## Roadmap

- 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

- 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

- 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?)

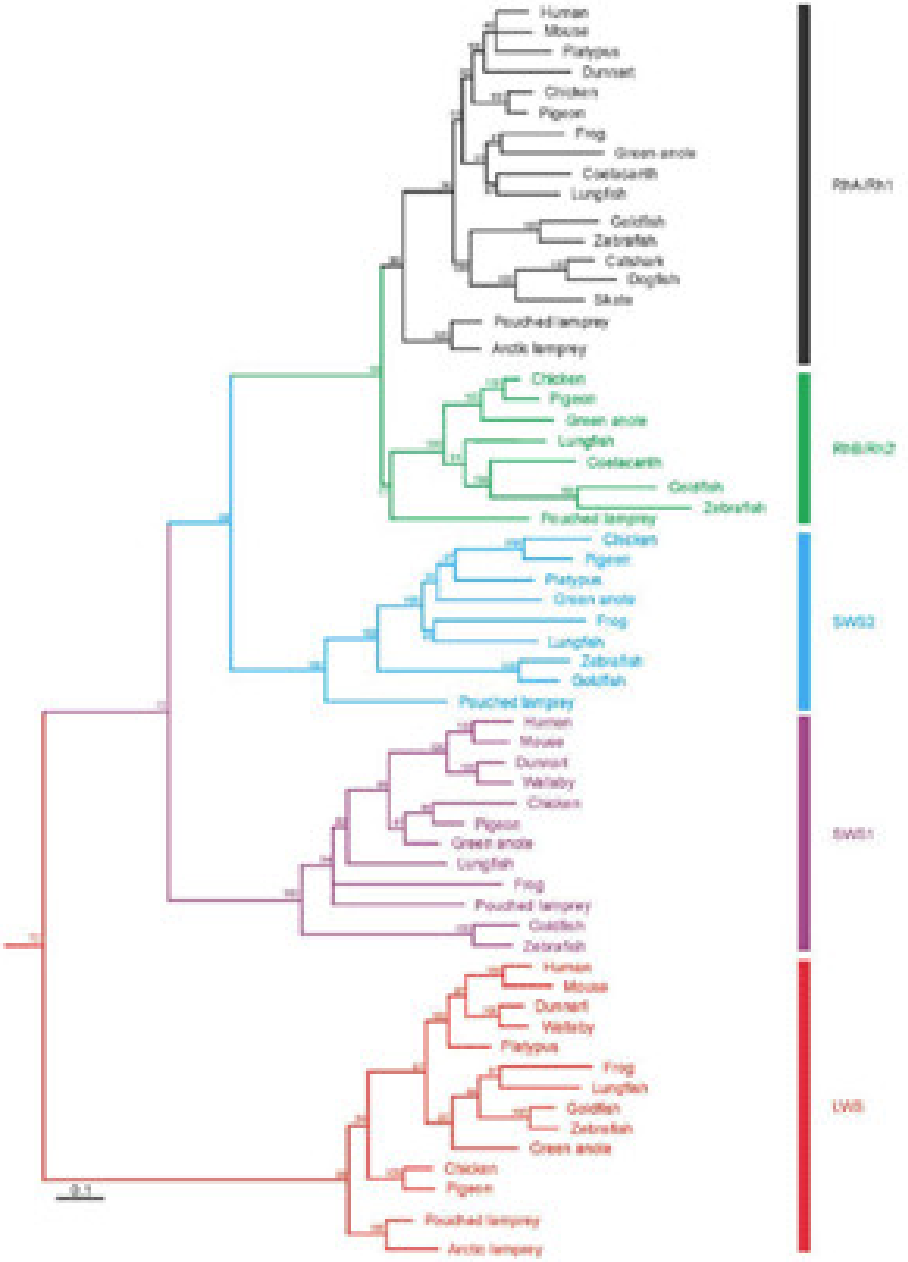


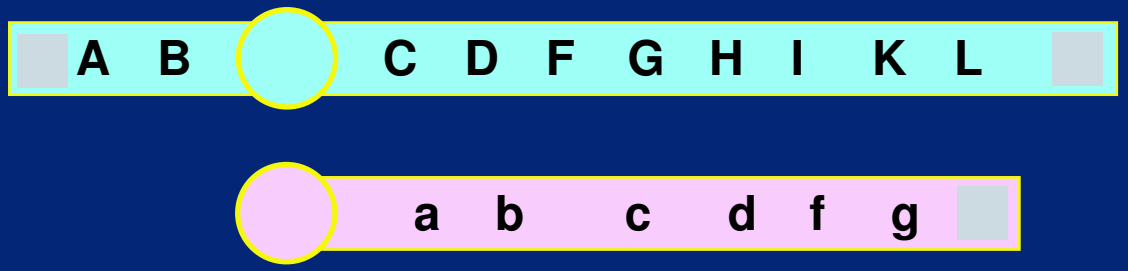

**Figure 22. Phylogeny of vertebrate cone and rod opsins**

## An opsin mystery

- 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....

