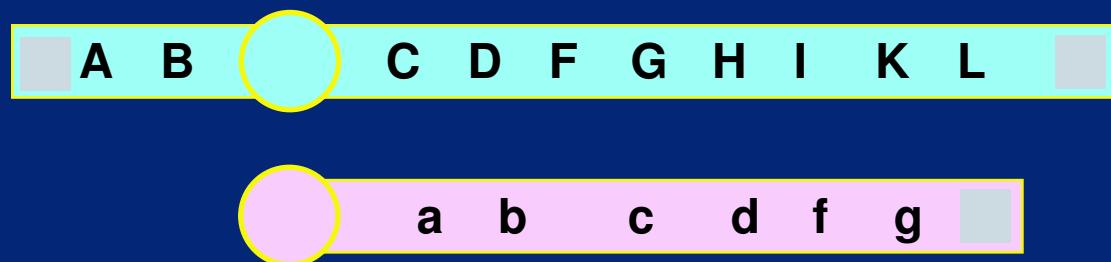


Chromosomes

- Bacterial chromosomes are circular
- Most higher organisms have linear chromosomes with a centromere that attaches them to the spindle
- Centromere can be in the center (metacentric), off-center (acrocentric) or at the end (telocentric)
- 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
- Meiotic pairing and segregation functions quite differently in this animal
- No one seems to know why this evolved

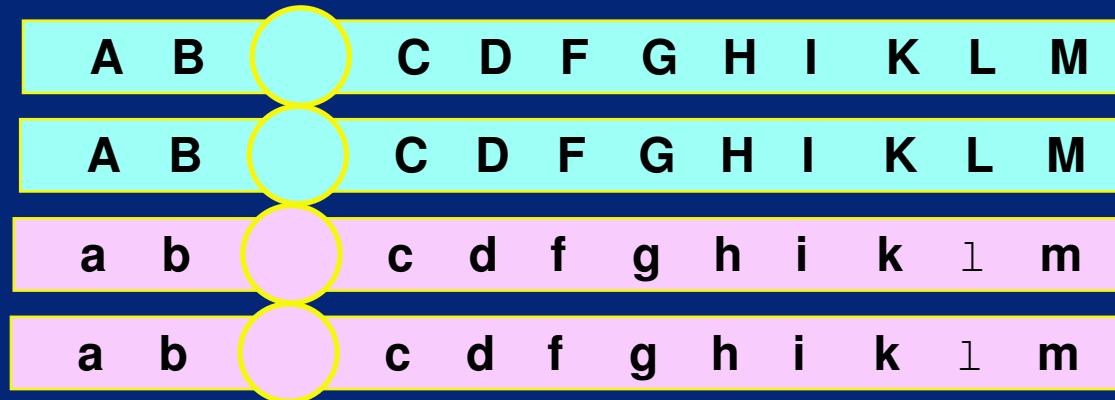
Chromosomes duplicate

- During early meiosis, each chromosome set duplicates.



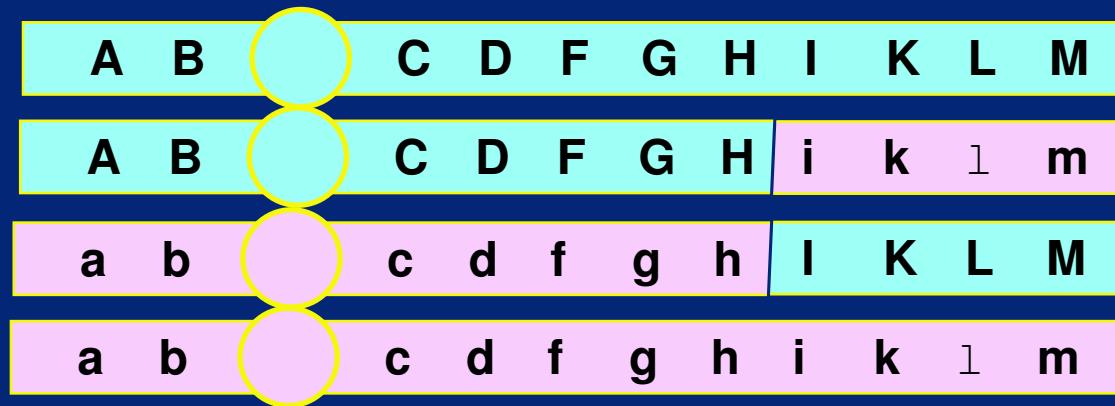
Crossing-over/recombination

- During meiosis, each chromosome set duplicates and the two sets form a tetrad

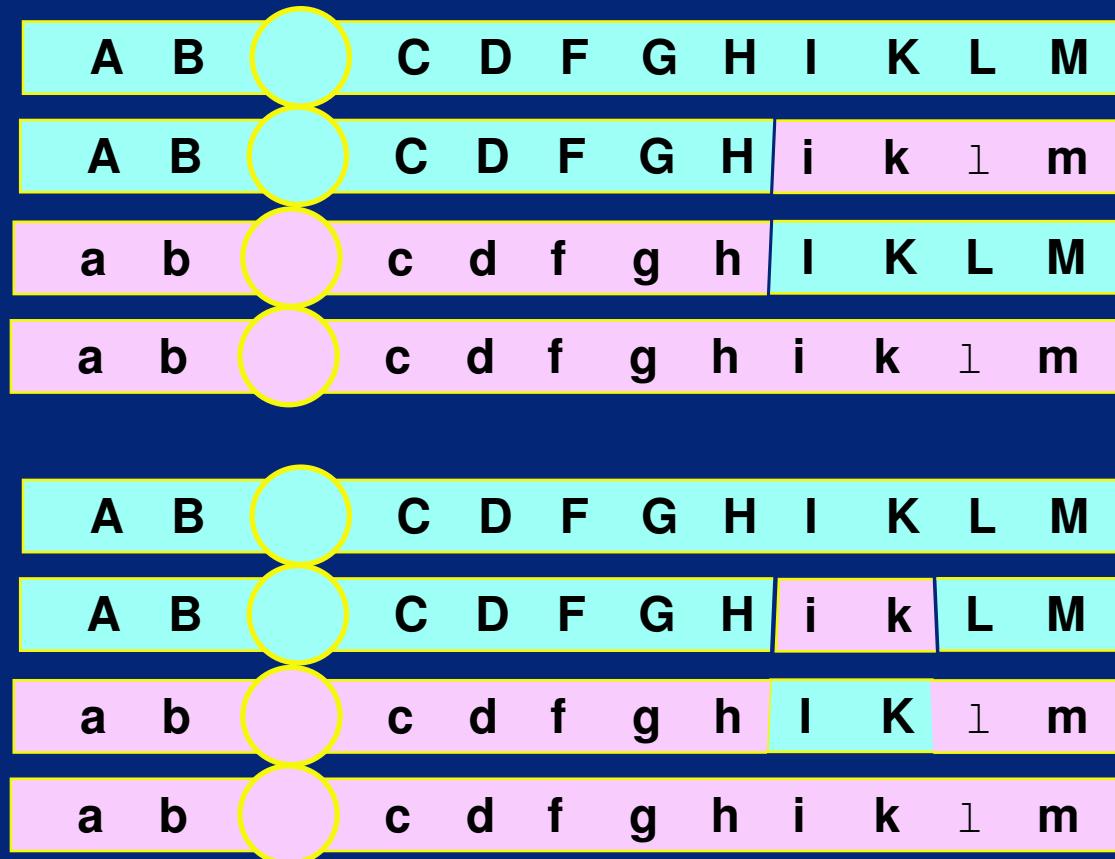


Crossing-over/recombination

- Through crossing-over chromosome strands can mix and recombine



Chromosomes



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.]

