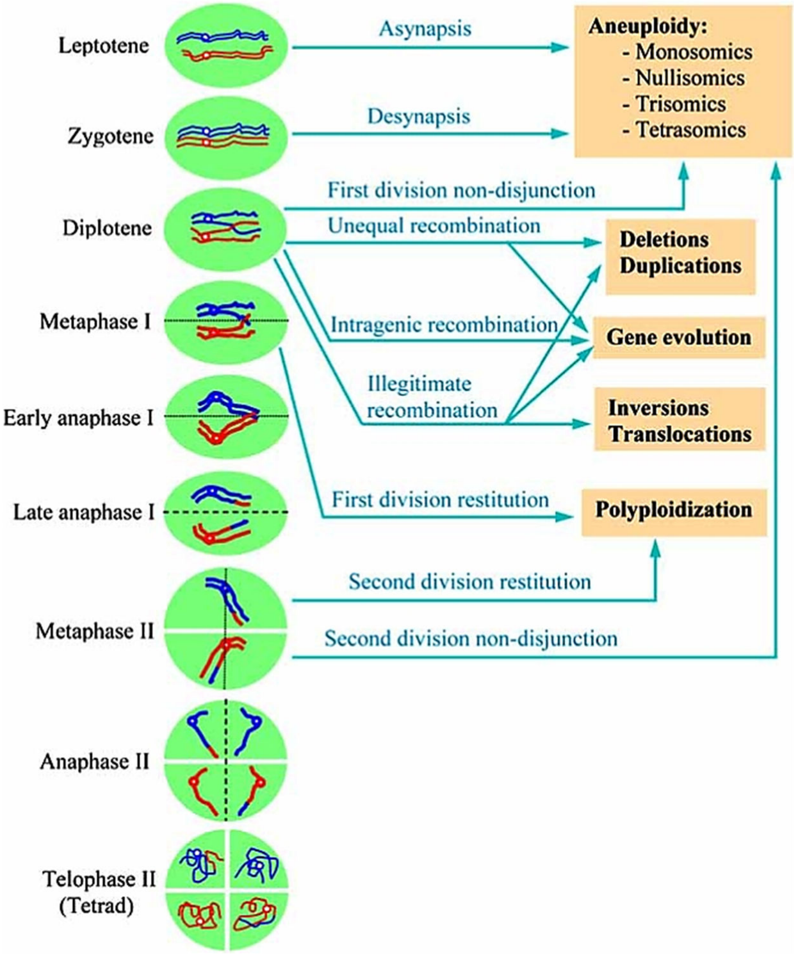
Meiotic alterations drive karyotype evolution

Cai & Xu, 2007
Review by **Heslop-Harrison and Schwarzacher, 2011**

“The genome is a metastable system, indeed an organelle, and not merely strings of DNA segments.”

- von Sternberg, 1996 [Acta Biotheoretica 44:95-118]

The point is that alterations in meiosis are what creates the changes that drive karyotypic evolution.



Synteny • Collinearity • Comparative mapping

"Syn: same; teny: threads" - the degree to which gene blocks have been conserved across species.

- Block of genes that have remained together in the same order.
- Highly related plants show more synteny than distant relatives

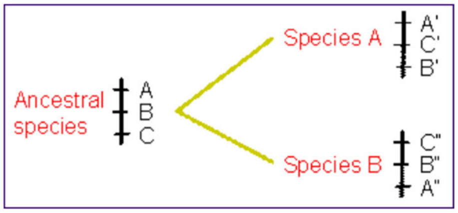
Initially, synteny determinations depended on chromosome pairing in wide hybrids (Vision, 2005)

- Then, genome-wide comparative maps in plants were constructed using molecular markers allowed synteny to be studied
  - Even if spp were not sexually compatible.

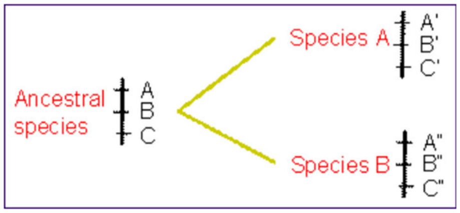
Some refer to sharing of blocks of genes as synteny and the sharing of the order of the genes as collinearity (Tang et al., 2008 Science 320, 486-488)

Synteny

Tang et al, 2008 (Paterson lab)



Collinearity



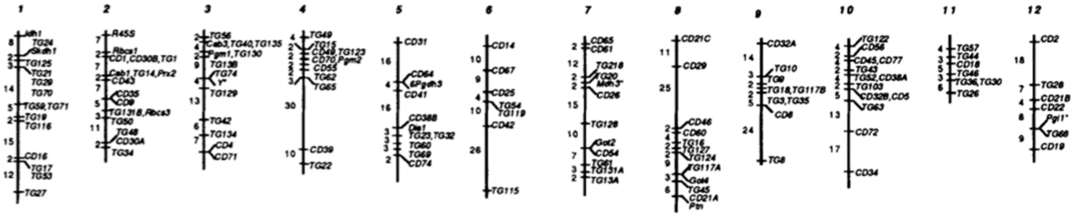
https://www.integratedbreeding.net/courses/genomics-and-comparative-genomics/www.generationcp.org/genomics/index42b0.html?page=1146

Alternatively, collinearity refers to genes at a small scale, while synteny is at a larger scale, such as a chromosome arm (Lovell et al, 2022).

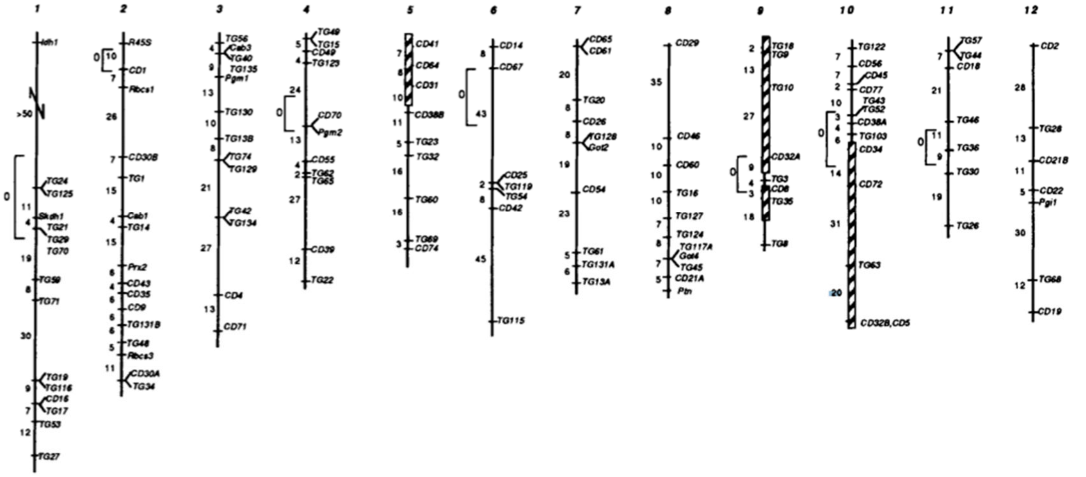
Macrosynteny

Bonierbale et al, 1988 (Tanksley lab)

Tomato and potato: A case of speciation without major chromosomal changes



Chromosomes of potato (top) and tomato (bottom)



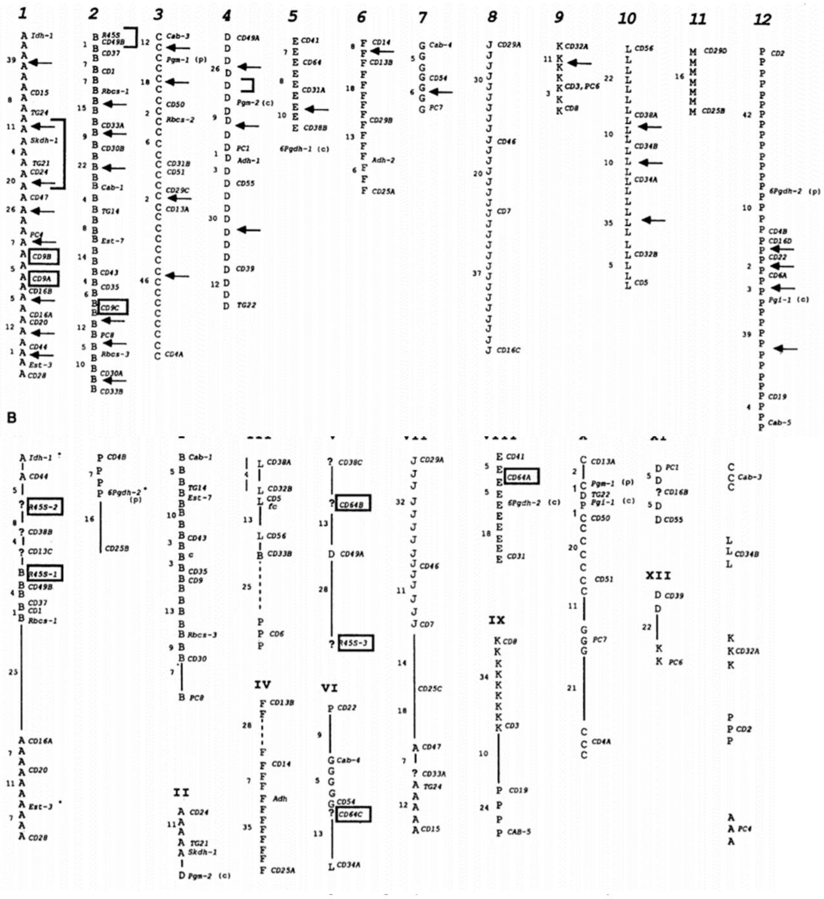
Separated by 2 paracentric & 1 pericentric inversions (hatched boxes)

