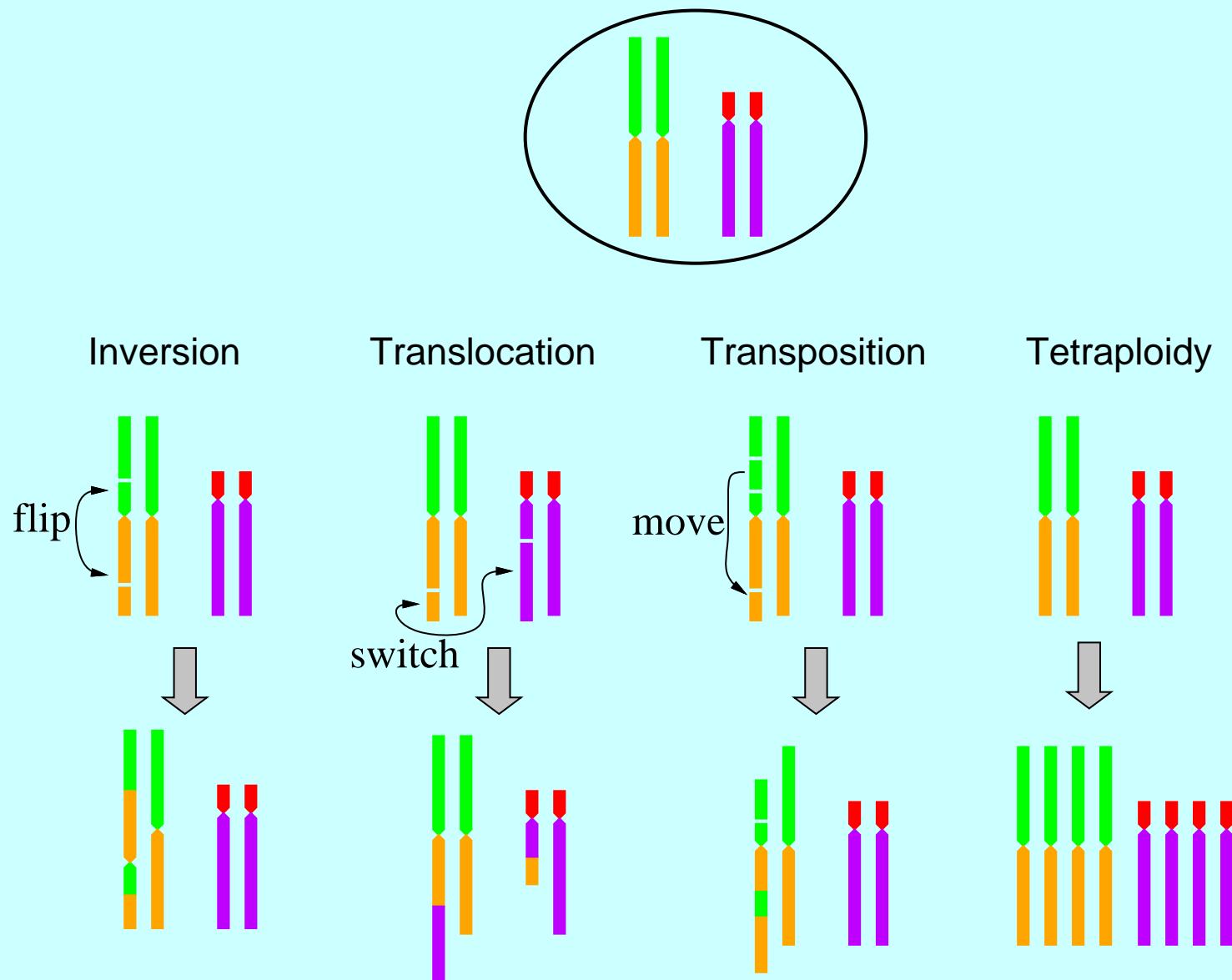


Evolution of chromosomes and genomes

Joe Felsenstein

GENOME 453, Autumn 2013

Chromosome rearrangements



Additional question: when these occur they are rare in a population: how do they spread?

Chironomus banded chromosomes

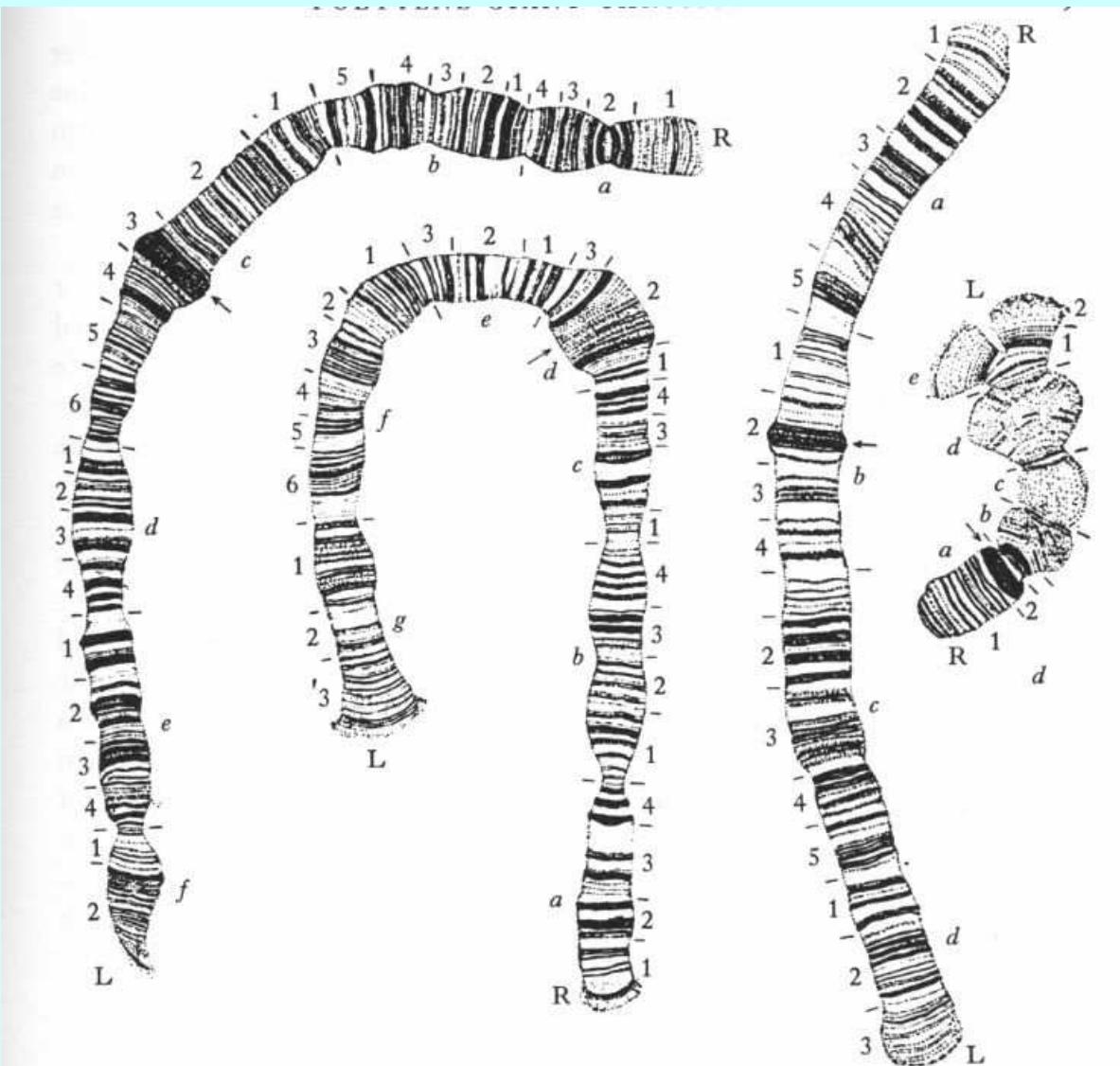
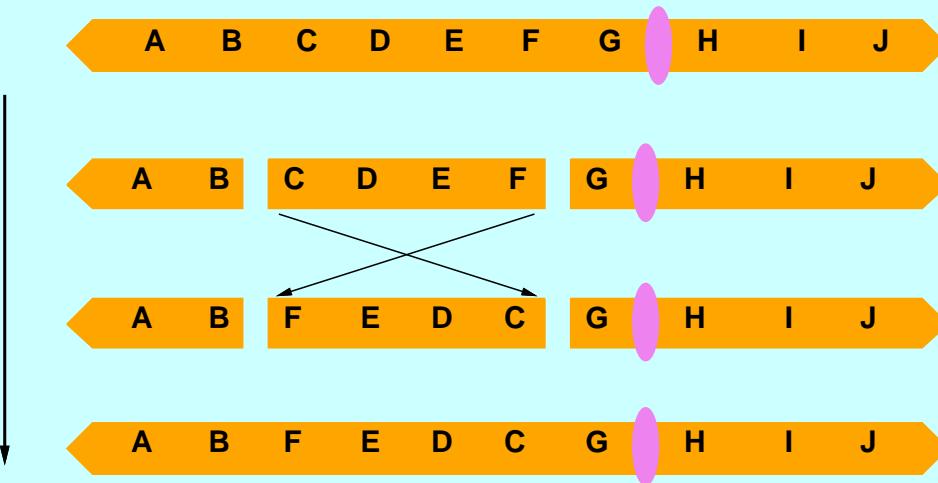


Fig. 4.1. The four polytene elements from a salivary gland nucleus of *Chironomus thummi*, showing the banding. Small arrows indicate the approximate positions of the centromeres. The shortest chromosome has its longer arm largely heterochromatic and the other three elements have short heterochromatic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