- Diptera (such as *Drosophila*, mosquitoes) have in their salivary glands giant chromosomes - polytene chromosomes.
- Chromosomes pair and then reduplicate themselves up to 1024 copies
- Useful tool to see pairing of rearrangement heterozygotes

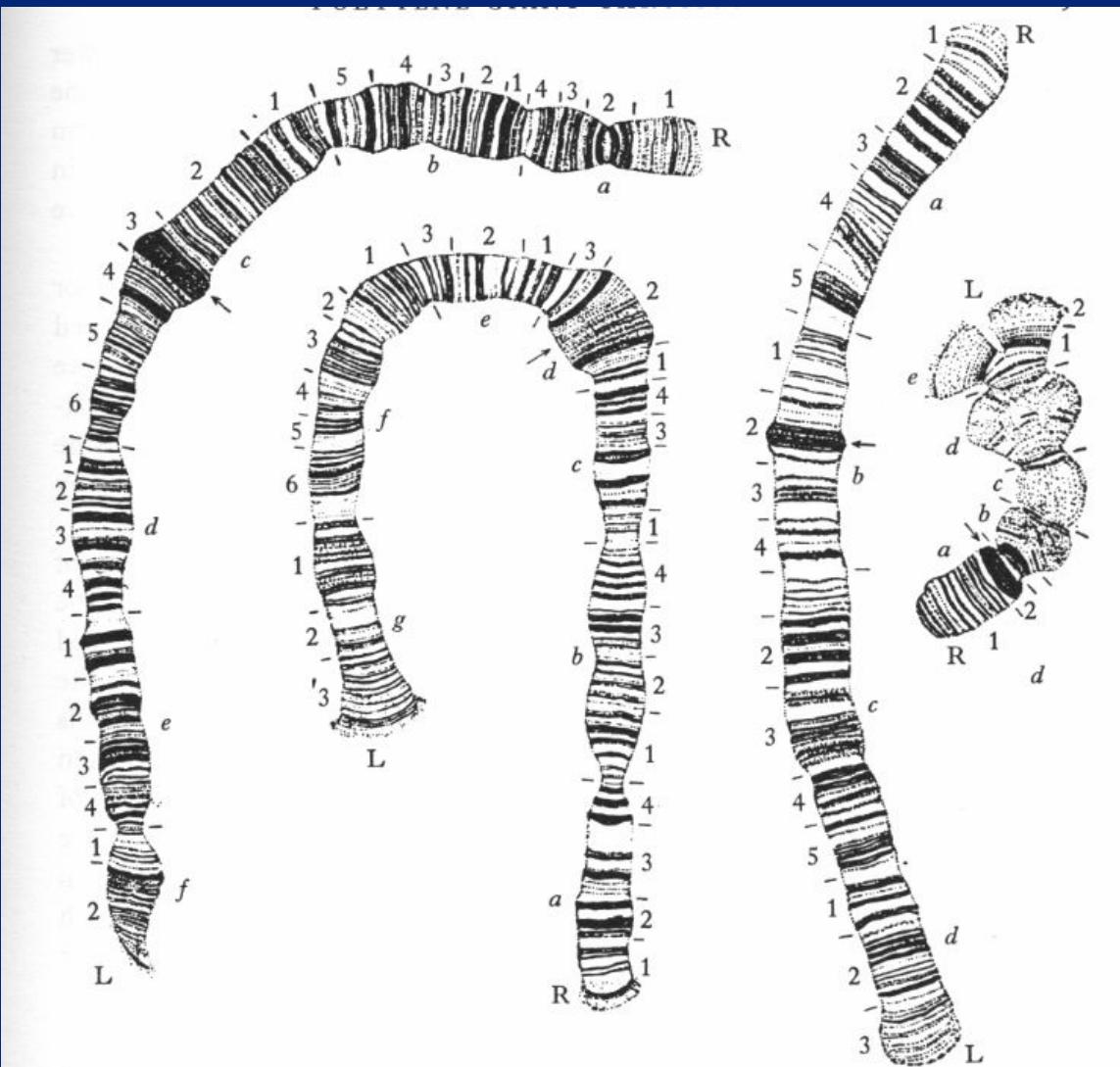


Fig. 4.1. The four polytene elements from a salivary gland nucleus of *Chironomus thummi*, showing the banding. Small arrows indicate the approximate positions of the centromeres. The shortest chromosome has its longer arm largely heterochromatic and the other three elements have short heterochromatic regions at the tips. From Bauer (1935).

Inversions

Paracentric inversion (does not include centromere)

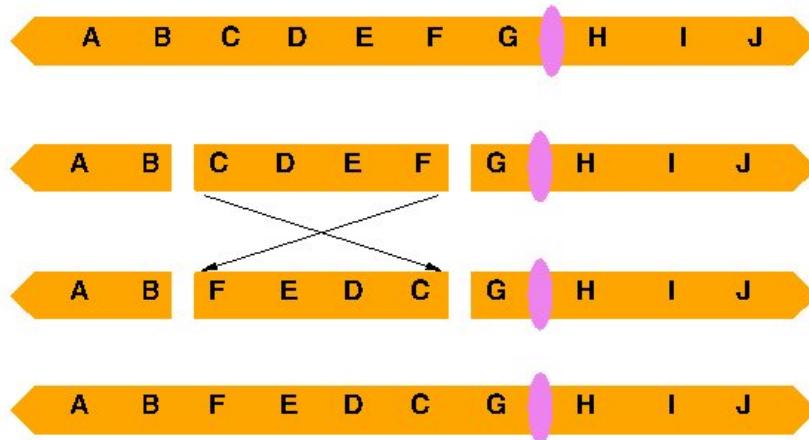


Pericentric inversion (includes centromere)

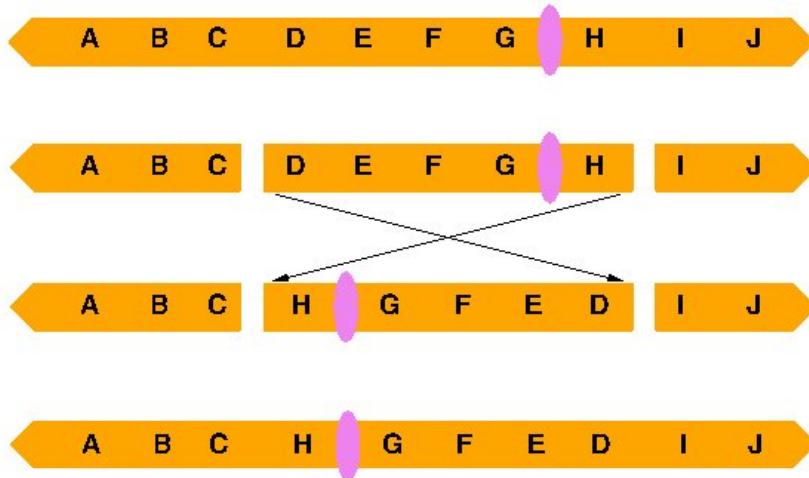


I prefer not to use these words as they sound too similar to me!