- Otherwise, linear order of linkage groups remains the same

The larger size of the tomato genome reflects increased frequency of crossovers that take place, not larger chromosome size

Tanksley et al, 1988; Prince et al., 1993

Tomato and bell pepper: A case of speciation with major chromosomal changes



Maps of tomato (top) and bell pepper (bottom). Arrows indicate breakpoints.

Chromosome number remains the same

At least 15 breakage (inversion and translocation) points were necessary to explain the observed rearrangements:

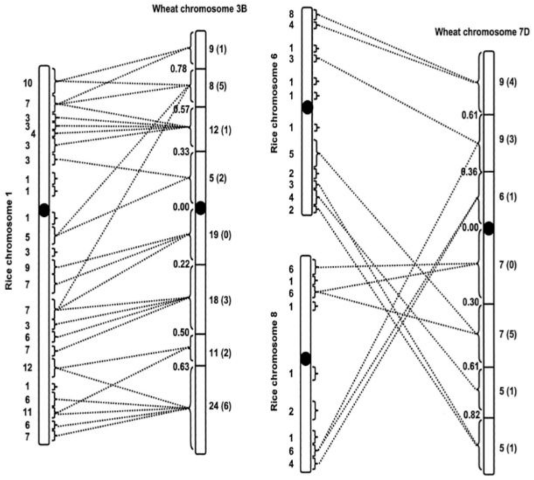
Pepper has 3-4× more DNA than tomato.

This is not reflected in map size. Long chromosomes have higher linkage.

31% of pepper map conserved relative to that of tomato

Besides translocations and inversions, centric fusions/fissions also took place

Some regions of the genome may be evolving more rapidly than others, to the point that heterologous probes no longer cross-hybridize, or sequences may have been lost during rearrangements



Wheat & rice synteny. Akhunov et al., 2003

Background on grasses

The best studied grasses (Poaceae), shared a common paleopolyploid ancestor about 42-47 million years ago.

Cereals show much collinearity, but have diverged in genome size, largely due to differential DNA amplification and elimination

Rice	430 million base pairs (Mbp)
Sorghum	760 Mbp
Maize	2,500 Mbp
Sugarcane	4,000 Mbp
Wheat	15,966 Mbp

Crop circles

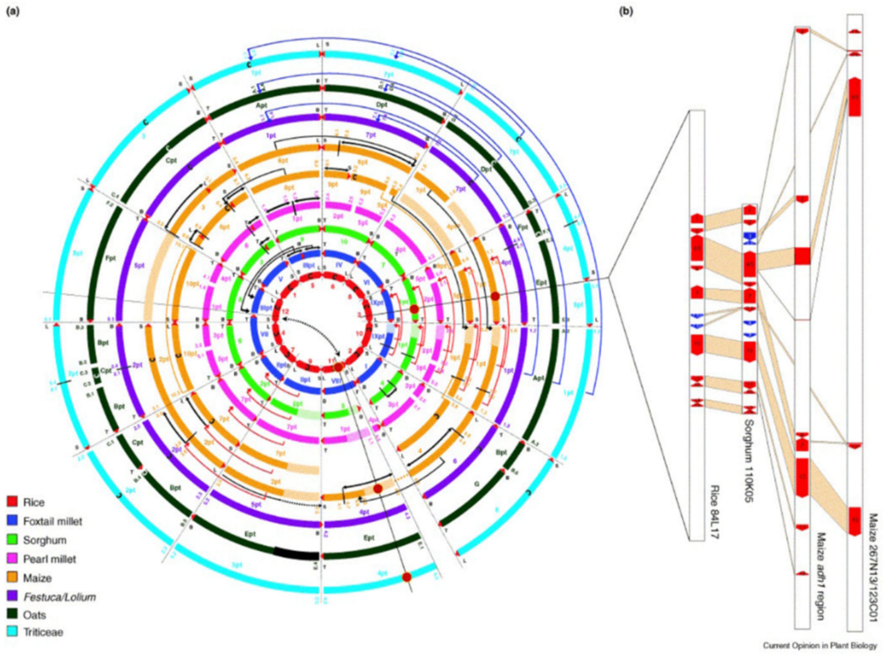
Devos, 2005

Originally aligned the genomes of eight major grass species: Grass chromosomes represented as ‘rice blocks’ on the basis of homology and/or conservation of gene order

At map level: identify genome segments or ‘linkage blocks’ consisting of large fragments that are largely colinear across species.

By comparing orthologous regions across multiple species, can infer the relative timing of the rearrangements and to assess the relative stability of the genomes analyzed.

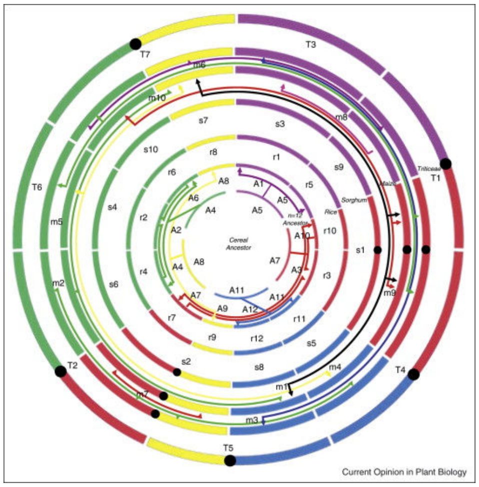
Most comparative applications originally used the rice genomic sequence as a source of markers to tag genes of interest in other grass crops.



Current Opinion in Plant Biology

Bolot et al., 2009

Now use the grass proto-chromosomes

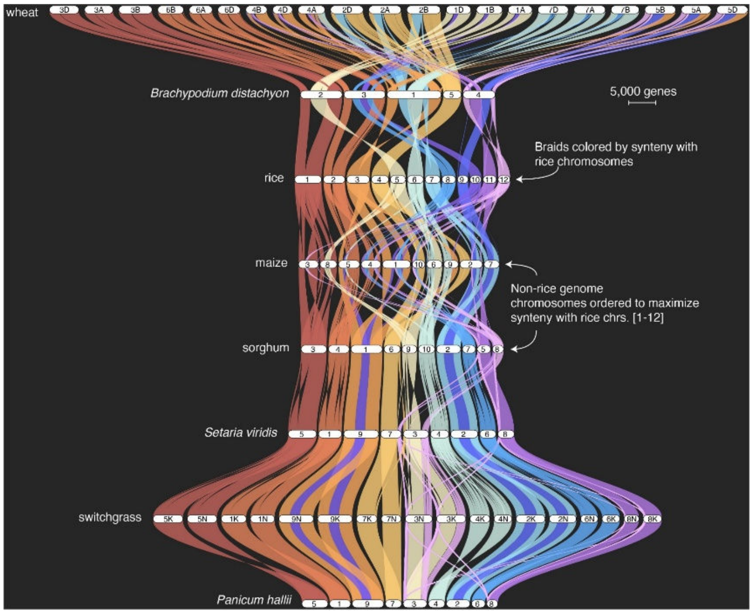


Current Opinion in Plant Biology

Lovell et al, 2022

GENESPACE

- Makes riparian plots



Evolution of chromosome number

Review by Mandáková & Lysak, 2018; Mayrose & Lyasak, 2021

Dysploidy

Babcock, Stebbins, & Jenkins, 1942

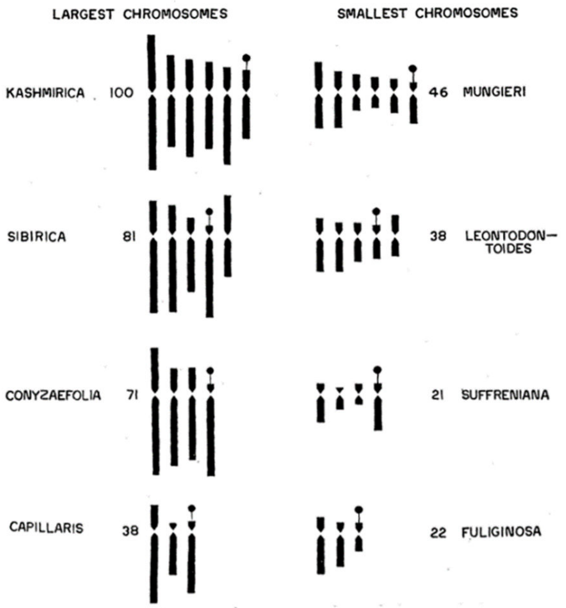
STEP-WISE VARIATIONS IN BASE CHROMOSOME NUMBER

