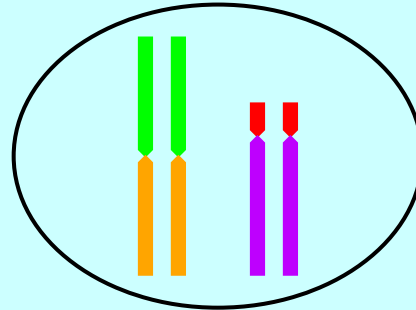


# Evolution of chromosomes and genomes

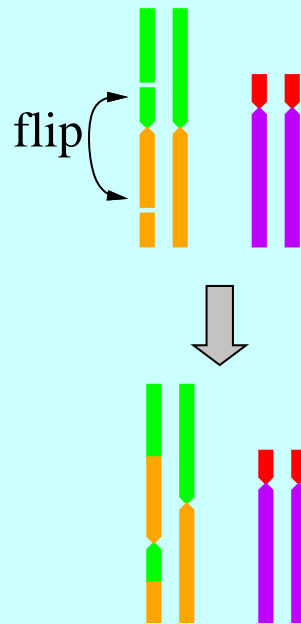
Joe Felsenstein

GENOME 453, Autumn 2013

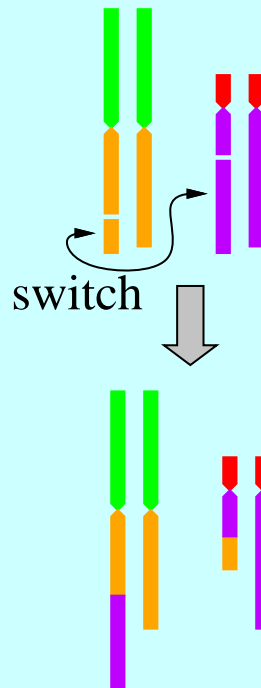
# Chromosome rearrangements



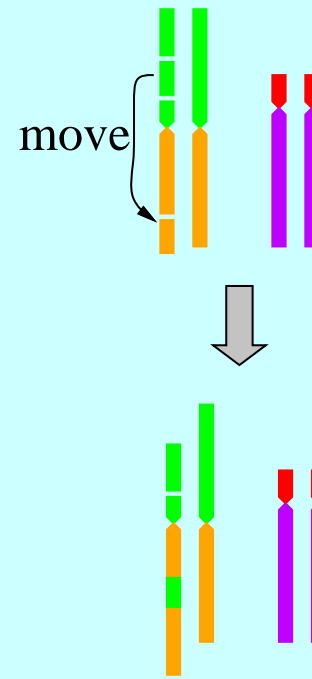
Inversion



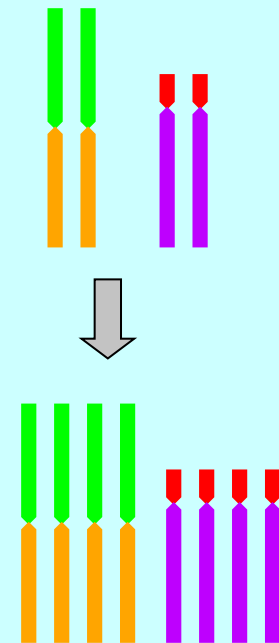
Translocation



Transposition



Tetraploidy



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

# Chironomus banded chromosomes

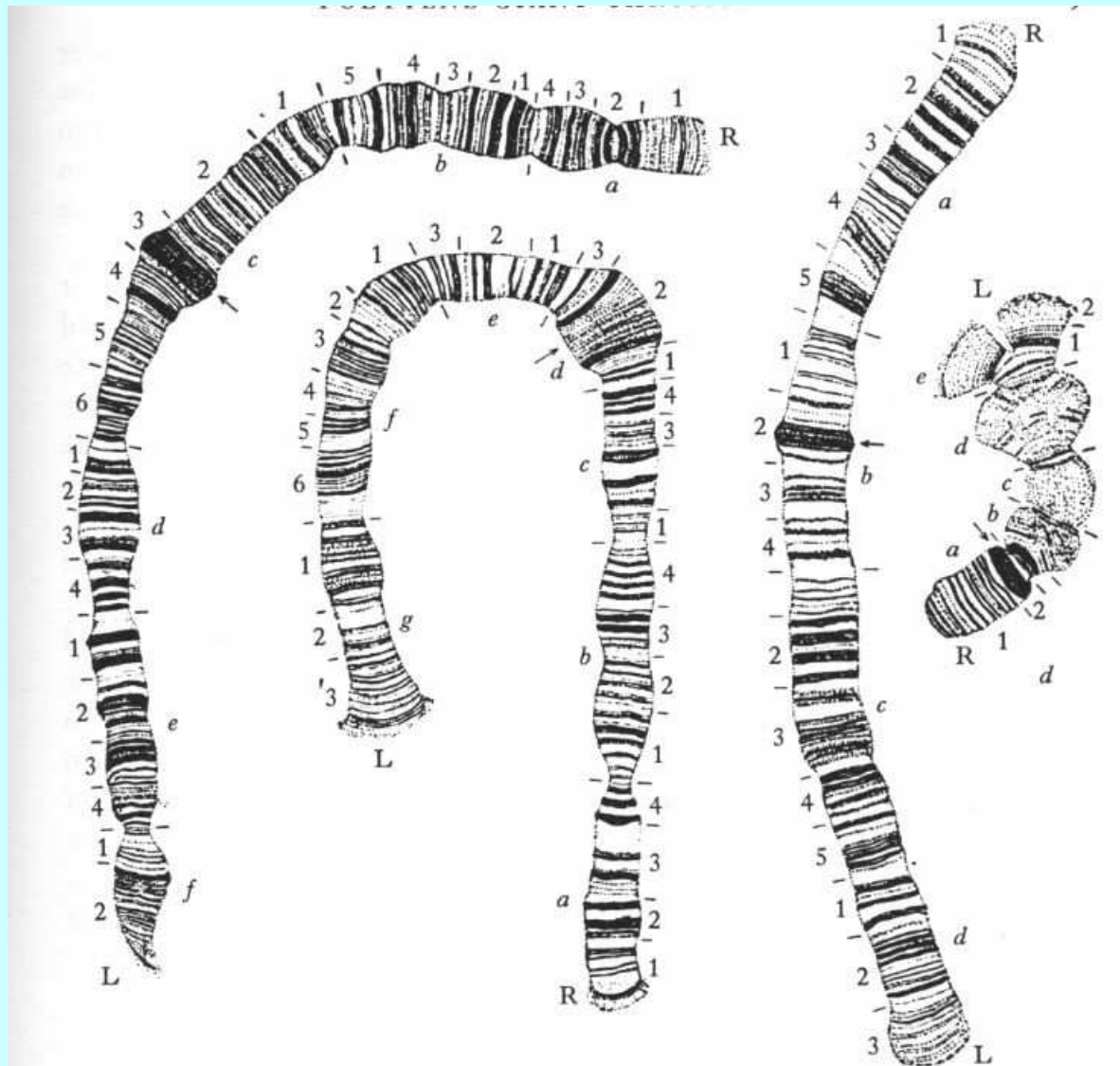
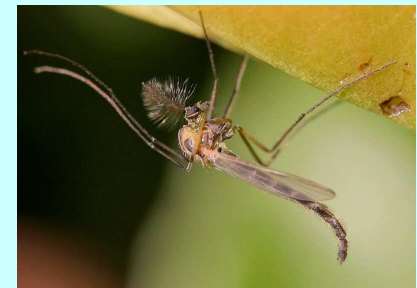
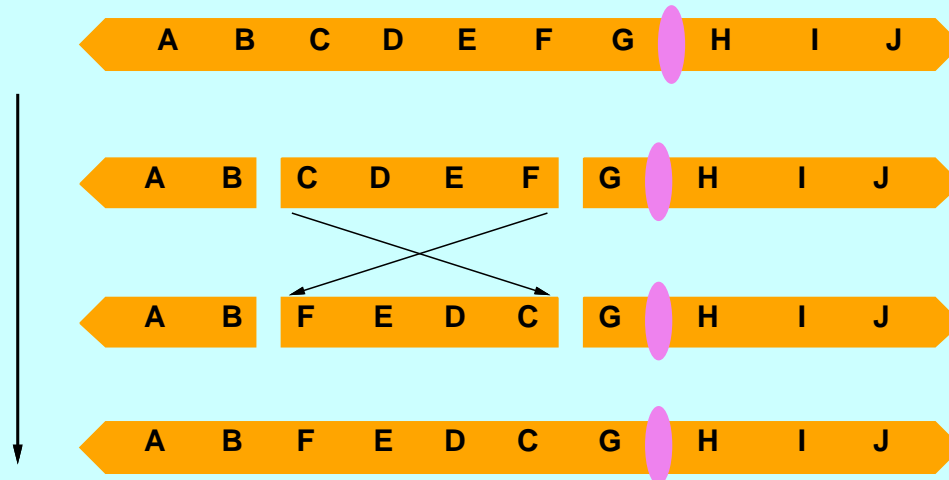


Fig. 4.1. The four polytene elements from a salivary gland nucleus of *Chironomus thummi*, showing the banding. Small arrows indicate the approximate positions of the centromeres. The shortest chromosome has its longer arm largely heterochromatic and the other three elements have short heterochromatic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

