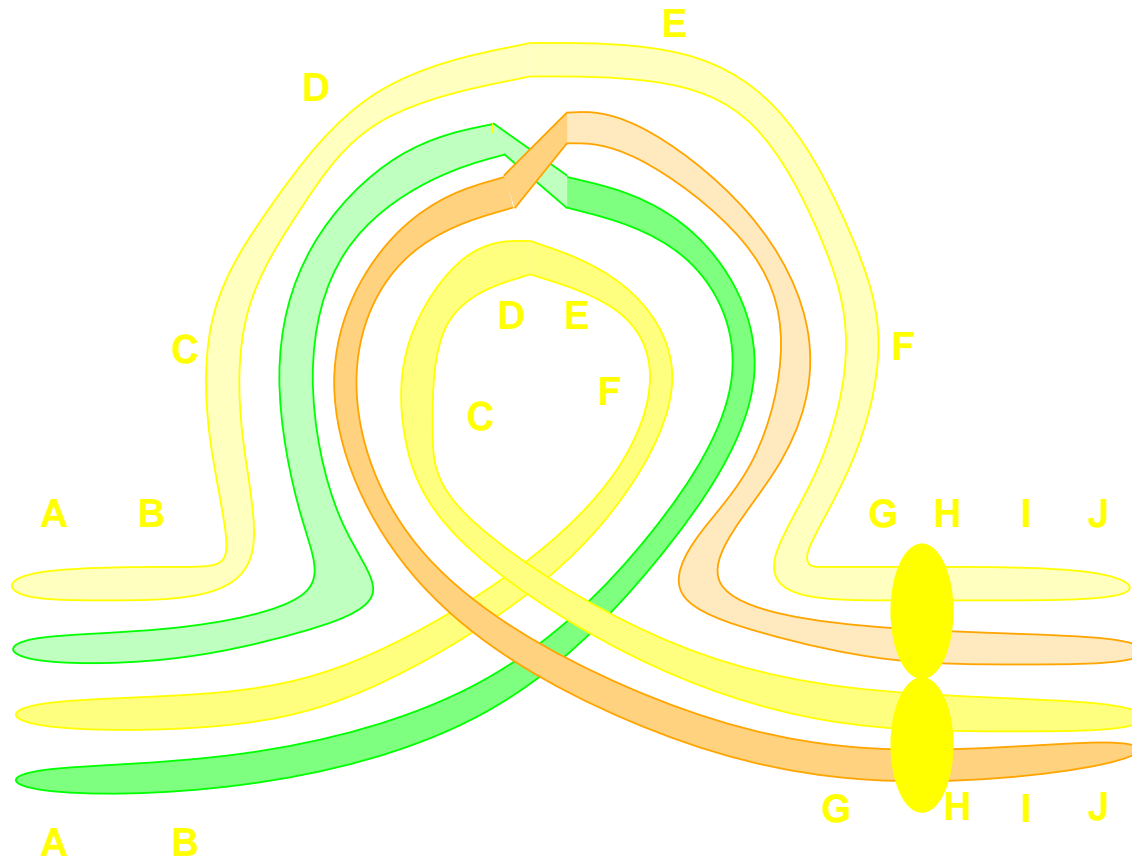


# Crossing-over in a paracentric inversion heterozygote





# Crossing-over in a paracentric inversion heterozygote



## Paracentric inversion heterozygote – Outcome

We produce one of each:

*Normal chromosome*

A-B-C-D-E-F-G-CEN-H-I-J

*Inverted chromosome*

A-B-F-E-D-C-G-CEN-H-I-J

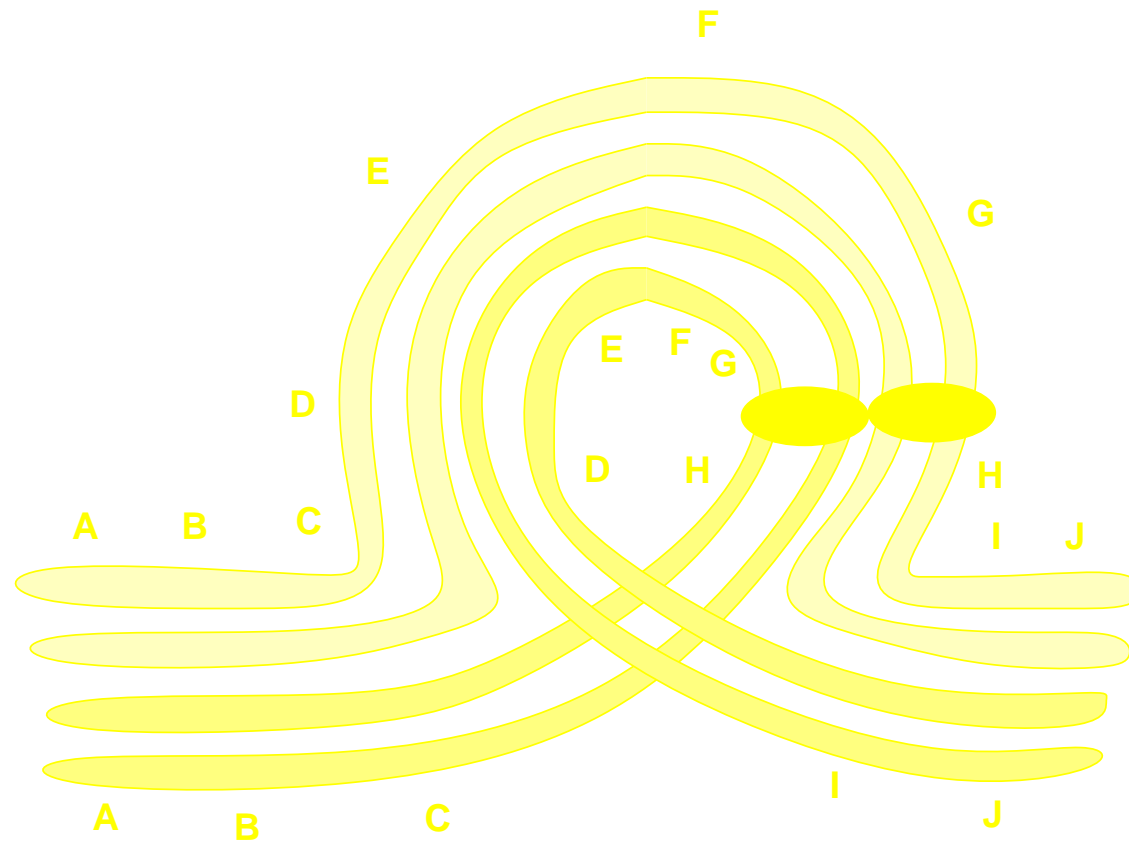
*No centromere*

A-B-C-D-E-F-G-B-A