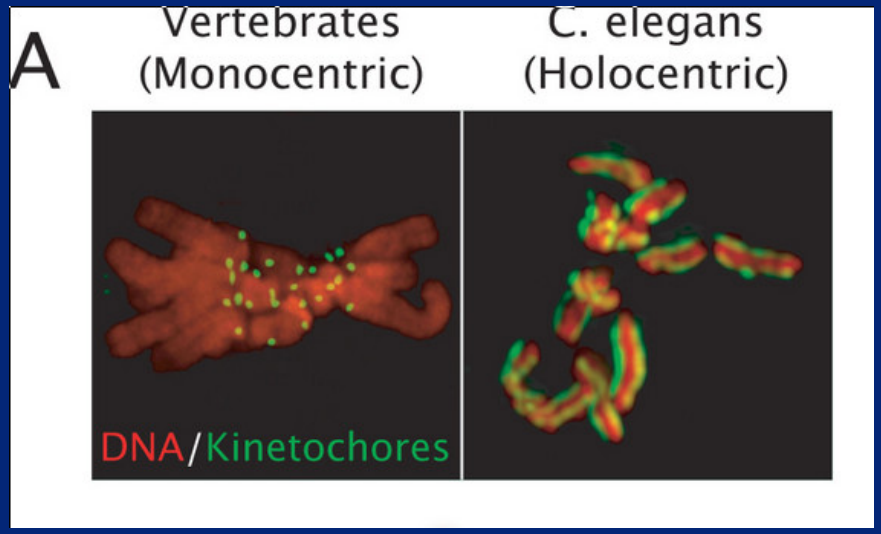
## Chromosomes

- 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



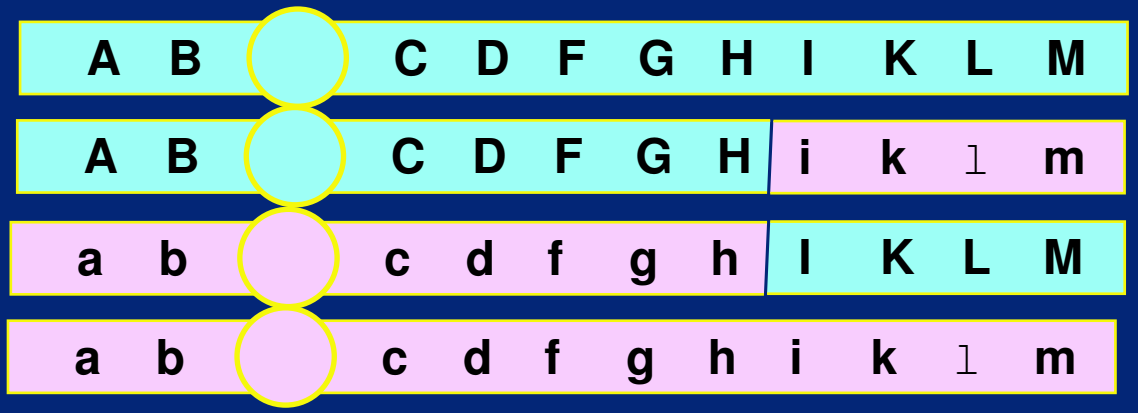
## An exception to the rule

- *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

- Through crossing-over chromosome strands can mix and recombine




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

## Chromosome rearrangements

- 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

**Figure 5.9** A photograph showing the aperiodic nature and synapsis of polytene chromosomes (chromosome III of *Drosophila mojavensis*). Although seemingly one, this is actually two homologous chromosomes intimately paired, band-for-band, along their lengths. Compare with Fig. 5.10, where the separate chromosomes may be detected. [Courtesy of Jean C. Brown and L. C. Saylor.]

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

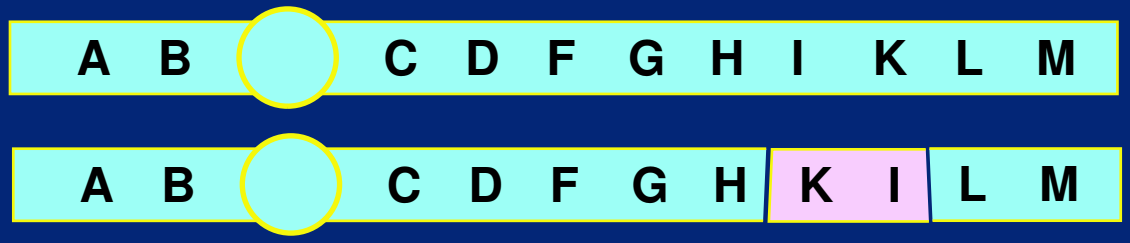
## Inversions

Paracentric inversion (does not include centromere)

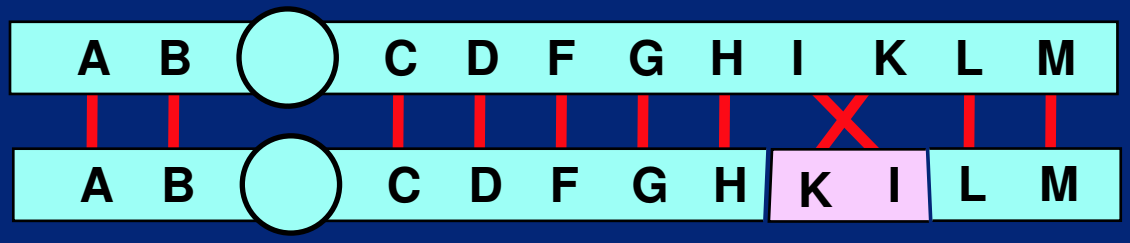

Pericentric inversion (includes centromere)