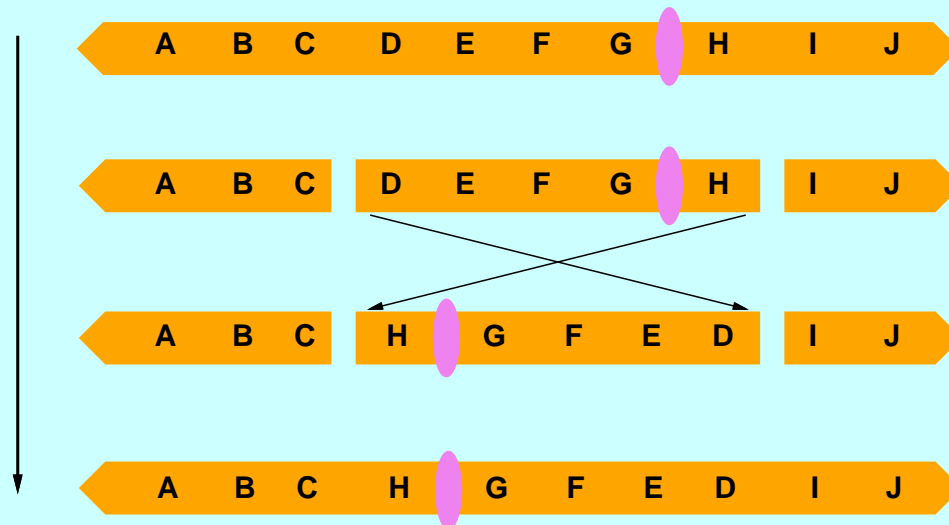


# Paracentric and pericentric inversions

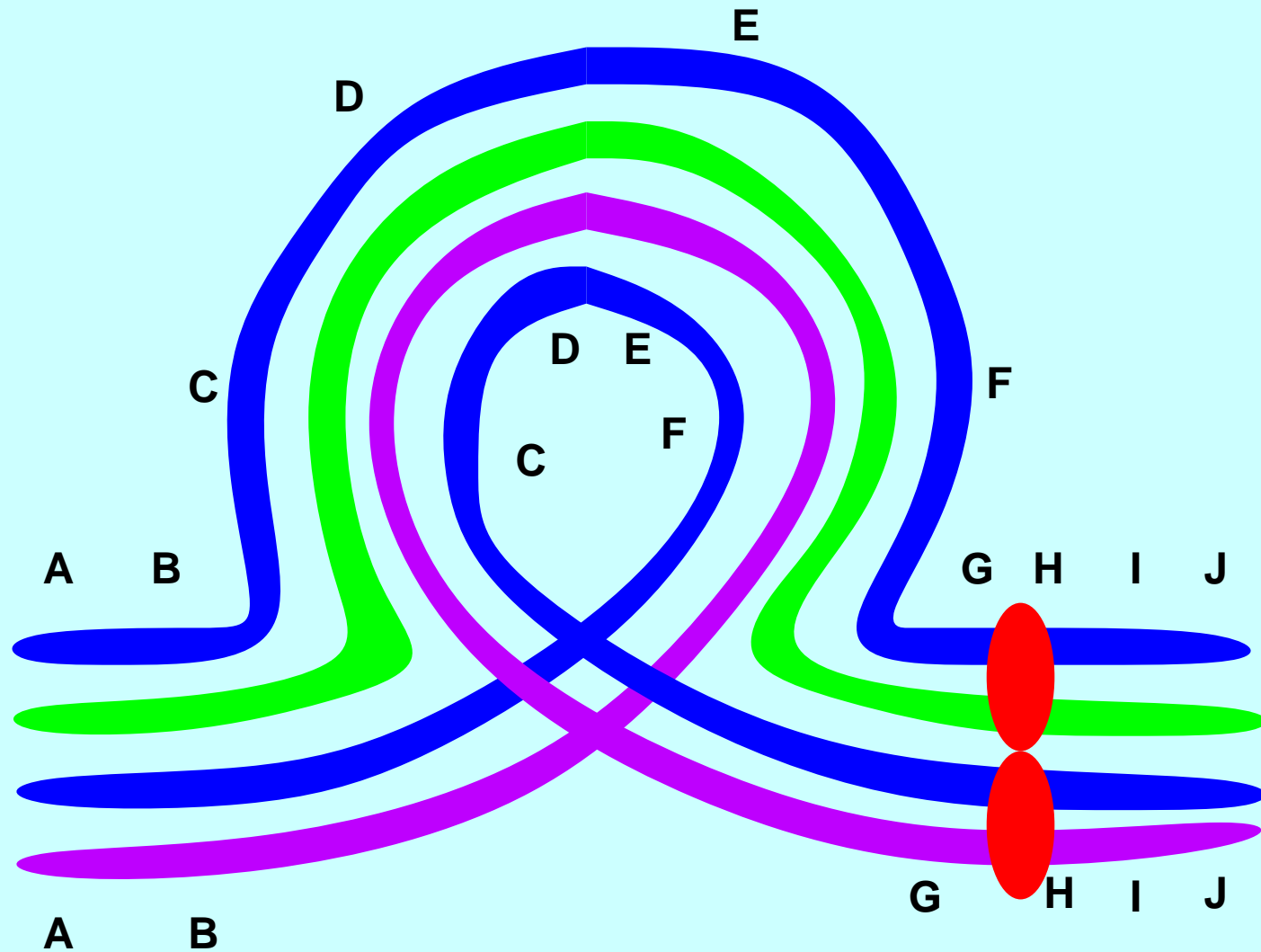
## A Paracentric Inversion



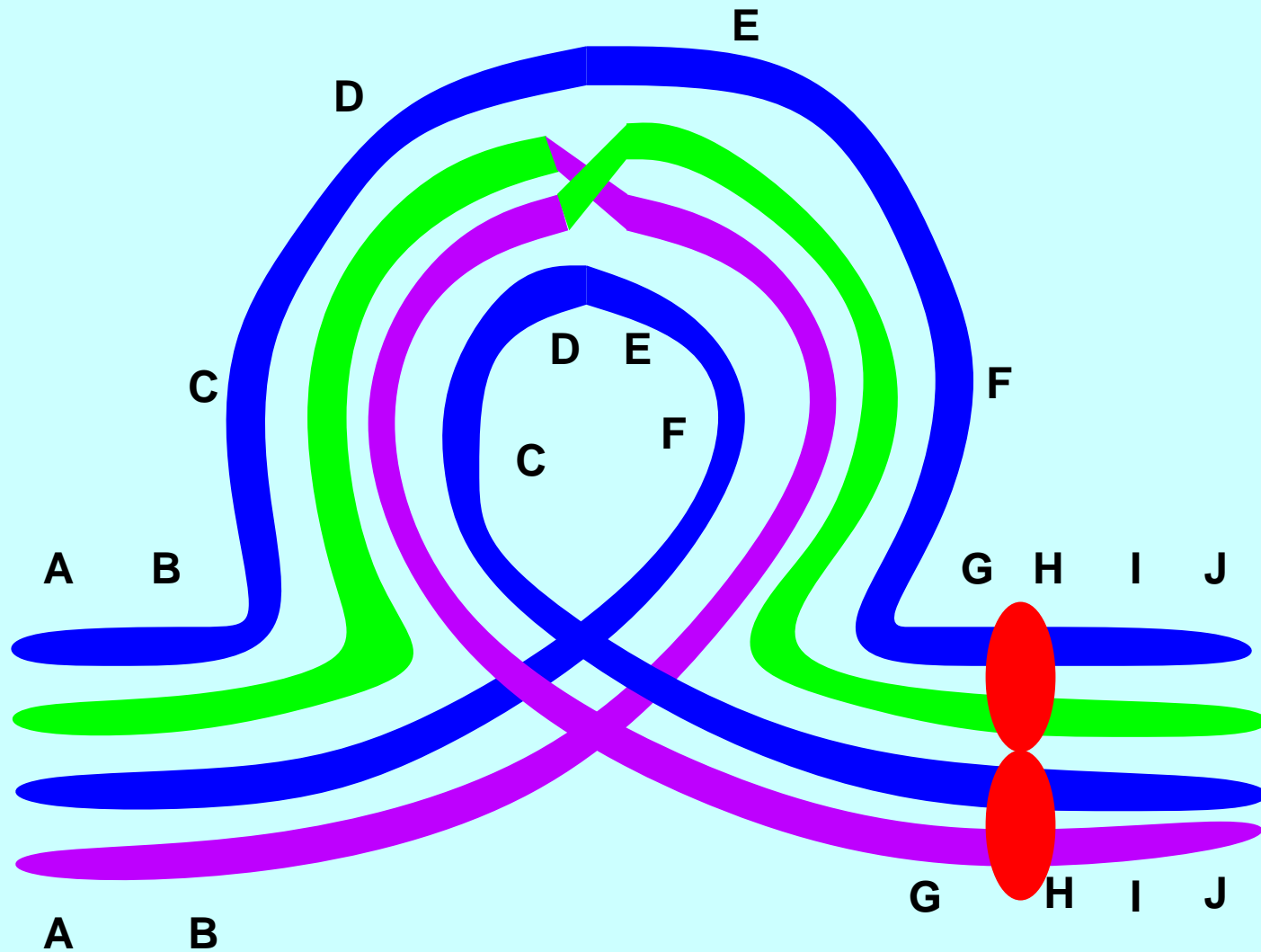
## A Pericentric Inversion



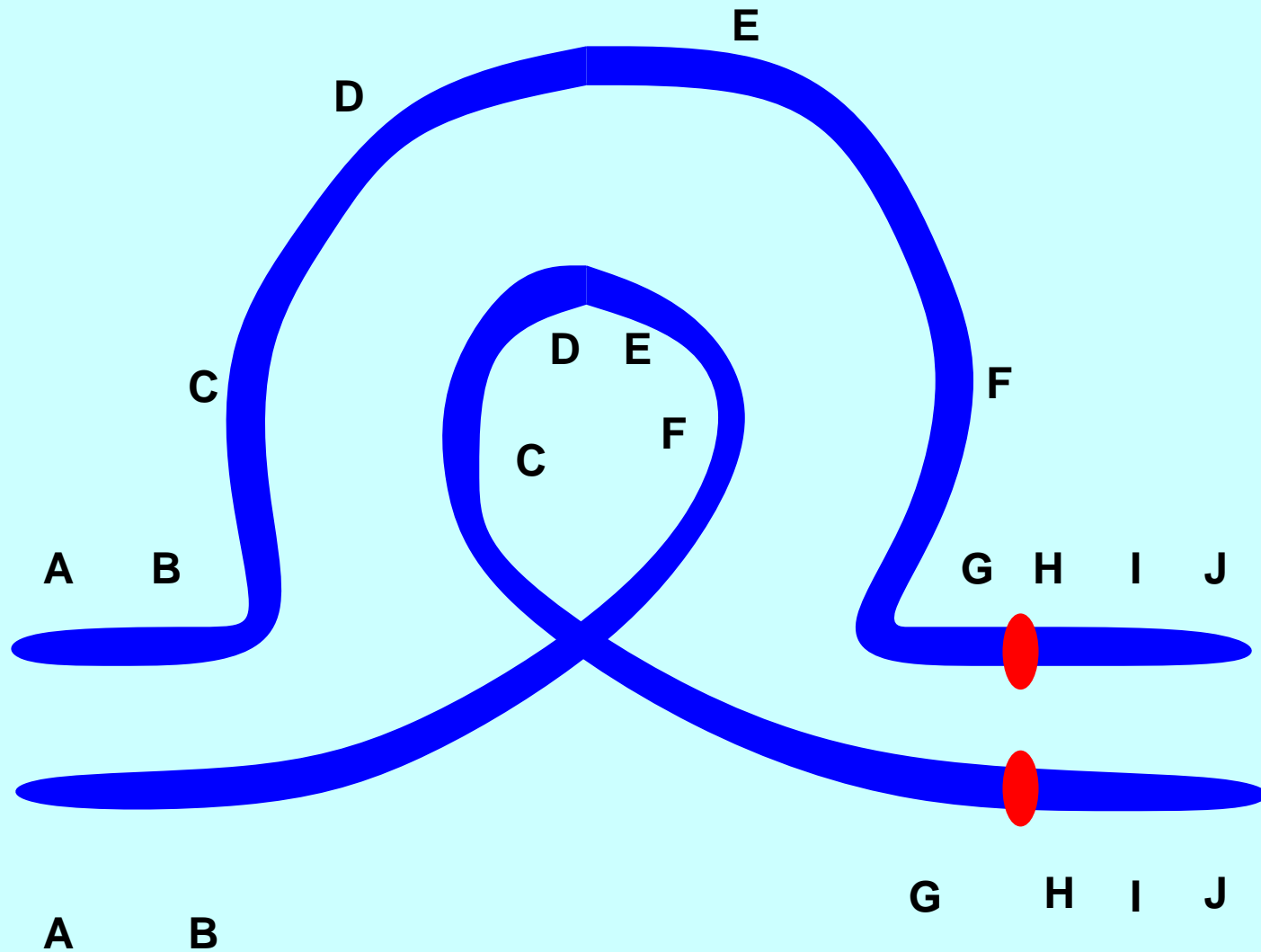
## Pairing in a paracentric inversion heterozygote



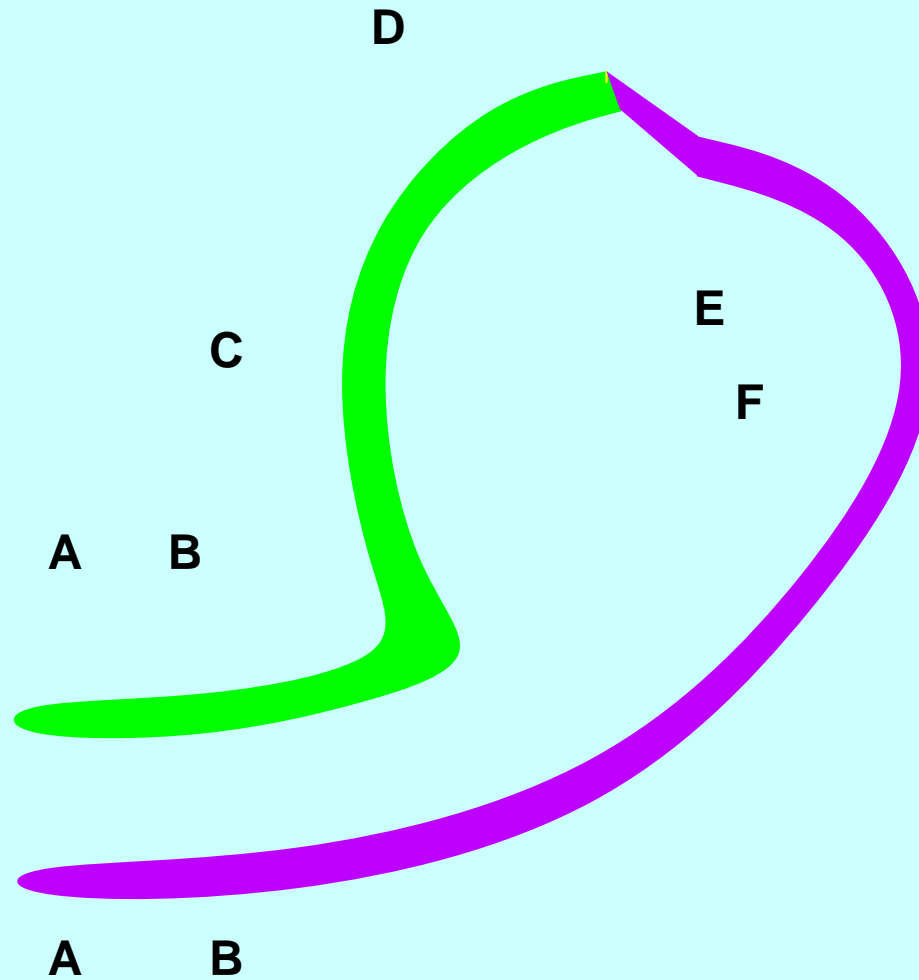
## Crossover in a paracentric inversion heterozygote