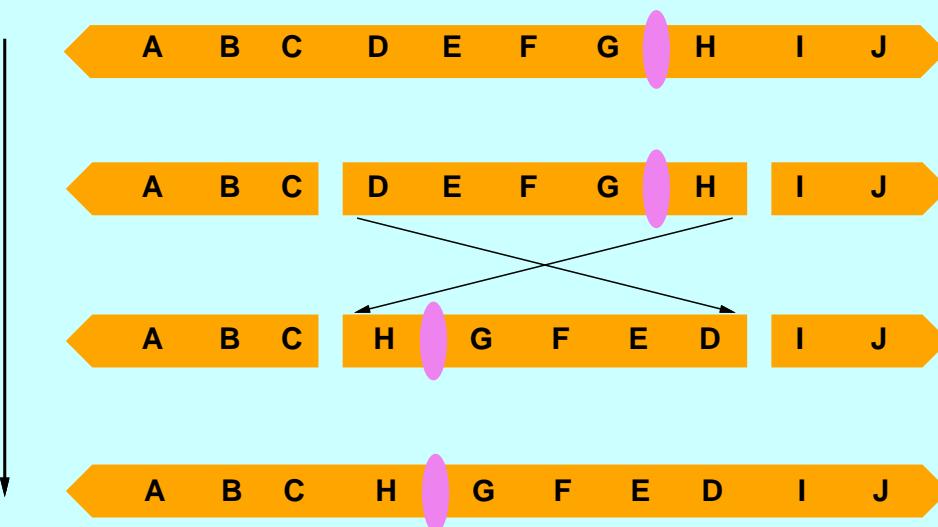


Paracentric and pericentric inversions

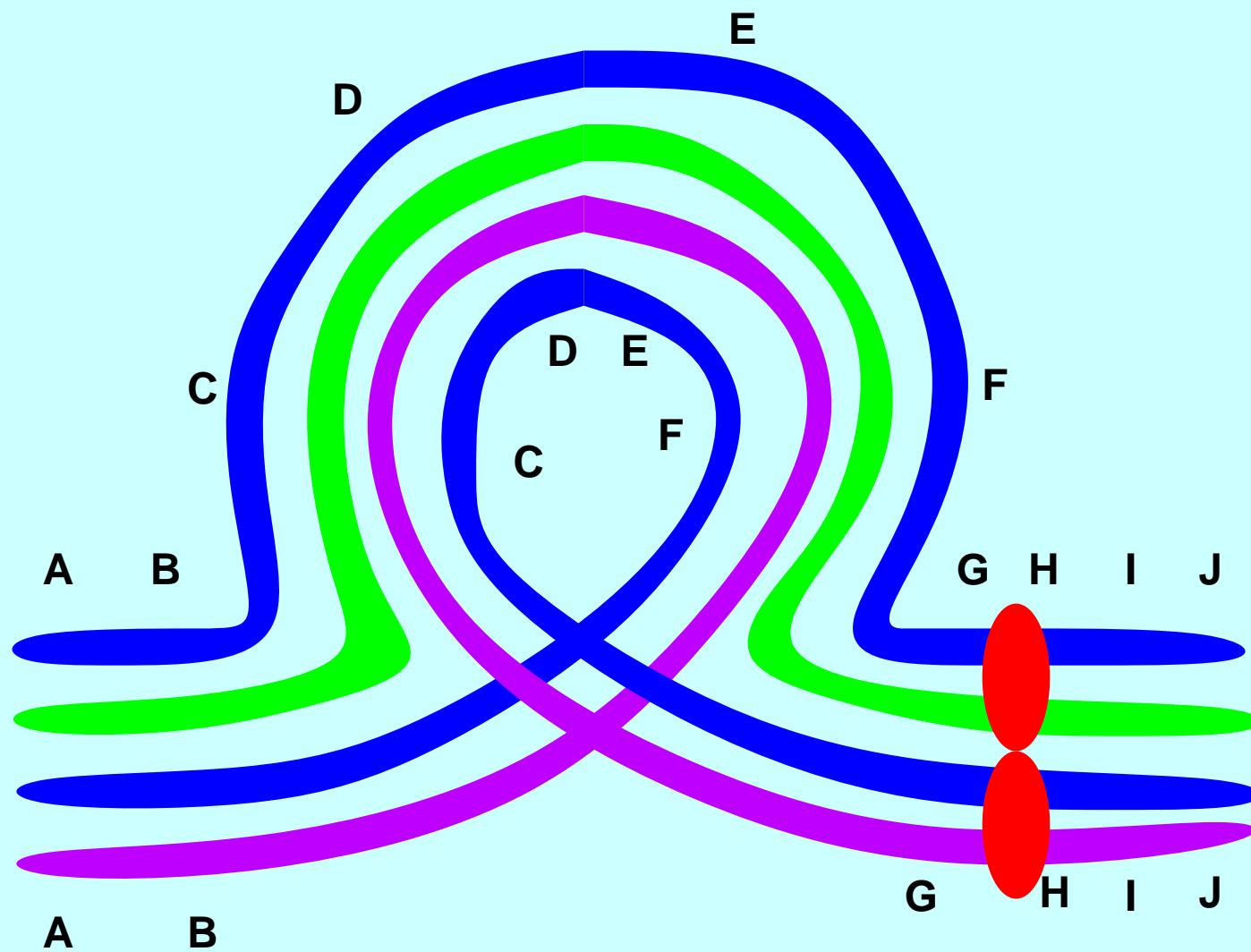
A Paracentric Inversion



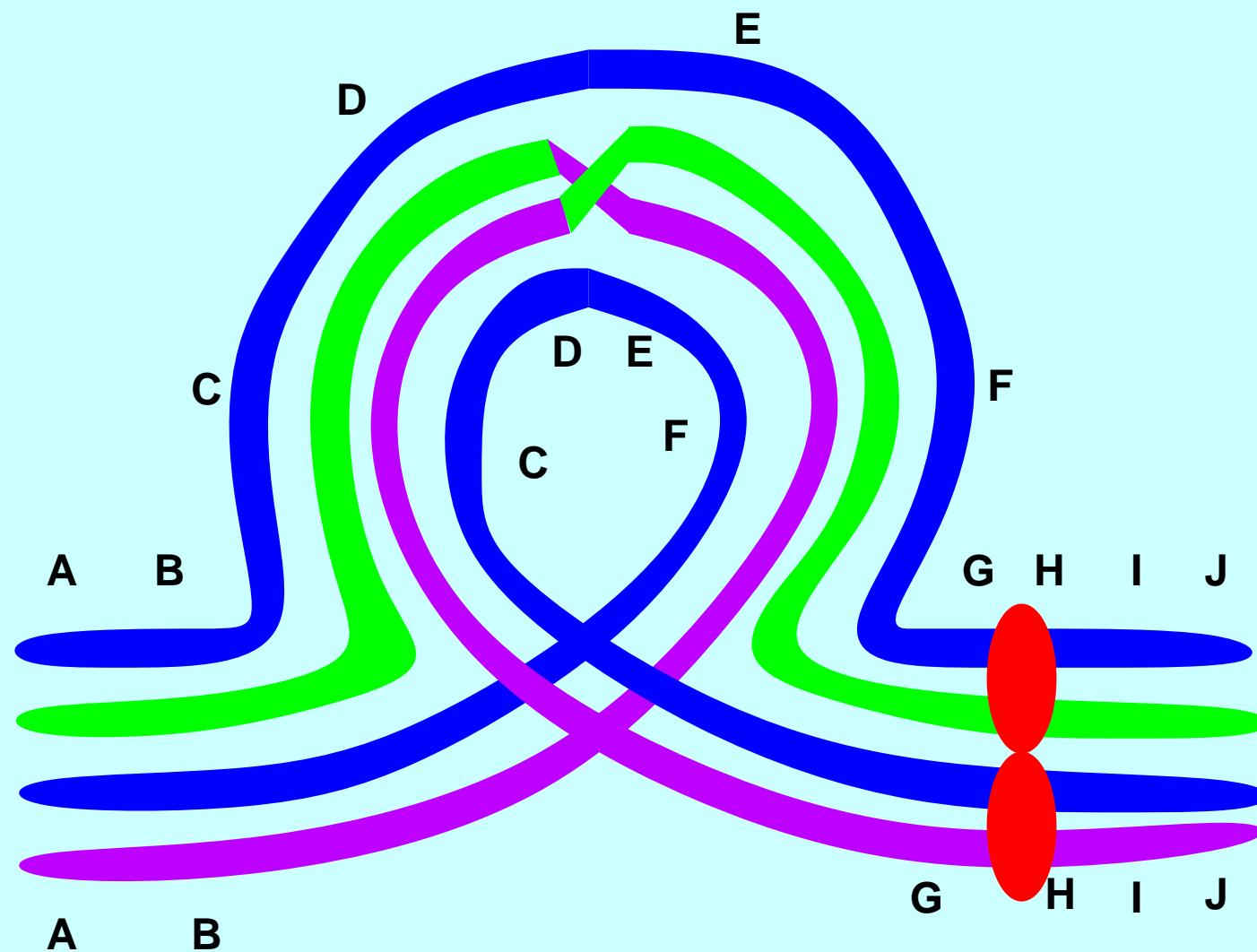
A Pericentric Inversion



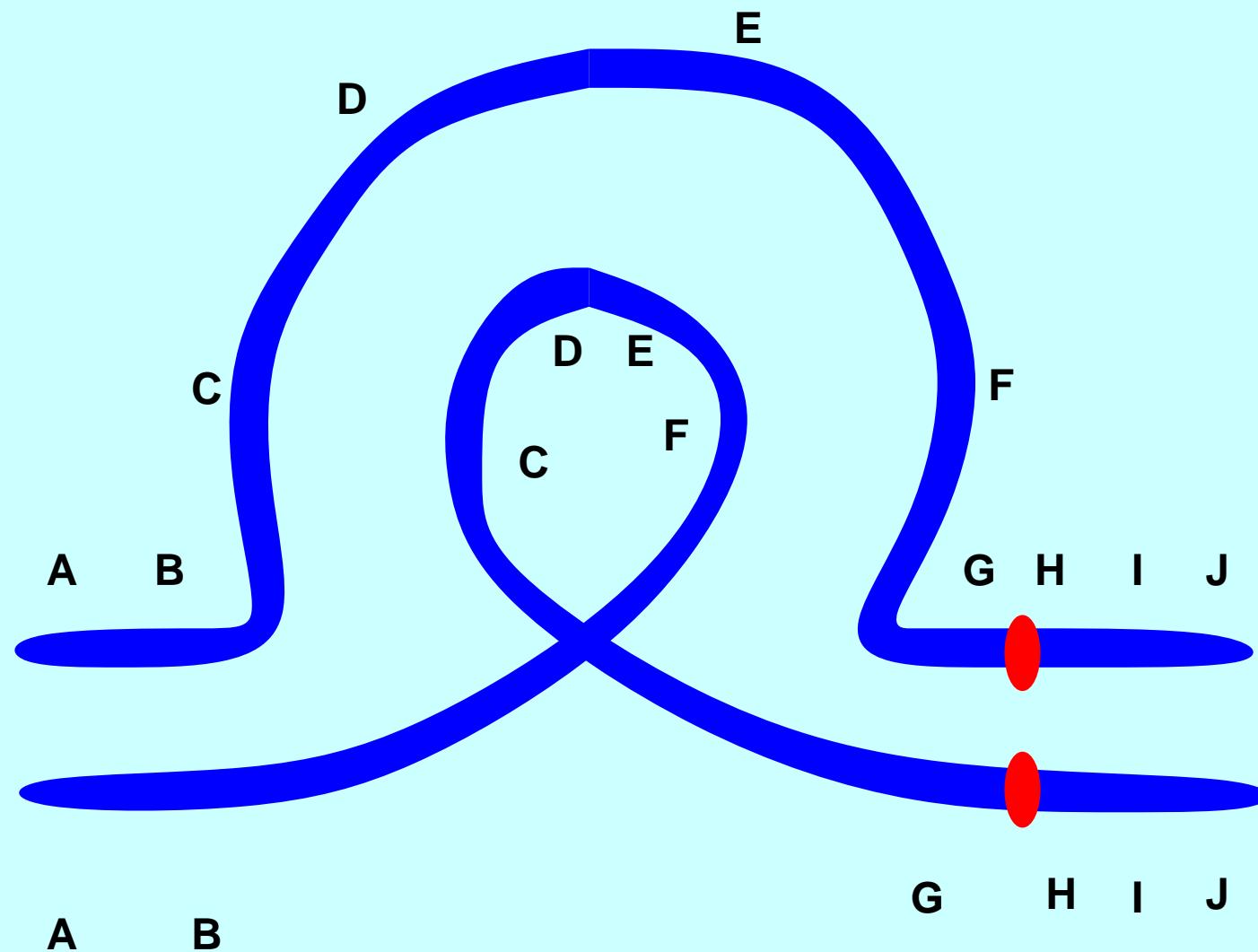
Pairing in a paracentric inversion heterozygote



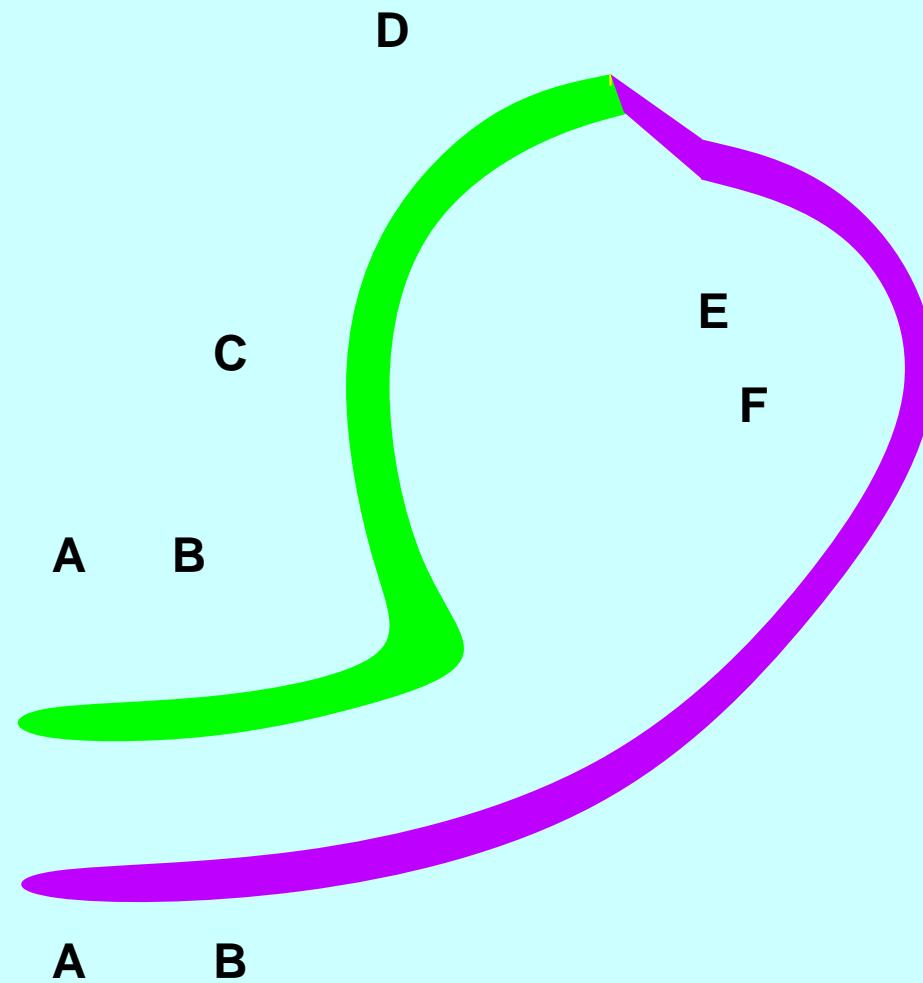
Crossover in a paracentric inversion heterozygote



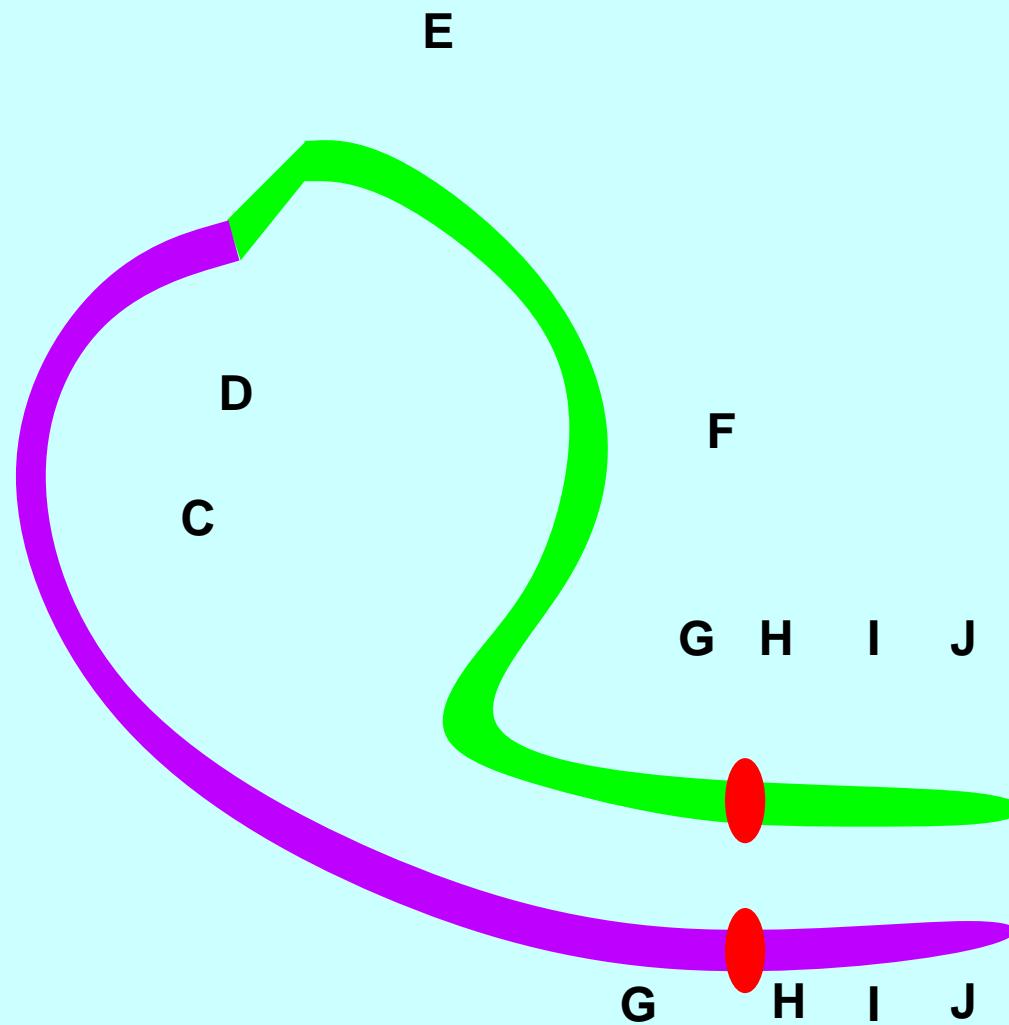
These two chromosomes did not cross over