- E.g., *Crepis,* base × = 6

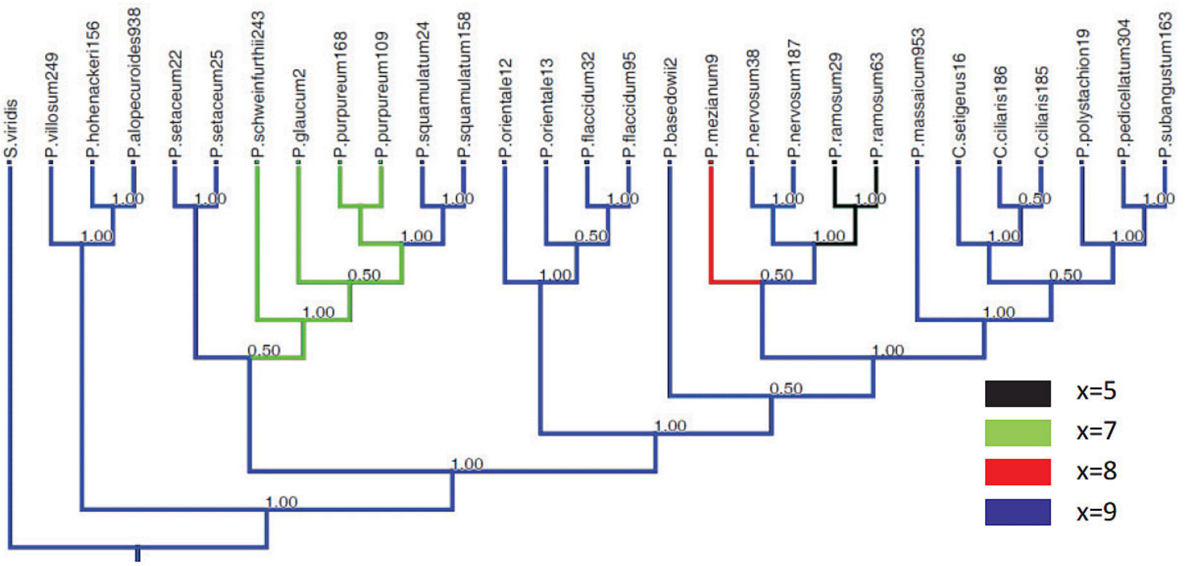
Dysploid series: a genus where step-wise variations in base chromosome number have occurred

- as evident by the chromosome number within the genus, eg., × = 5, 4, 3, or 7

Dysploids differ from aneuploids in that there is no major loss/gain in gene copy number in a dysploid



Dysploid series in Crepis; base × = 6. Babcock, Stebbins, & Jenkins, 1942



Dysploidy in the genus Pennisetum. Akiyama et al., 2011

Trends (and only trends) within a genus

Stebbins

Ancestral species tend to be:

- Perennial
- Cross pollinated
- High base number (x = 7 to 9)
- Symmetrical karyotype

As species invade less favorable environments, they evolve from the ancestral types into derived types adapting to:

- Wet/dry seasons
- Lack of pollinators
- Etc.

The new species tend to be highly adapted and have a short life span, they tend to:

- Be annuals
- Be self-pollinated
- Have a lower × number
- Asymmetrical karyotype

Eg, within *Happlopapus* species (compositae):

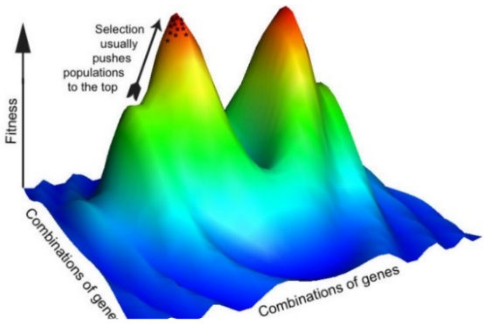
Woody perennials	× = 9
Herbaceous annuals	× = 6
Annuals	× = 2, 3 or 4

The effect of lower chromosome number is to increase the amount of linkage, which in turn is more likely to preserve favorable genetic combinations for unfavorable environments.

Chromosome number vs linkage

Stucky & Jackson, 1975

n	gamete number
4	256
5	1024
6	4096
7	16,384
8	65,536
9	262,144
10	1,048,576



https://www.discovermagazine.com/health/a-thousand-little-adaptive-platoons- *Adaptive peaks and valleys, after Sewell Wright*

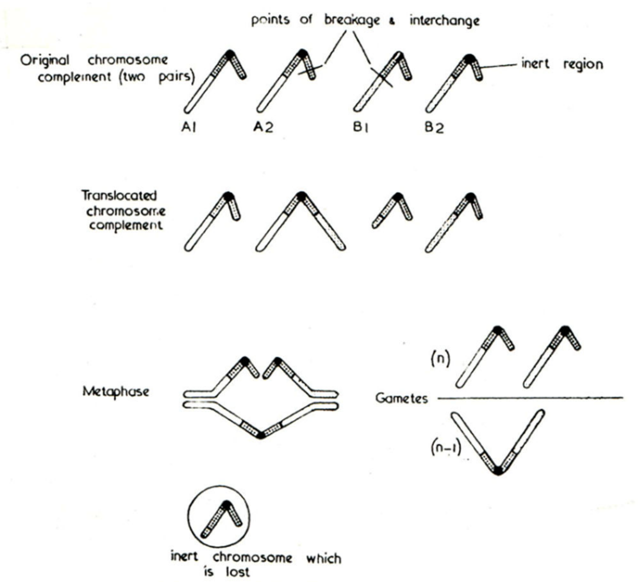
- If each II is heterozygous for 1 locus, then the number of gametes possible is 2^n
- If ∃ 1CO/II, then gametic number = 4^n (what is shown in the table above)

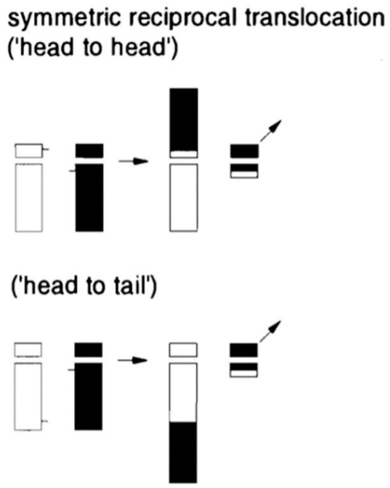
Mechanisms of dysploidy

Darlington, 1937

1) Progressive reduction or descending basic aneuploidy

Darlington, 1937 **Schubert et al, 1995**





Happens at the genus level.

The events leading to head-to-head or tail-to-tail fusion were previously explained by Darlington:

- Start with acrocentric chromosomes
- Unequal translocation of material to other chromosomes
- Loss of the remaining centromere and surrounding heterochromatin

Though Darlington called it inert, it would now be called pericentromeric chromatin, and some genes would be present on it.