**These two chromosomes did not cross over**

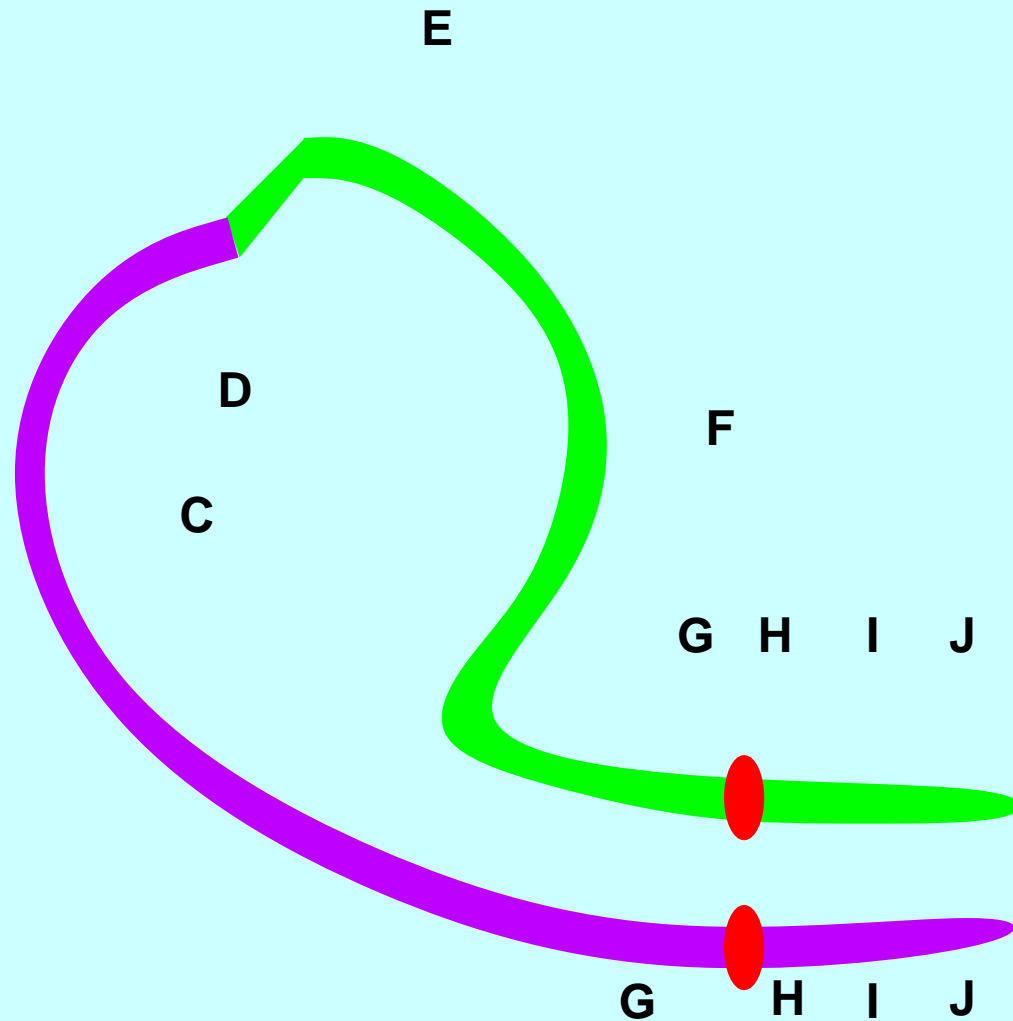


**This one did. How will it segregate?**

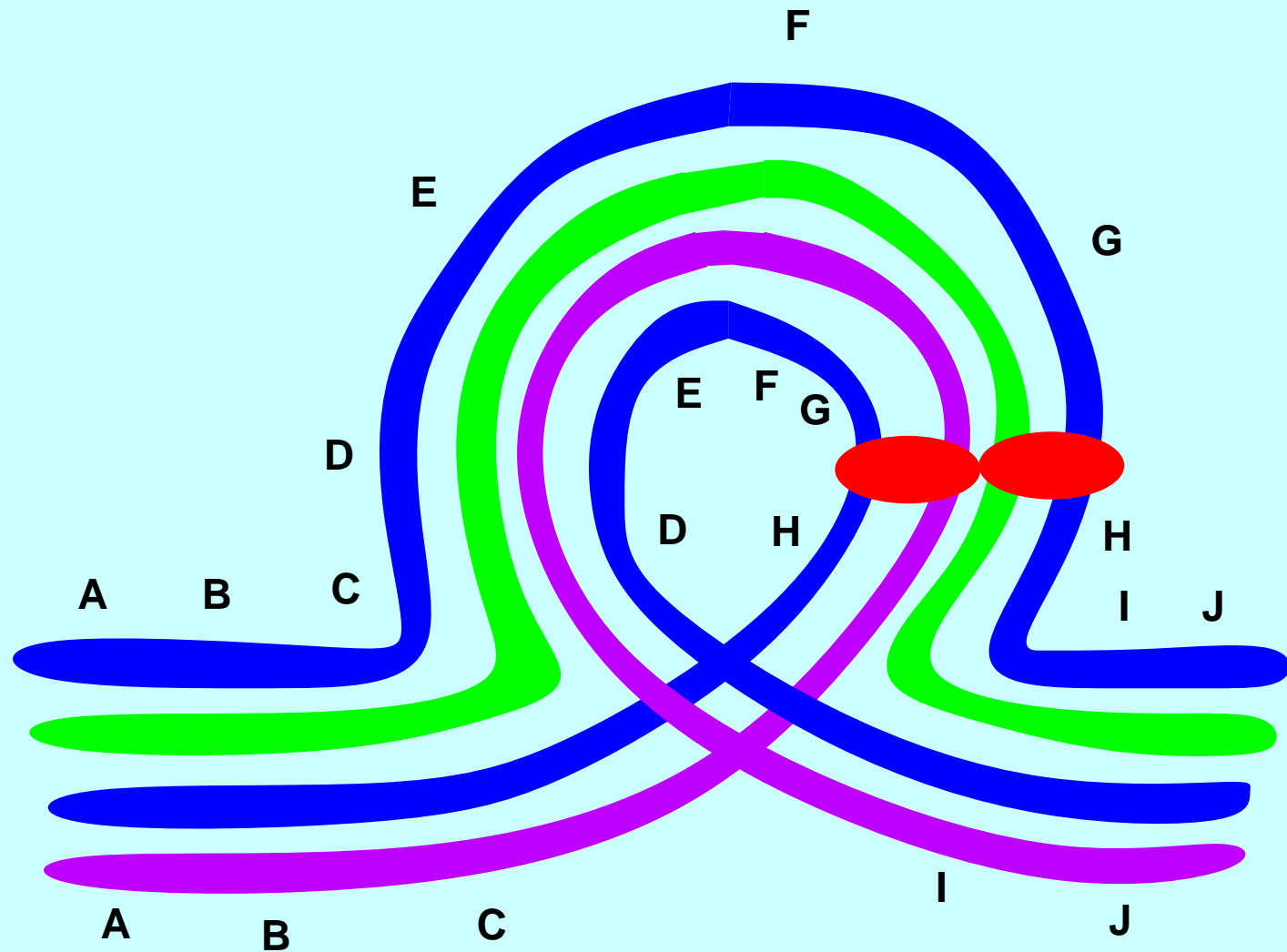




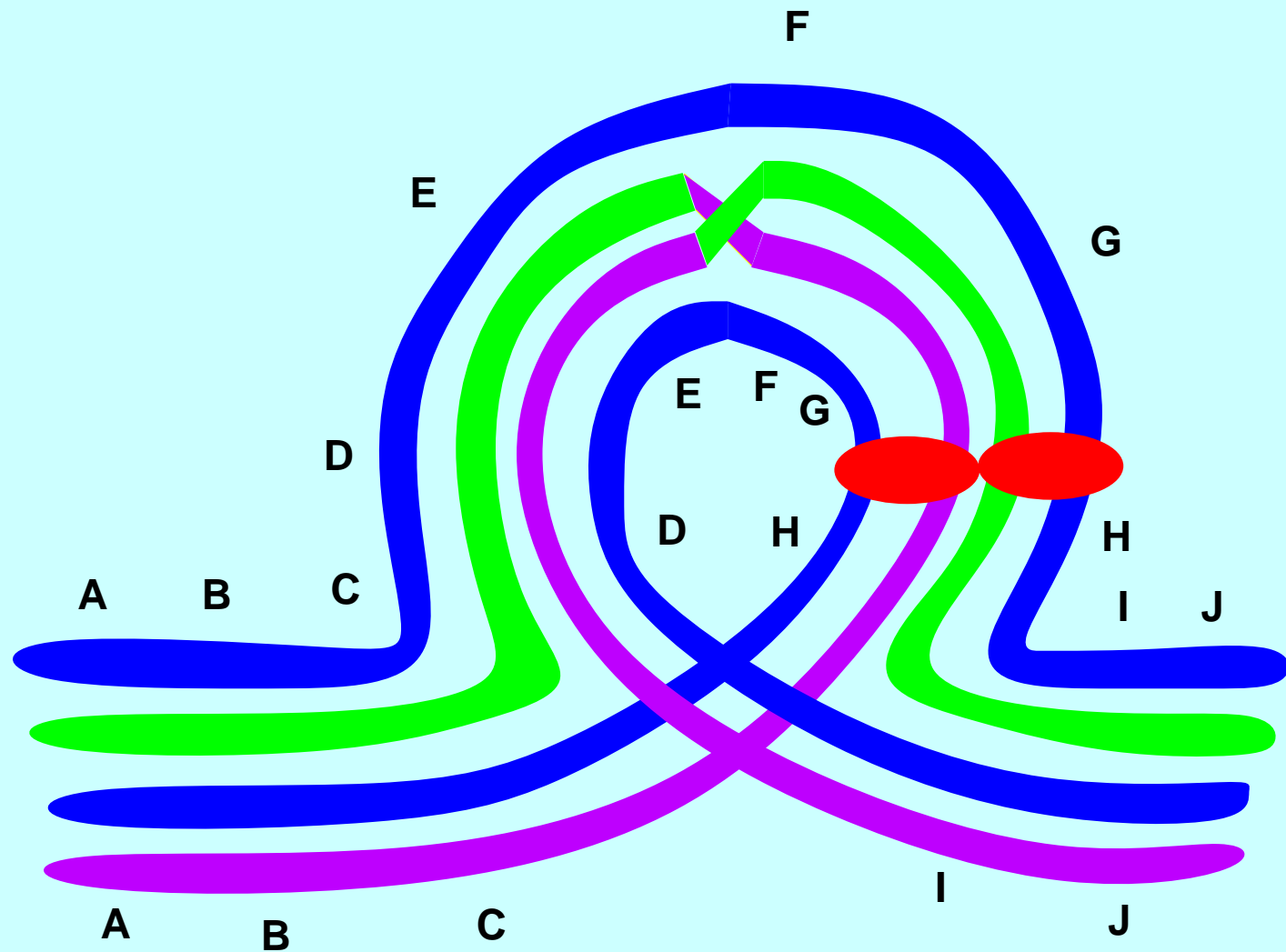
**This one did too. How will it segregate?**



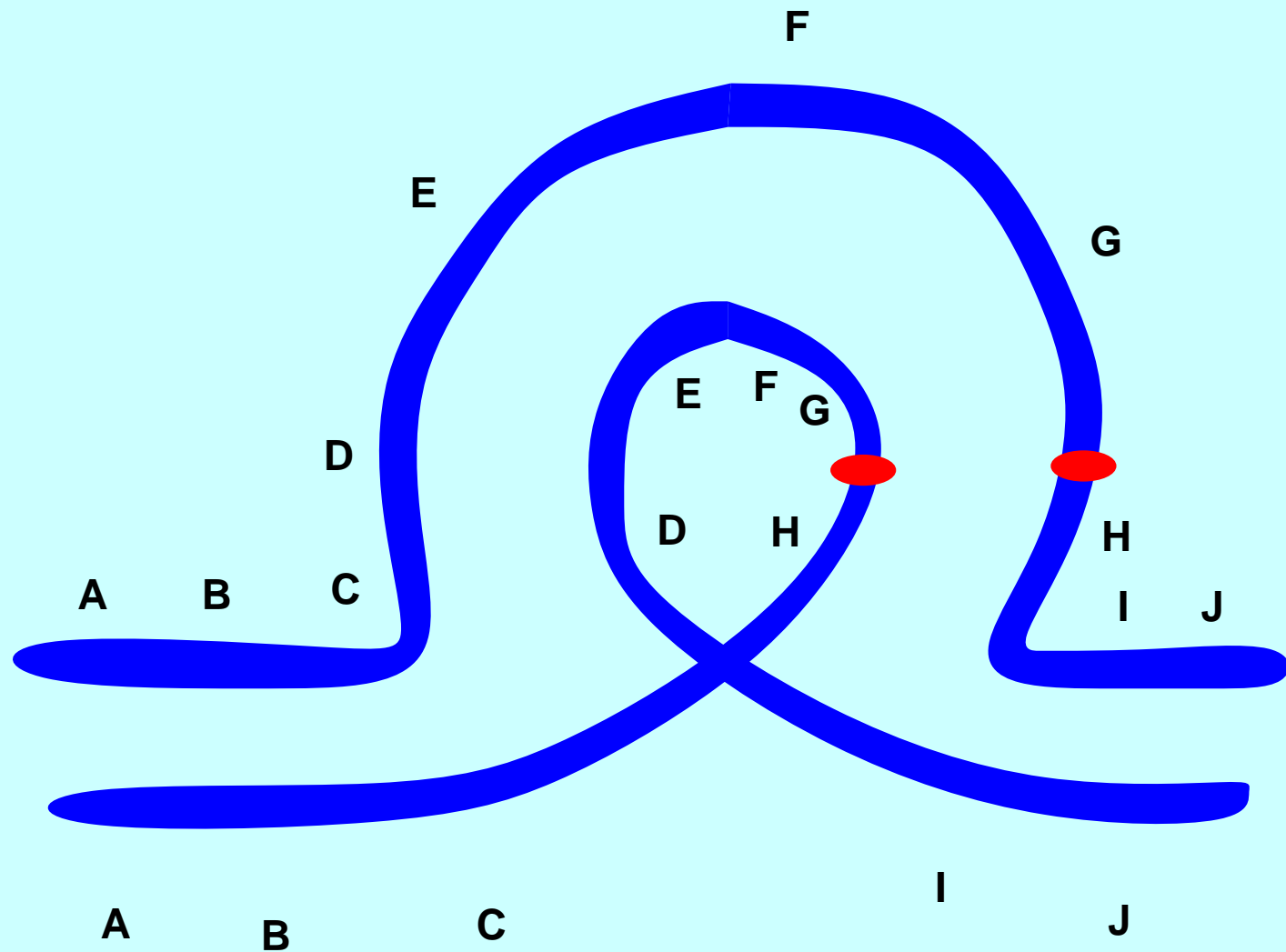
## Pairing in a pericentric inversion heterozygote



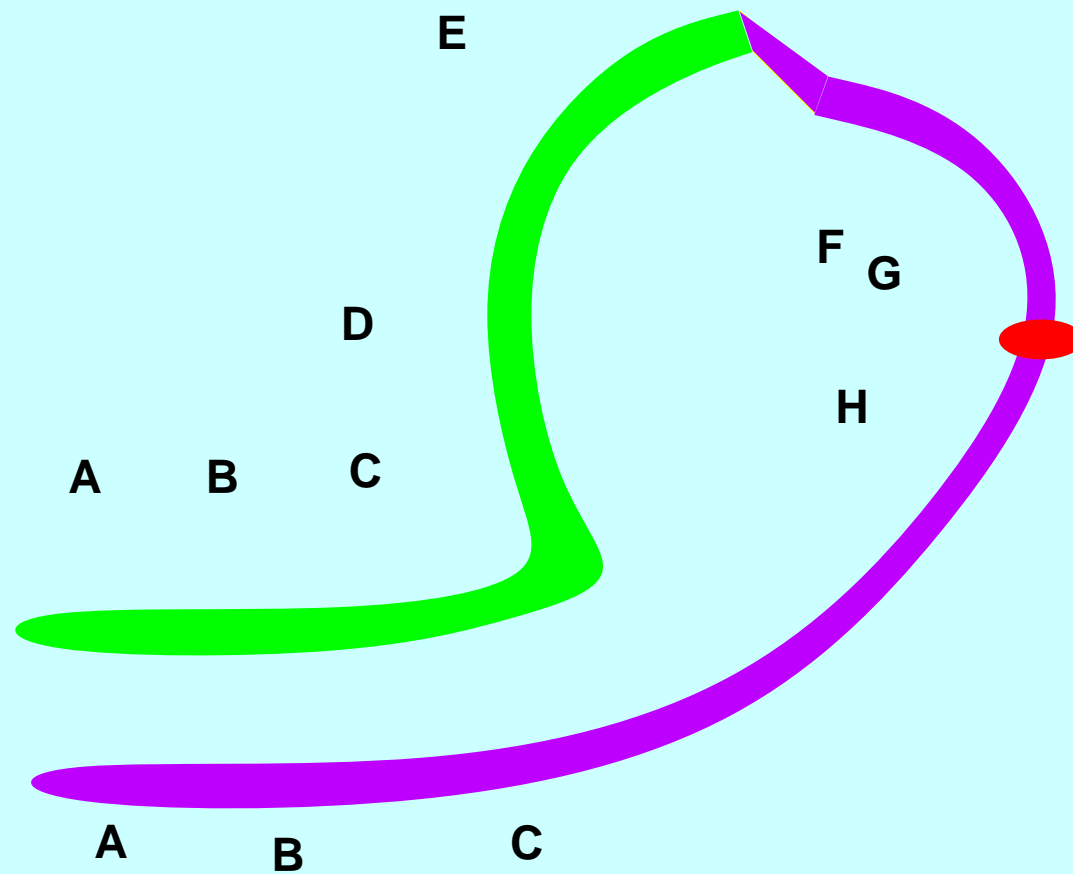
## Crossover in a pericentric inversion heterozygote