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

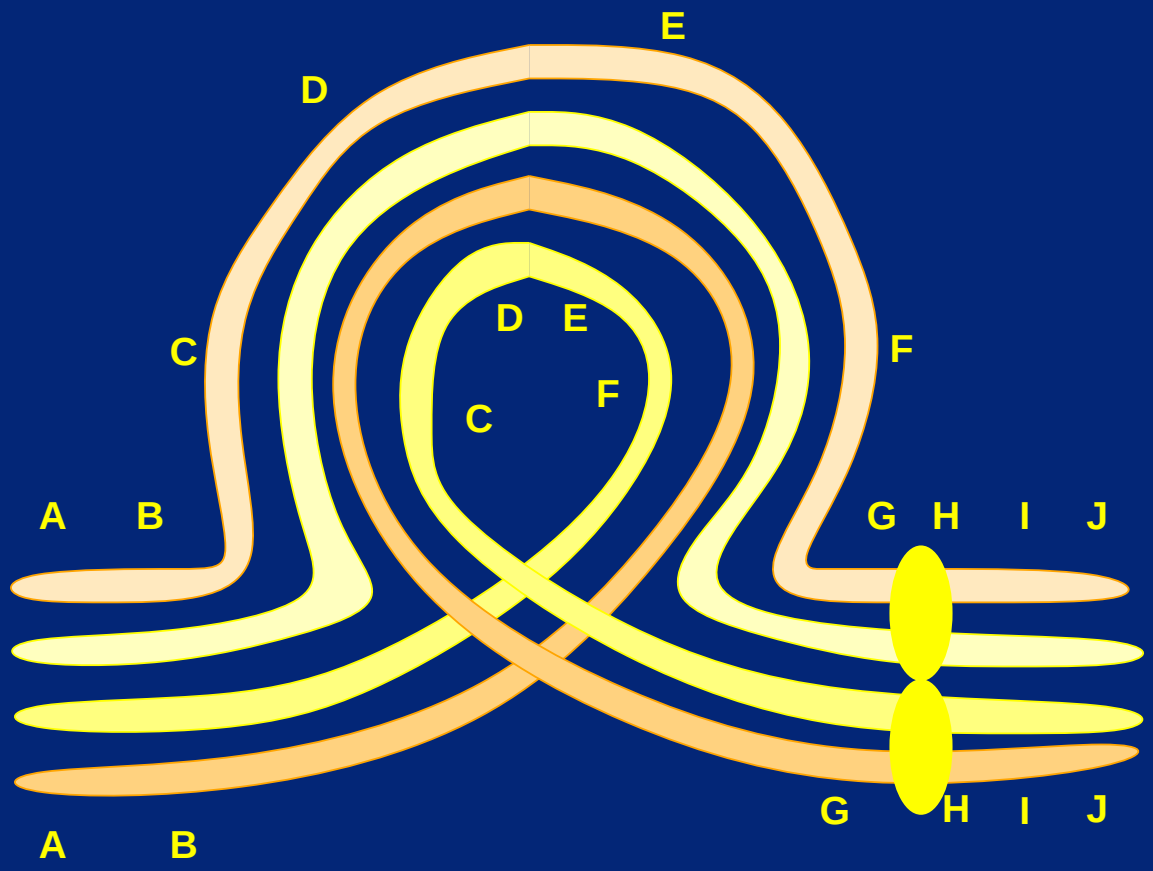
# Paracentric inversion heterozygote

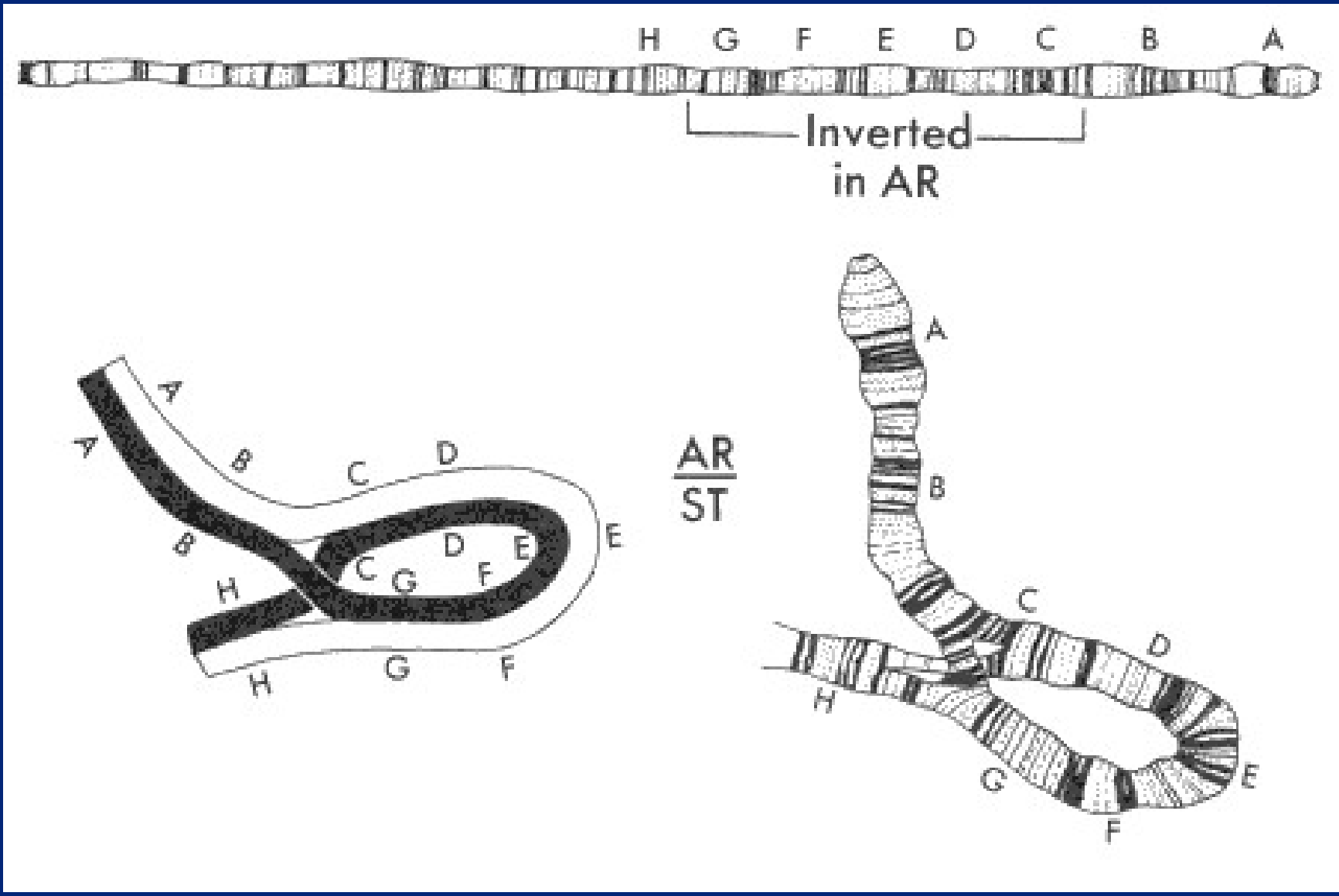