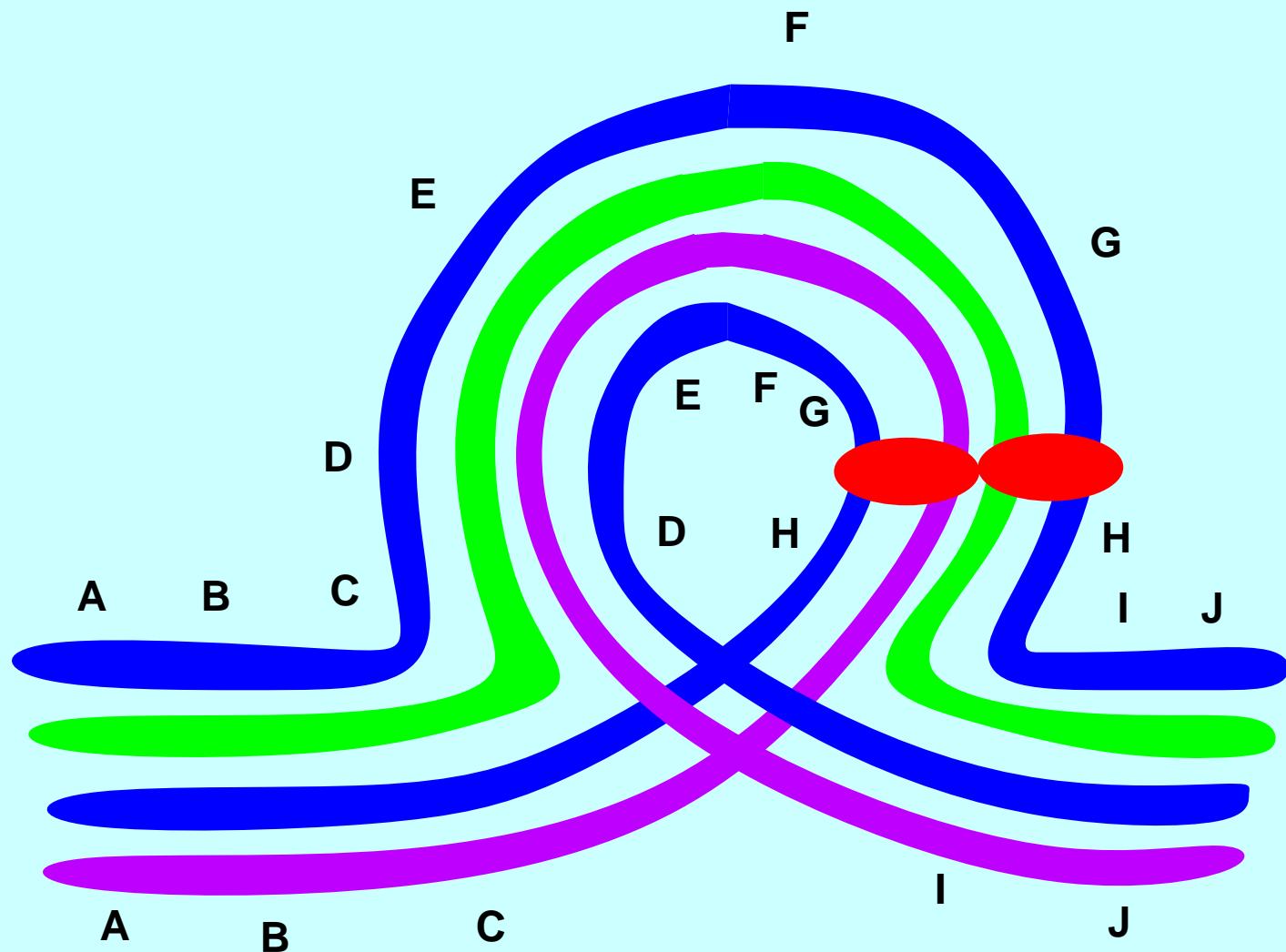
This one did. How will it segregate?



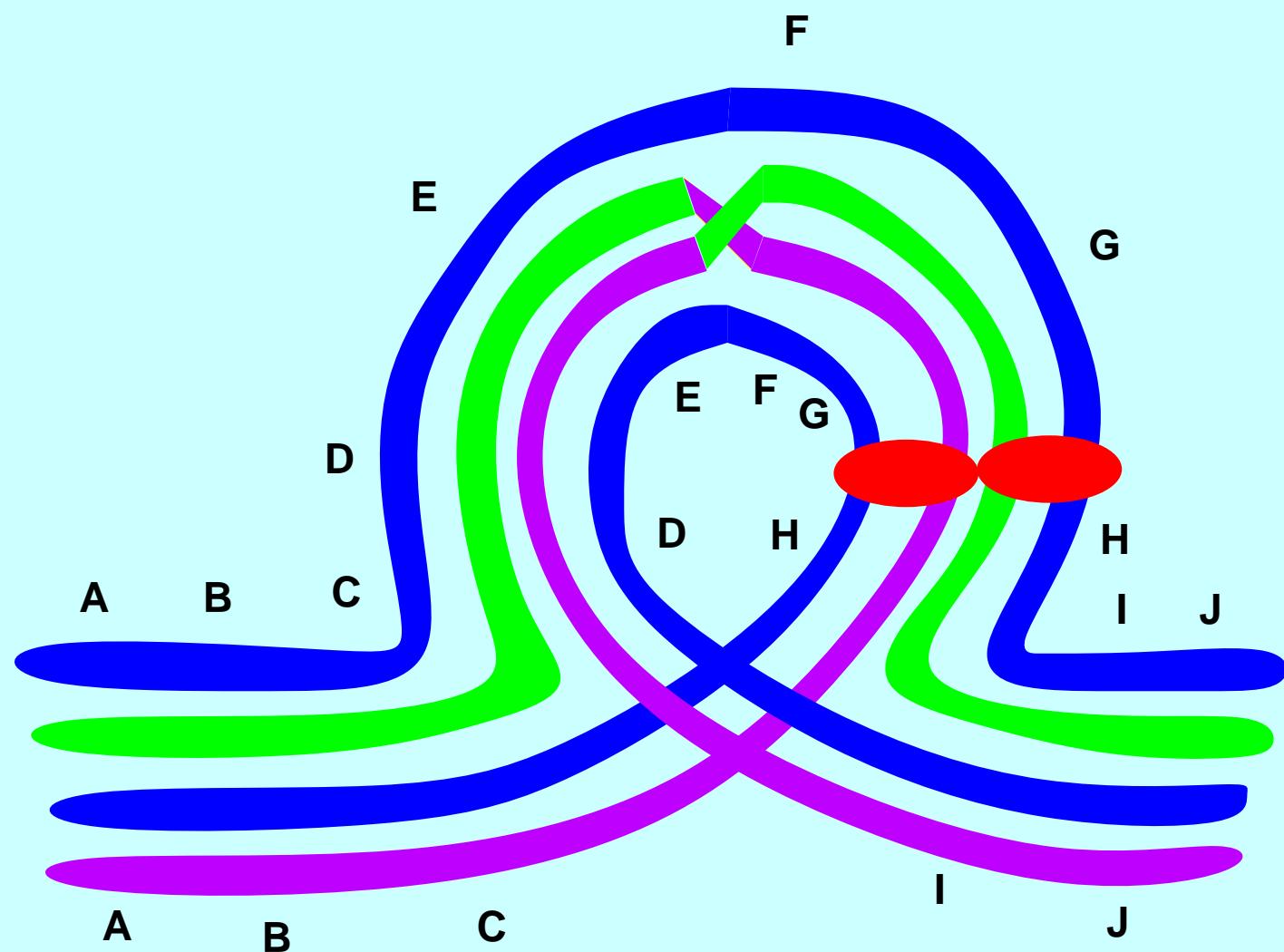
This one did too. How will it segregate?



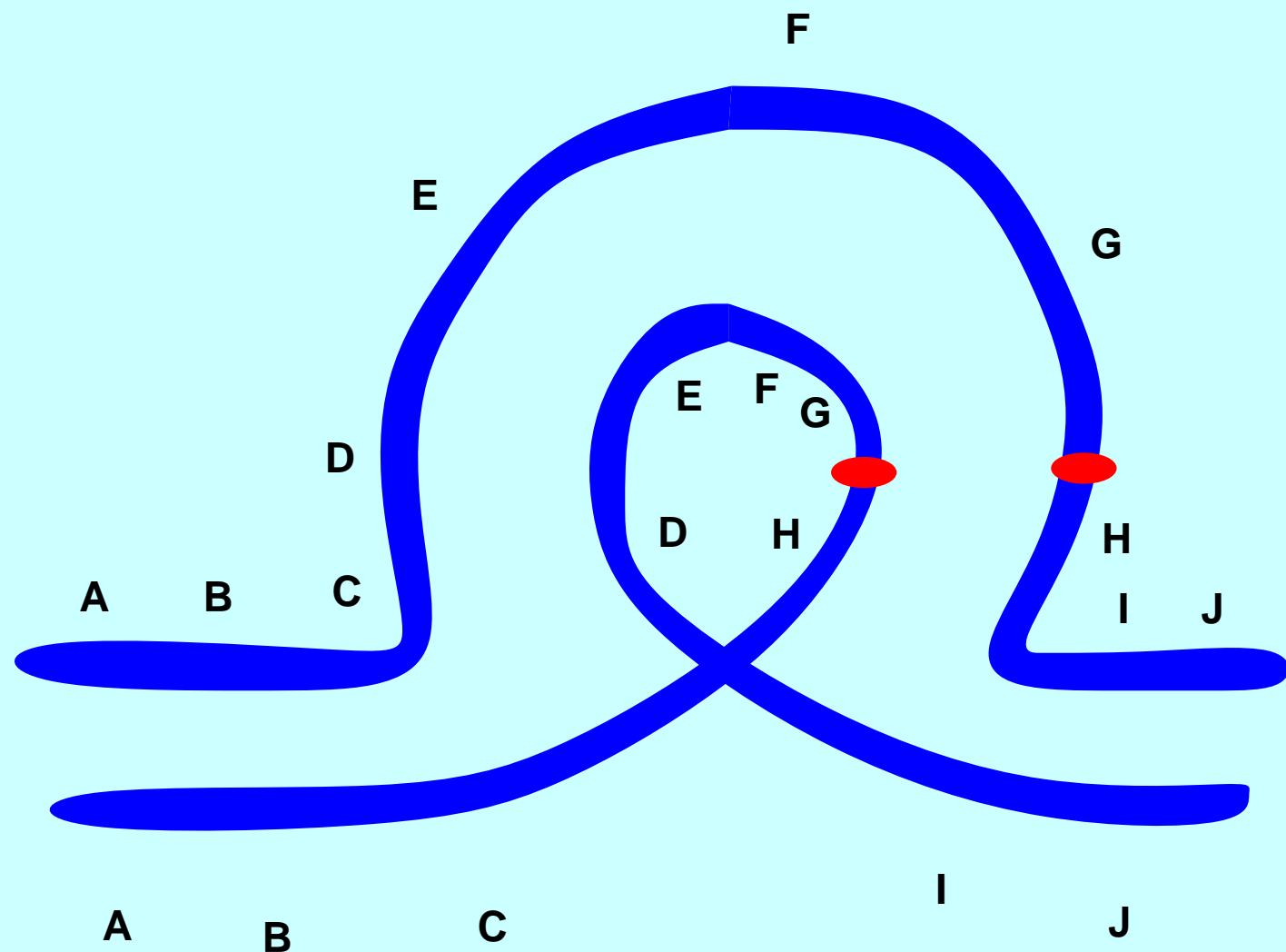
Pairing in a pericentric inversion heterozygote