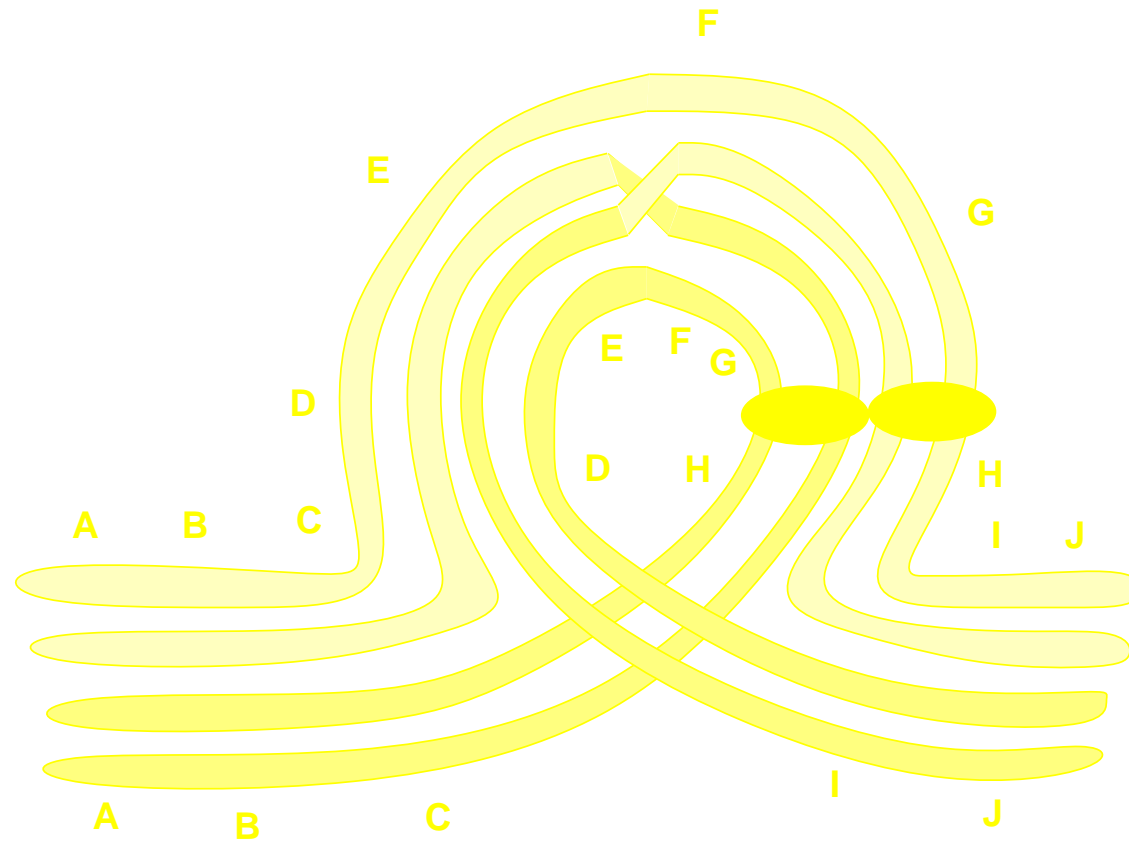
*Two centromeres (breaks randomly)*

J-I-H-CEN-G-C-D-E-F-G-CEN-H-I-J

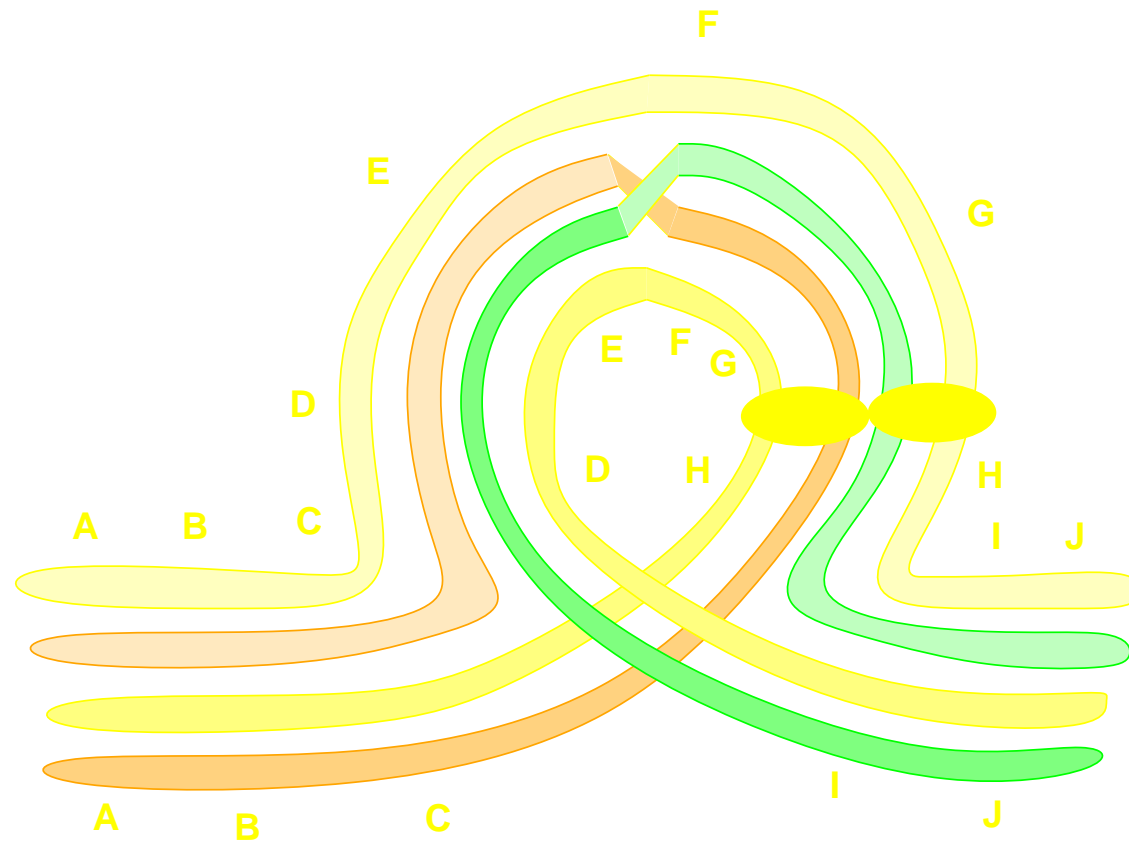
# Pairing in a pericentric inversion heterozygote