# Paracentric inversion heterozygote

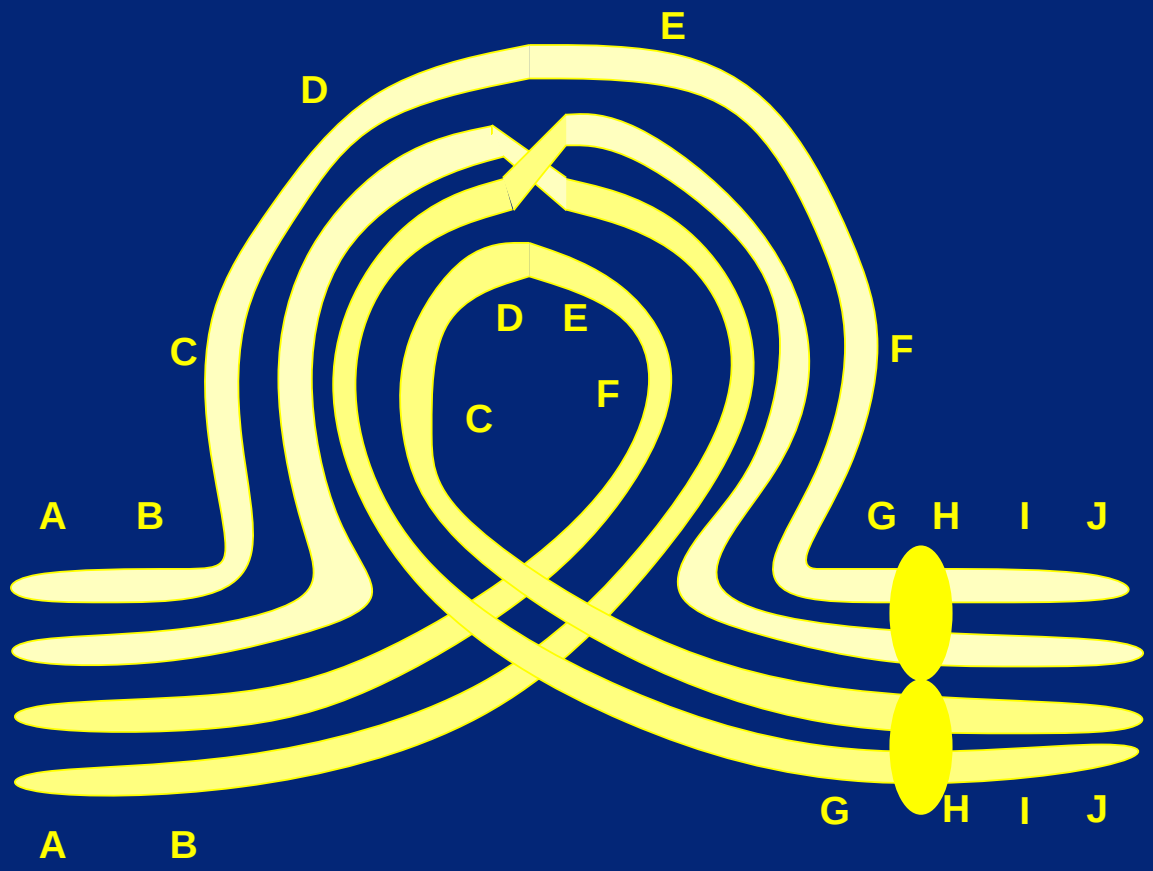


# Pairing in a paracentric inversion heterozygote

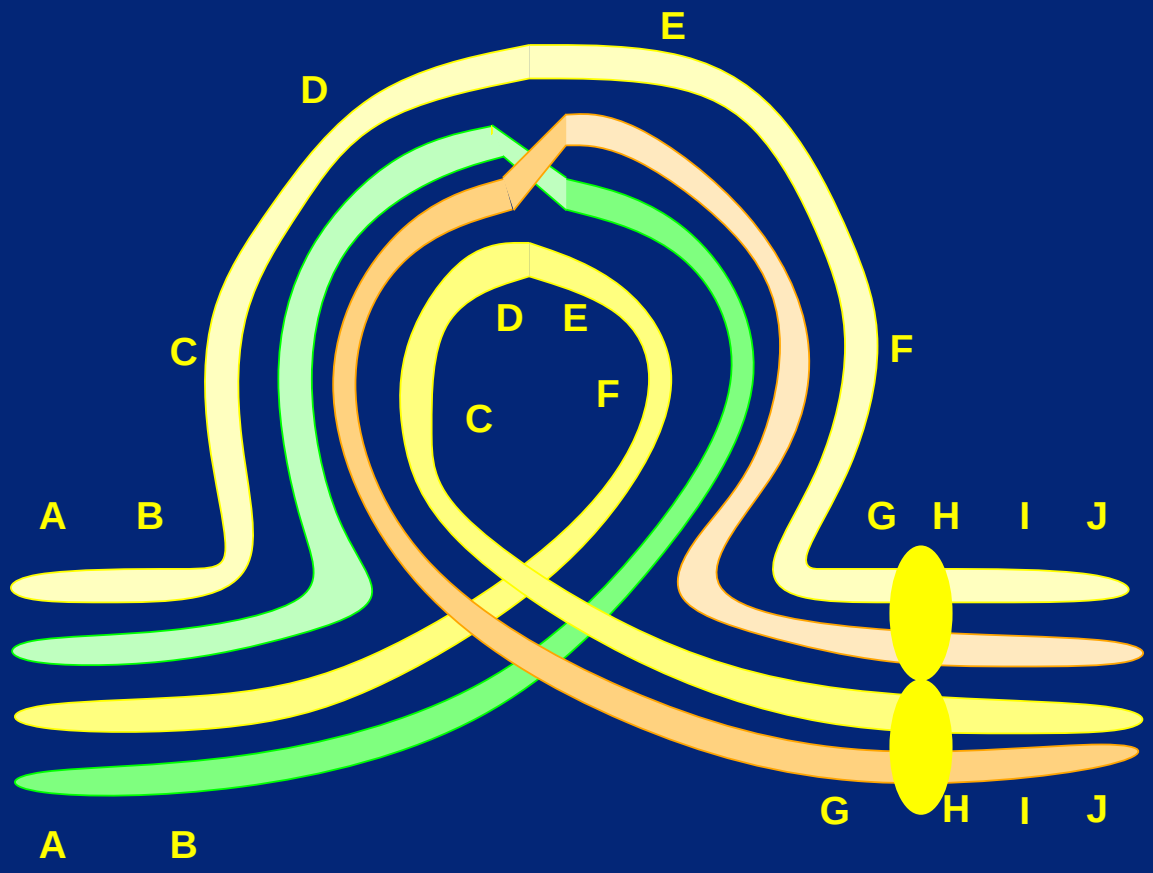



H G F E D C B A
Inverted
in AR
A B C D E F G H
AR
ST