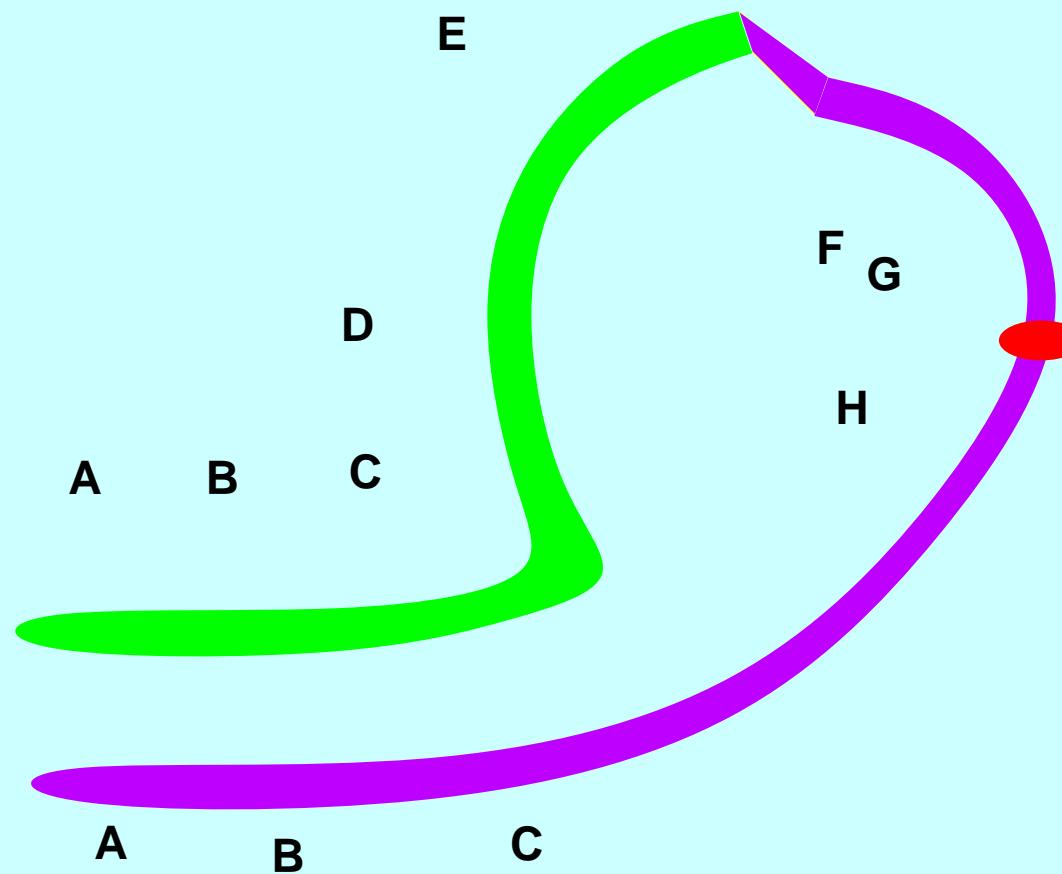
Crossover in a pericentric inversion heterozygote



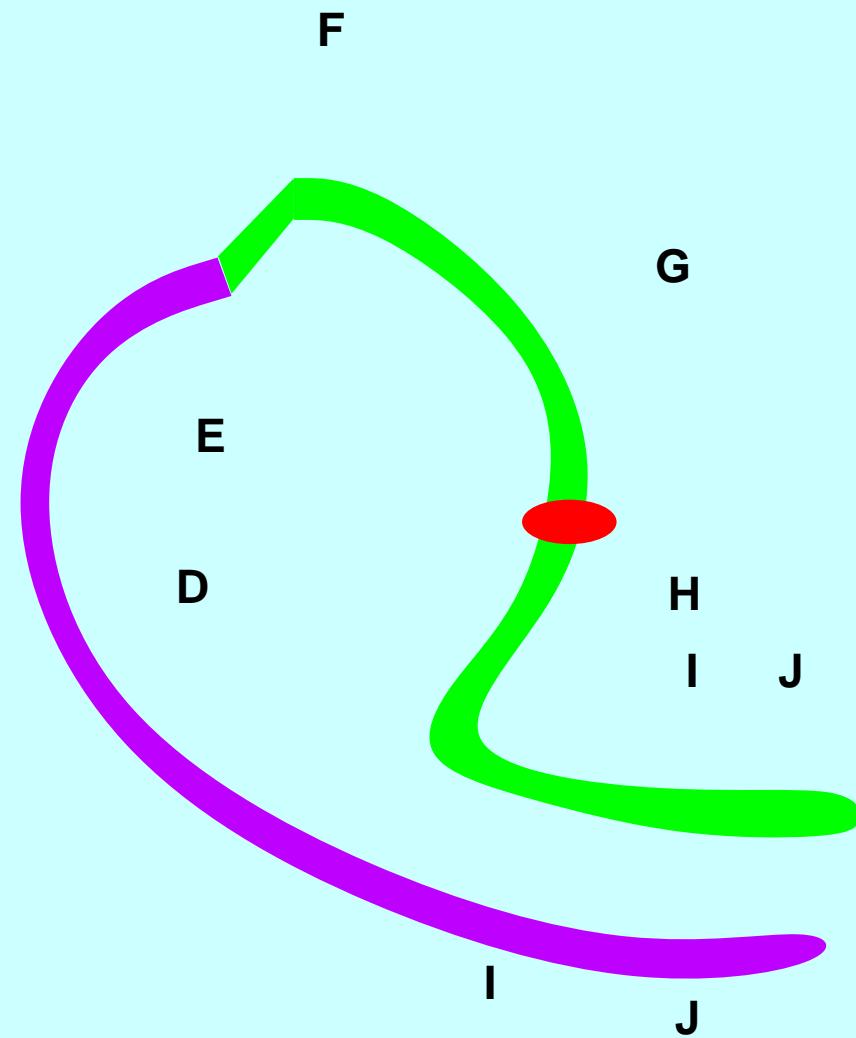
These two products didn't undergo crossing-over



One that did. Is anything deleted? Duplicated?



The other one that did. Anything deleted, duplicated?



A translocation

Before



Breaks



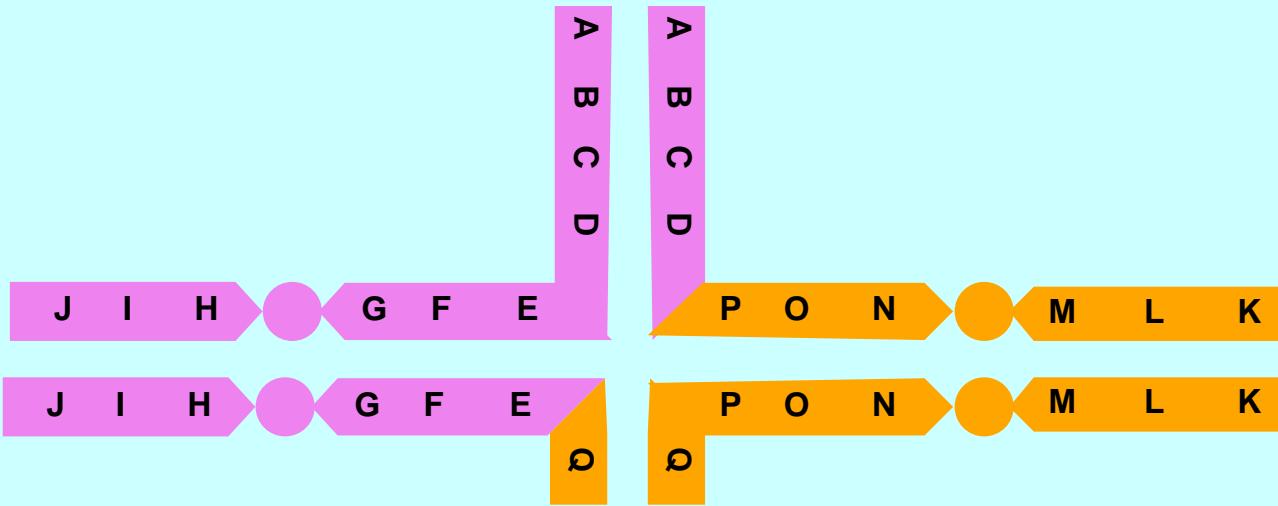
Rearrangement



After

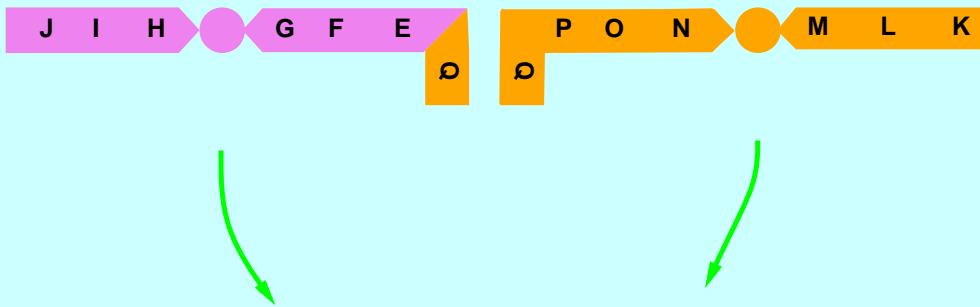
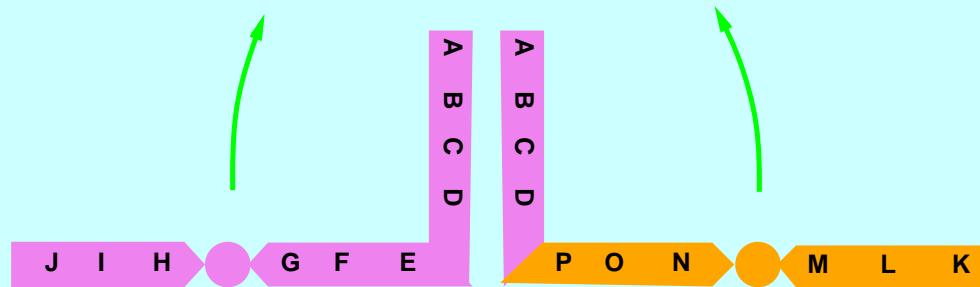


Segregation of translocation heterozygote at first division of meiosis metaphase



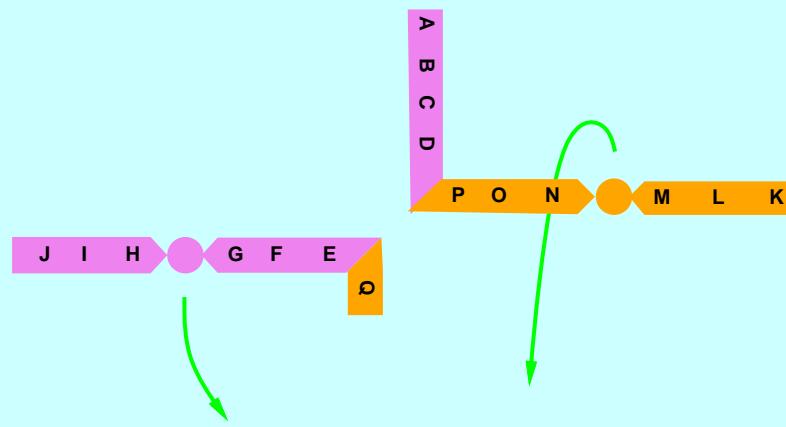
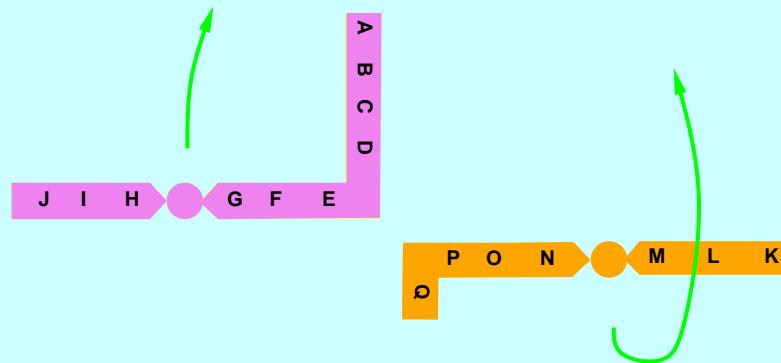
A pair of translocated chromosomes
pairs with a pair of untranslocated chromosomes

Adjacent segregation in translocations



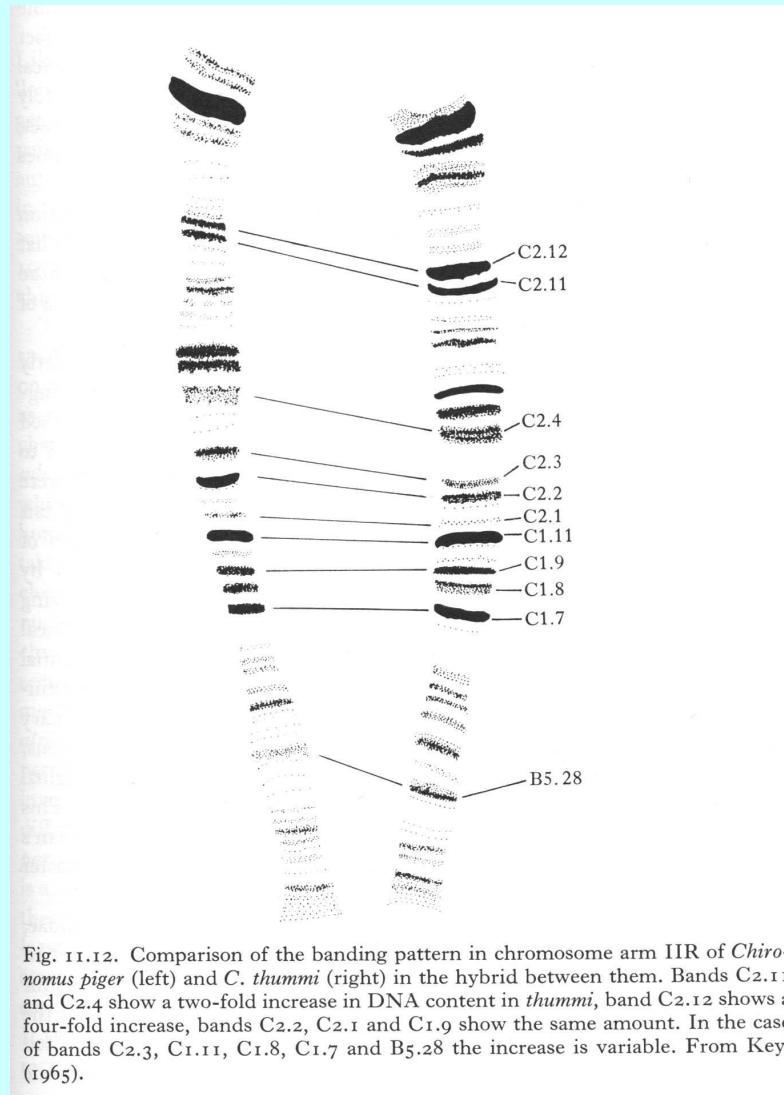
... Leads to deletions and duplications.