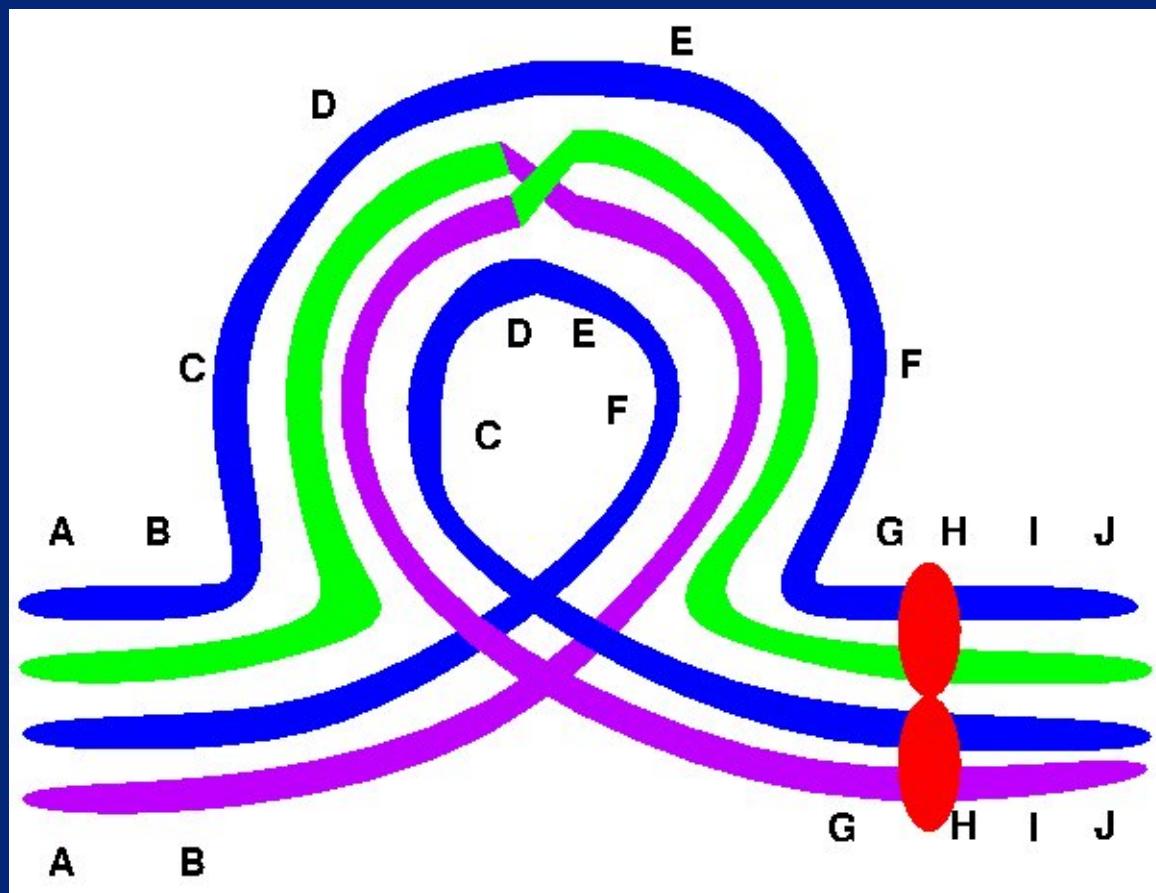
A Paracentric Inversion



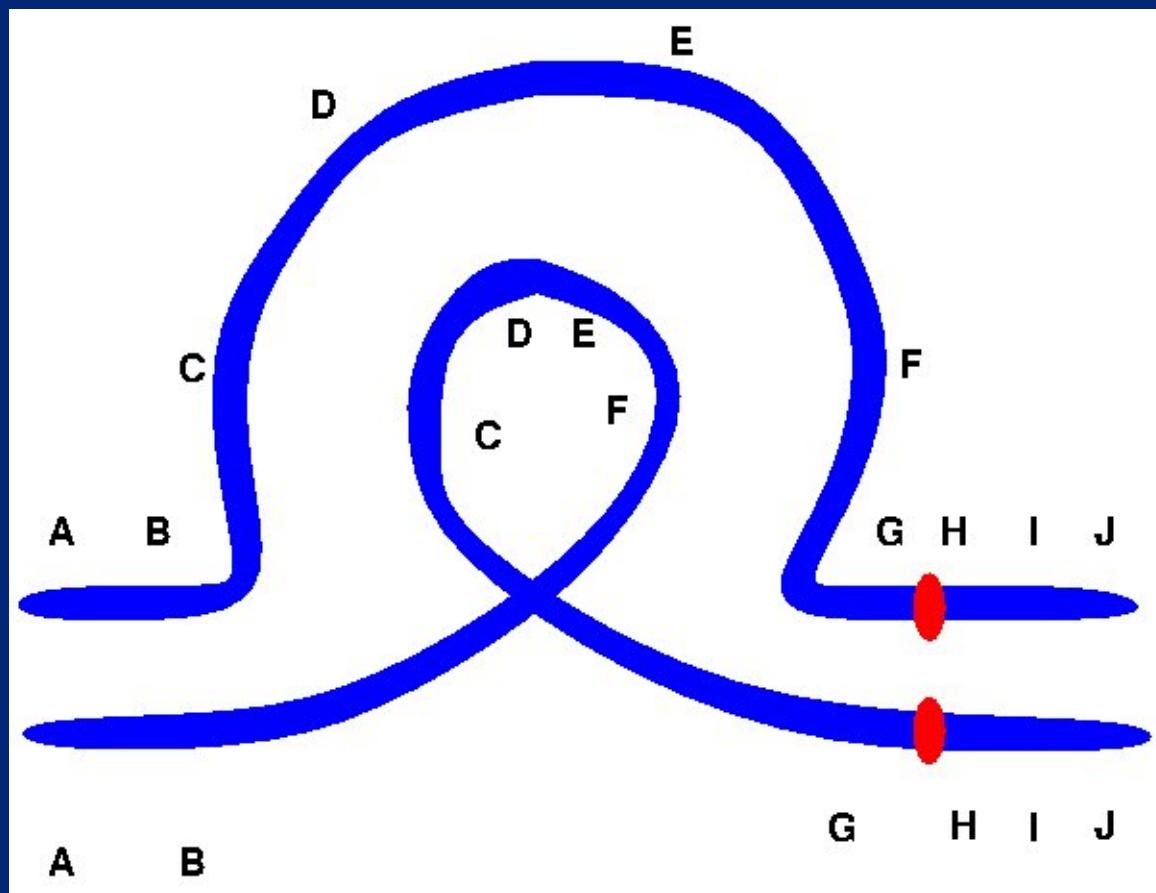
A Pericentric Inversion



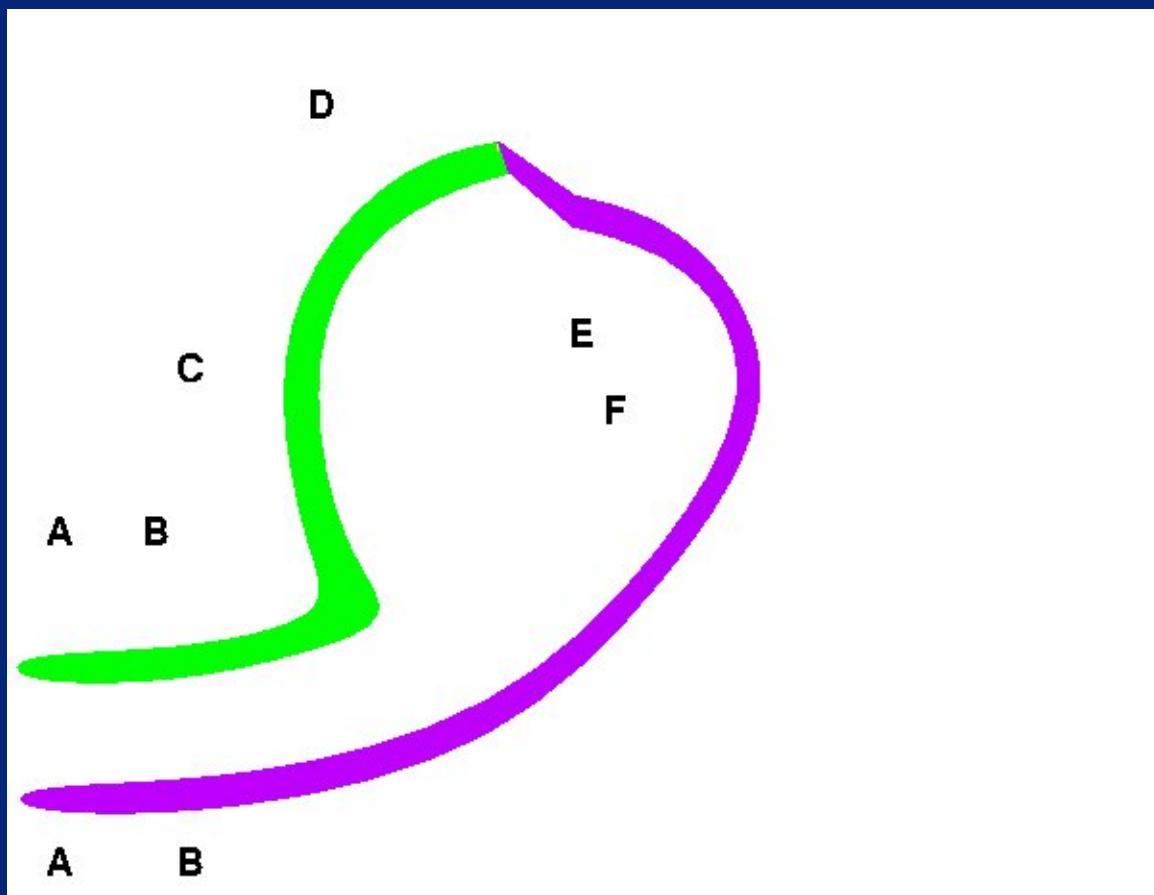
Paracentric inversion