A B C D E F G H

# 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`

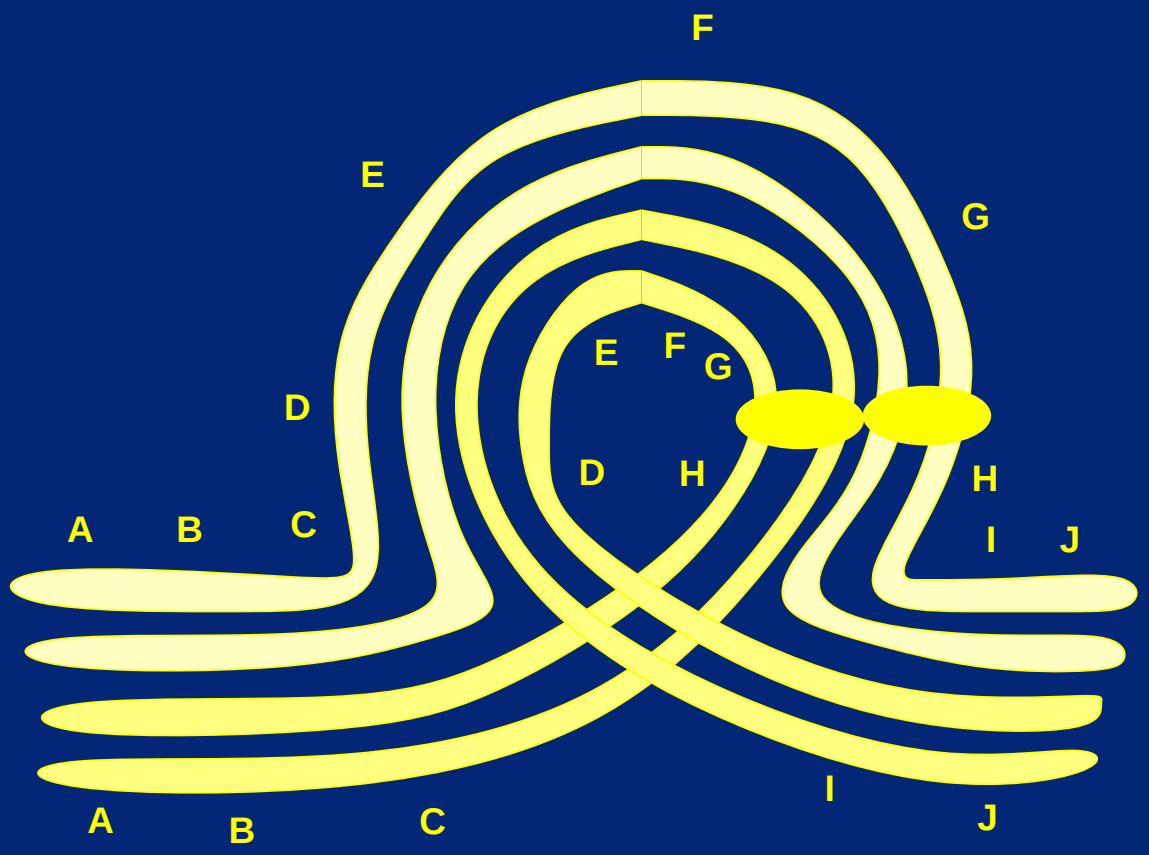
*No centromere*

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

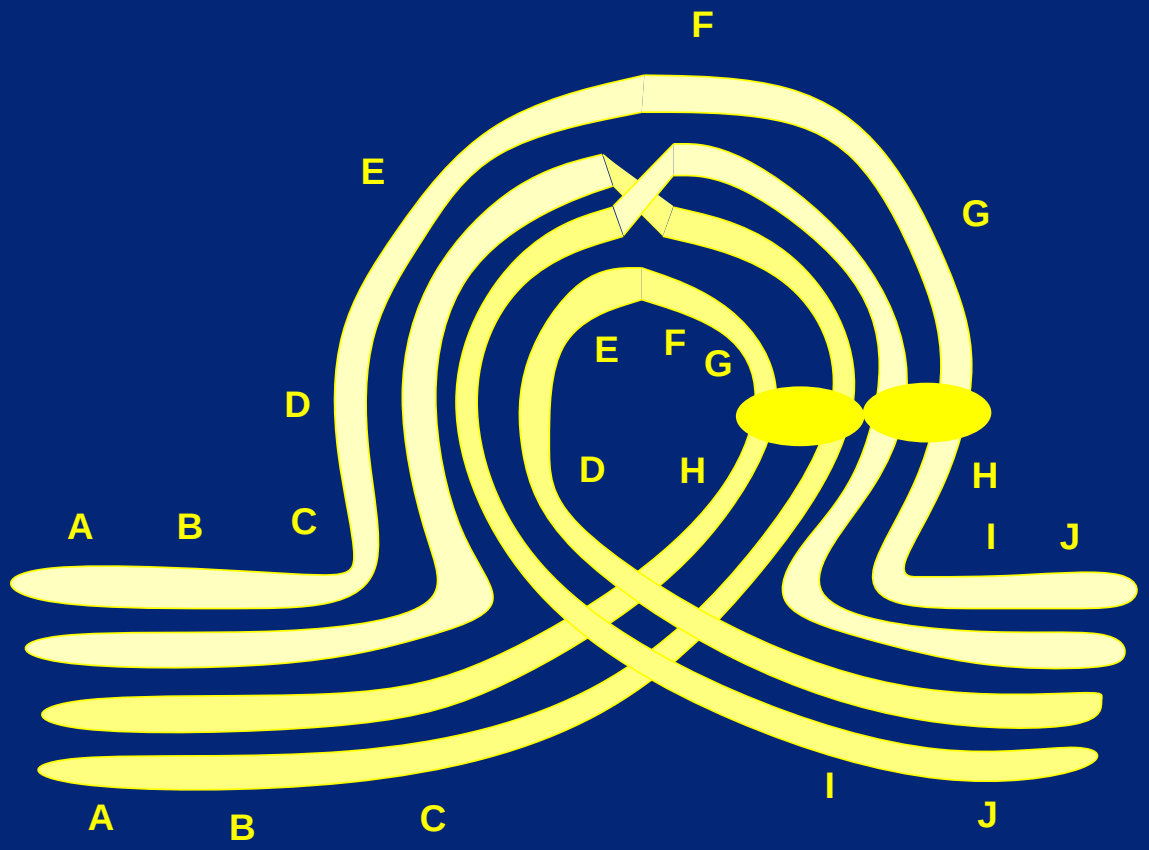