Schubert et al, 1995

Chromosome number decrease via symmetric reciprocal translocation

- Notice that head-to-head translocations lead to formation of a metacentric chromosome.
- Notice that a head to tail translocation leads to the formation of another telocentric chromosome

End to end fusions - overview

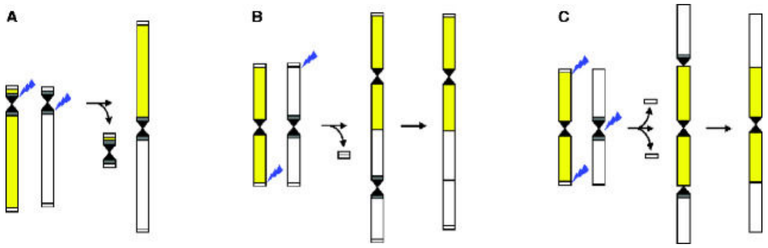
Mayrose & Lysak, 2021; Lysak, 2022

DSB's in telomeres during bouquet stage

- Such that a non-homologous chromosome is used as the repair template, leading to translocations

The result is dicentric or tricentric chromosomes

- Need to delete or inactivate the extra centromeres



Schubert & Oud, 1997

There is an upper limit to chromosome length

- ½ the length of the spindle axis at telophase
- Longer chromosomes cannot be pulled apart prior to completion of cell division
    - Leads to DNA loss

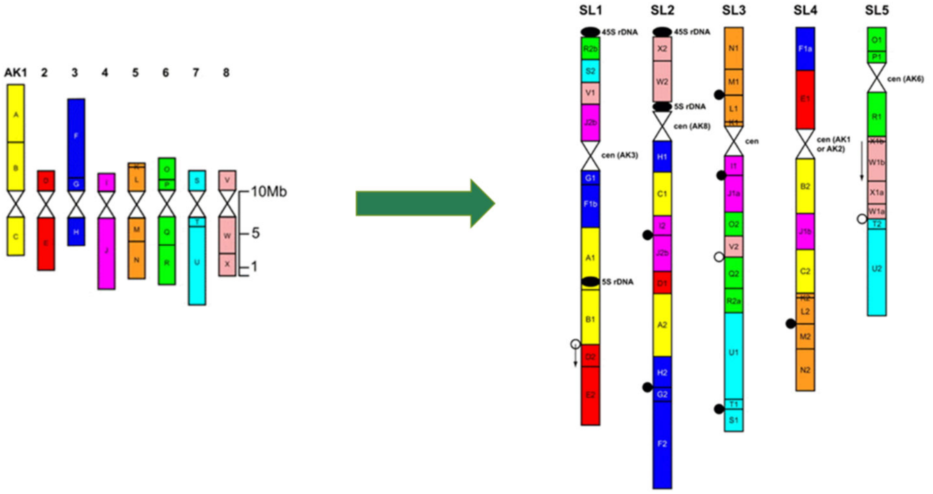
Centromere fate

Han et al, 2006

It is becoming evident that the centromere does not have to be eliminated in this process.

- When a chromosome has more than one centromere, it is able to silence the extra centromeres

Mandáková et al, 2010



The left idiogram is of the ancestral genome for Brassica. The larger one is for *Stenopetalum lineare.*

Black circles represent silenced centromeres

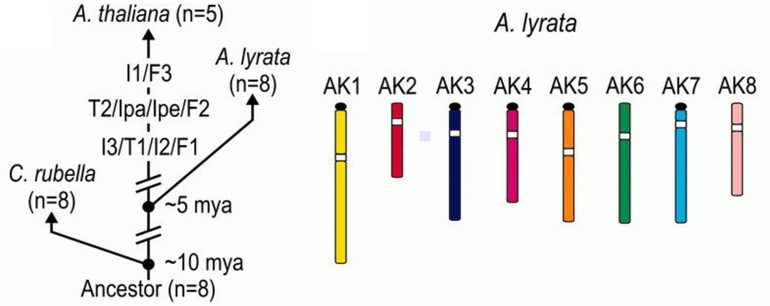
White circles are where different chromosome arms have fused together and the centromere has disappeared.

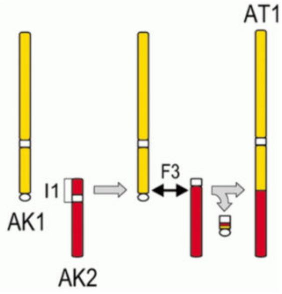
Example

Lysak et al, 2006 (Shubert Lab)

Arabidopsis ancestor had 2n = 8. Arabidopsis is 2n =5:

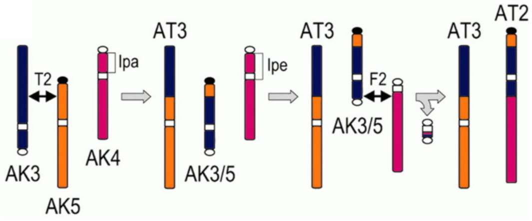
I = inversion; Ipa = paracentric inversion; Ipe = pericentric inversion; T = translocation; F = fusion



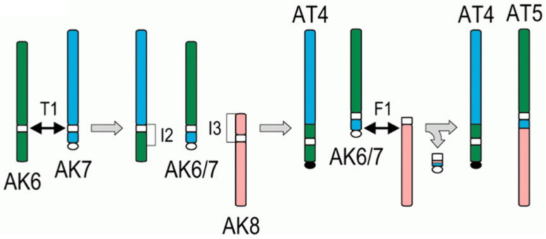


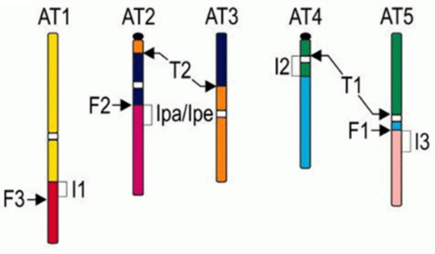
The AT1 chromosome is created when AK1 & 2 undergo a head to tail fusion, after a pericentric inversion in AK2 creates an acrocentric chromosome.

Next, the AT2 and AT3 chromosomes are created from the AK2, 4, and 5 chromosomes through translocations & inversions. As before, a pericentric inversion is needed to create an acrocentric chromosome



Finally, the process is repeated to create the AT 4 and 5 chromosomes from AK 6, 7, and 8. AT4 is acrocentric, as would be expected from a Head to Head fusion





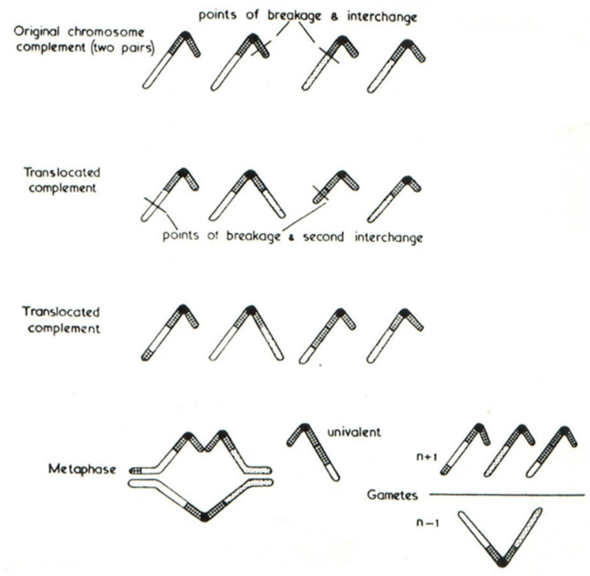
Pattern

- Inversions create telocentric chromosomes which then fuse
- So, the Darlington/Schubert model appears to be correct, at least for arabidopsis

Progressive increase

Darlington, 1937

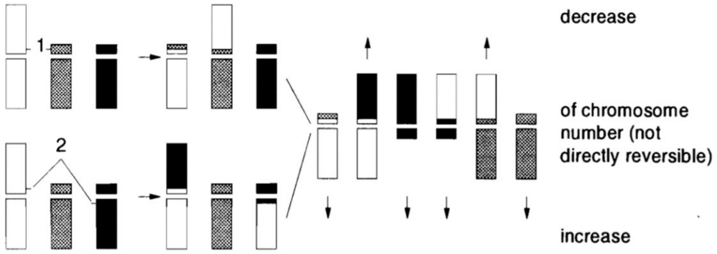
Not very common



Multiple translocations

Schubert et al, 1995

Notice that multiple translocations can lead to either an increase or a decrease



2) Progressive reduction + polyploidy

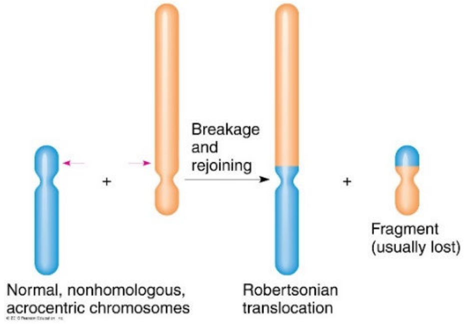
- Same result as progressive increase

3) Polyploidy → progressive reduction

- Also called the polyploid drop

- Also looks like a progressive increase

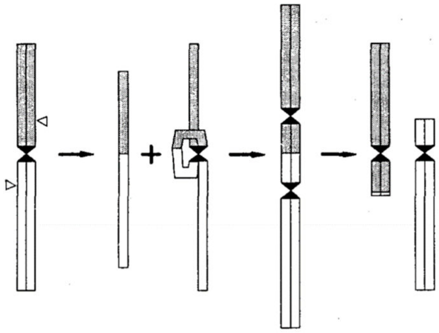
5) Robertsonian fusion/fission



- The fusion/fission of 2 telocentric chromosomes to form a metacentric chromosome.
- Is reversible