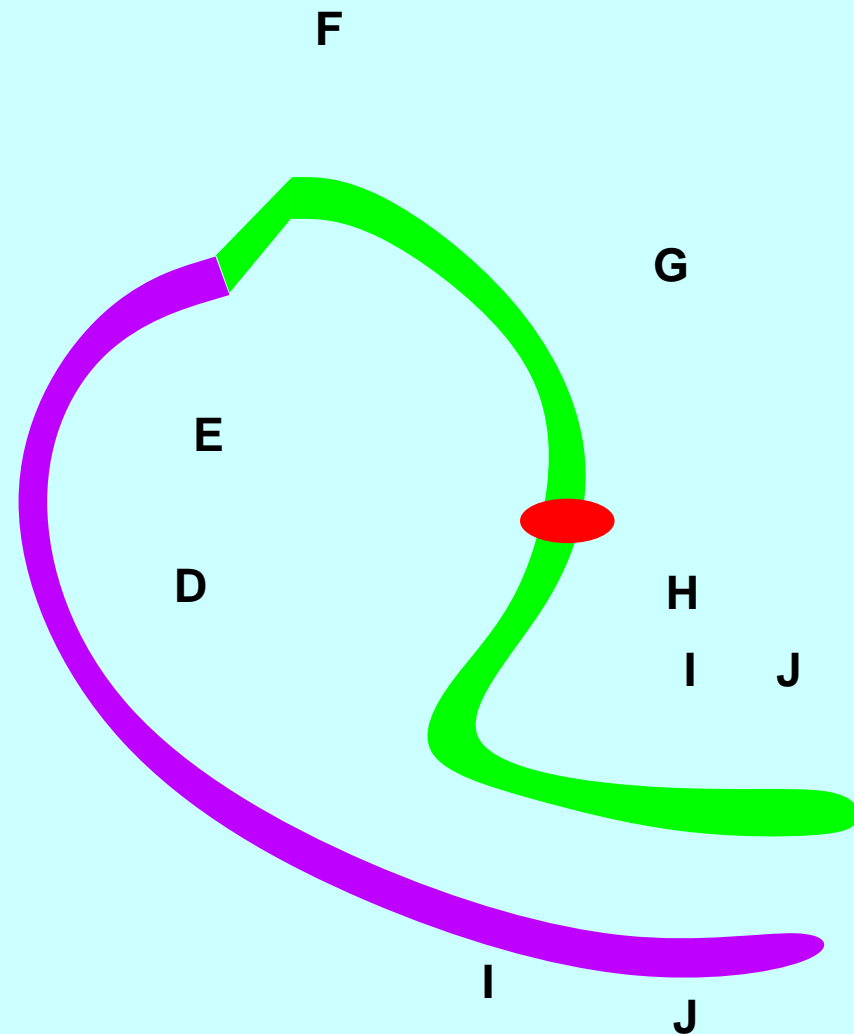
These two products didn't undergo crossing-over



One that did. Is anything deleted? Duplicated?

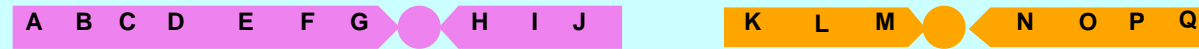


**The other one that did. Anything deleted, duplicated?**

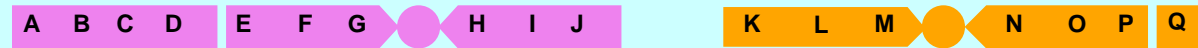


# A translocation

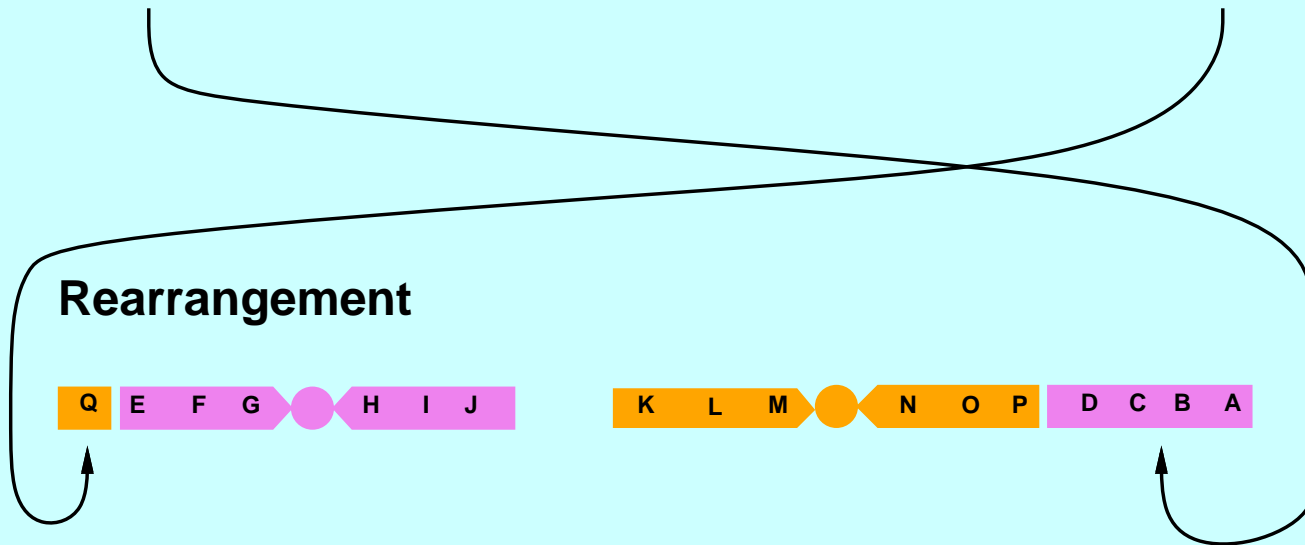
Before



Breaks



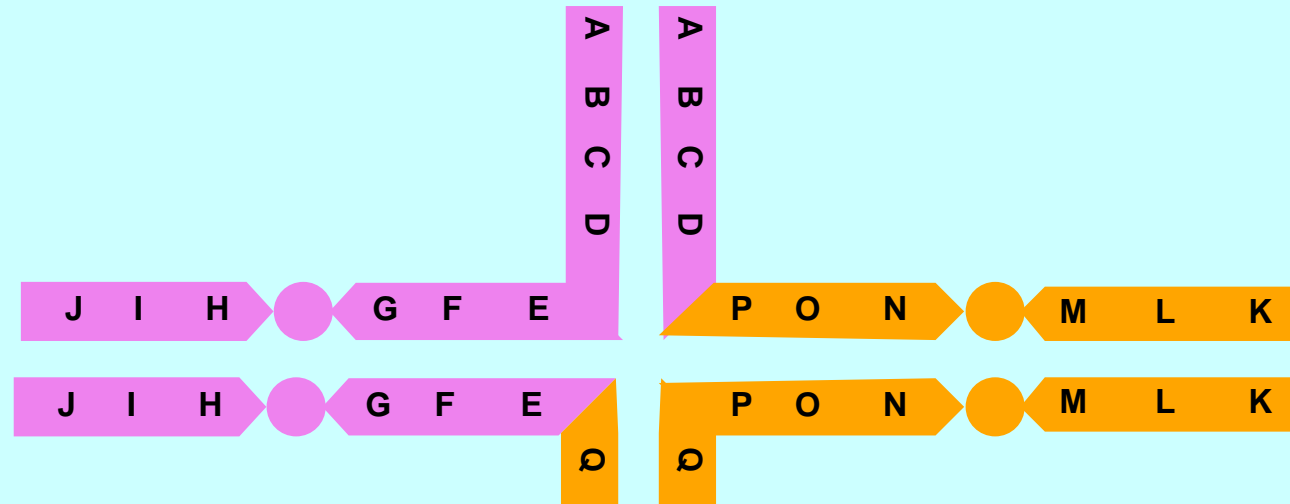
Rearrangement



After



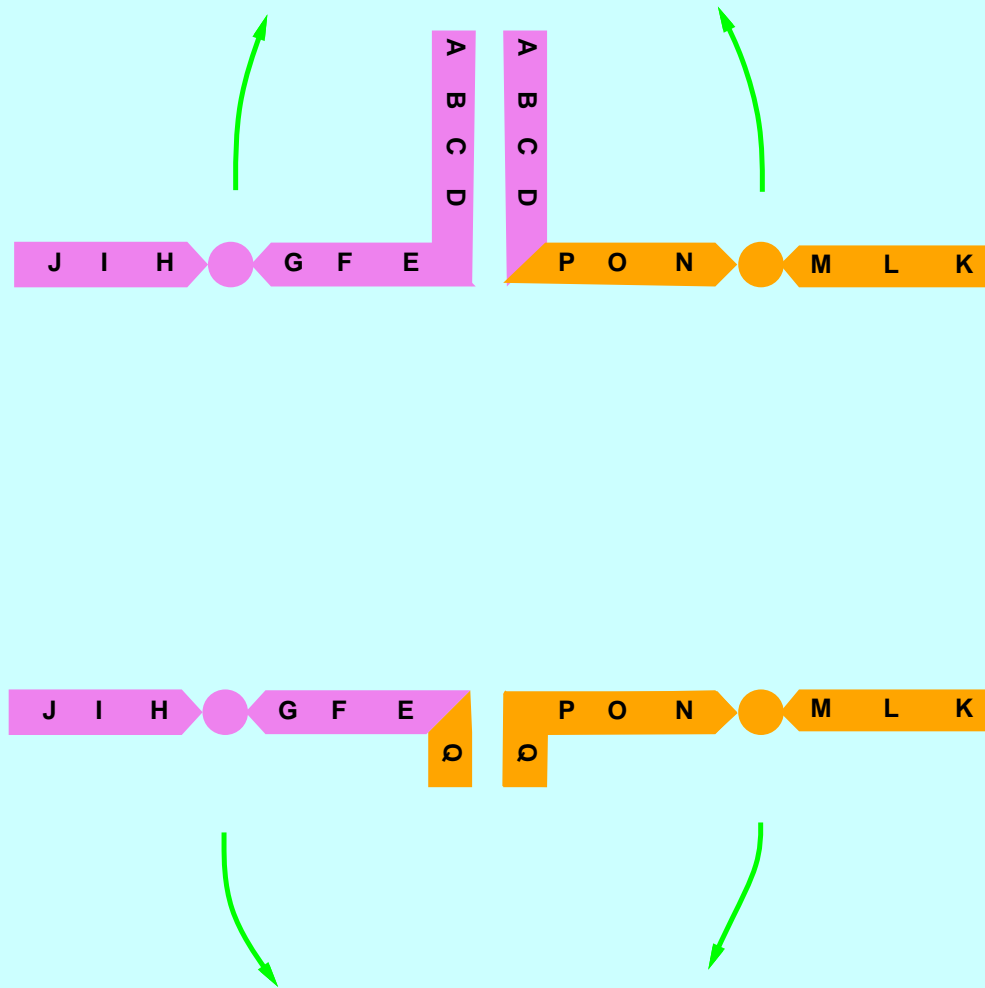
# Segregation of translocation heterozygote at first division of meiosis metaphase