*Two centromeres (breaks randomly)*

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

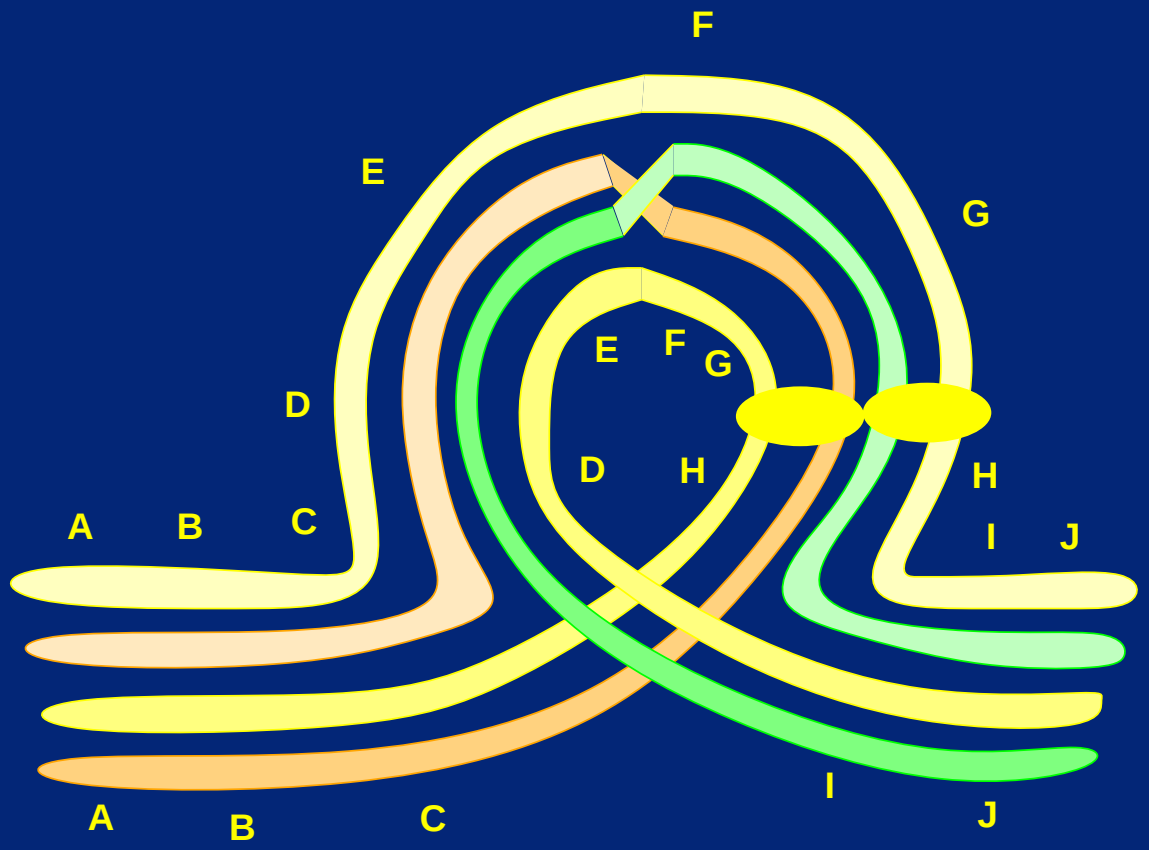
# Pairing in a pericentric inversion heterozygote