Alternate segregation in translocations



... Leads to euploid (normal gene complement) gametes

Banding pattern changes



in two species of Chironomus midges

Polytene Drosophila chromosomes



Chromosome phylogeny of *Drosophila pseudoobscura* etc

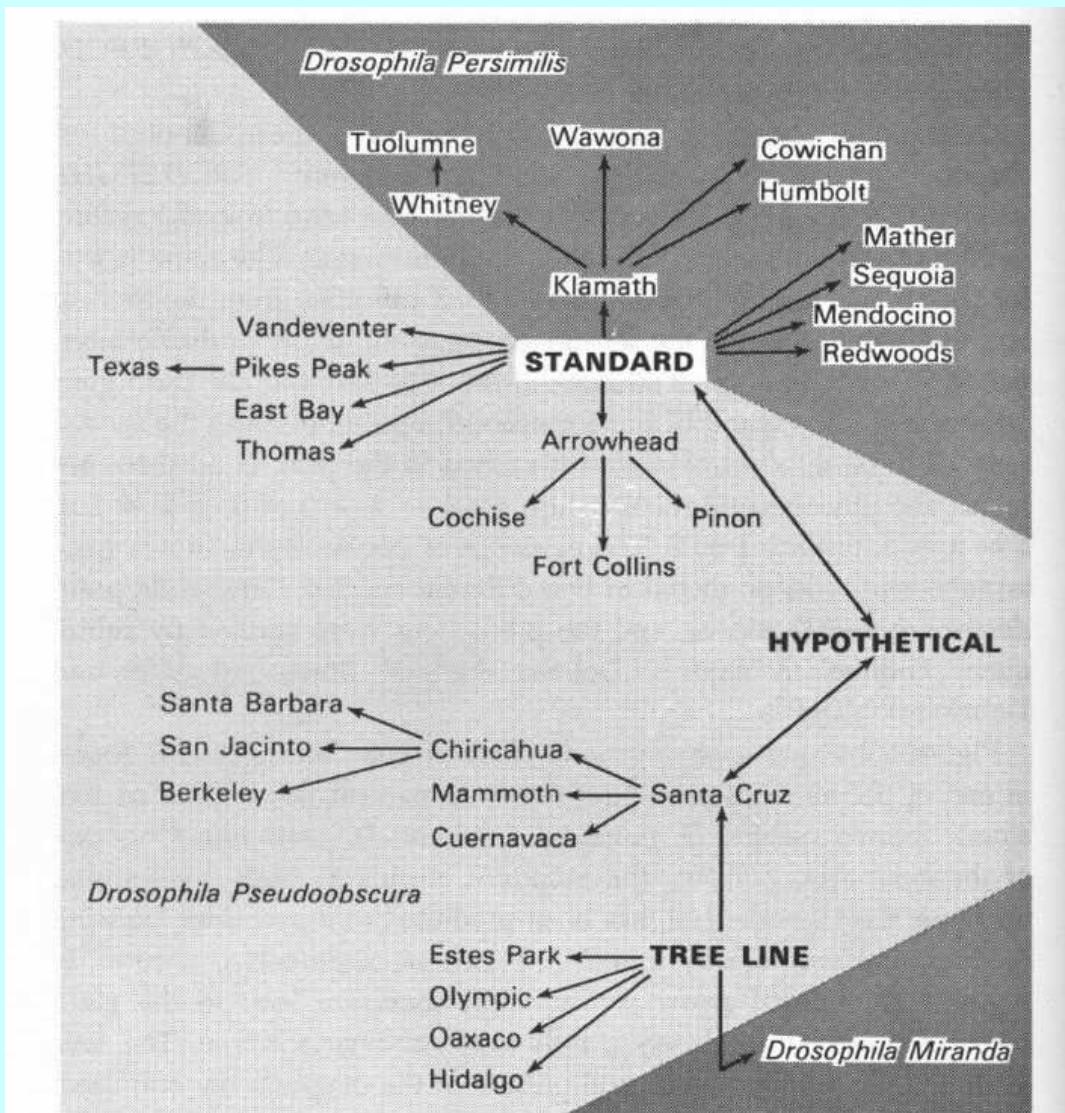
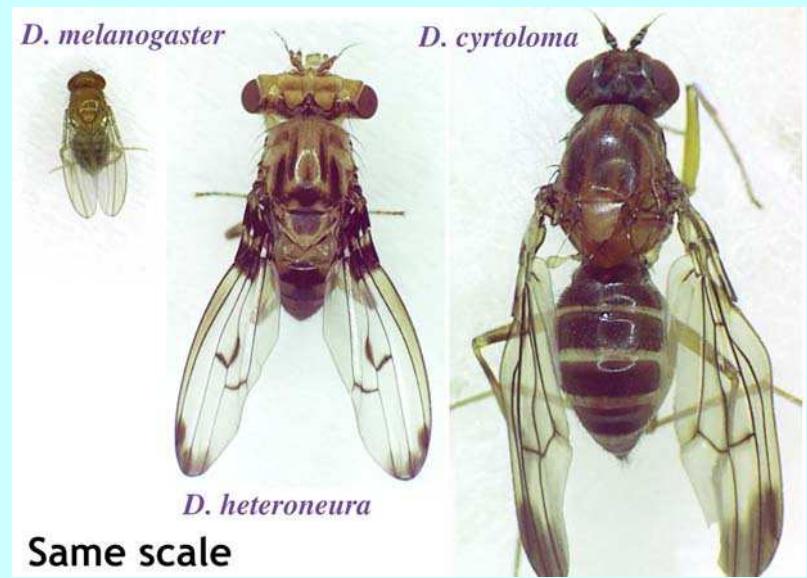
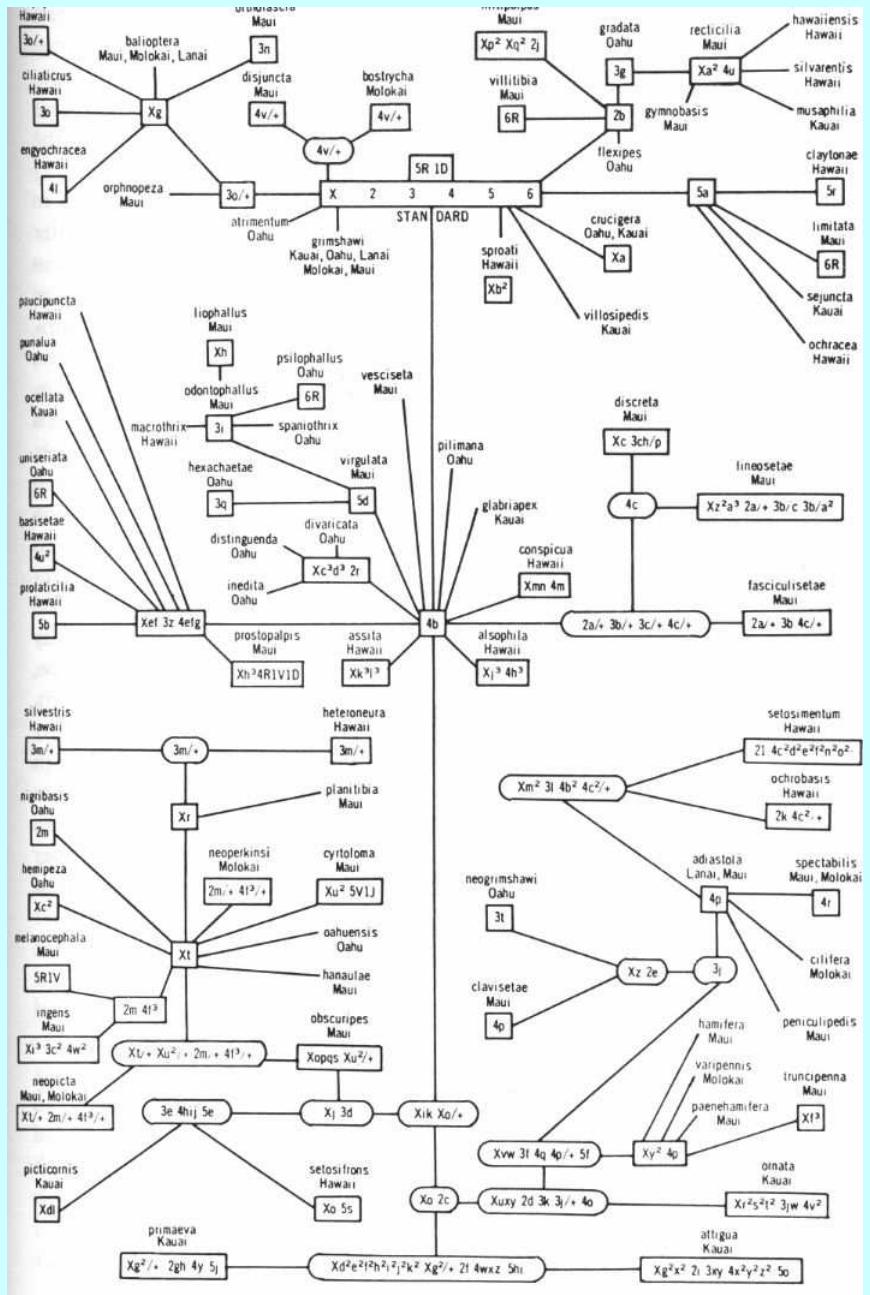


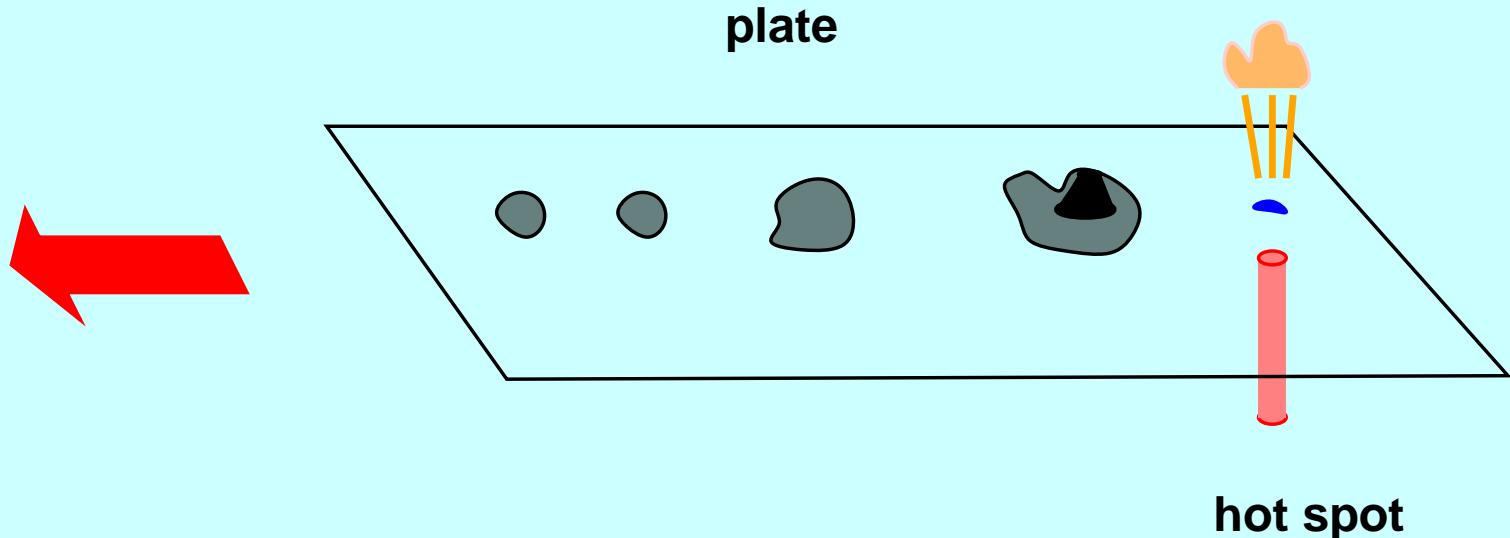
FIGURE 5.2

Phylogenetic relationships of the gene arrangements in the third chromosomes of *Drosophila pseudoobscura*, *D. persimilis*, and *D. miranda*.