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

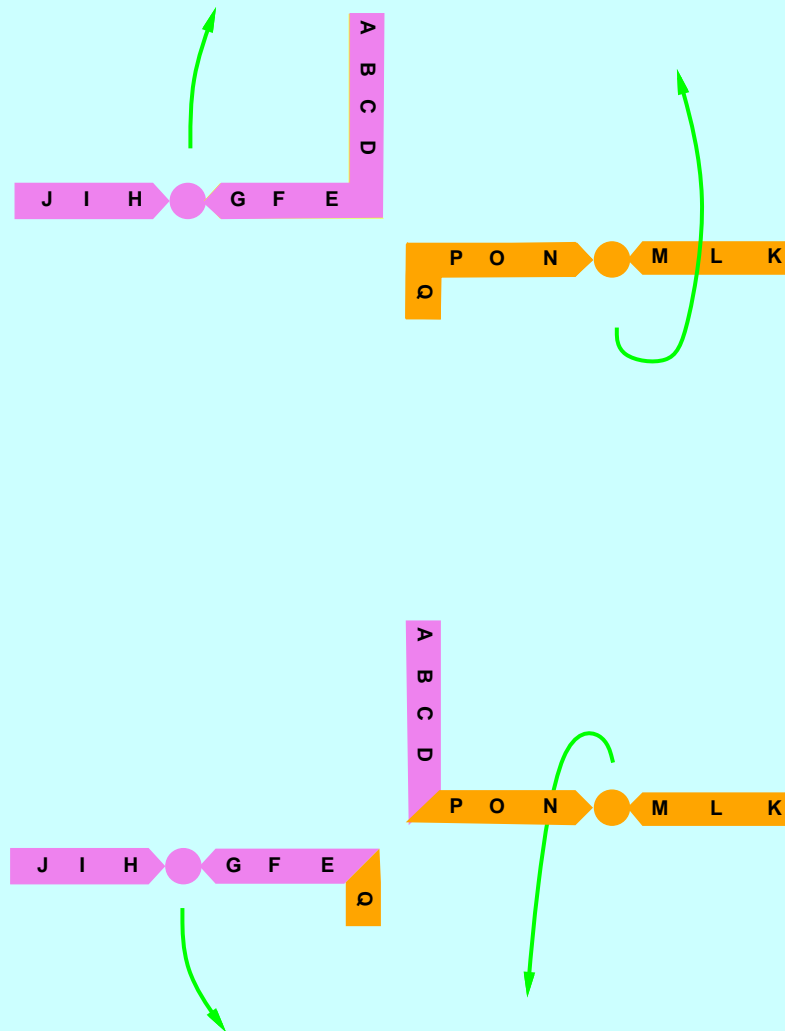


# Adjacent segregation in translocations



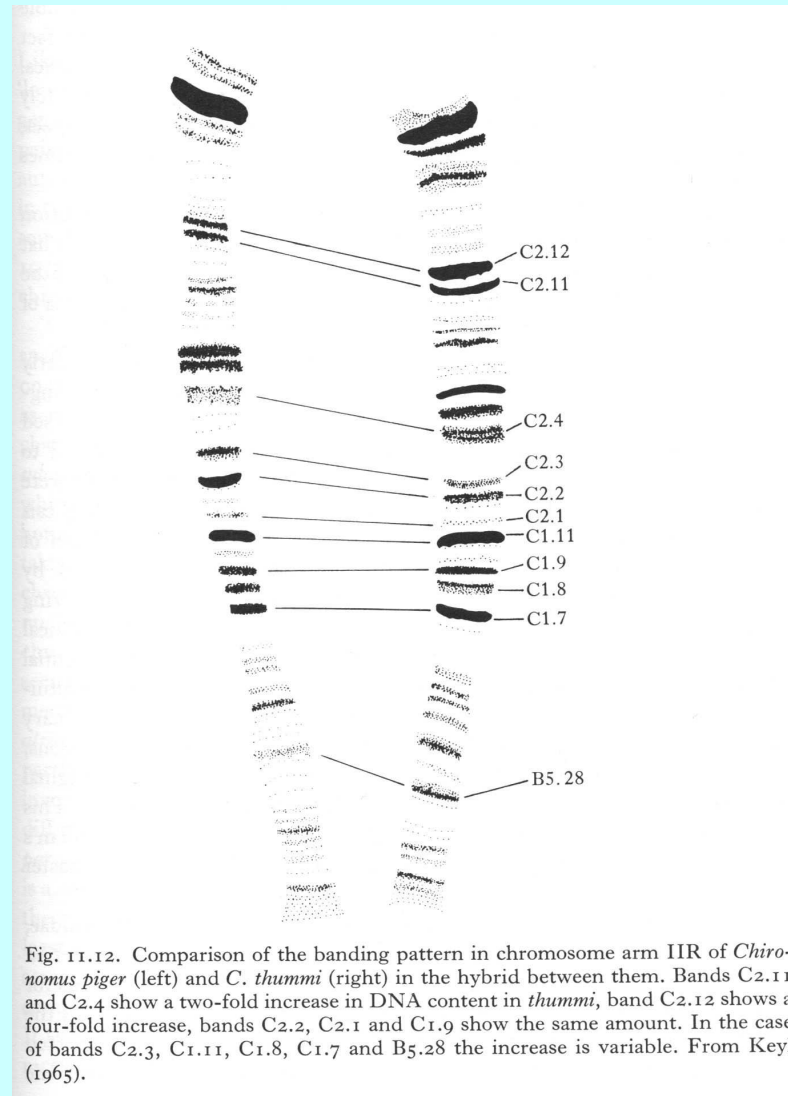
... Leads to deletions and duplications.

# Alternate segregation in translocations



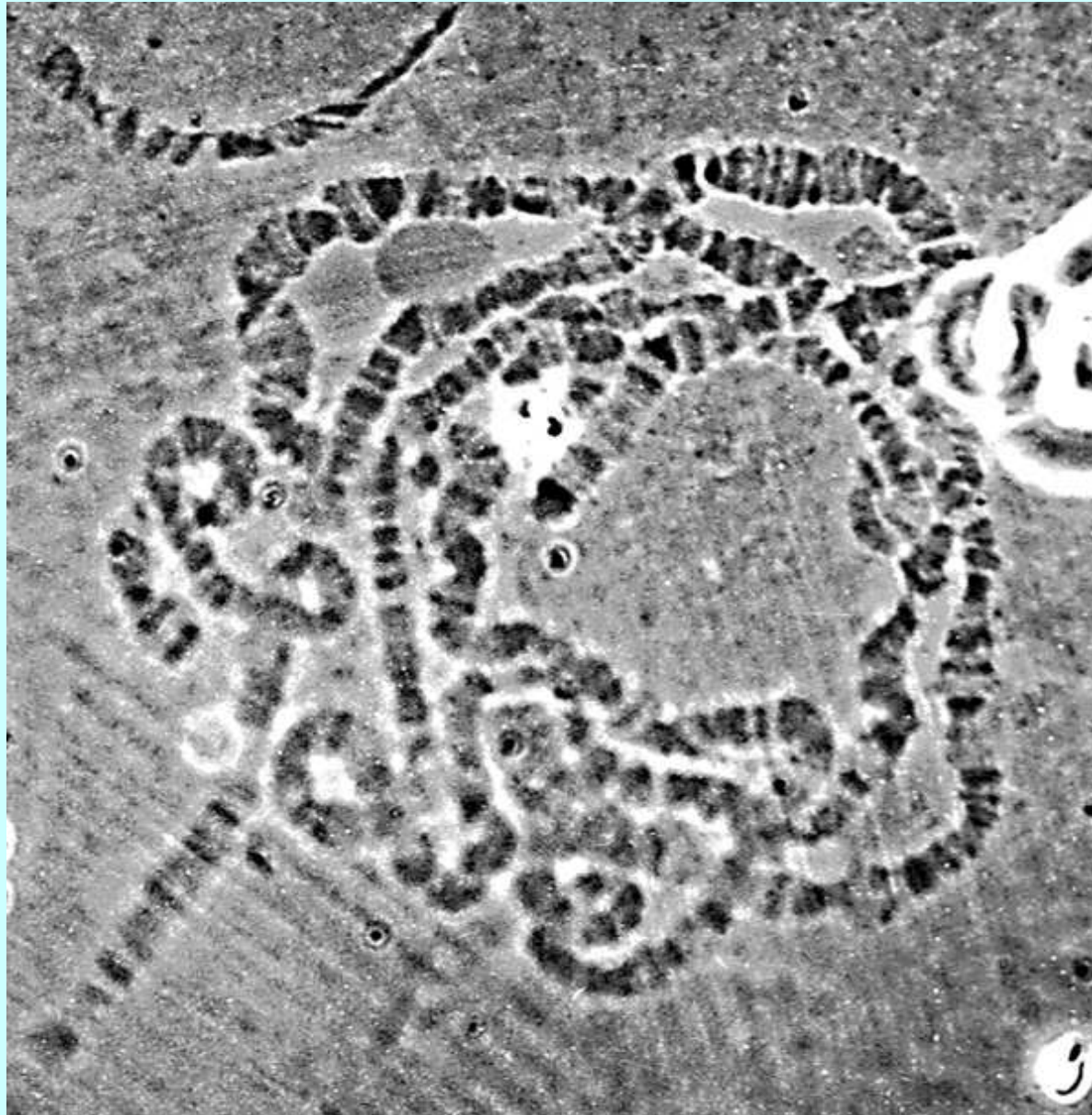
... Leads to euploid (normal gene complement) gametes

# Banding pattern changes

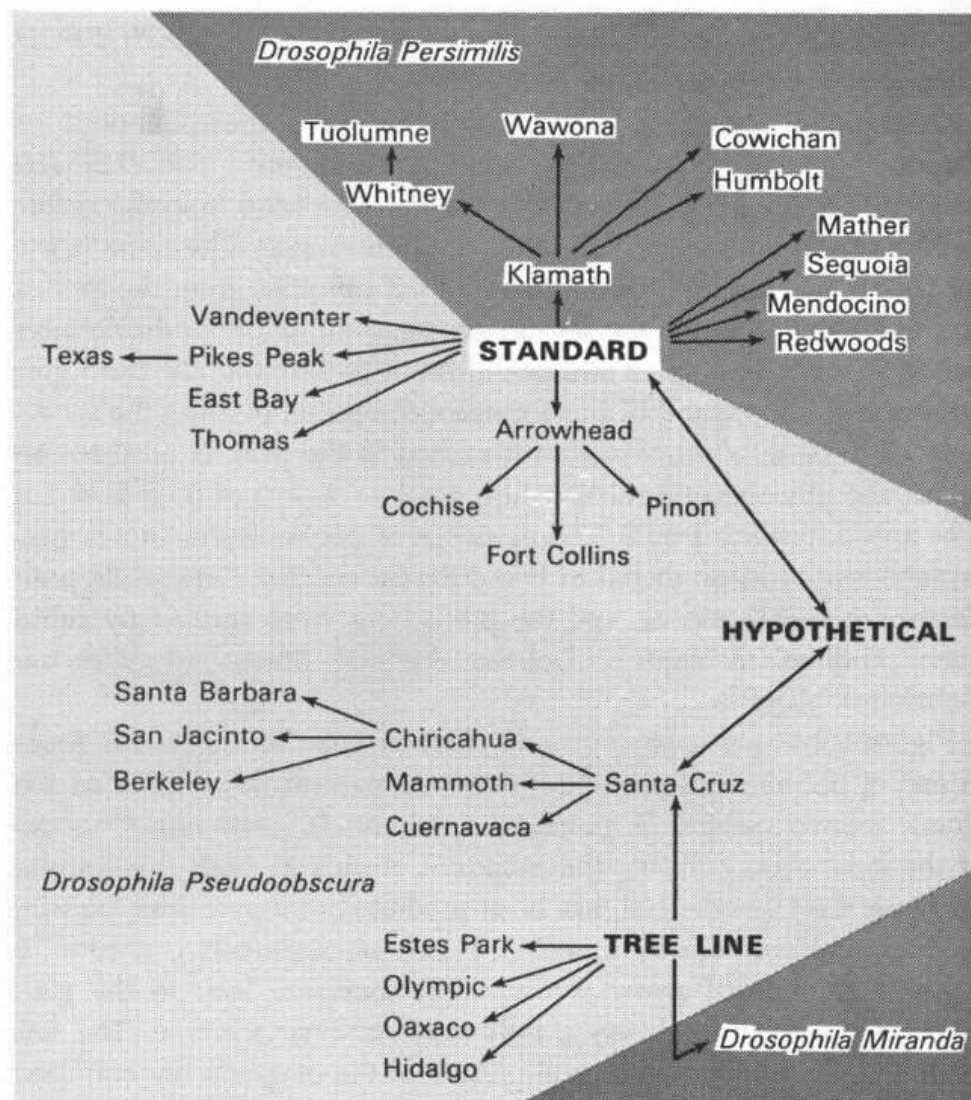


in two species of Chironomus midges

## Polytene Drosophila chromosomes



# Chromosome phylogeny of *Drosophila pseudoobscura* etc

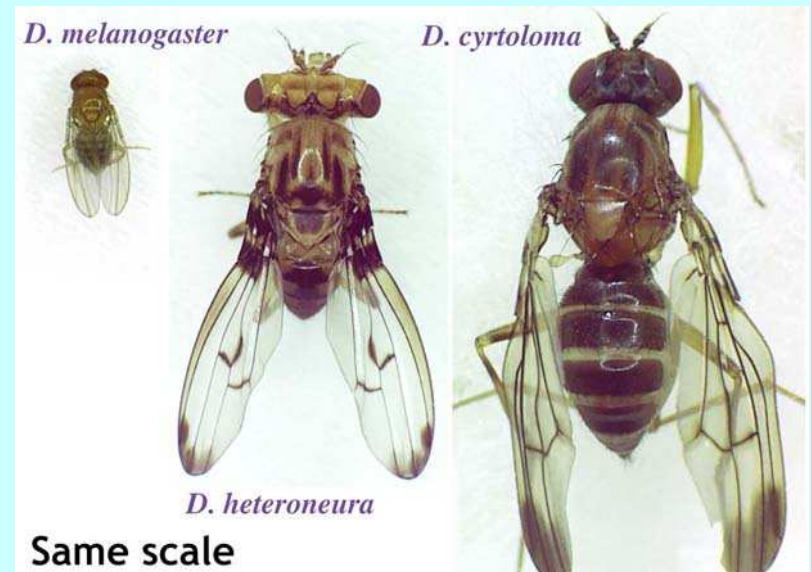
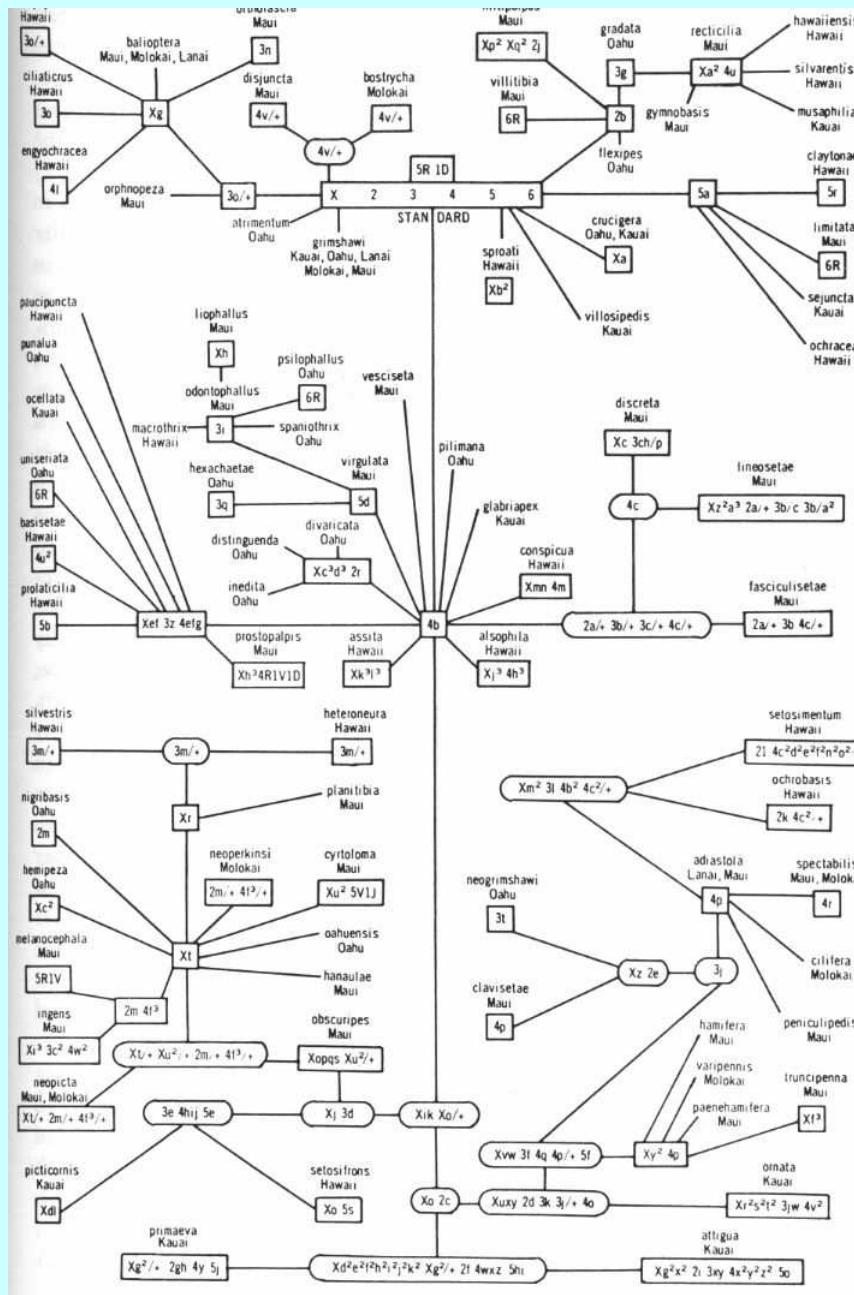