# 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*

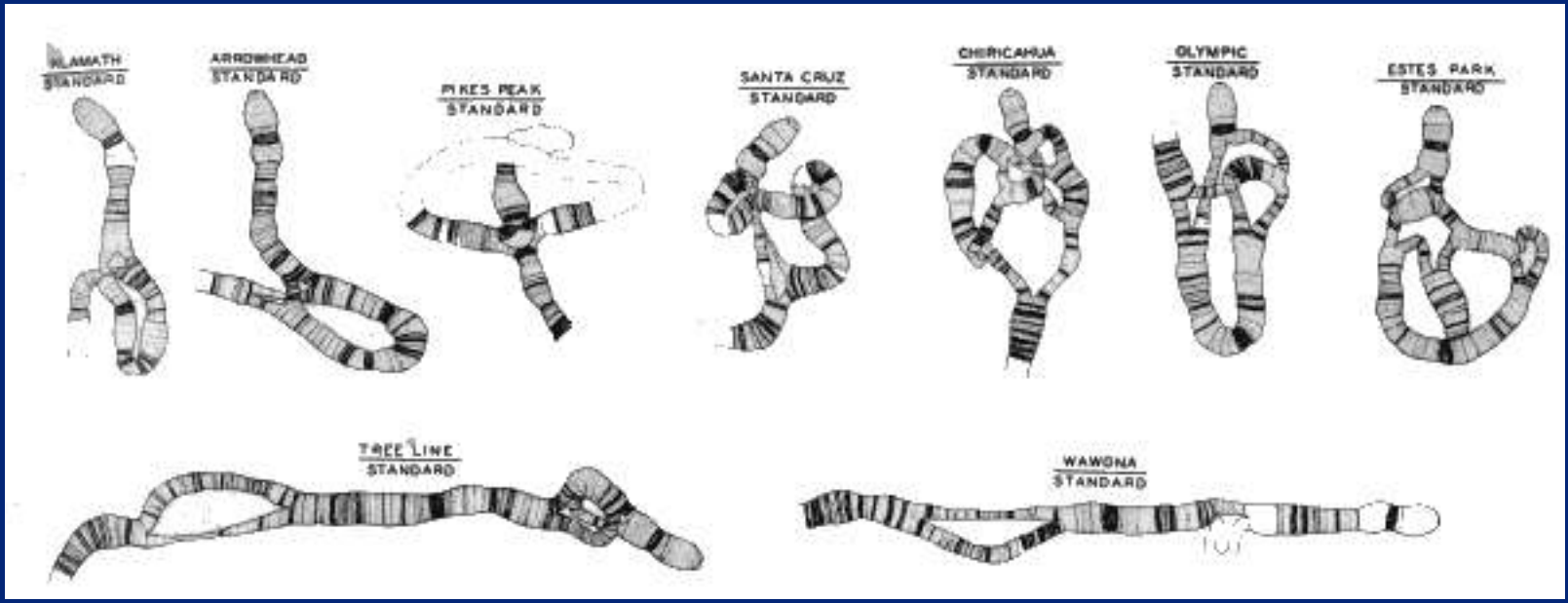
J-I-D-E-F-G-CEN-H-I-J

## Inversion summary

- 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

- 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

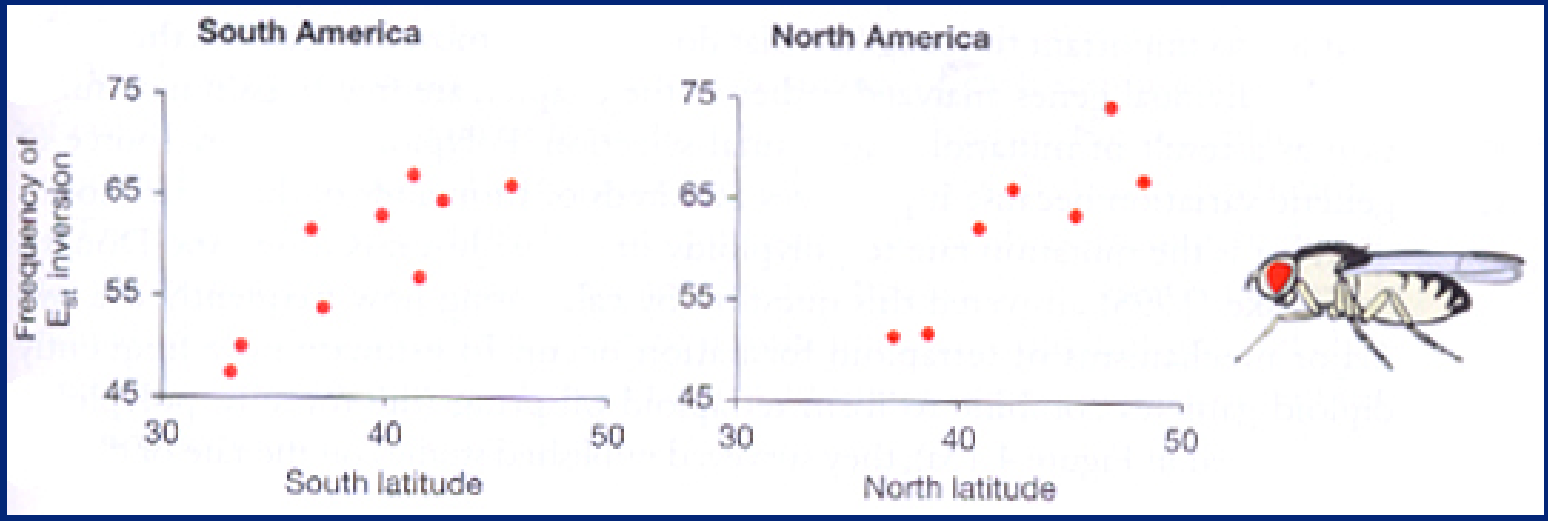



(c) Steven M. Carr

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

- 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

What, if any, problems arise for:

- A male Drosophila inversion heterozygote?
- A female Drosophila inversion heterozygote?
- A human inversion heterozogyote?
- A human inversion $homozogyote$?

Assume no genes were damaged by creation of the inversion

## Practice problem

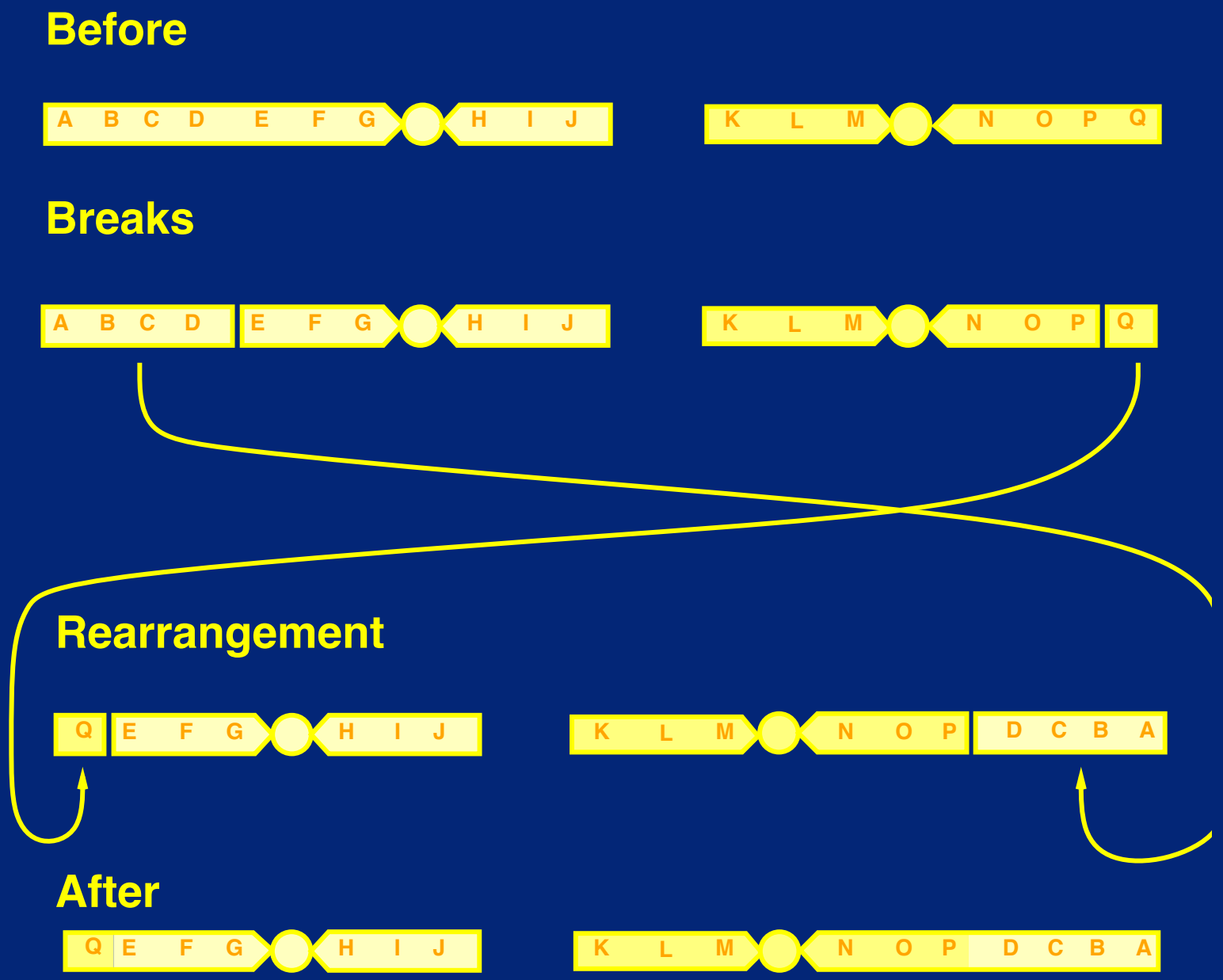
What, if any, problems arise for:

- 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 heterozogyote? *Fertility reduction*
- A human inversion *homozogyote*? *Should be fine*