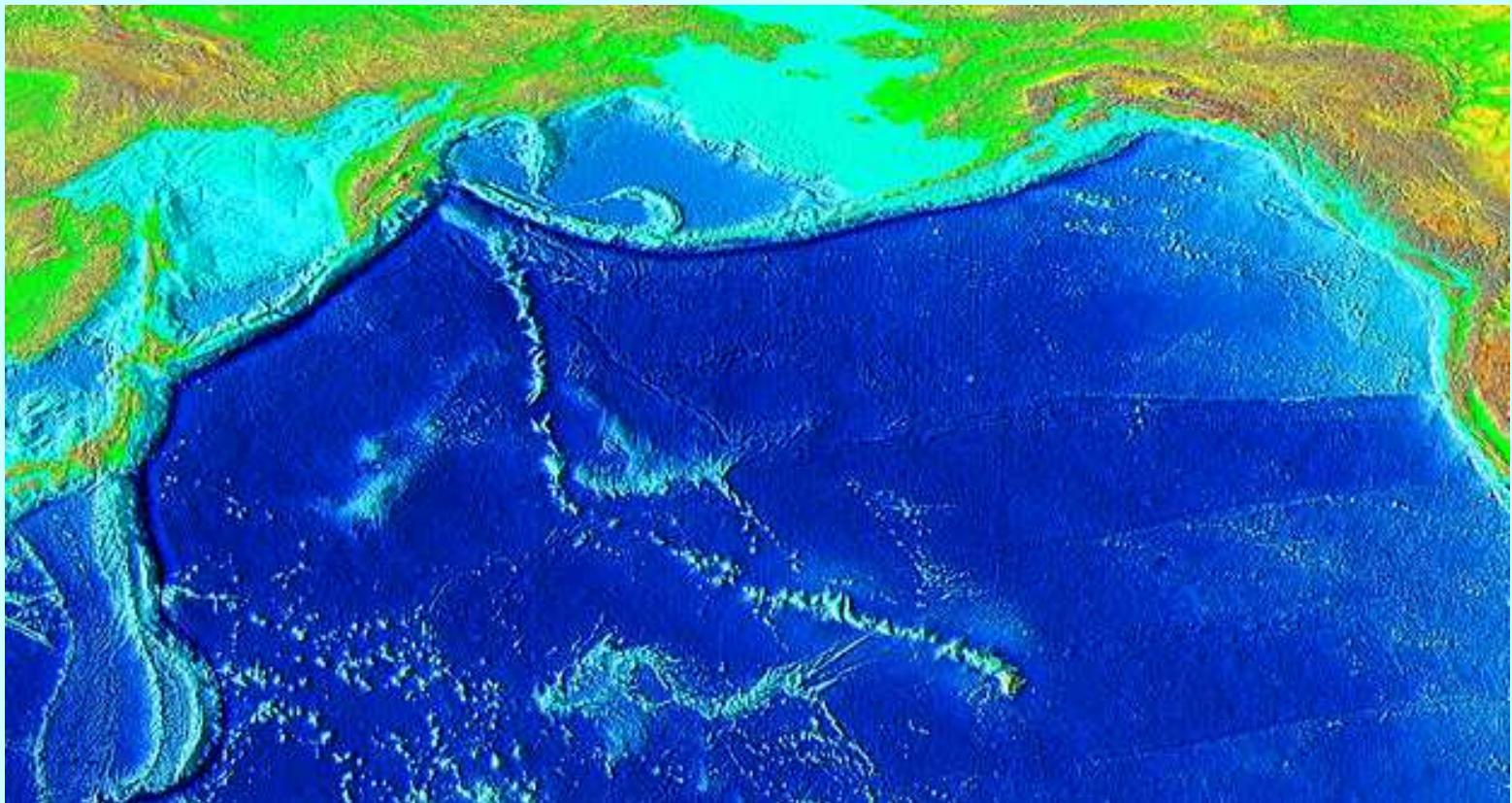
Chromosome phylogeny of Hawaiian Drosophila



Formation of the Hawaiian islands

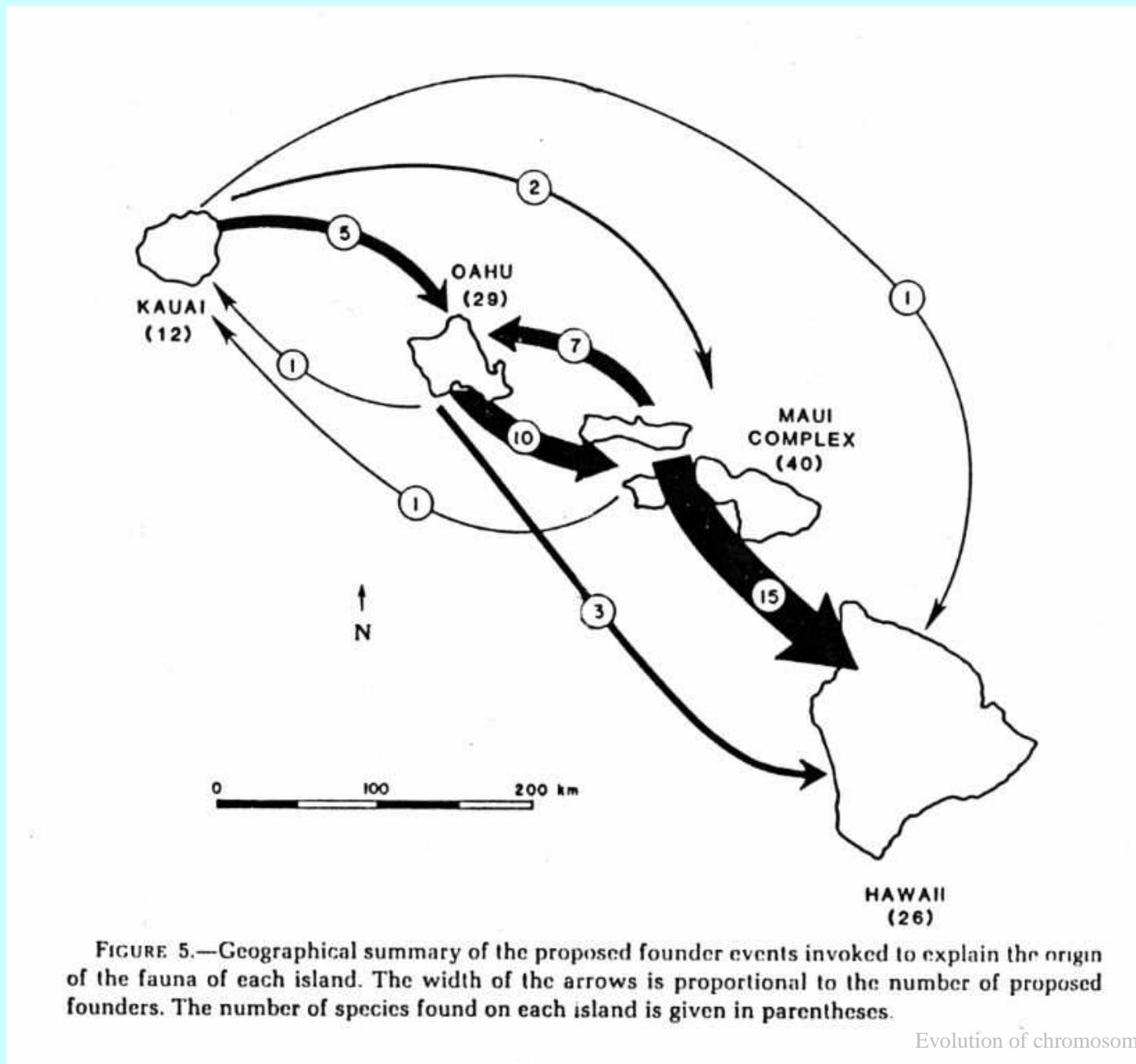


Geology of the Hawaiian islands

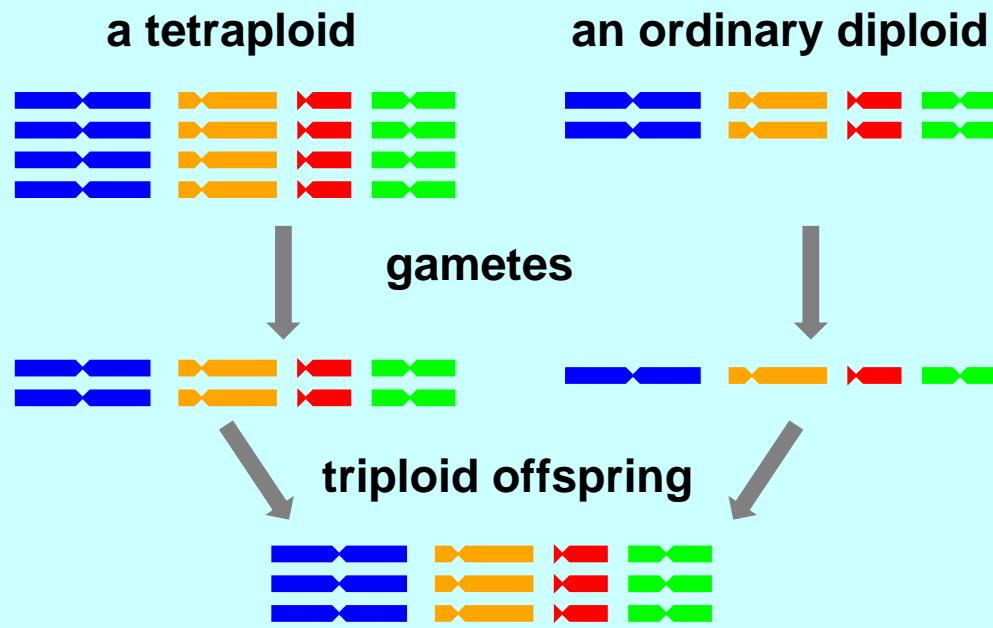


A chain of seamounts leads northwest back across the Pacific as far as the Kamchatka peninsula of Siberia, an estimated 85 million years.

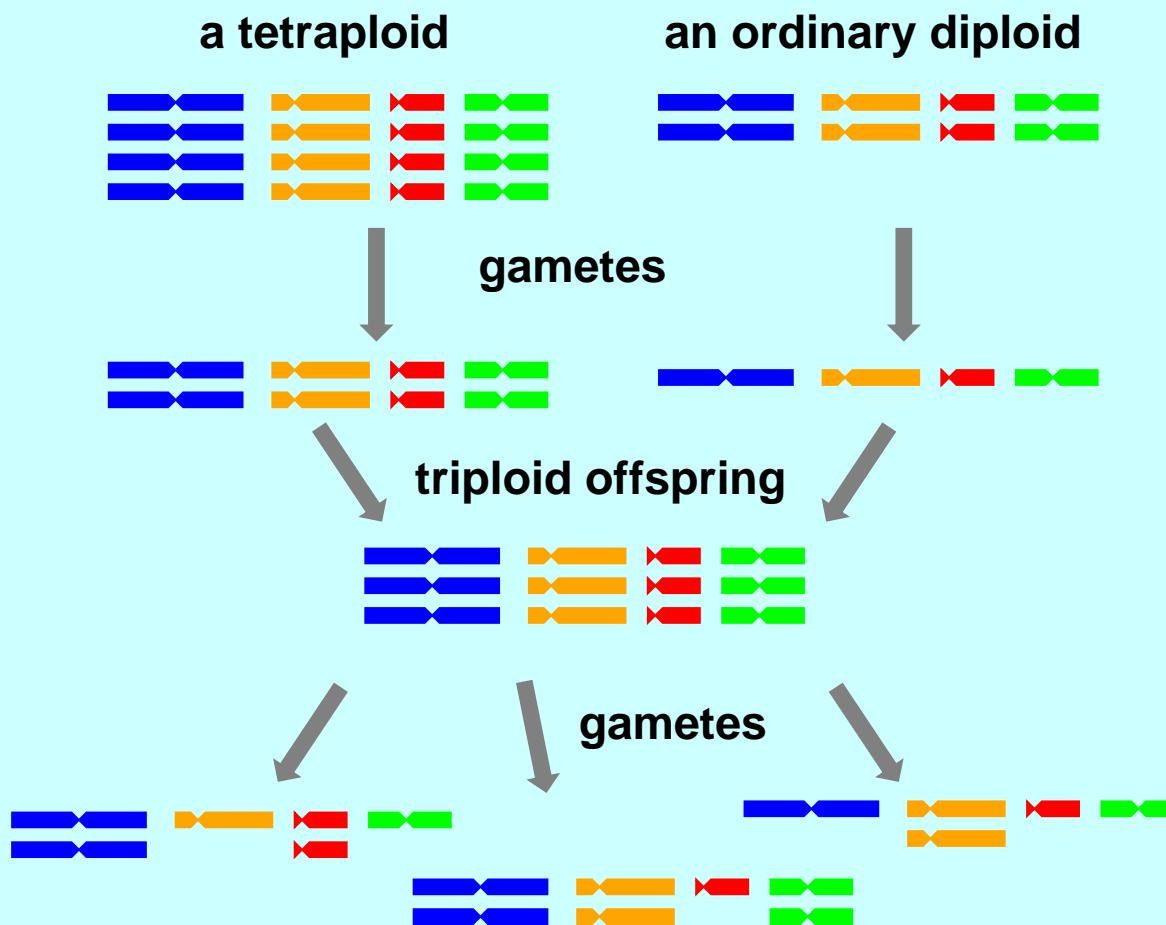
Inferred migration events on the Hawaiian islands