6) Number increase via a Breakage-Fusion-Bridge Cycle

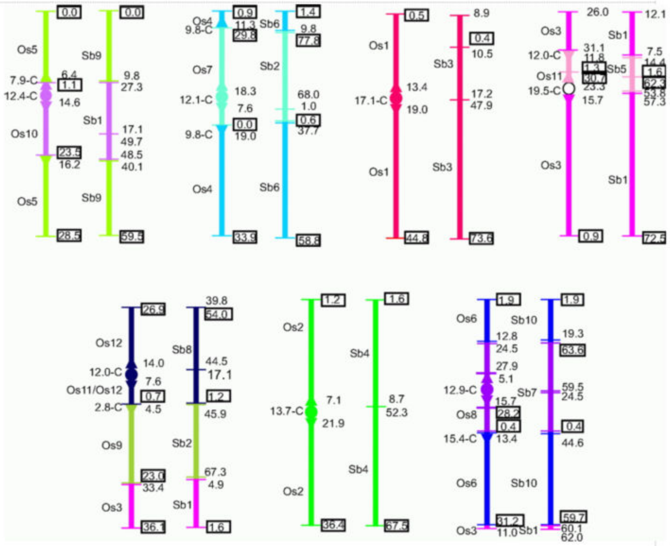
Fuchs et al, 1995

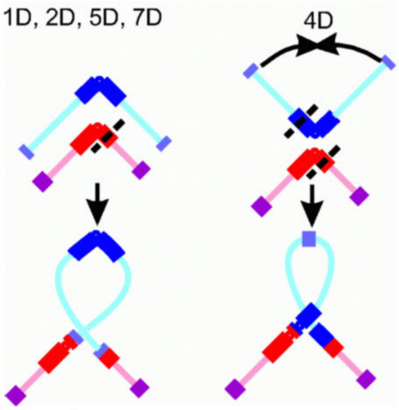


7) Chromosomal insertion into centromeres

Luo et al., 2009

Is how the monocot ancestral number of 12 got reduced to 7 in the triticeae and some other grasses

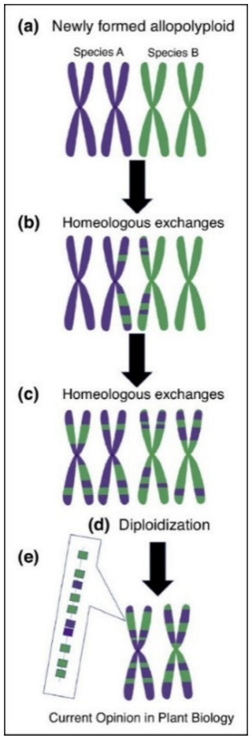




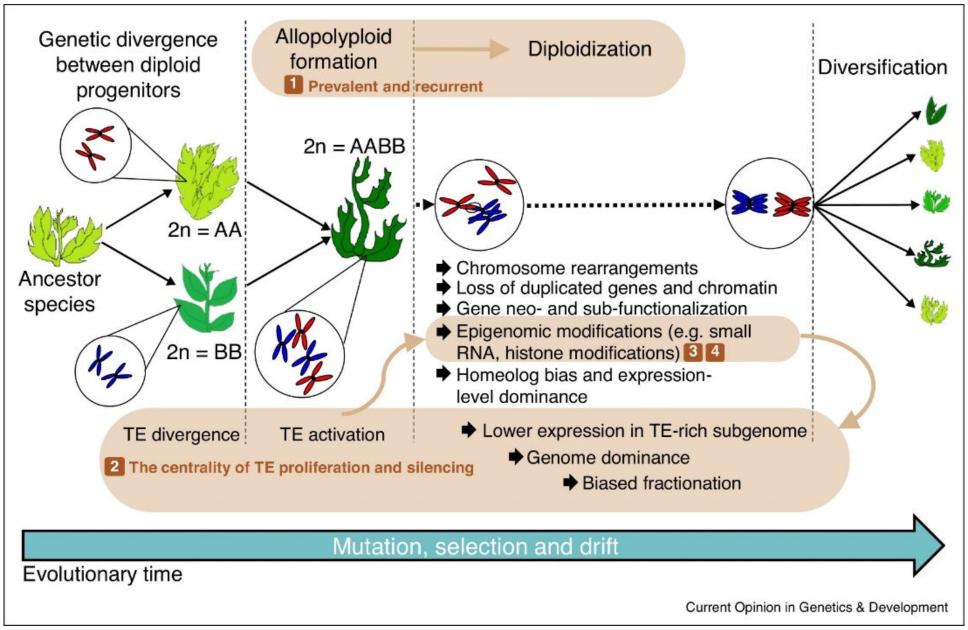
2 versions of the chromosome insertion model explain the current triticeae chromosomes

Diploidization

Dysploidy + Fractionation → diploidization



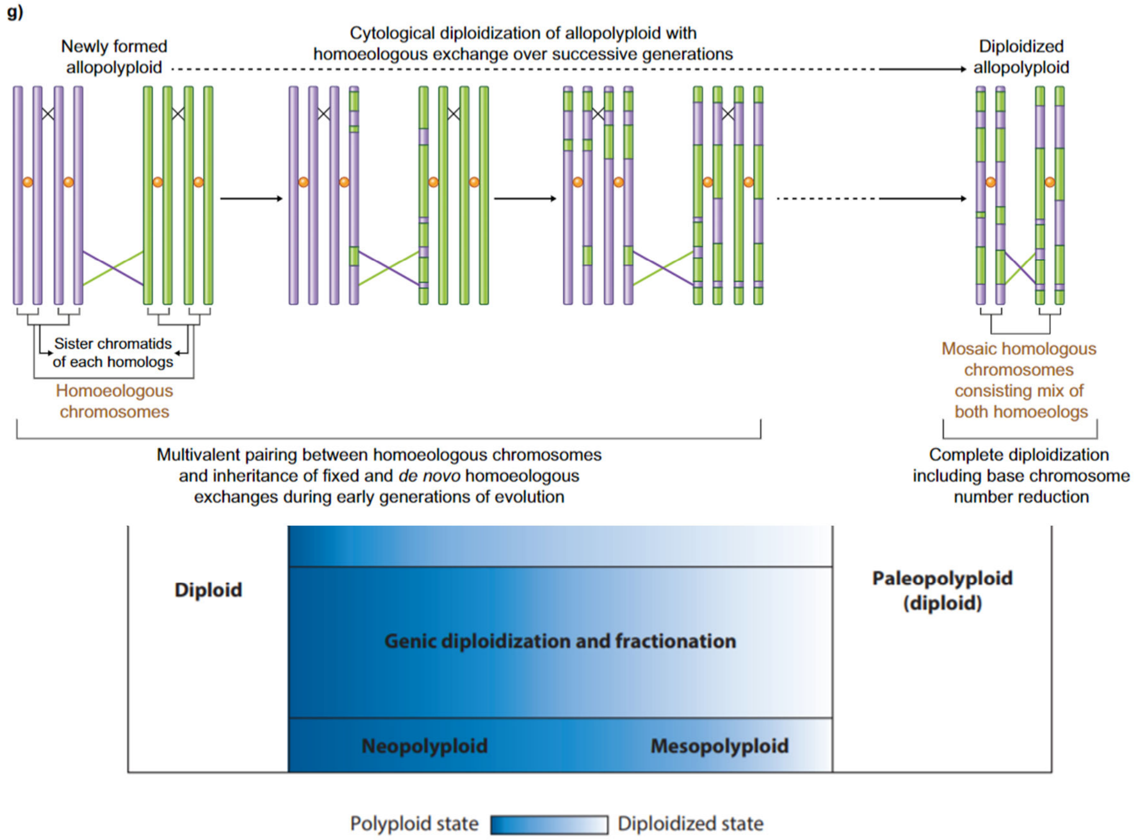
From Edger et al, 2018



From Wendel et al, 2018

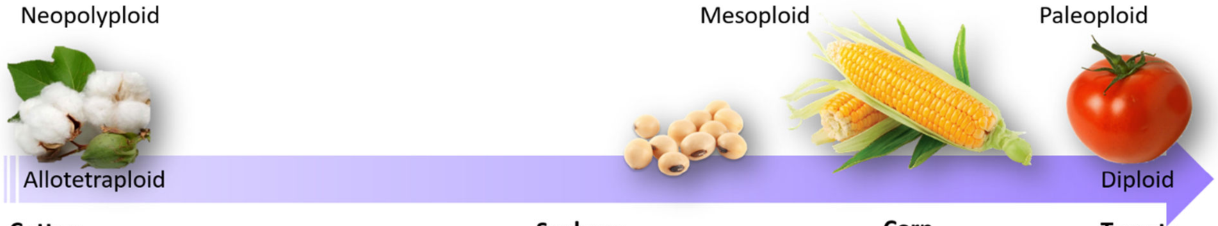
Deb et al, 2023

- In addition to fractionation, homoeologous exchange contributes to diploidization