Assume no genes were damaged by creation of the inversion

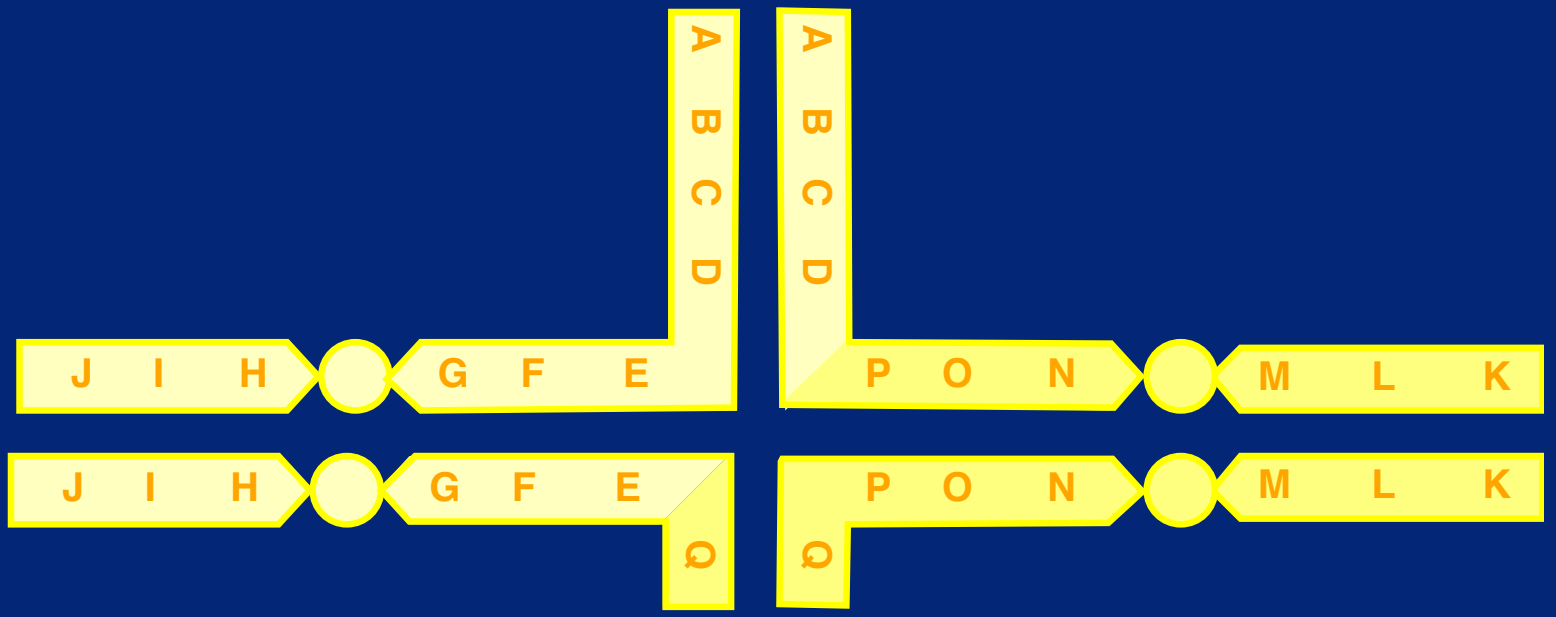
# Translocation

# A translocation



# A translocation heterozygote

**at first division of meiosis metaphase**



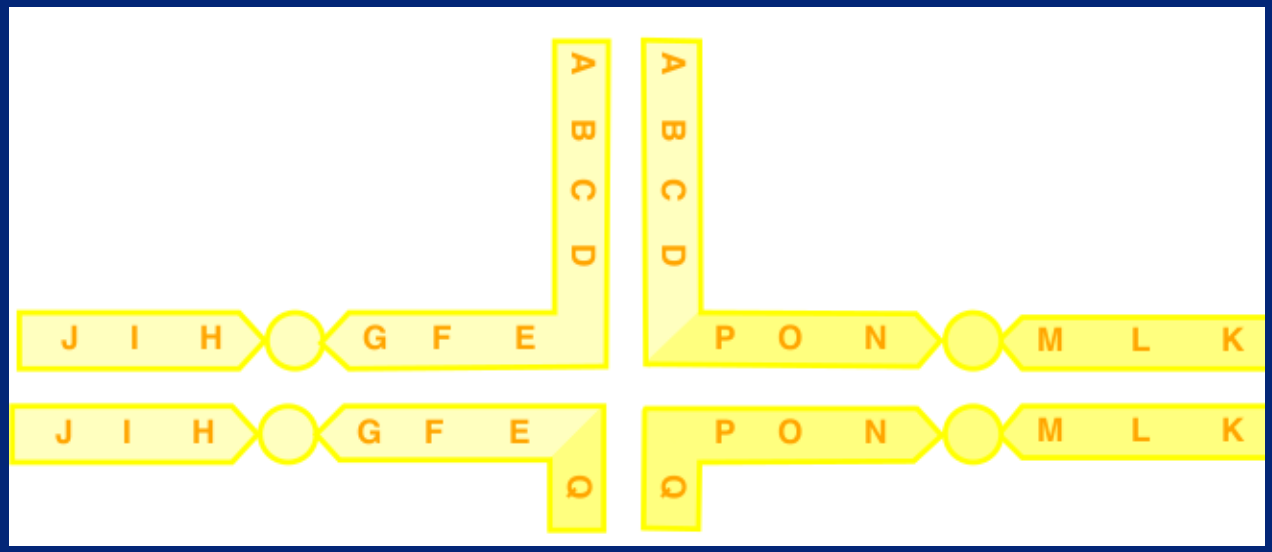

**A pair of translocated chromosomes**
**pairs with a pair of untranslocated chromosomes**

## Translocation

- 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 | Possibility 2 |
|---|---|
| A-B-C-D-E-F-G-CEN-H-I-J | A-B-C-D-E-F-G-CEN-H-I-J |
| K-L-M-CEN-N-O-P-Q | Q-E-F-G-CEN-H-I-J |
| A-B-C-D-P-O-N-CEN-M-L-K | A-B-C-D-P-O-N-CEN-M-L-K |
| Q-E-F-G-CEN-H-I-J | K-L-M-CEN-N-O-P-Q |

## Practice problem

What, if any, problems arise for:

- A male Drosophila translocation heterozygote?
- A female Drosophila translocation heterozygote?
- A human translocation heterozogyote?
- A human translocation $homozogyote$?

## Practice problem

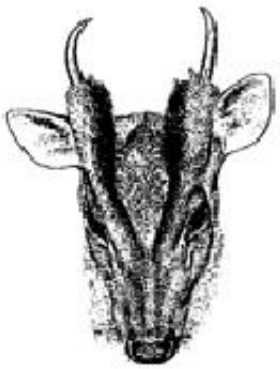
What, if any, problems arise for:

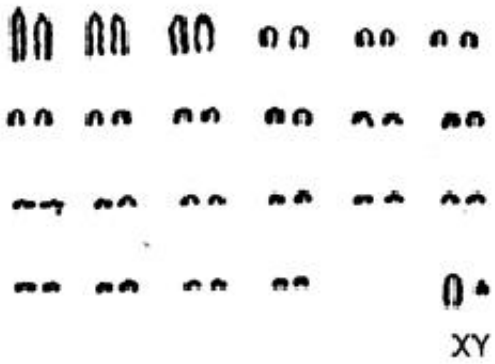
- A male Drosophila translocation heterozygote? *Fertility reduction due to mis-segregation*
- A female Drosophila translocation heterozygote? *Fertility reduction due to mis-segregation*
- A human translocation heterozogyote? *Fertility reduction due to mis-segregation; this is one cause of Down's Syndrome*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

## Chromosome fission and fusion

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

- 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

- 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