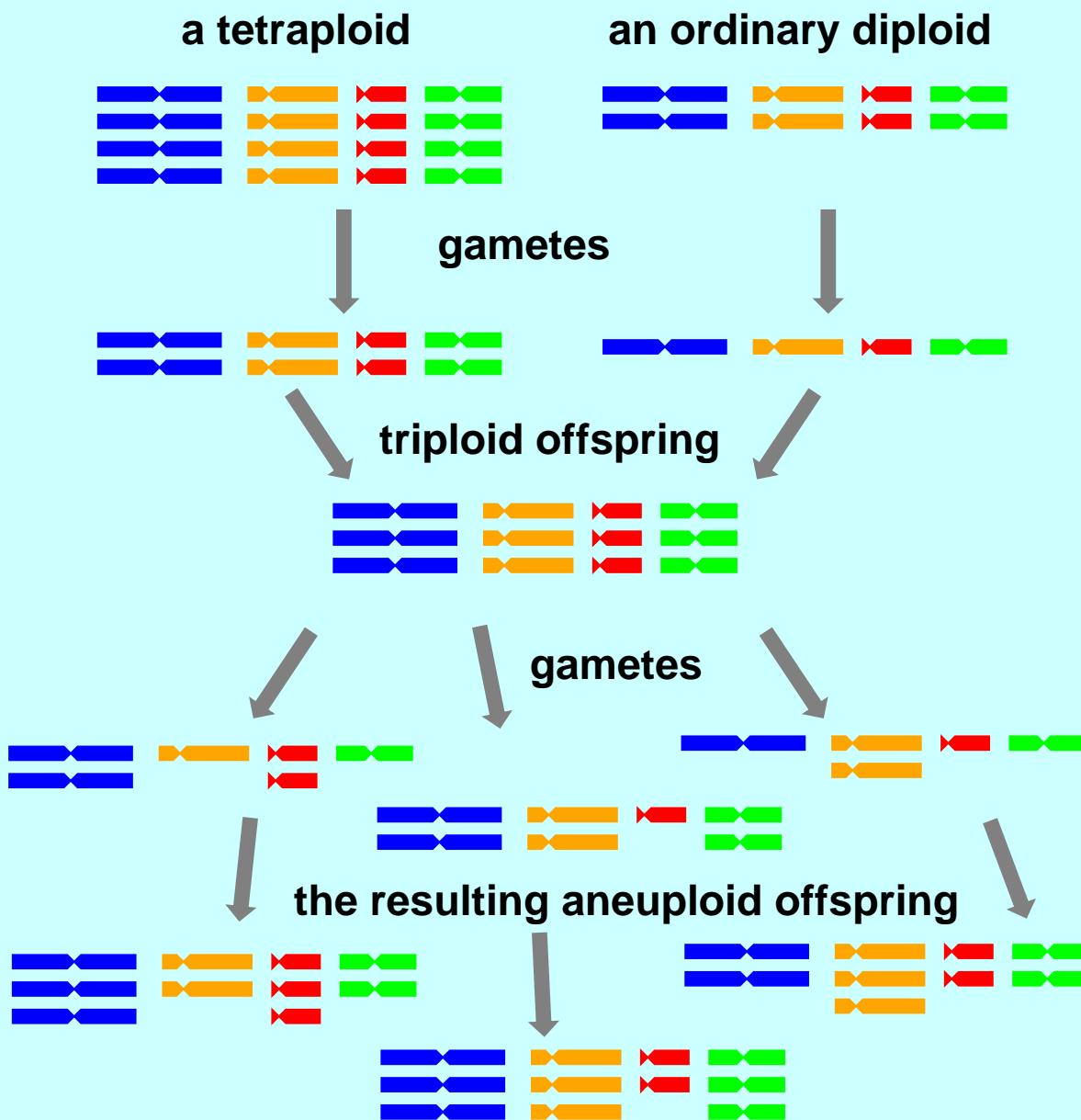
When a tetraploid mates with a diploid



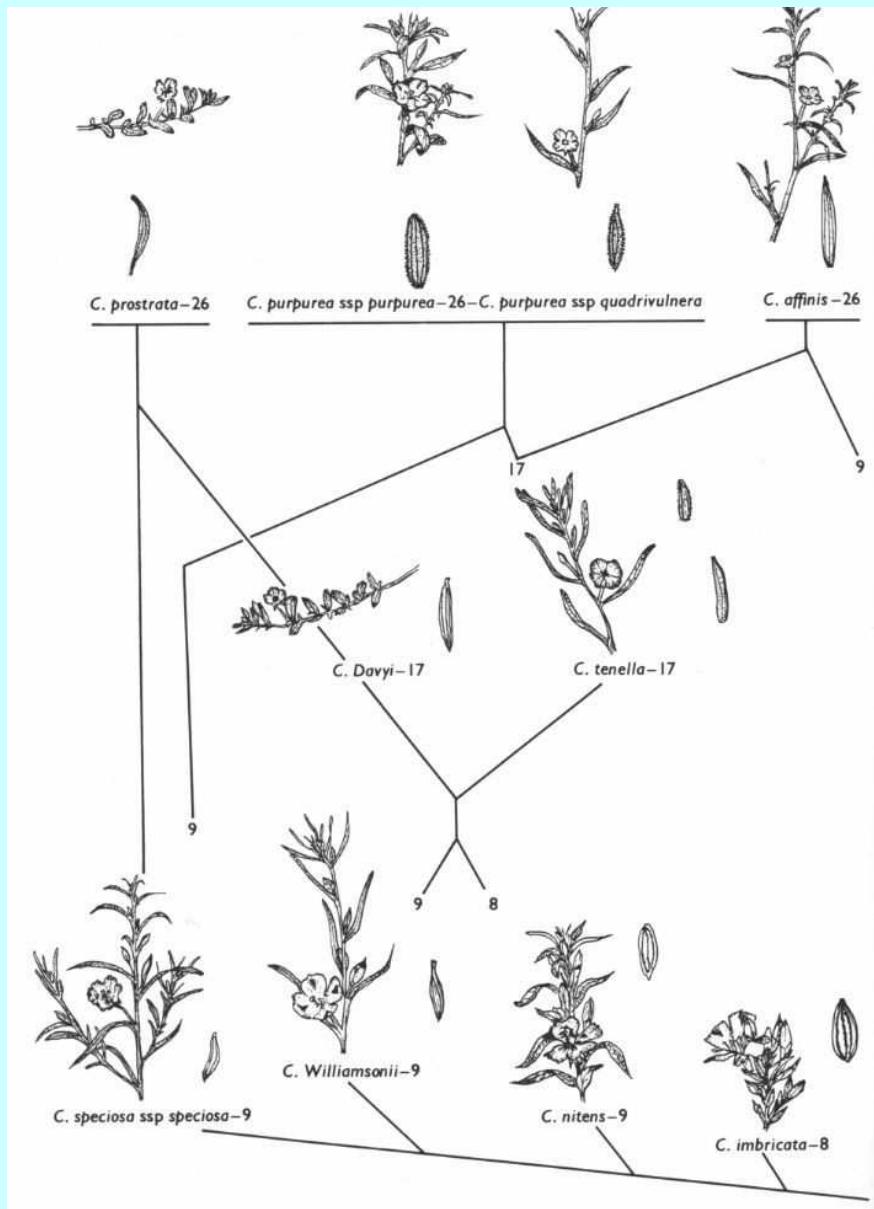
The gametes are aneuploid



And so are the offspring



Polyplloid evolution in Clarkia



C. Williamsonii
Fort Miller Clarkia

The fern Ophioglossum, high number champion



Fig. 2.11 Meiotic prophase (diakinesis) in a sporocyte of *Ophioglossum reticulatum*, showing about 630 bivalents. (From Ninan.¹⁷⁴)



Adders-tongue fern

Highest number is in this genus, about 1260 chromosomes, or 630 pairs.

The ant Myrmecia, low chromosome number champion

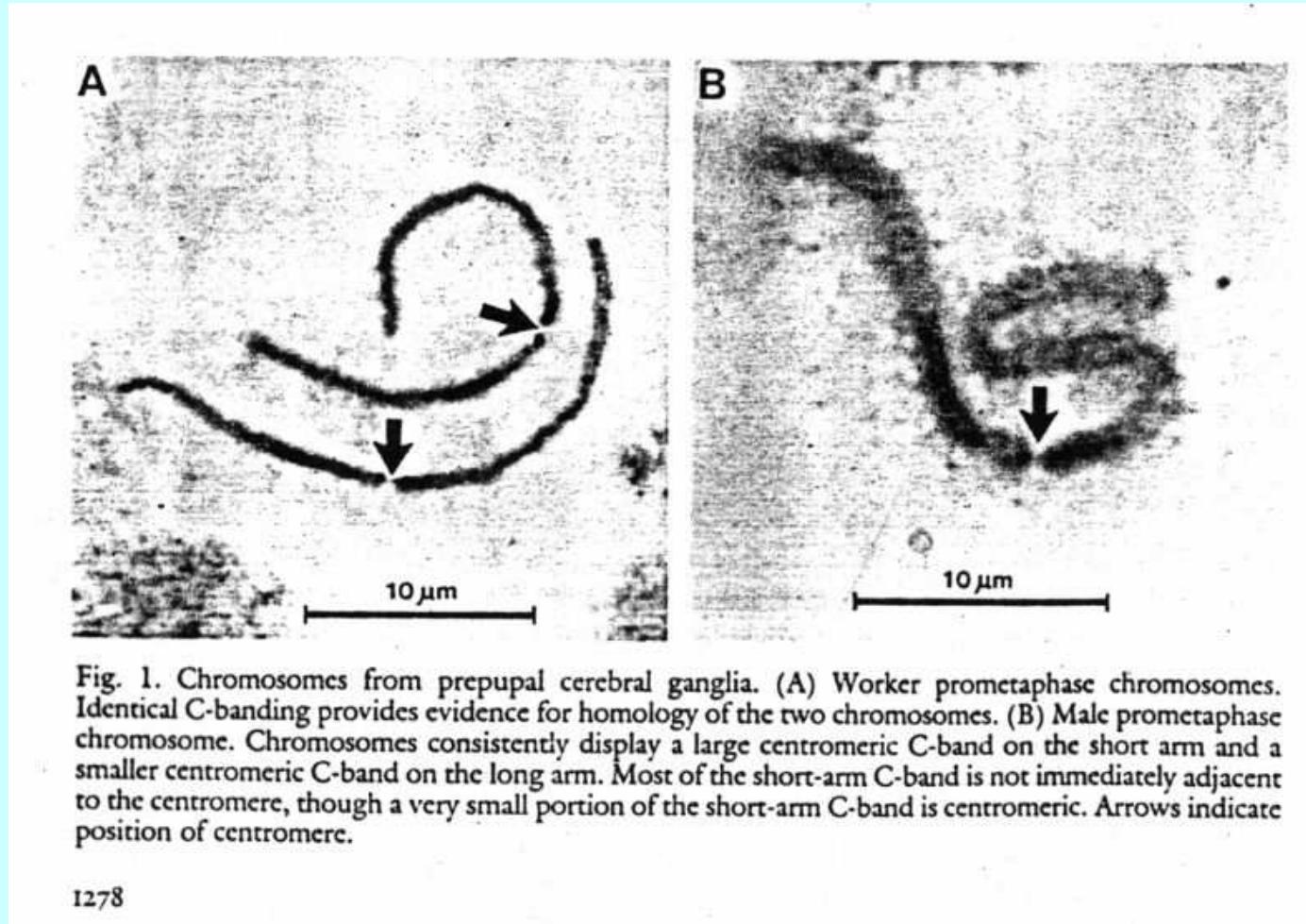


Fig. 1. Chromosomes from prepupal cerebral ganglia. (A) Worker prometaphase chromosomes. Identical C-banding provides evidence for homology of the two chromosomes. (B) Male prometaphase chromosome. Chromosomes consistently display a large centromeric C-band on the short arm and a smaller centromeric C-band on the long arm. Most of the short-arm C-band is not immediately adjacent to the centromere, though a very small portion of the short-arm C-band is centromeric. Arrows indicate position of centromere.