**FIGURE 5.2**

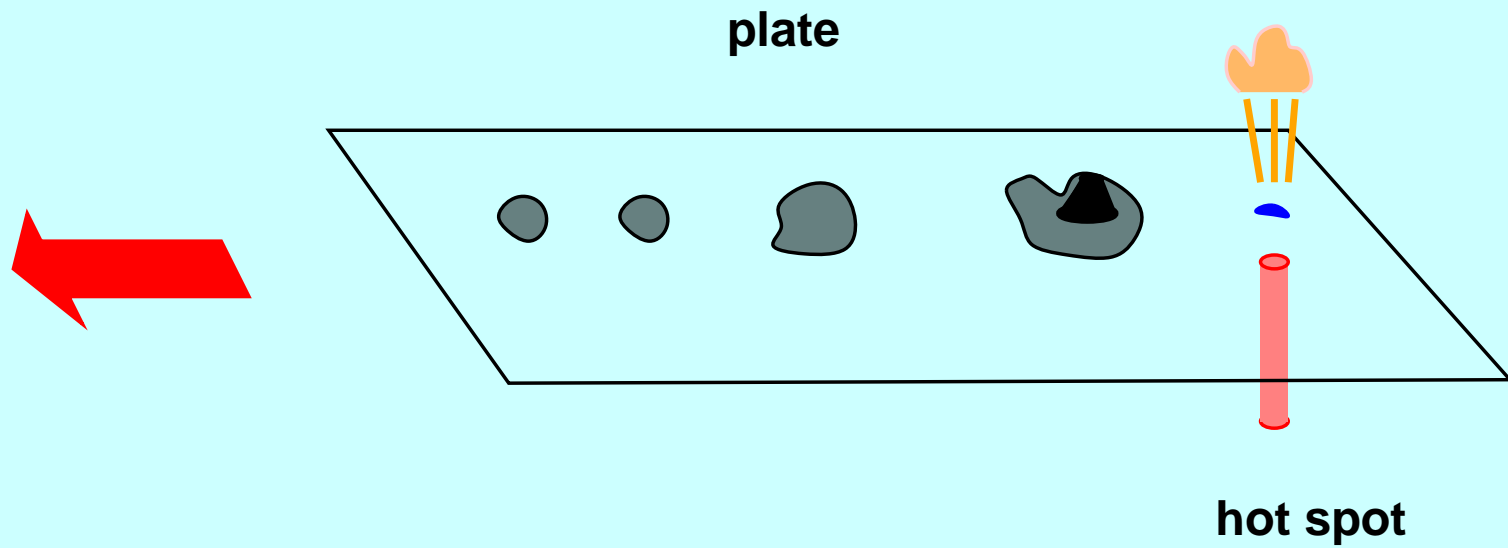
Phylogenetic relationships of the gene arrangements in the third chromosomes of *Drosophila pseudoobscura*, *D. persimilis*, and *D. miranda*.



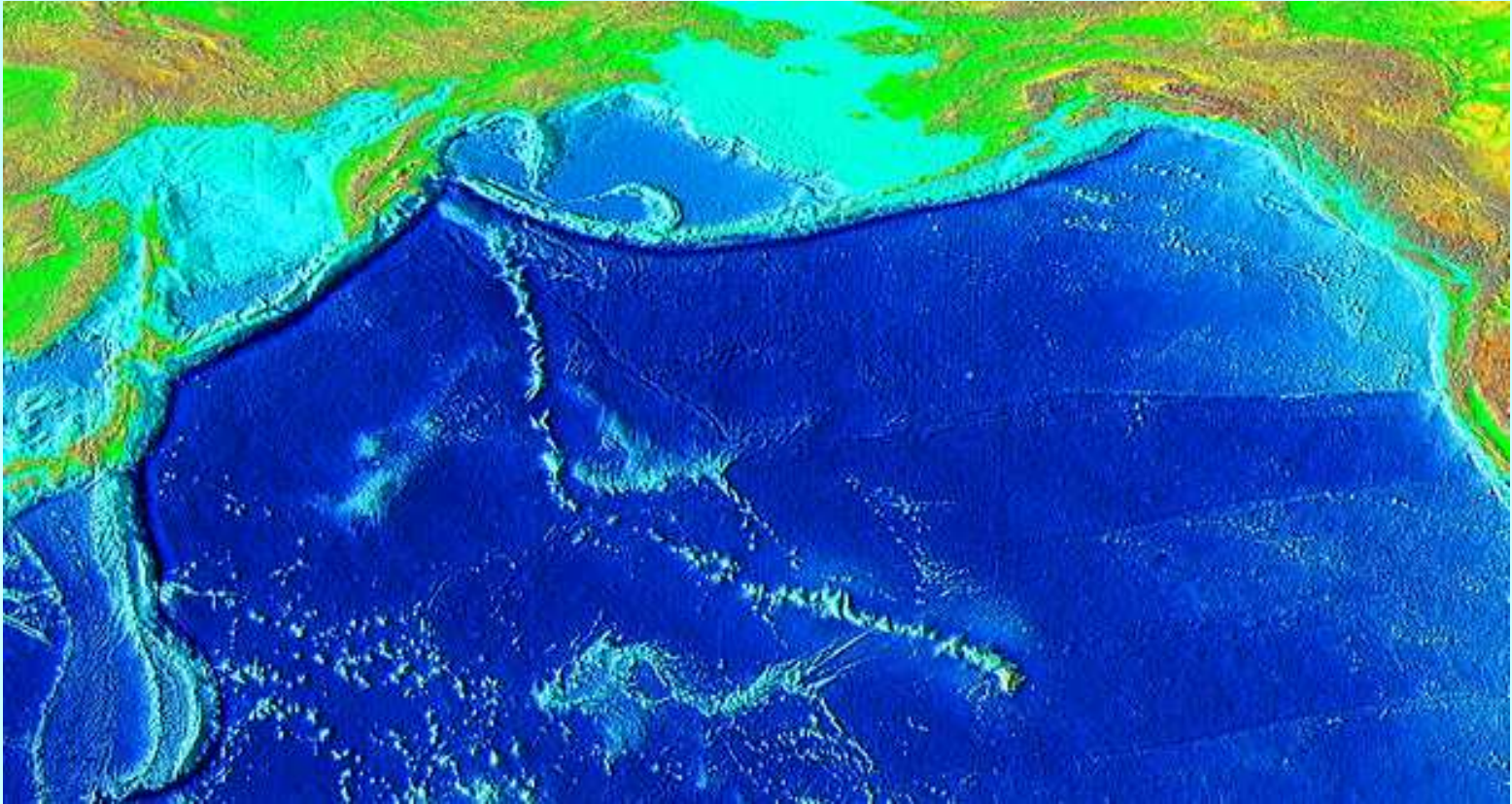
# Chromosome phylogeny of Hawaiian *Drosophila*



# Formation of the Hawaiian islands



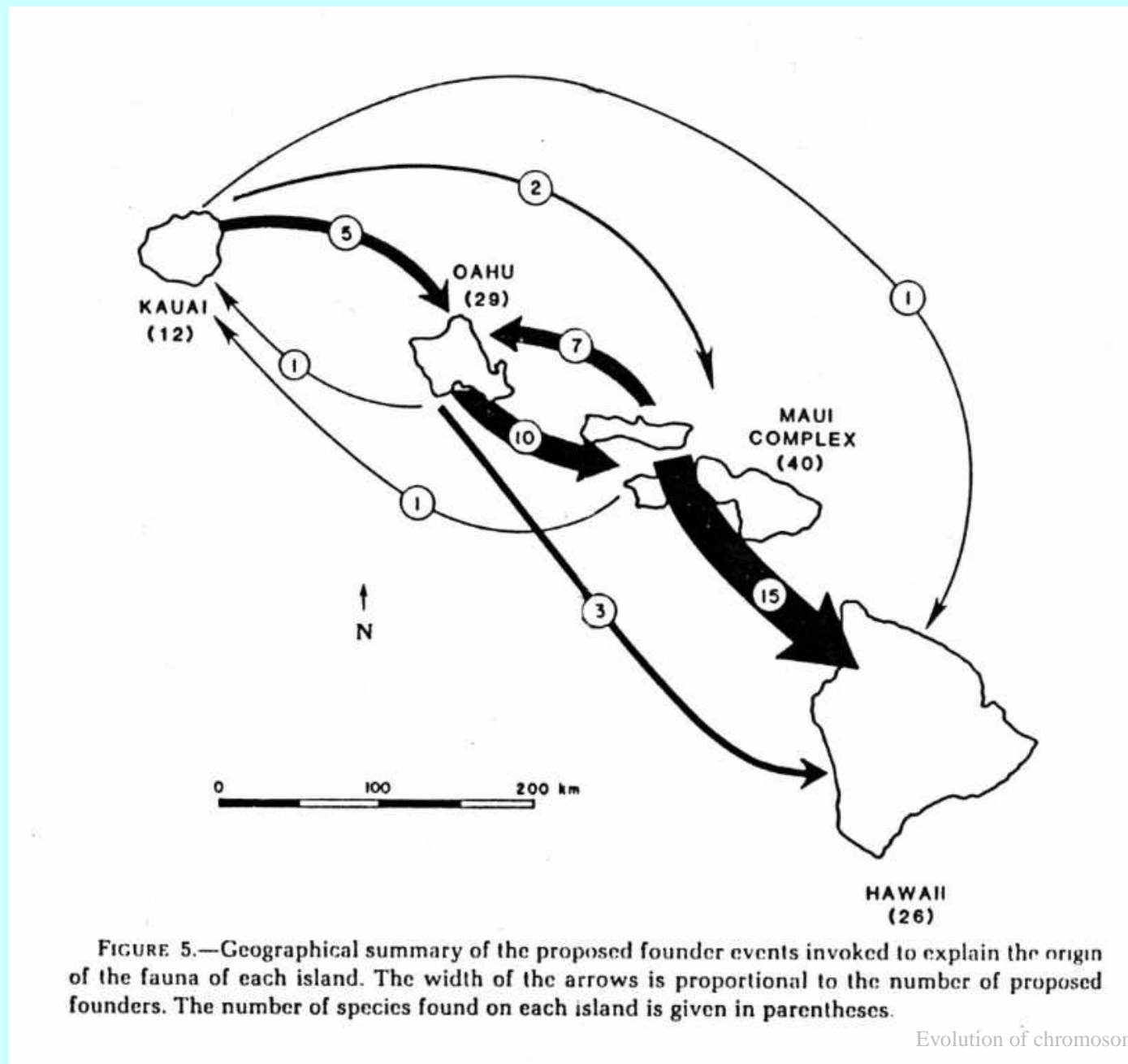
## Geology of the Hawaiian islands



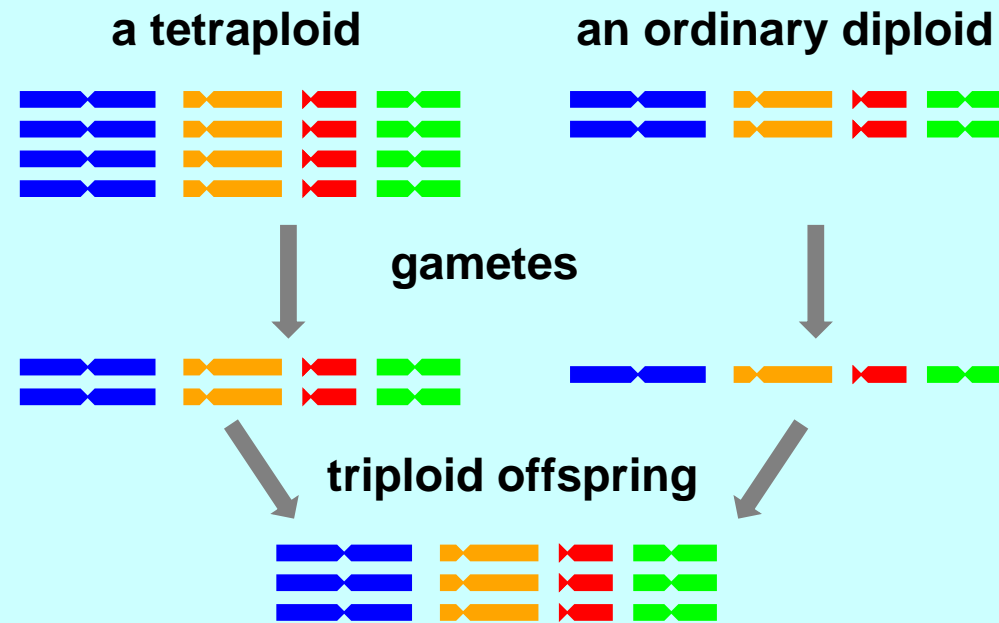
A chain of seamounts leads northwest back across the Pacific as far as the Kamchatka peninsula of Siberia, an estimated 85 million years.