# Crossing-over in a pericentric inversion heterozygote



# Crossing-over in a pericentric inversion heterozygote



## Pericentric inversion heterozygote – Outcome

We produce one of each:

*Normal chromosome*

A-B-C-D-E-F-G-CEN-H-I-J

*Inverted chromosome*

A-B-C-H-CEN-G-F-E-D-I-J

*Duplicate ABC, delete IJ*

A-B-C-D-E-F-G-CEN-H-C-B-A

*Duplicate IJ, delete ABC*

J-I-D-E-F-G-CEN-H-I-J

## **Inversion summary**

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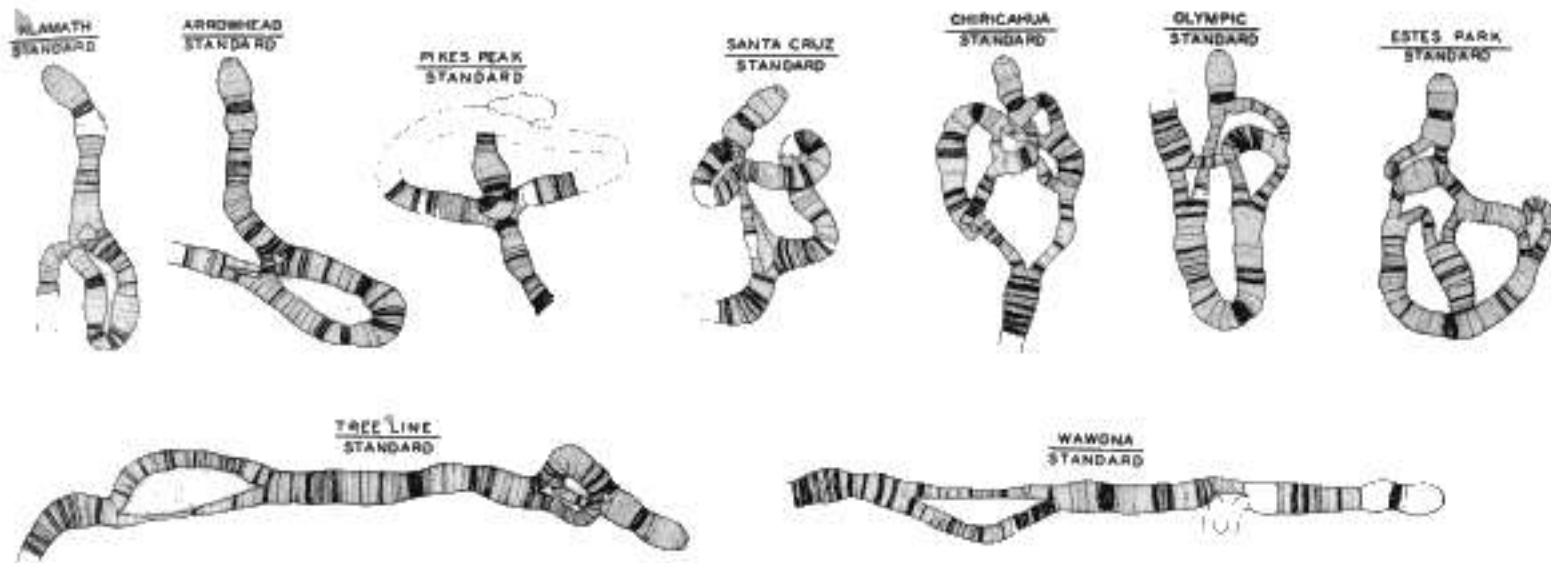
- Paracentric (doesn't include centromere)
  - Recombination produces two-centromere and no-centromere chromosomes
  - Recombinant gametes almost always die
- Pericentric (includes centromere)
  - Recombination produces duplications and deletions
  - Recombinant gametes might be viable if affected area is small
- If there is only one recombination, two of the four chromatids will be okay (but non-recombinant)