1278



Myrmecia pilosula, the “Jack-jumper ant”

The (temporary?) death of junk DNA

An integrated encyclopedia of DNA elements in the human genome

The ENCODE Project Consortium

Affiliations | Contributions Corresponding author

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PDF



Citation



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Article metrics

Abstract

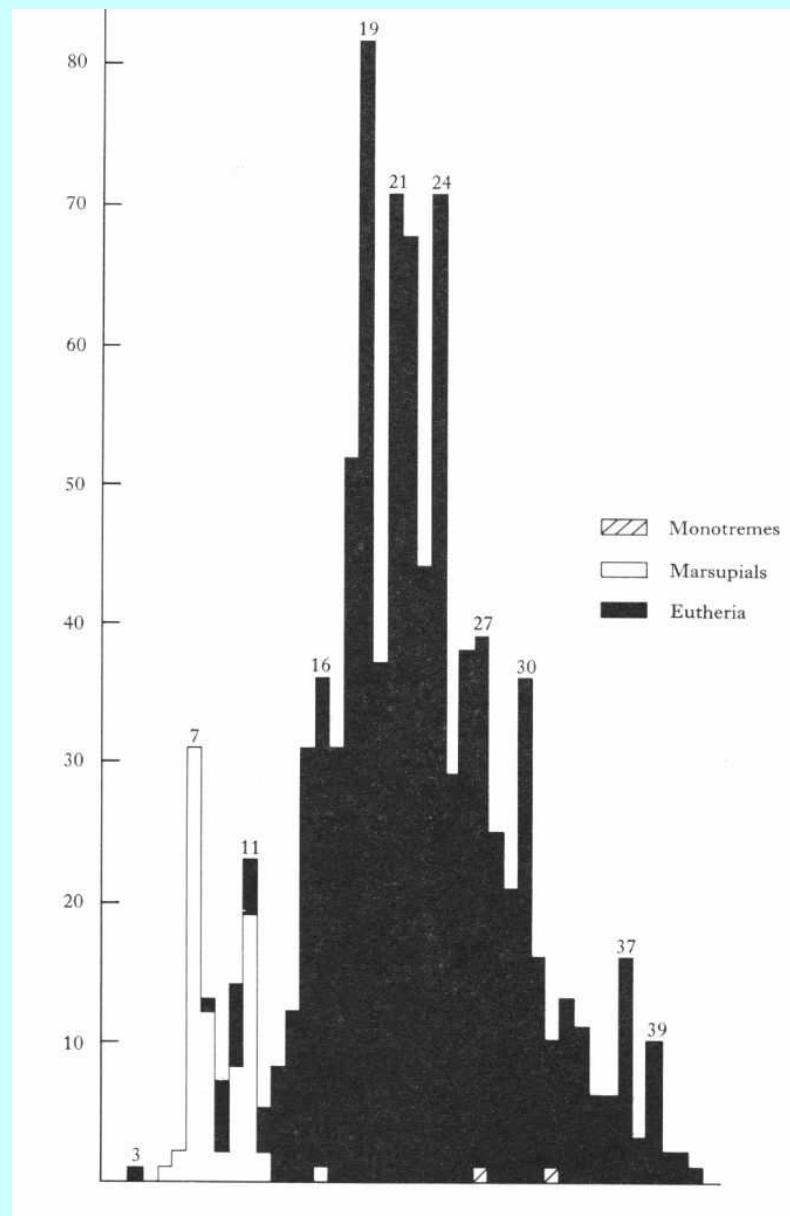
Abstract · Introduction · ENCODE data production and initial analyses · ENCODE data integration with known genomic features · Genome-wide integration · Insights into human genomic variation · Rare variants, individual genomes and somatic variants · Common variants associated with disease · Concluding remarks · Methods · References · Acknowledgements · Author information · Supplementary information · Comments

The human genome encodes the blueprint of life, but the function of the vast majority of its nearly three billion bases is unknown. The Encyclopedia of DNA Elements (ENCODE) project has systematically mapped regions of transcription, transcription factor association, chromatin structure and histone modification. These data enabled us to assign biochemical functions for 80% of the genome, in particular outside of the well-studied protein-coding regions. Many discovered candidate regulatory elements are physically associated with one another and with expressed genes, providing new insights into the mechanisms of gene regulation. The newly identified elements also show a statistical correspondence to sequence variants linked to human disease, and can thereby guide interpretation of this variation. Overall, the project provides new insights into the organization and regulation of our genes and genome, and is an expansive resource of functional annotations for biomedical research.

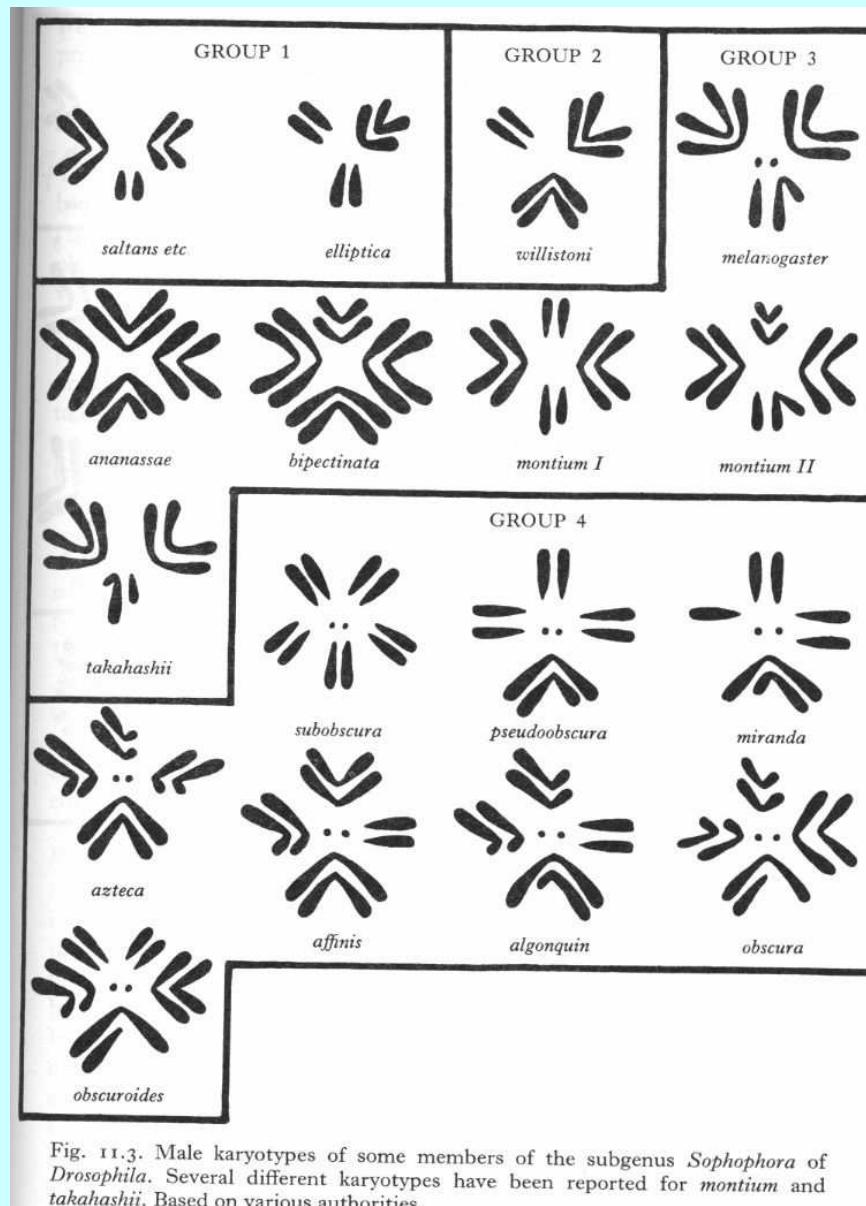
Pros and cons

| Pro | Con |
|--|---|
| <ol style="list-style-type: none">1. Much of the genome is transcribed.2. Transcription factor binding sites are found throughout the genome.3. Just think of all the grants we can apply for to work out the functions of all that DNA! | <ol style="list-style-type: none">1. The Onion Test: do onions really need $5\times$ as much DNA as we do?2. Mutational load would be excessive if the whole genome needs to be kept in its current state3. Most of the genome isn't conserved evolutionarily.4. Annotatable "selfish" DNA elements make up about half of the human genome. |

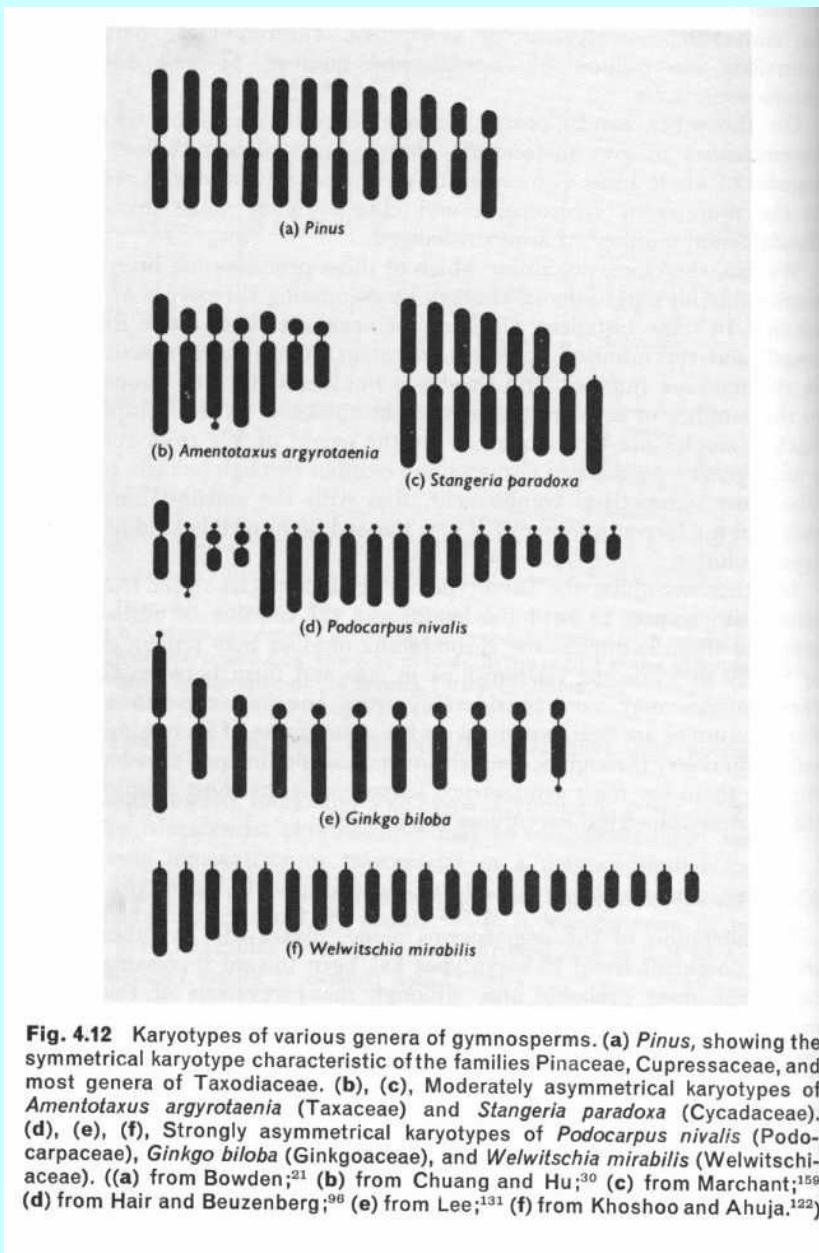
Distribution of chromosome numbers in mammals



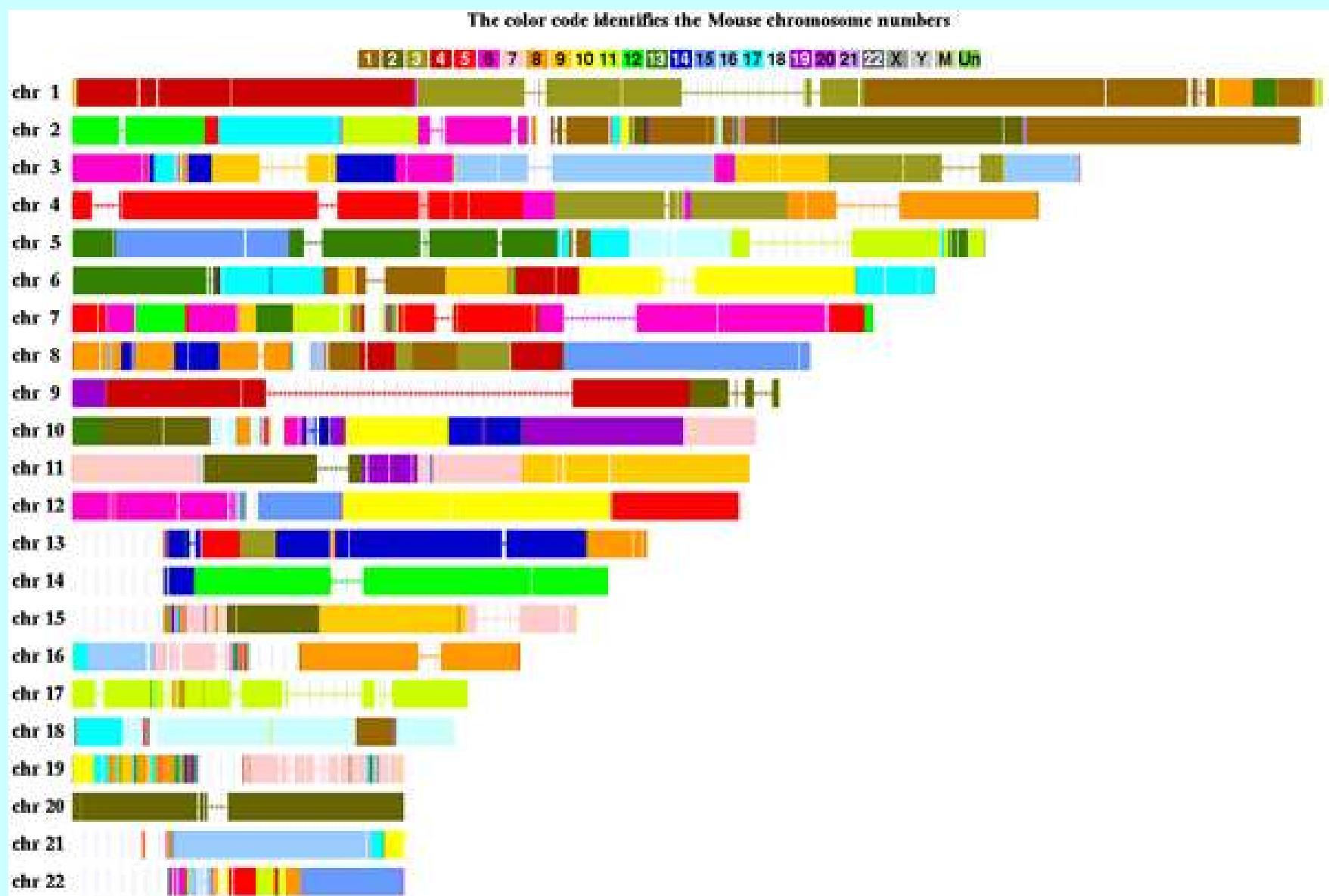
Karyotypes of *Drosophila* species



Karyotypes of gymnosperms



Human and mouse genomes compared



Human and chimp karyotypes compared