Diploidization is a gradual process

Some crops are more diploidized than others



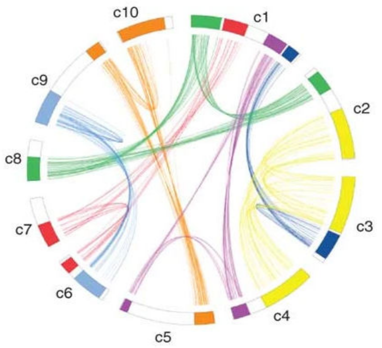
Cotton	**Soybean**	**Corn**	**Tomato**
2n = 4x = 52 *or* n¼ = x = 13	2n = 4x = 40 *or* 2n = 2x = 40	2n = 2x = 20	2n = 2x = 24
(40K x 2) genes	55K genes	n = x = 10	n = x = 12
		42K genes	31K genes

Evidence of polyploid ancestry in modern diploids

A diploidized polyploid

Cacao, 2n = 2x = 20

Argout et al, 2011



A mesopolyploid (in the process of becoming a diploid)

Soybean 2n = 2x/4x = 40

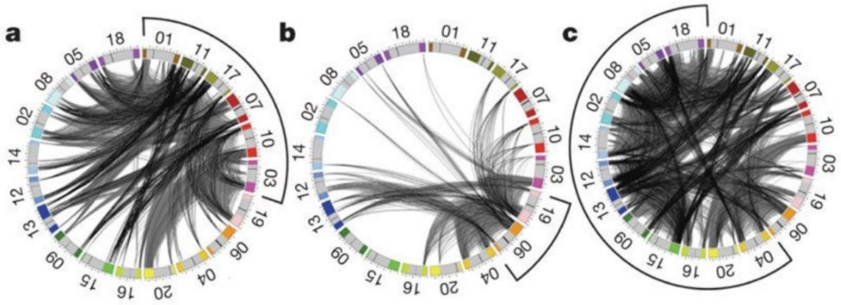
Schmutz et al, 2010

Compared duplicated segments in soybean

20 chromosomes arranged in a circle

- Colored areas are the gene-rich regions

- Grey represents 13-Myr genome duplication
- Black represents 59-Myr genome duplication



Cycles of whole genome doubling

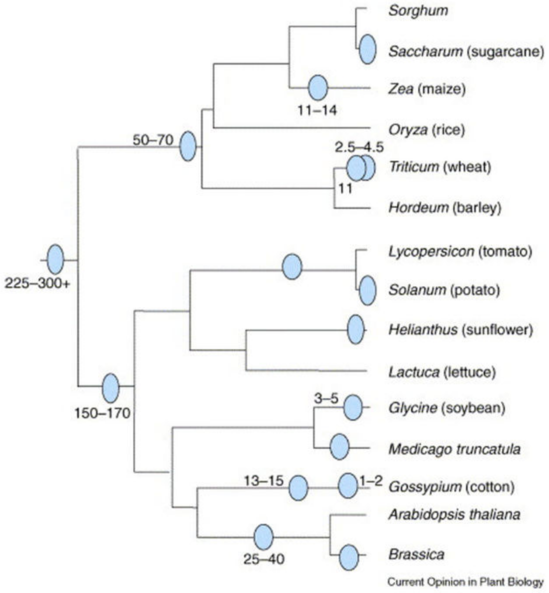
Adams & Wendel, 2005

Inferred polyploidy events during the evolution of angiosperms

Blue shaded ovals indicate suspected large-scale duplication events

Branch lengths are not to scale

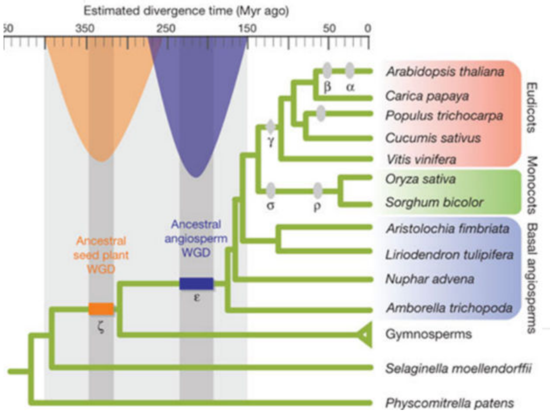
Numbers indicate roughly estimated dates (in millions of years) since the duplication event