## **Evolutionary consequences of inversion**

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- Inversions “suppress recombination” – really they kill recombinants
  - Reduced fertility – bad
  - Preserve favorable groupings of alleles – possibly good
- In most species, fixing an inversion requires drift or strong positive selection
- Some species have genetic systems more permissive of inversions



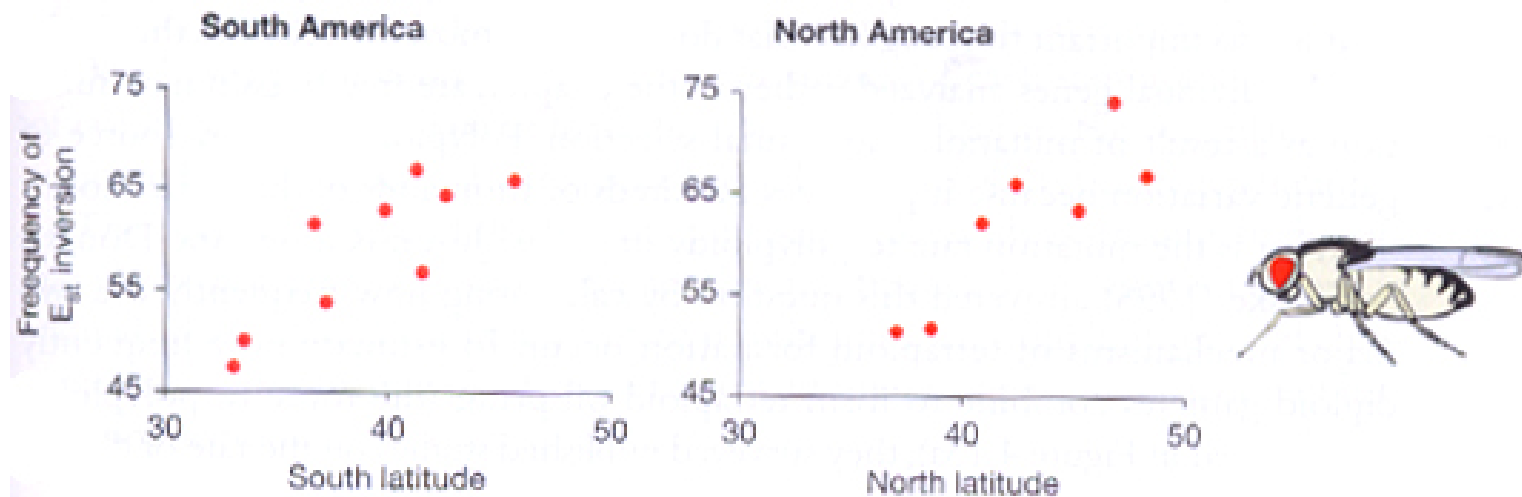


(c) Steven M. Carr

Multiple inversions have happened between different lineages  
of *Drosophila*

## Drosophila inversion clines

- *Drosophila subobscura* shows many inversions in the south relative to the north
- This cline was recreated in Chile and the West Coast of the USA after introduction of flies from Spain.



## **Drosophila inversion clines**

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- Inversions are unusually common in *Drosophila* – why?
  - No recombination in males
  - Female oogenesis preferentially uses a cell with no broken chromosomes
  - Populations often established by a single female – strong genetic drift

## Practice problem

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What, if any, problems arise for:

- A male *Drosophila* inversion heterozygote?
- A female *Drosophila* inversion heterozygote?
- A human inversion heterozygote?
- A human inversion *homozygote*?