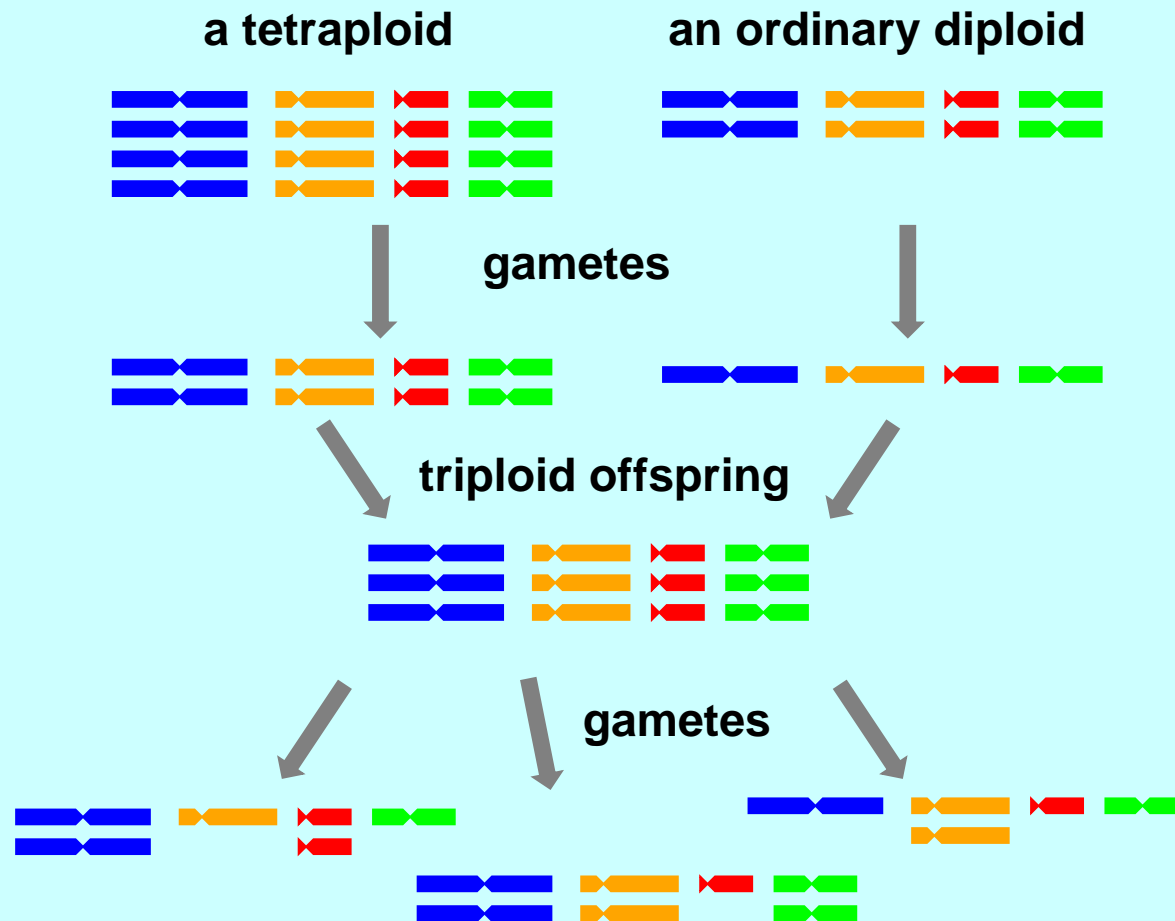
# Inferred migration events on the Hawaiian islands



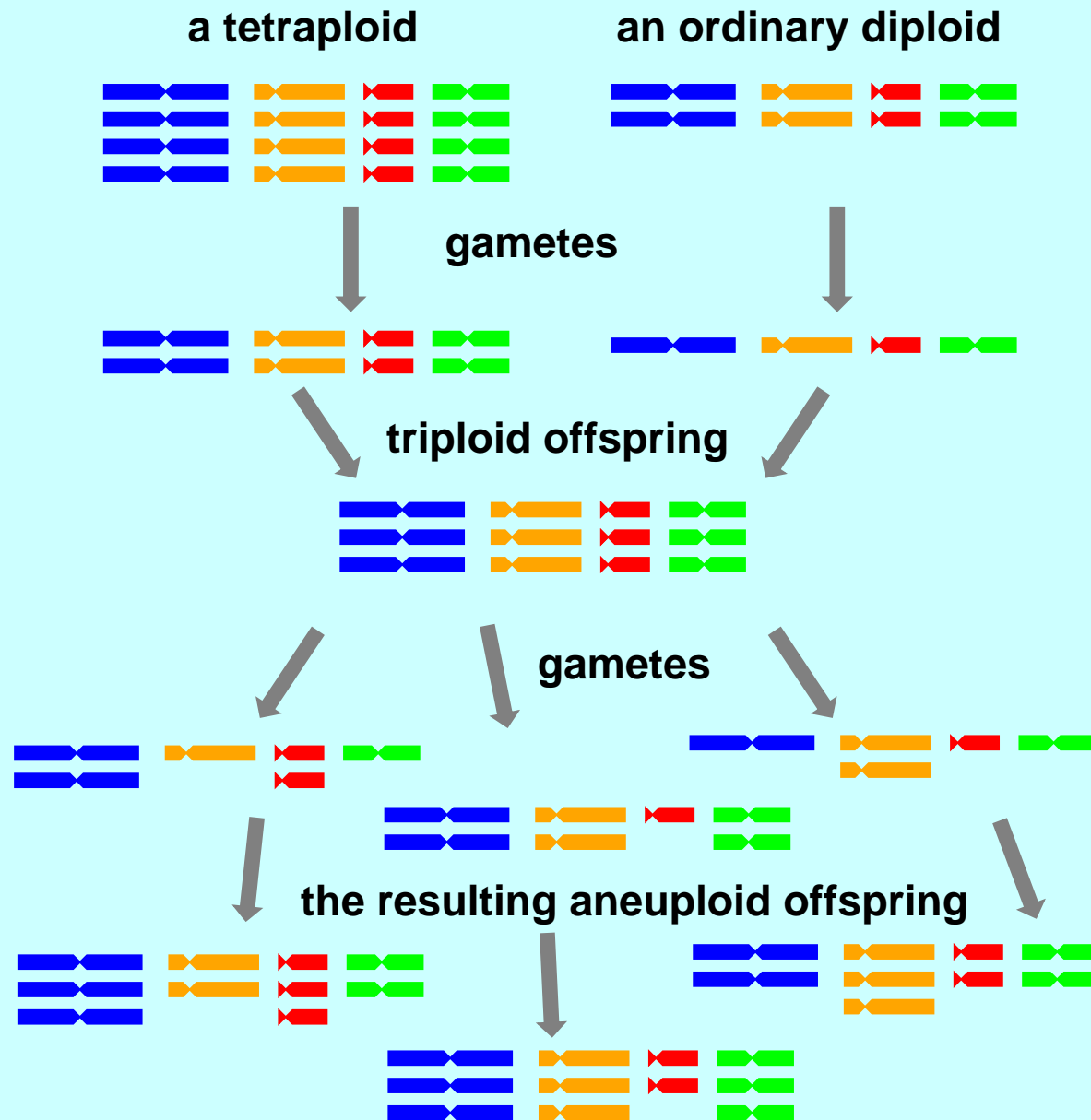
# When a tetraploid mates with a diploid



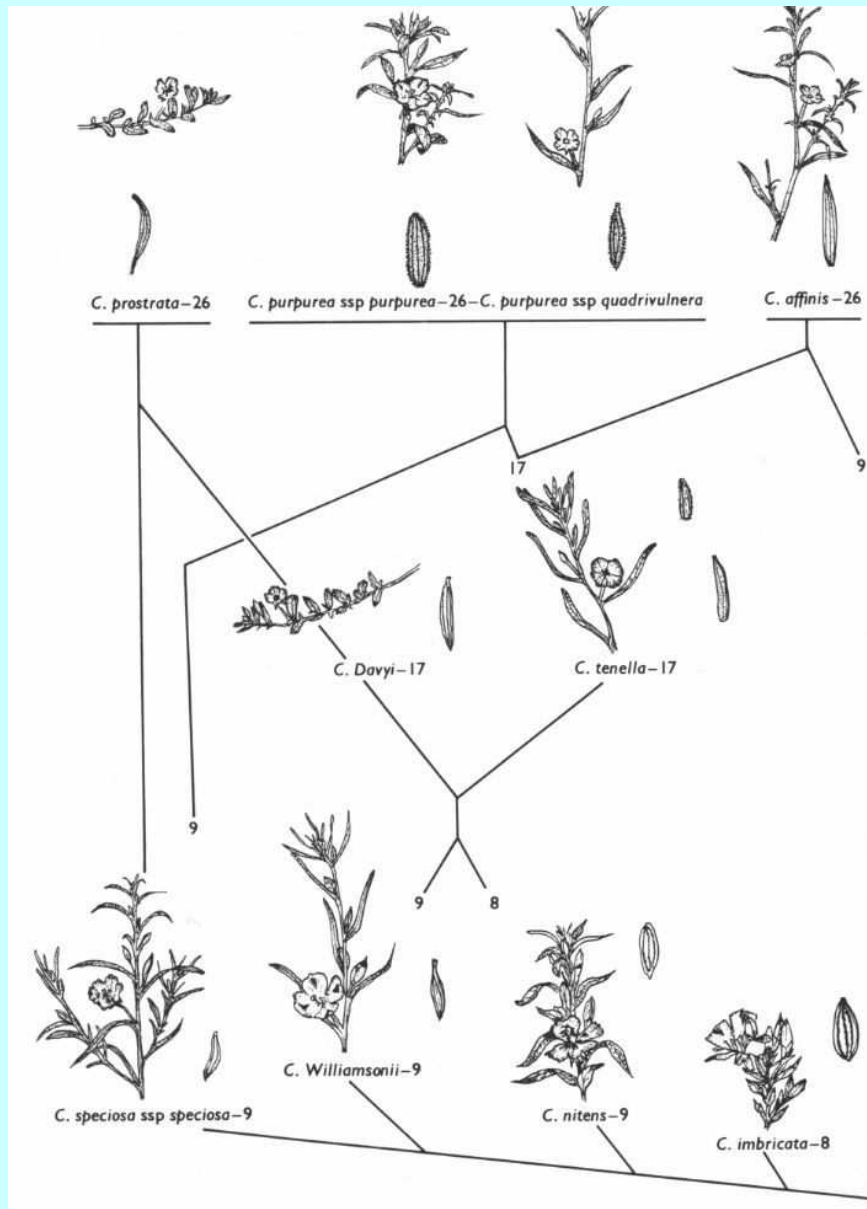
# The gametes are aneuploid



## And so are the offspring



# Polyploid evolution in Clarkia

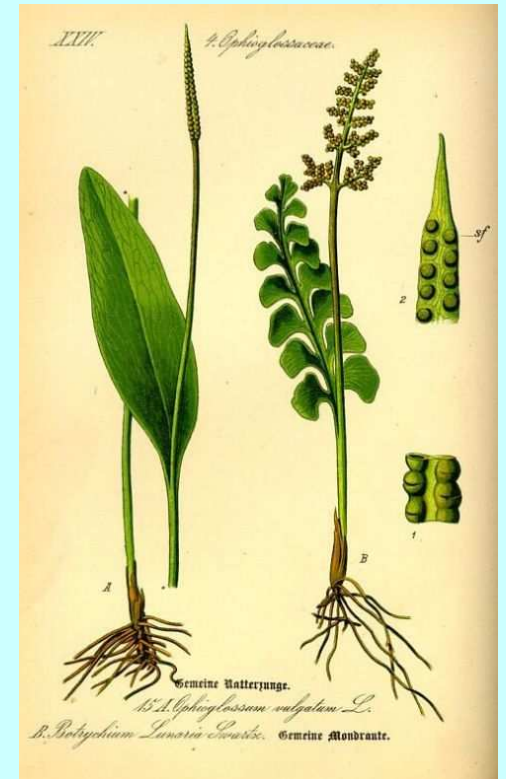


*C. Williamsonii*  
Fort Miller Clarkia

# The fern *Ophioglossum*, high number champion



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



Adders-tongue  
fern

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



# The ant *Myrmecia*, low chromosome number champion

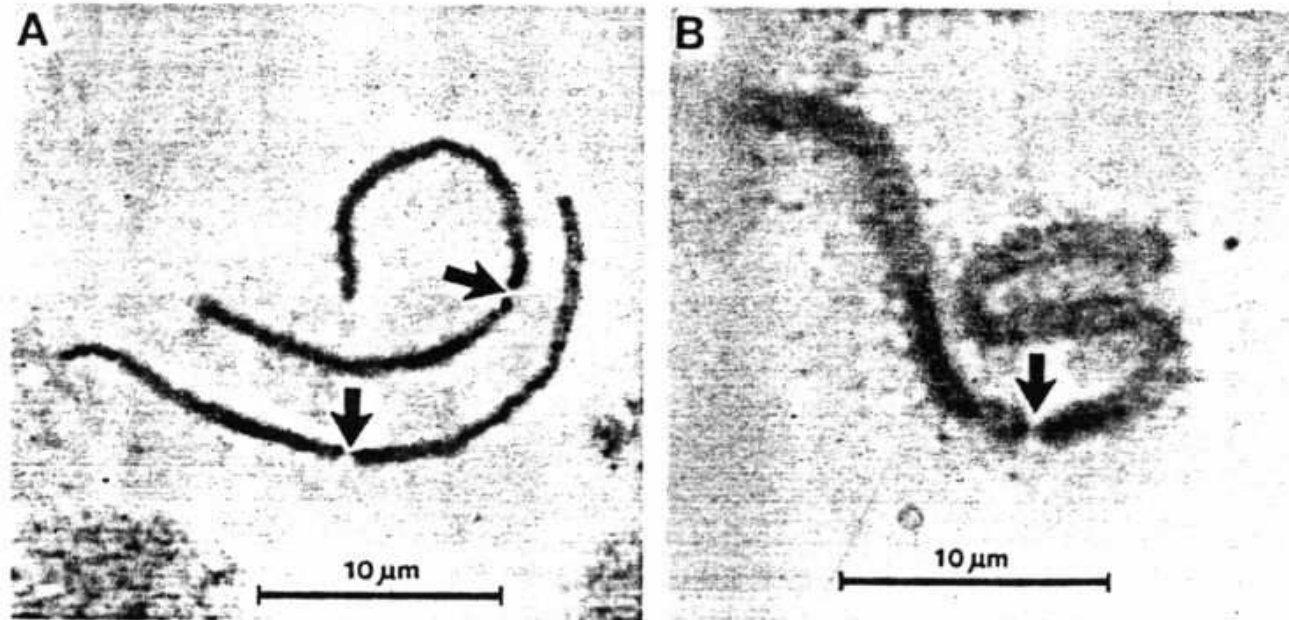


Fig. 1. Chromosomes from prepupal cerebral ganglia. (A) Worker prometaphase chromosomes. Identical C-banding provides evidence for homology of the two chromosomes. (B) Male prometaphase chromosome. Chromosomes consistently display a large centromeric C-band on the short arm and a smaller centromeric 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](#)