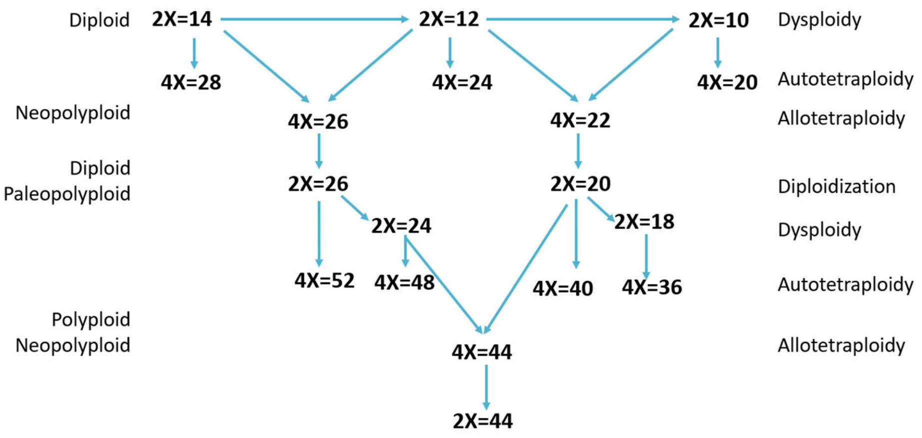
Current Opinion in Plant Biology

These WGDs have been given names

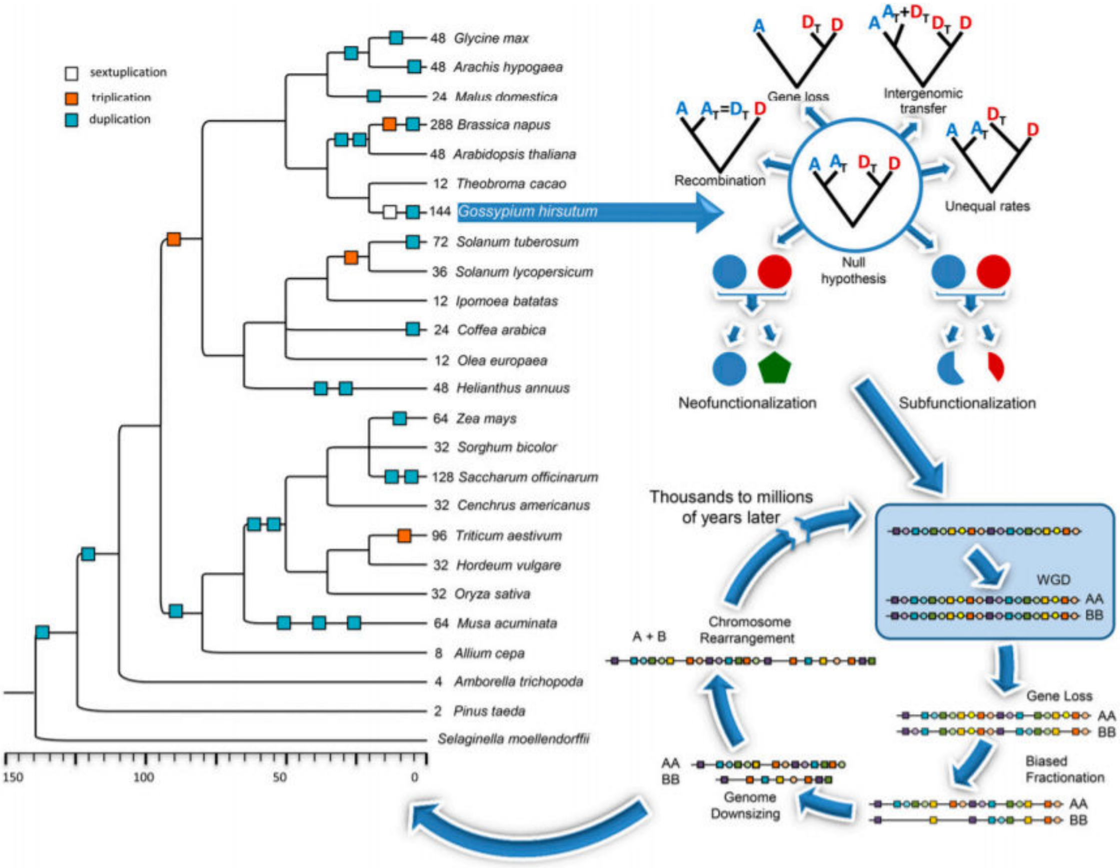
Jiao et al, 2011



Putting all the mechanisms into action



Wendel, 2015

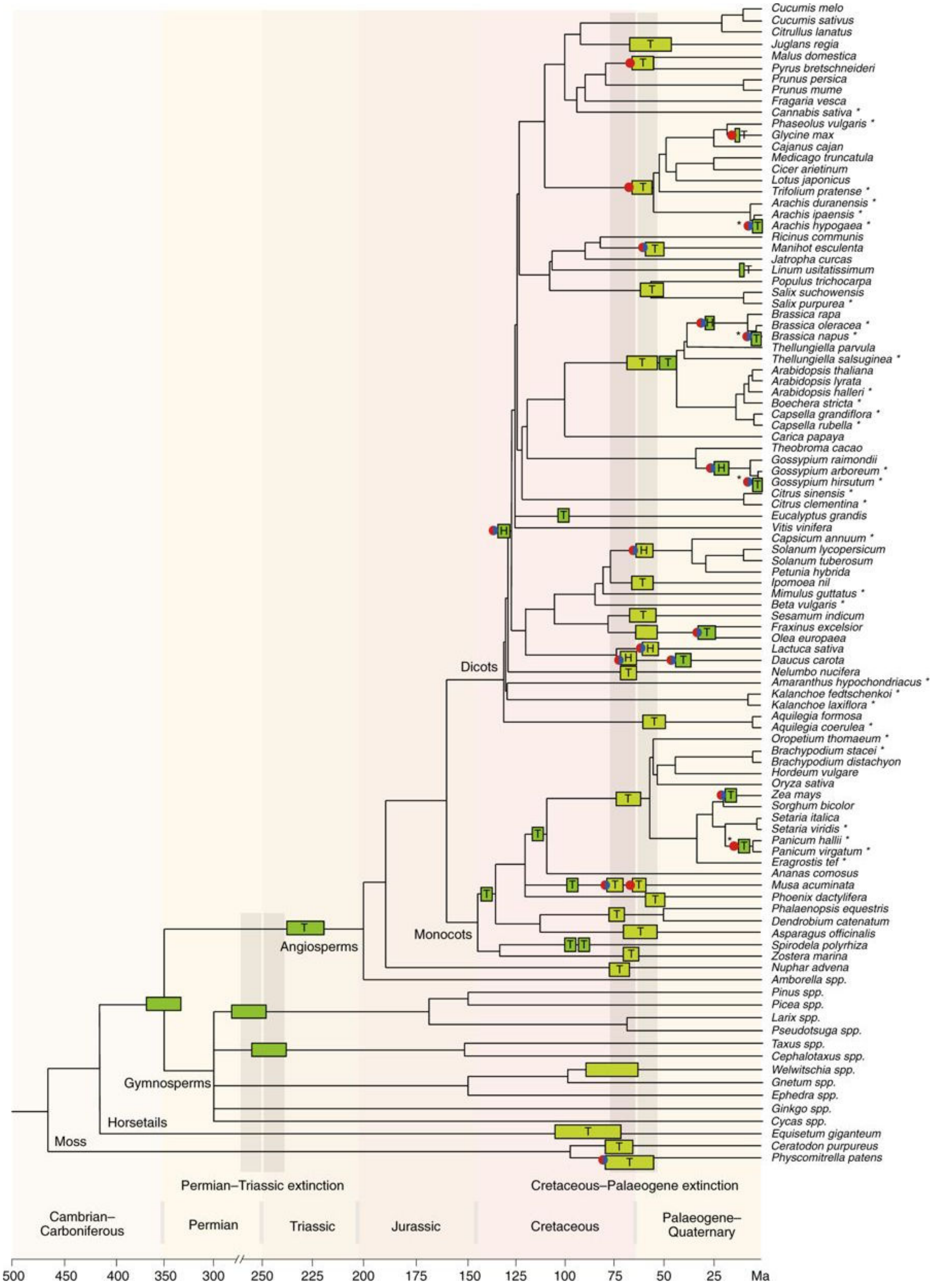


- Currently, × ranges from 2 to at least 20
- The ancestor to the angiosperms probably had × = 7
- Since that time, repeated cycles of polyploidization → dysploidy → diploidization have occurred, giving a wide range of base numbers
- In this case, the original polyploid is called a paleoploid, and the derived diploid is called a neoprimary diploid or a neoploid.
    - Not all authors agree on the terminology, and all varieties are found in the literature

- Plants with × numbers of 9 or greater are probably in this category
    - Homoeologous chromosomes no longer exist due to rearrangements
    - Duplicate loci have diverged into different functions