Assume no genes were damaged by creation of the inversion

## Practice problem

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What, if any, problems arise for:

- A male *Drosophila* inversion heterozygote? *Should be fine*
- A female *Drosophila* inversion heterozygote? *If she has multiple crossovers in the inversion, she may not make a viable egg, so she will have some fertility reduction*
- A human inversion heterozygote? *Fertility reduction*
- A human inversion homozygote? *Should be fine*

Assume no genes were damaged by creation of the inversion

# Translocation

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# A translocation

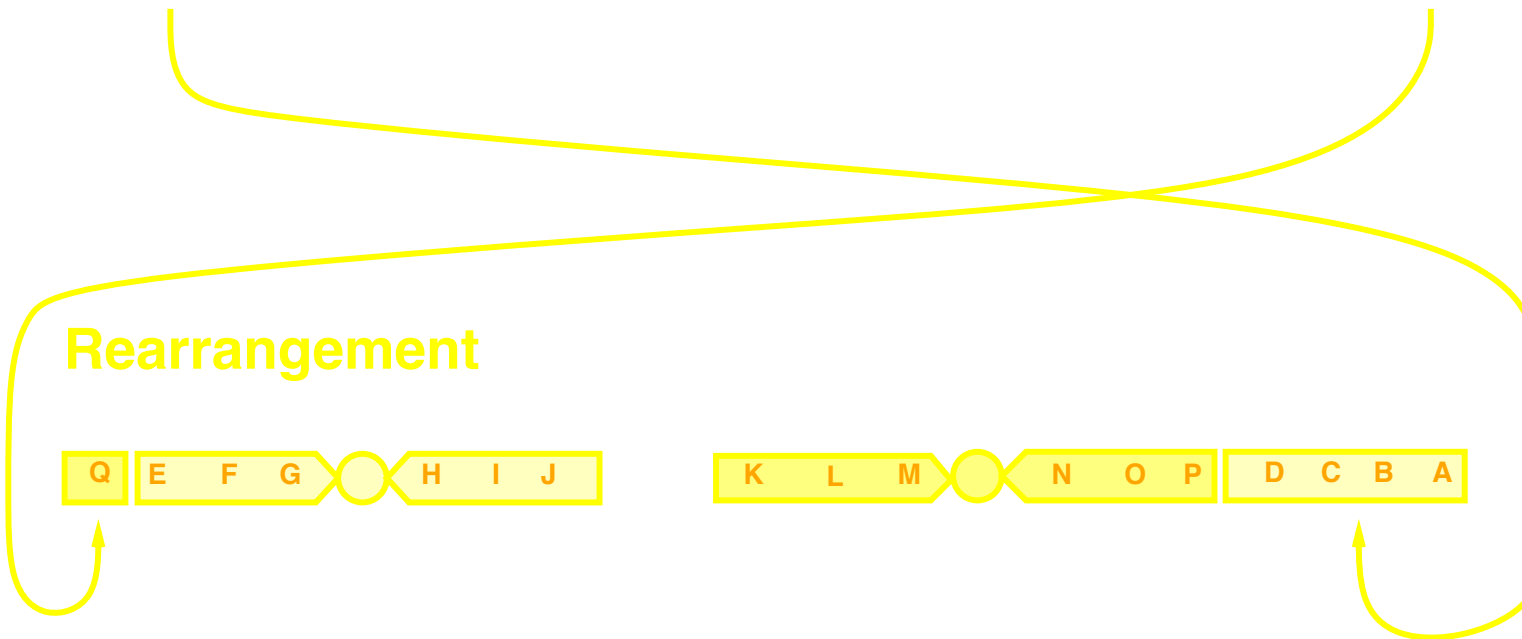
Before



Breaks



Rearrangement

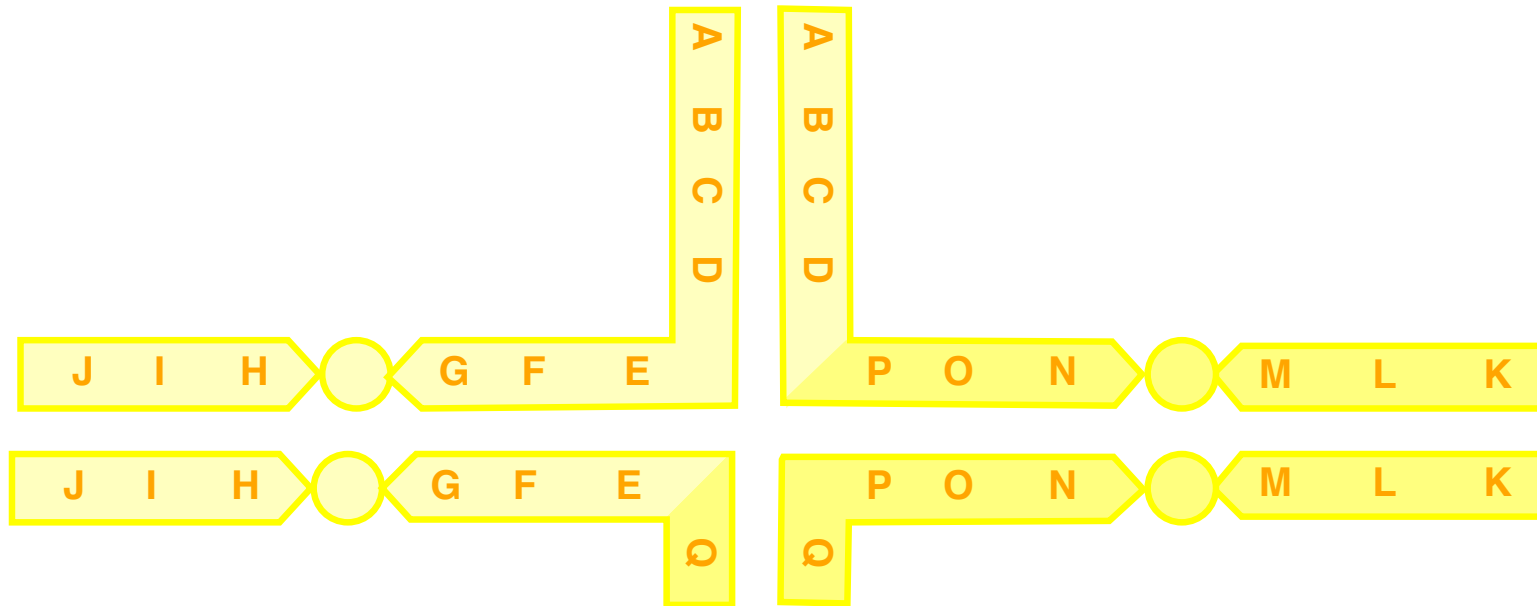


After



# A translocation heterozygote

at first division of meiosis metaphase



A pair of translocated chromosomes  
pairs with a pair of untranslocated chromosomes