*Nature* **489**, 57–74 (06 September 2012) | doi:10.1038/nature11247

Received 24 November 2011 | Accepted 29 May 2012 | Published online 05 September 2012



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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](#)

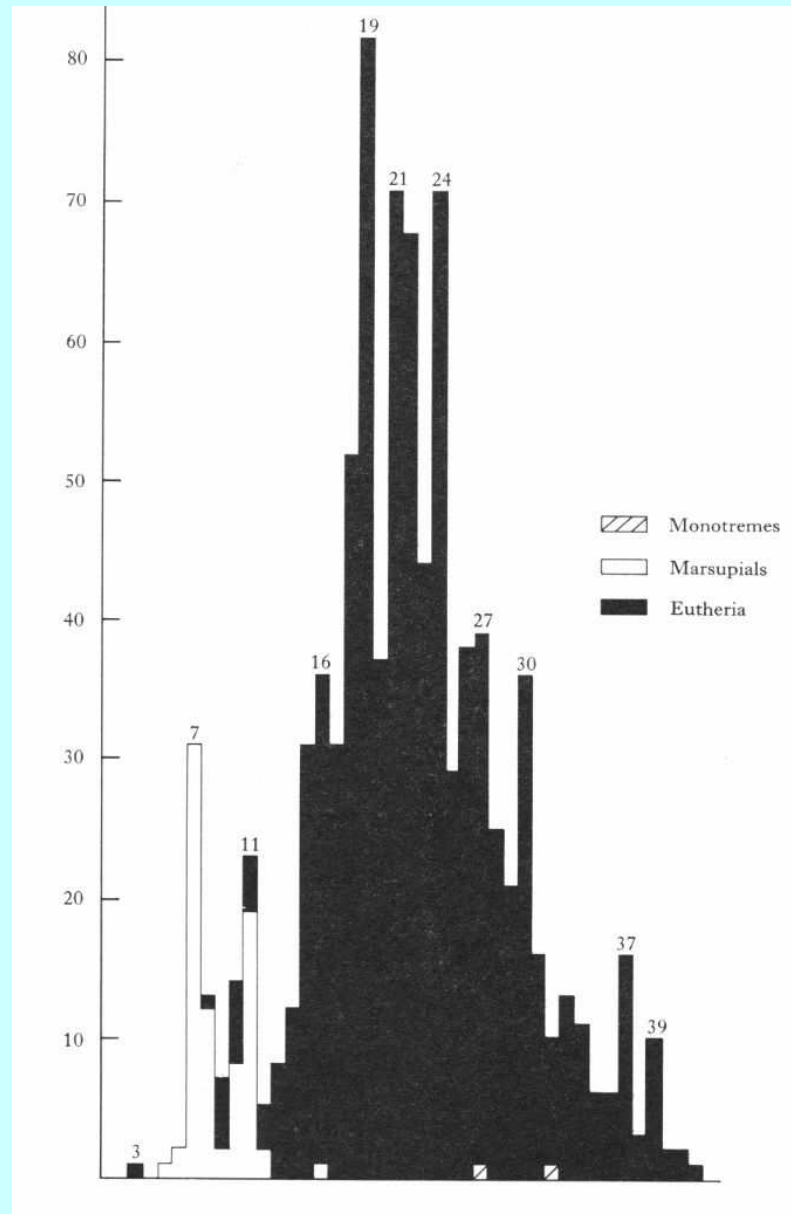
The human genome encodes the blueprint of life, but the function of the vast majority of its nearly three billion bases is unknown. The Encyclopedia of DNA Elements (ENCODE) project has systematically mapped regions of transcription, transcription factor association, chromatin structure and histone modification. 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.



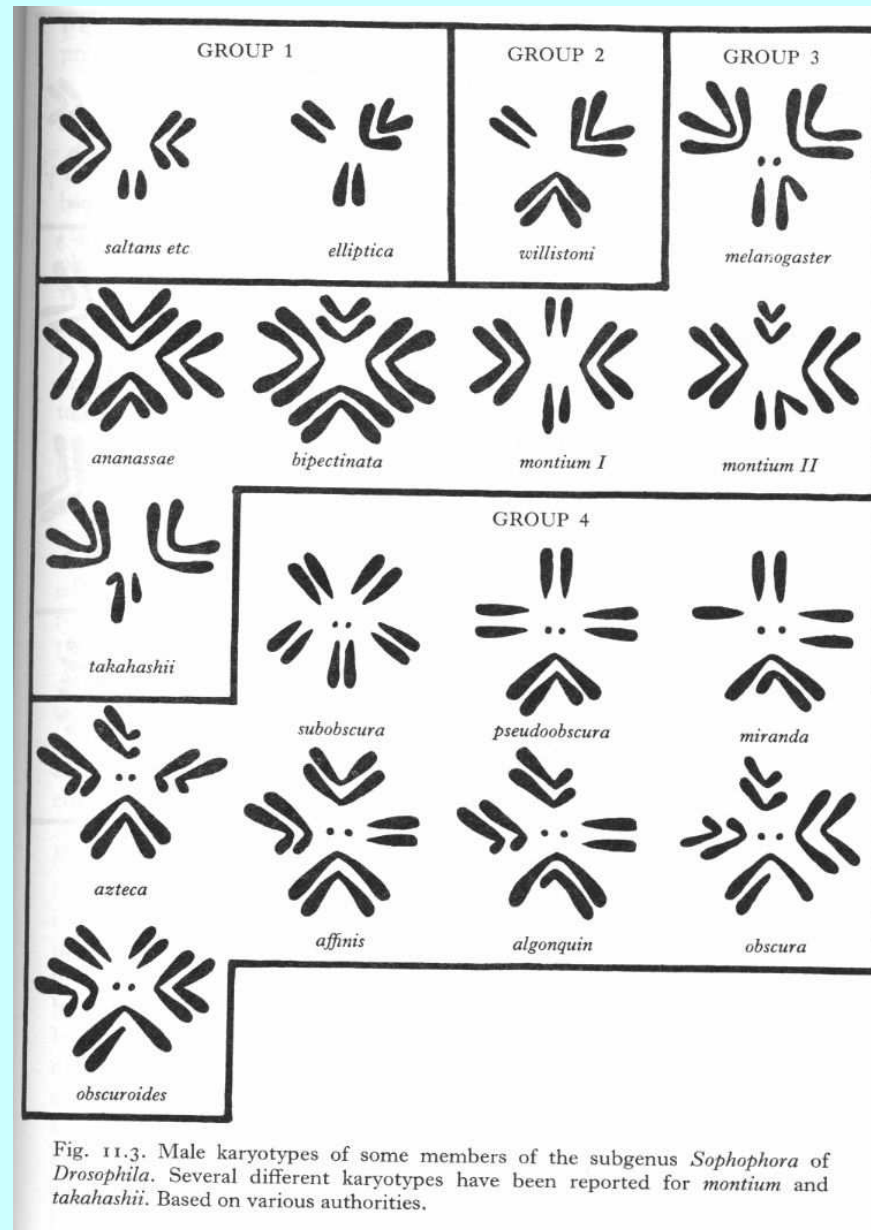
## Pros and cons

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

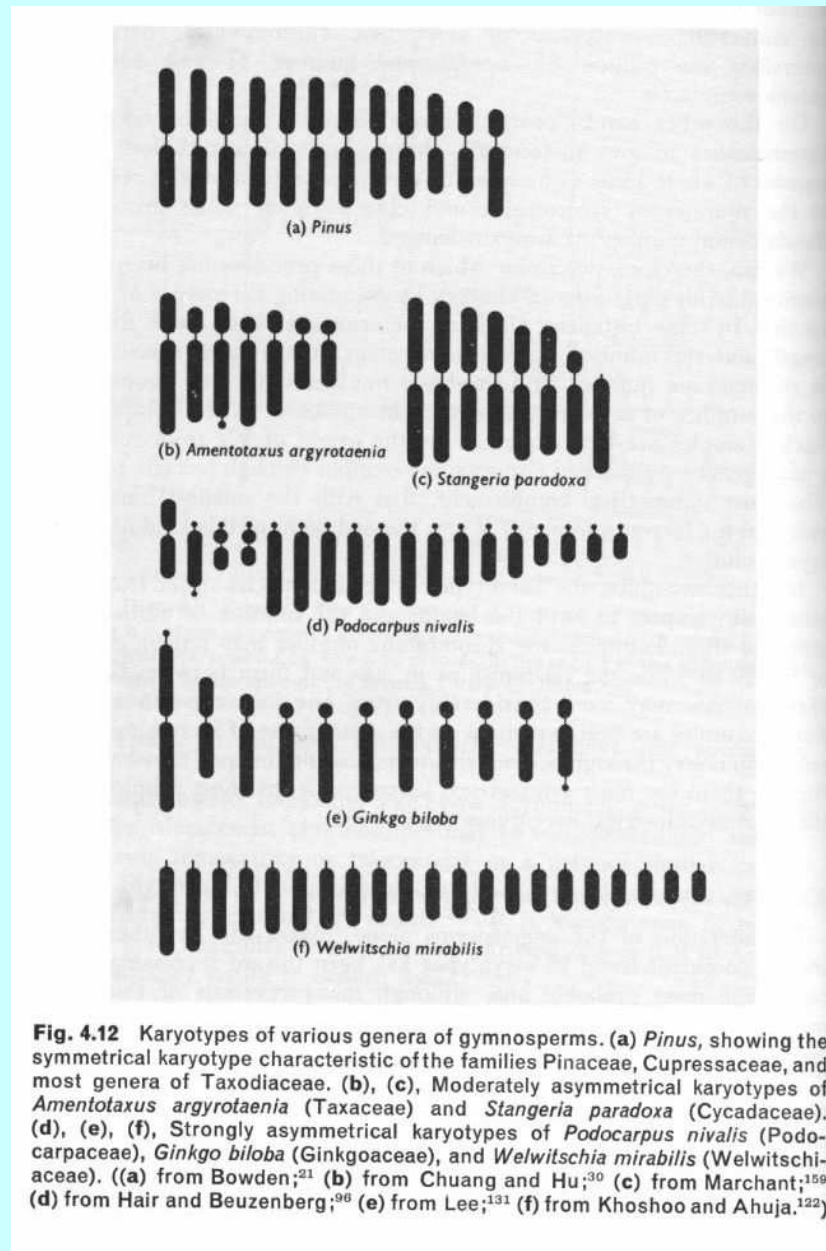
# Distribution of chromosome numbers in mammals



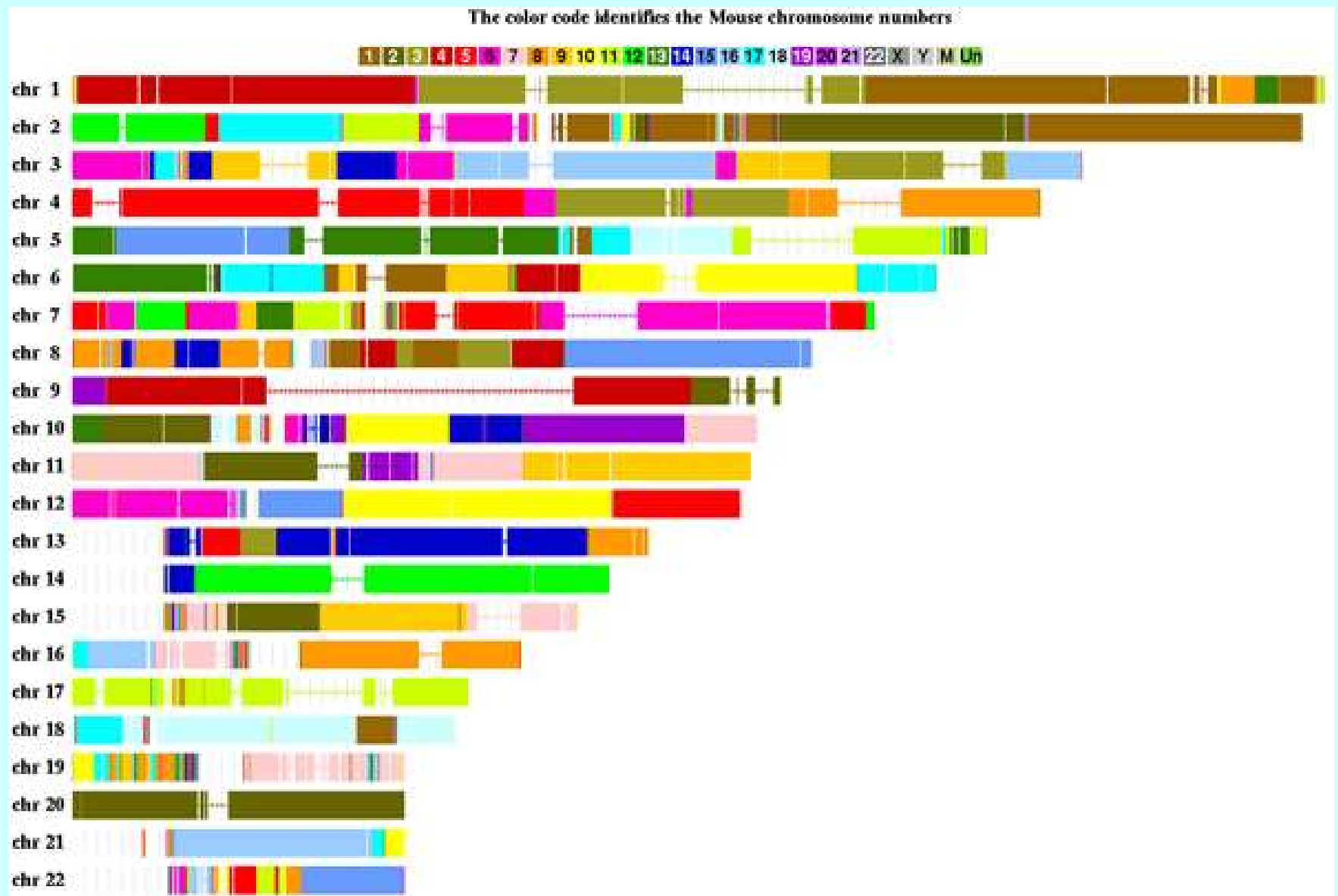
# Karyotypes of *Drosophila* species



# Karyotypes of gymnosperms



# Human and mouse genomes compared



# Human and chimp karyotypes compared