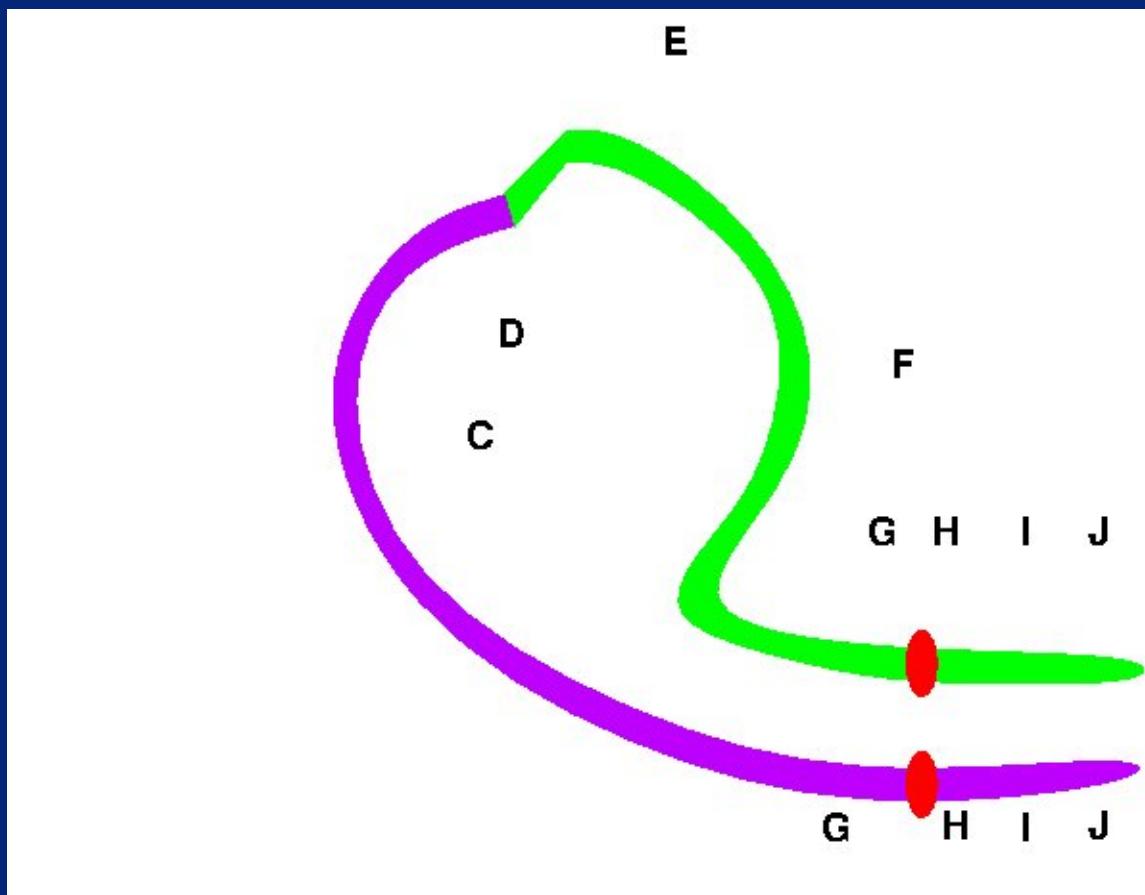
Paracentric inversion



Paracentric inversion



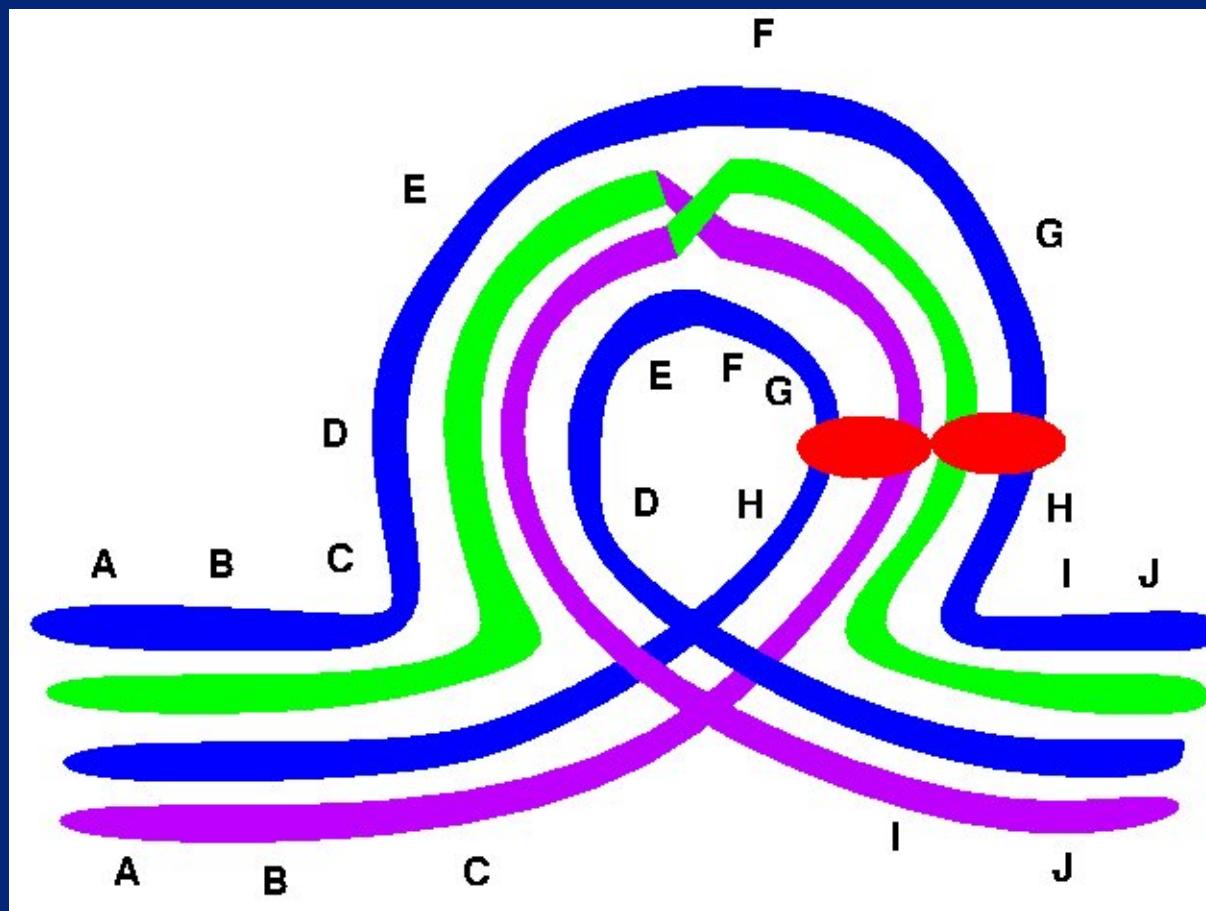
Paracentric inversion



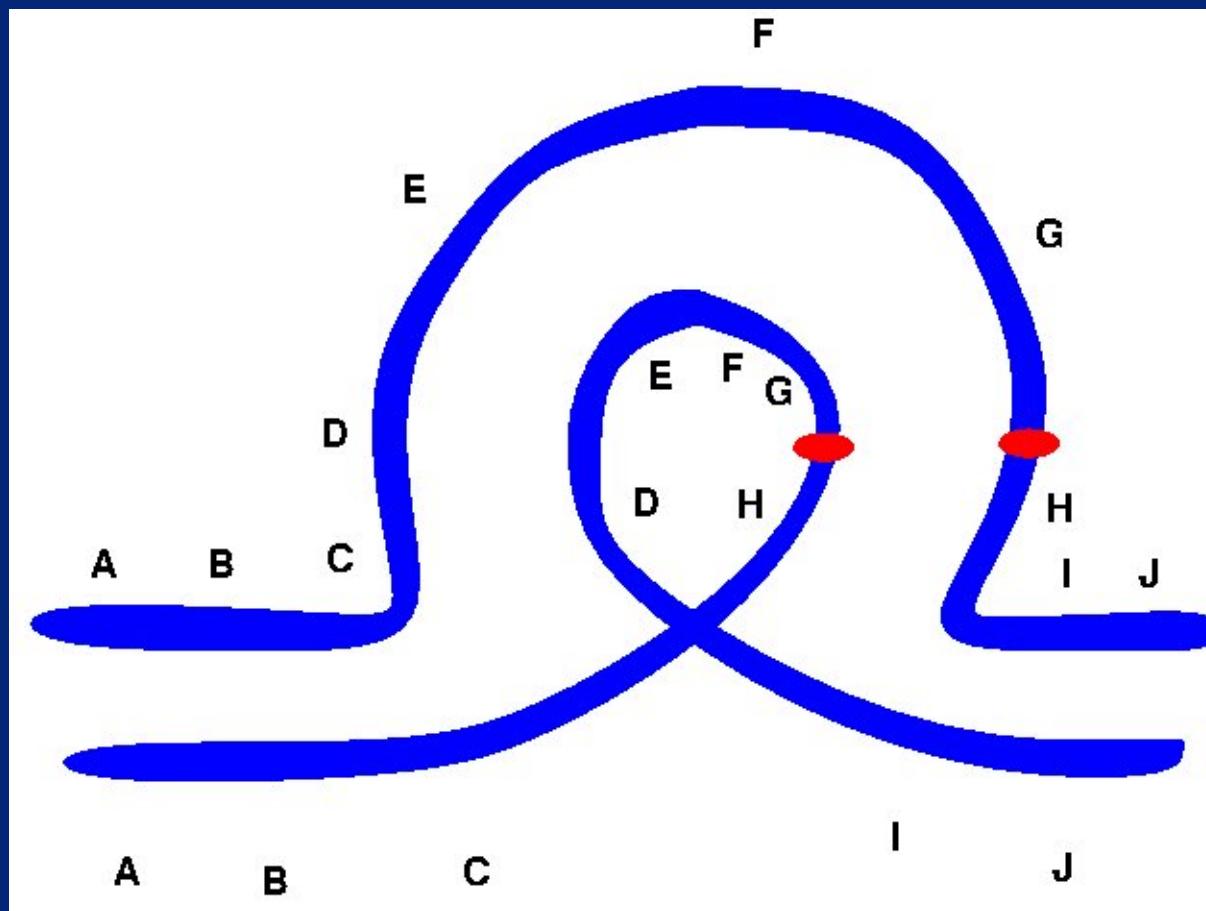
Paracentric inversion outcome

A-B-C-D-E-F-G-centromere-H-I-J	functional
A-B-F-E-D-C-G-centromere-H-I-J	functional
A-B-C-D-E-F-B-A	not functional
J-I-H-centromere-G-C-D-E-F-G-centromere-H-I-J	breaks randomly