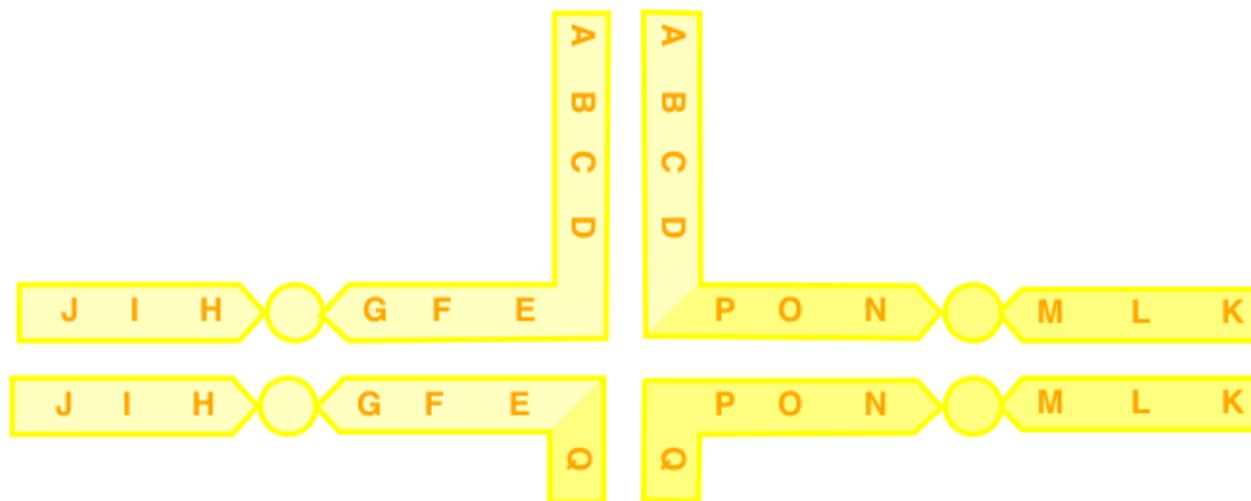
# Translocation

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- Translocation arises from:
  - Breakage and repair
  - Illegitimate recombination between different chromosomes
- Translocation is usually reciprocal because every chromosome end must have a telomere
- *Even without recombination*, translocation heterozygotes mis-segregate their chromosomes in meiosis
- Translocations almost always underdominant
- Only drift or strong selection can allow one to spread

## **Why is translocation a problem?**

- The strange pairing is fine in itself
- Recombination is fine
- Separation of the chromosomes in meiosis can have two outcomes:
  - Two translocated chromosomes go to one daughter and two normal to the other – fine
  - Each daughter gets one translocated and one not – not good
  - This is a 50/50 chance



Possibility 1

A-B-C-D-E-F-G-CEN-H-I-J

K-L-M-CEN-N-O-P-Q

A-B-C-D-P-O-N-CEN-M-L-K

Q-E-F-G-CEN-H-I-J

Possibility 2

A-B-C-D-E-F-G-CEN-H-I-J

Q-E-F-G-CEN-H-I-J

A-B-C-D-P-O-N-CEN-M-L-K

K-L-M-CEN-N-O-P-Q

## Practice problem

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What, if any, problems arise for:

- A male *Drosophila* translocation heterozygote?
- A female *Drosophila* translocation heterozygote?
- A human translocation heterozygote?
- A human translocation *homozygote*?

## Practice problem

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What, if any, problems arise for:

- A male *Drosophila* translocation heterozygote? *Fertility reduction due to mis-segregation*
- A female *Drosophila* translocation heterozygote? *Fertility reduction due to mis-segregation*
- A human translocation heterozygote? *Fertility reduction due to mis-segregation; this is one cause of Down's Syndrome*
- A human translocation homozygote? *Should be fine*

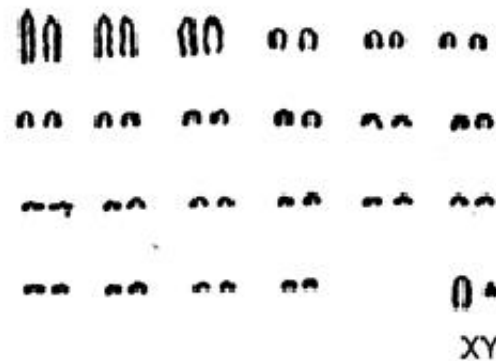
# Chromosome fission and fusion

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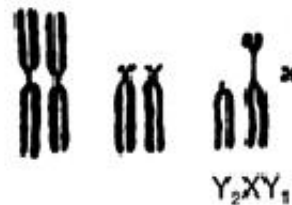
Chromosome fusion probably explains why domestic horses have 64 chromosomes. . .



. . . and the closest living wild species and probable ancestor of domestic horses, Przewalski's horse, has 66 chromosomes.



Chinese muntjac deer  
*Muntiacus reevesi*



Indian muntjac deer  
*Muntiacus muntjak*

Translocation,  
chromosome fusion,  
and/or fission explain  
why these two very  
similar species of  
hoofed mammal, the  
Chinese and Indian  
muntjac deer, have  
such different  
karyotypes.

## Chromosome fission and fusion

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- Fissions and fusions may begin as translocations where the reciprocal product is lost
- Generally underdominant
- This contributes to the sterility of mules (hybrids between two closely related species which differ in chromosome number)
- Closely related species often differ in chromosome number
- This may represent an early step in reproductive isolation



## One-minute responses

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- Tear off a half-sheet of paper
- Write one line about the lecture:
  - Was anything unclear?
  - Did anything work particularly well?
  - What could be better?
- Leave at the back on your way out