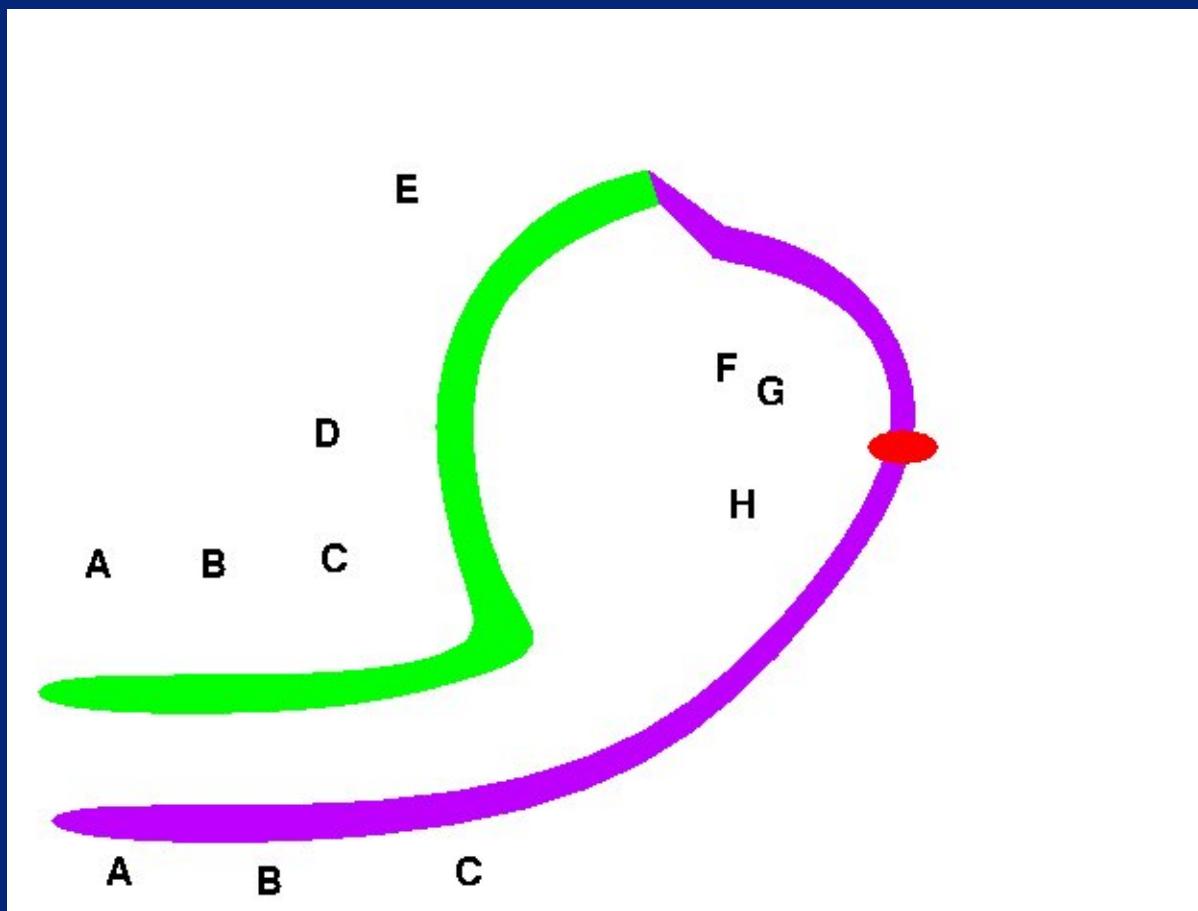
Pericentric inversion



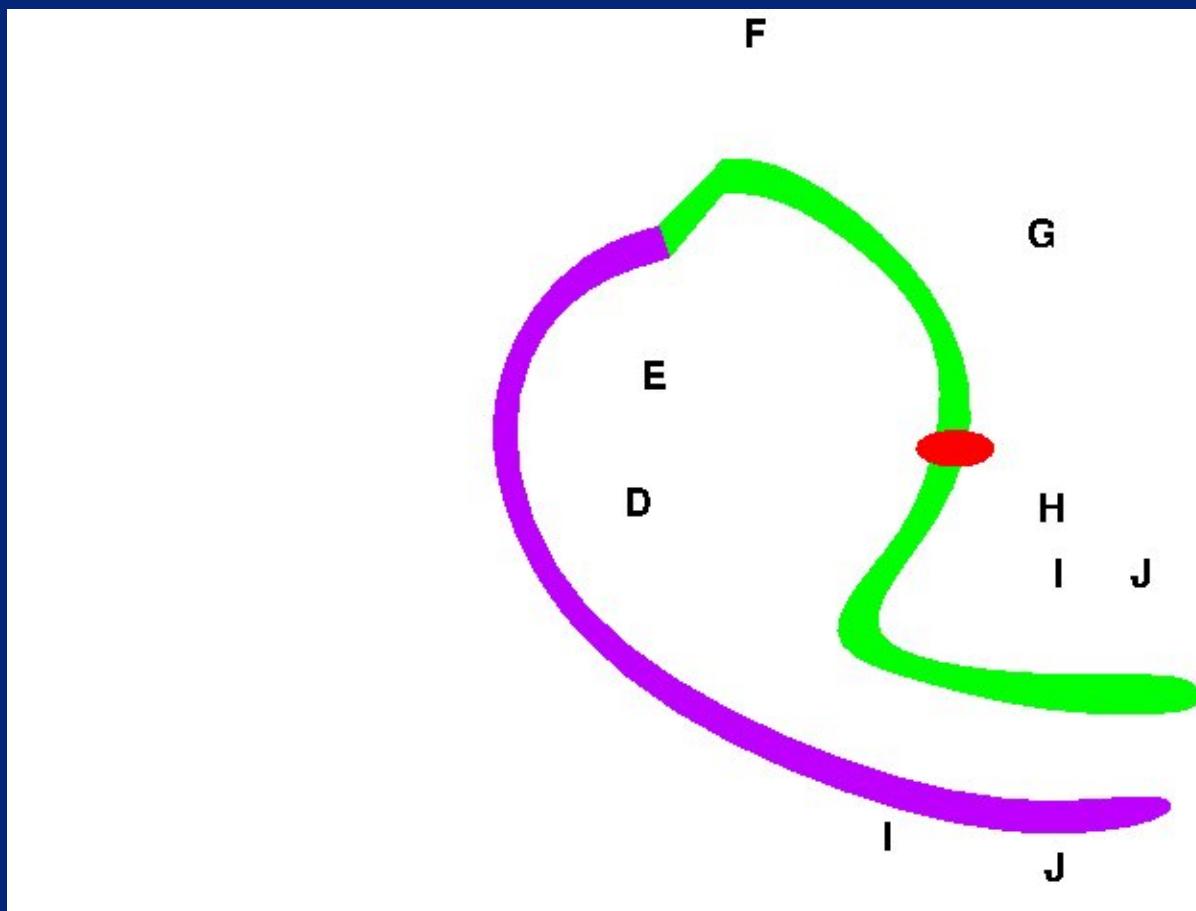
Pericentric inversion



Pericentric inversion



Pericentric inversion



Pericentric inversion outcome

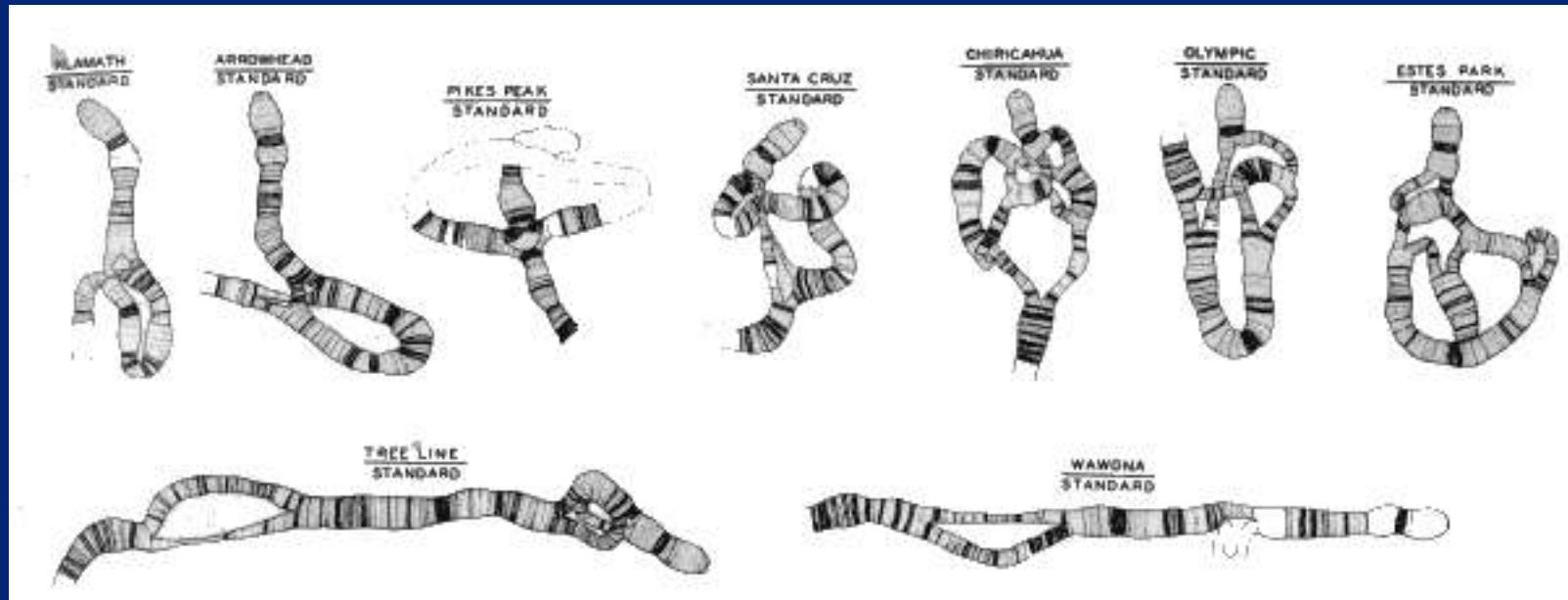
A-B-C-D-E-F-G-	centromere	-H-I-J	functional
A-B-C-H-	centromere	-G-F-E-D-I-J	functional
A-B-C-D-E-F-G-	centromere	-H-C-B-A	duplication, deletion
J-I-D-E-F-G-	centromere	-H-I-J	duplication, deletion

Inversion summary

- Inversions come into existence when the chromosome breaks twice and is reversed while being repaired
- Recombination in an inversion heterozygote that does not include the centromere produces two-centromere and no-centromere chromosomes
- Such gametes will almost never be viable
- Recombination in an inversion heterozygote that does include the centromere produces duplications and deletions
- These are generally not viable either
- In all cases, if there is only one recombination two of the four chromatids will still be okay (but non-recombinant)

Inversion summary

- Inversions are often said to suppress recombination
- In most species they kill recombinants
- This might be an advantage by preventing a favorable group of genes from being broken up by recombination
- Most of the time, however, inversions act as underdominant traits
- They can establish themselves only if helped by drift or a strong selective advantage
- Some species have genetic systems more permissive of inversions

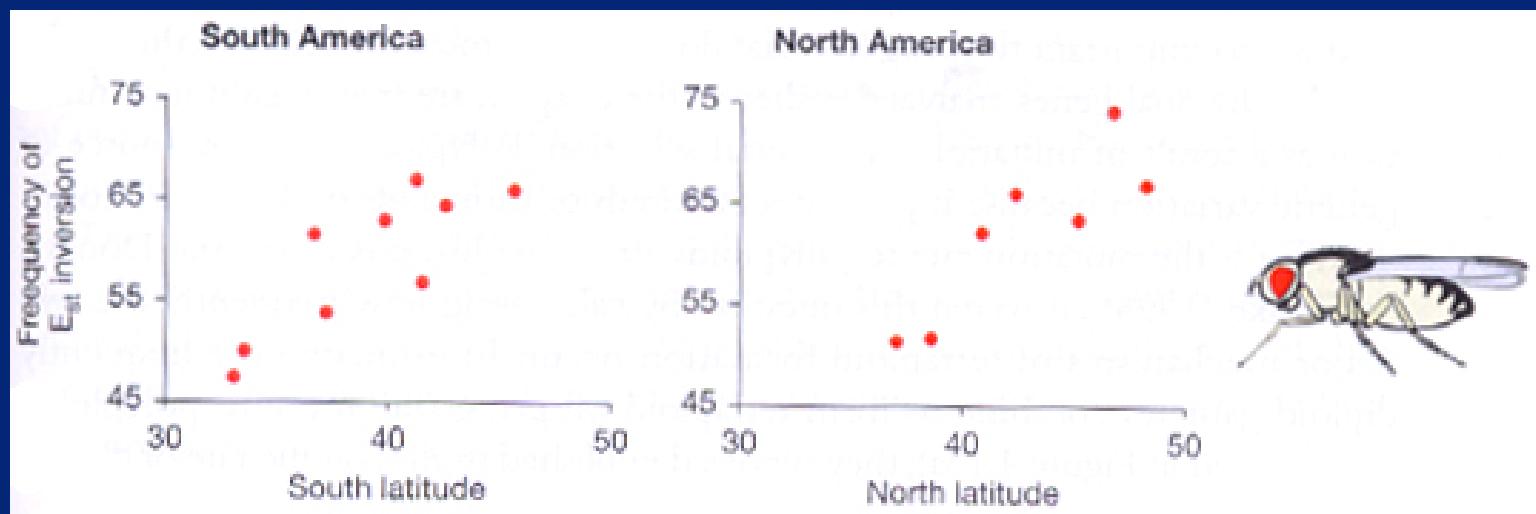


(c) Steven M. Carr

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
- Three factors may allow this:
 - Male Drosophila have no recombination, so there are no harmful effects on a male heterozygote
 - Female Drosophila use only one meiotic product to make the egg, and preferentially use ones where there are no broken chromosomes
 - Drosophila populations may often be established by a single fertilized female, so genetic drift is very powerful

Practice problem

What, if any, problems arise for:

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

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

Assume no genes were damaged by creation of the inversion

Translocation



A translocation

Before



Breaks



Rearrangement

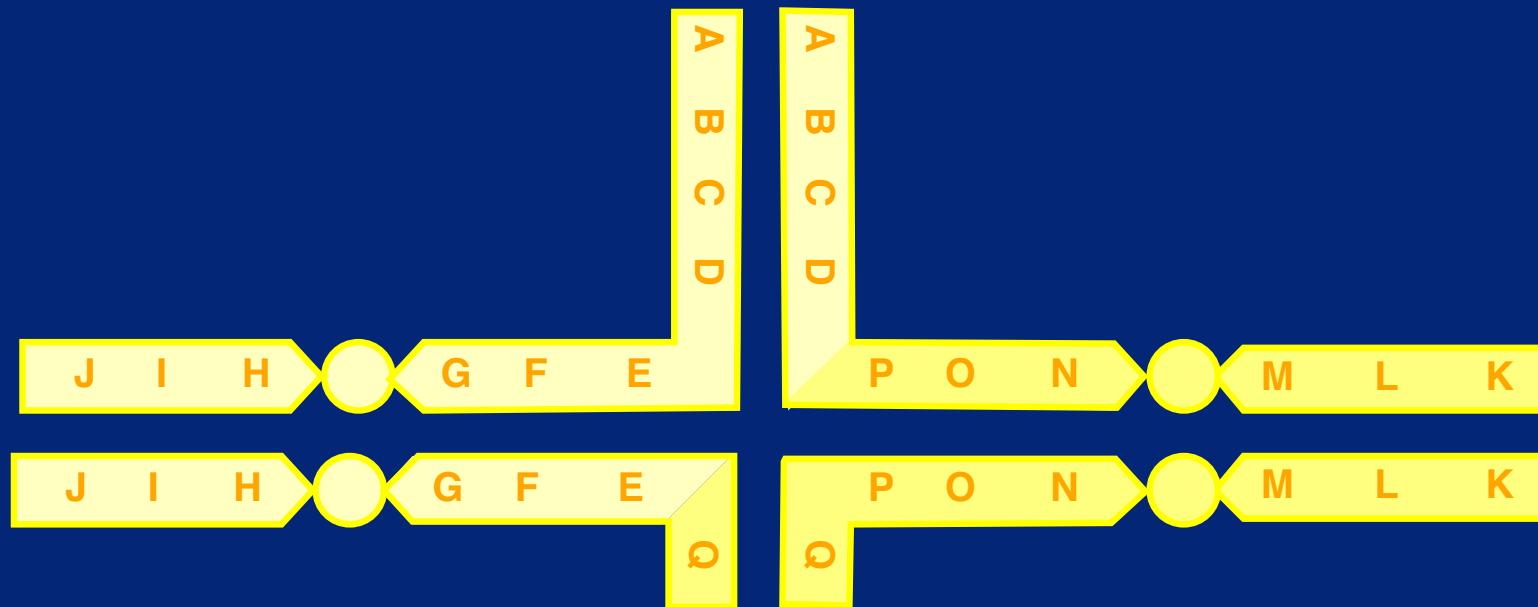


After



A translocation heterozygote

at first division of meiosis metaphase



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

Translocation

- Translocation occurs when chromosomes break and rejoin
- It can also happen by wrong chromosome pairing followed by recombination
- Translocation is usually reciprocal because every chromosome end must have a telomere
- Chromosome ends without telomeres are quickly destroyed; this is usually lethal
- Even without recombination, translocation heterozygotes mis-segregate their chromosomes in meiosis
- Translocations behave as underdominant traits
- Only drift or strong selection can allow one to spread

Practice problem

What, if any, problems arise for:

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

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 heterozygote? *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

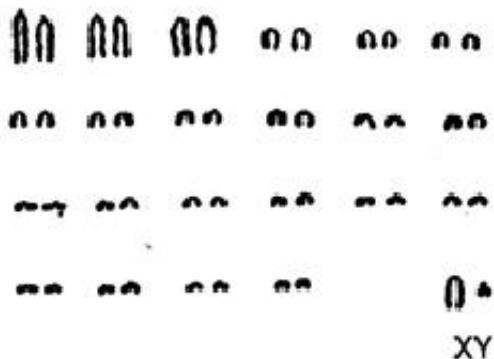
Chromosome fusion probably explains why domestic horses have 64 chromosomes. . .



. . . 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

- Chromosome fissions and fusions may begin as translocations where the reciprocal product is lost
- Like other chromosomal rearrangements, they are generally underdominant
- The heterozygote has trouble segregating its chromosomes in meiosis
- 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

A thought about speciation

- Conventional idea about speciation:
 - Populations adapt differently, so that gene flow between them is disadvantageous
 - They then evolve reproductive isolation because crossbreeding is unfavorable
- Possible alternative idea:
 - Reproductive isolation arises by chance, for example via chromosome rearrangements fixed by drift
 - Now that the gene pools are separate, the populations are free to adapt differently
 - In this view, chromosomal evolution may drive speciation