Polyploidy in plant lineages

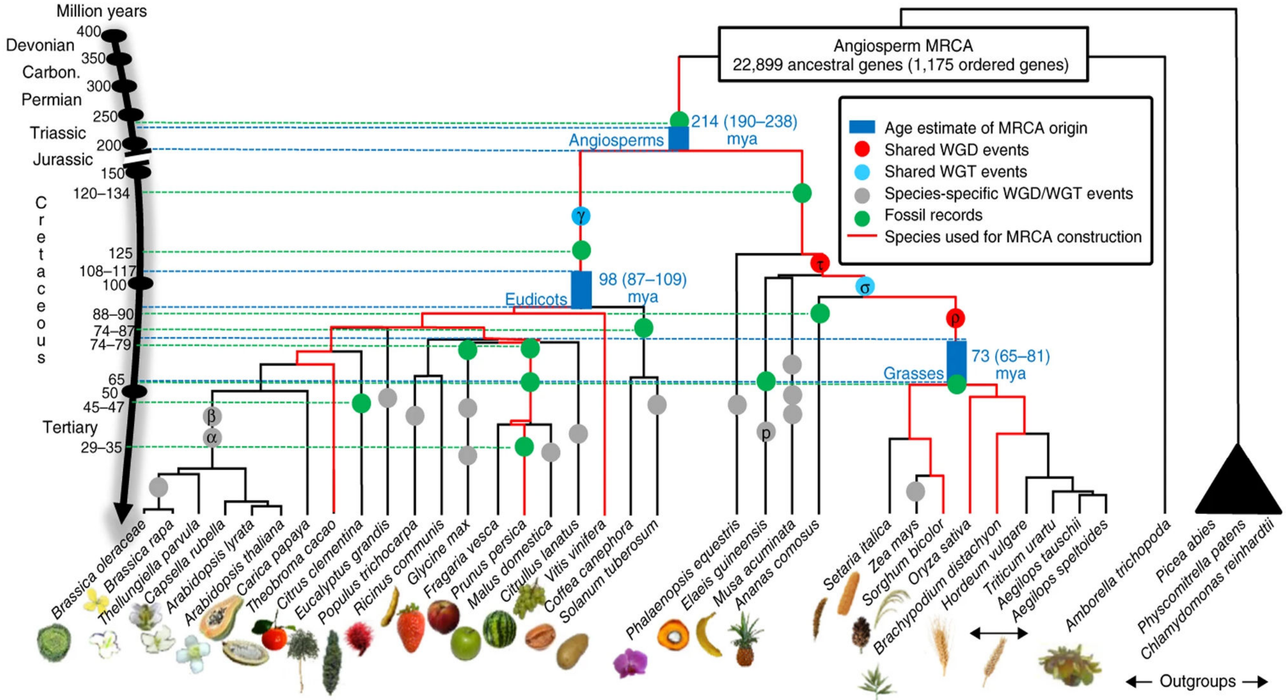
Cheng et al, 2018



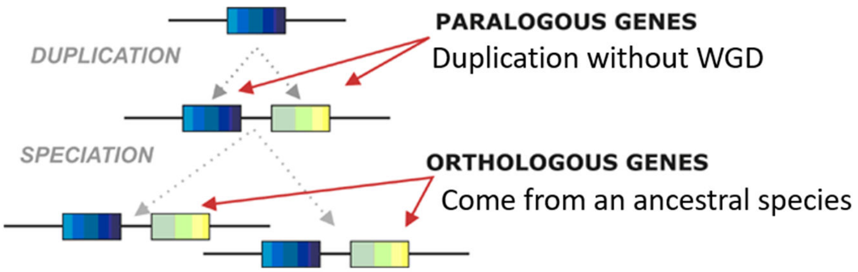
Murat et al., 2017

Shows the timing of whole genome duplications across the angiosperm tree of life

- Authors leveraged sequenced genomes to time duplications



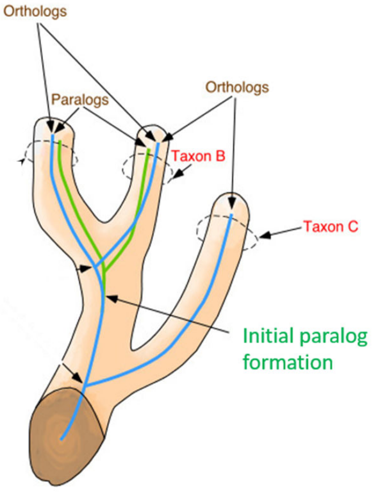
Terminology



Duplication without WGD

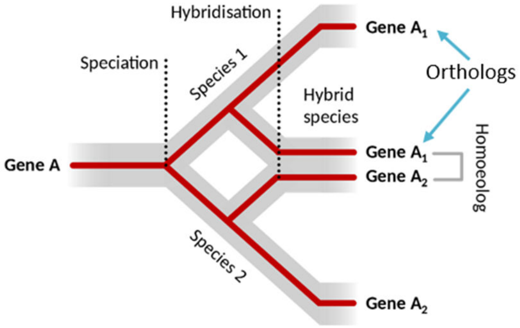
Come from an ancestral species

http://biol.lf1.cuni.cz/ucebnice/en/glossary.htm



Genes that started out as paralogs in an ancestral species remain paralogs.

electronica.org/2000_1/retinal/fig17.htm



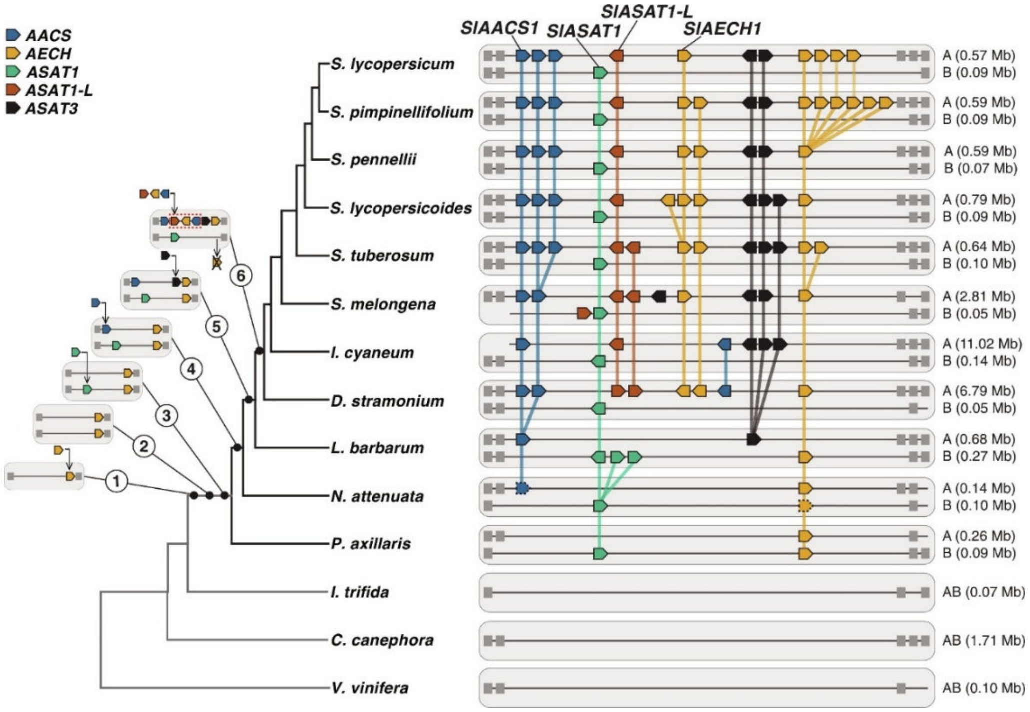
Orthologs become homoeologs upon allopolyploidization
https://en.wikipedia.org/wiki/Sequence_homology#/media/File:Homoeology.svg

- Paralogous genes– evolve through duplication of individual genes followed by divergence
  - Whole genome duplication is not involved
  - Paralogs may have originated in a given species, or in the ancestor of a species.
- Orthologous genes- evolves through divergence as species with common ancestor diverge.
- Orthologous genes after allopolyploid formation are called homoeologous genes
  - Whole genome duplication is involved.

Role of paralogues

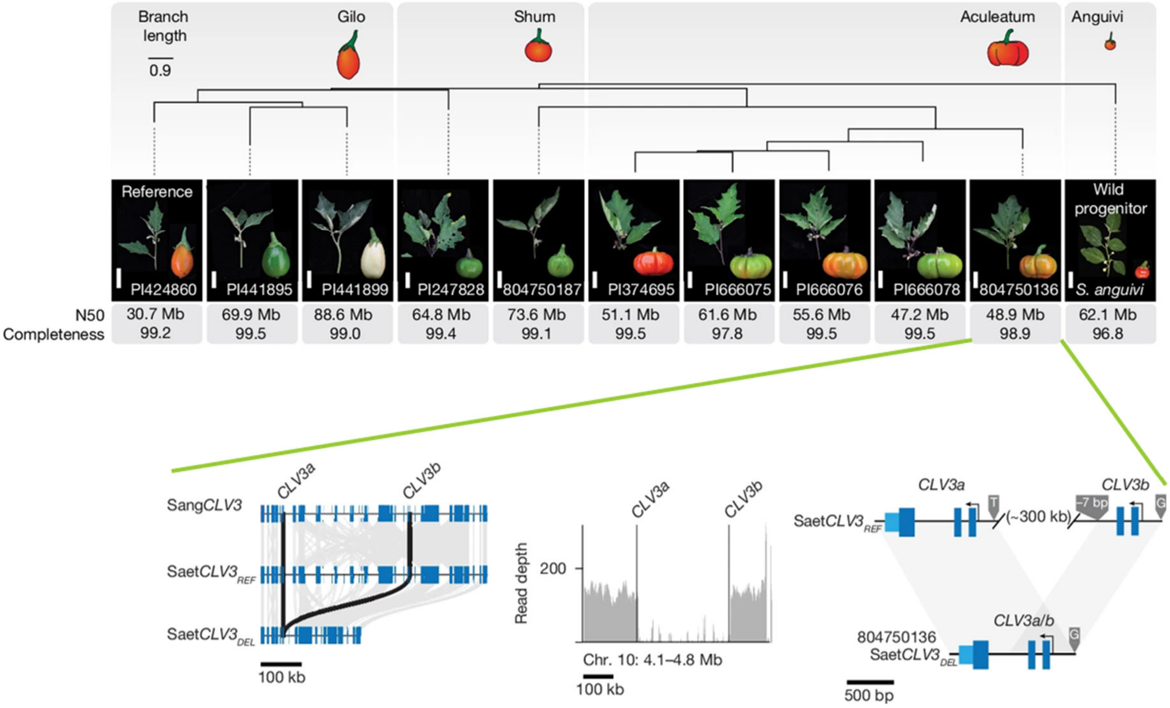
Kerwin et al, 2024

- Expansion allows evolution of metabolic pathways. In this case, it is production of acyl sugars in different solanaceous spp.



Benoit et al, 2025 (https://doi.org/10.1038/s41586-025-08619-6)

- In this case in an Afrida eggplant (*S. aethiopicum*), one paralog lost function due to a deletion.
- Then, a 300 kb second deletion of the DNA between the paralogs formed a functional, chimeric gene.

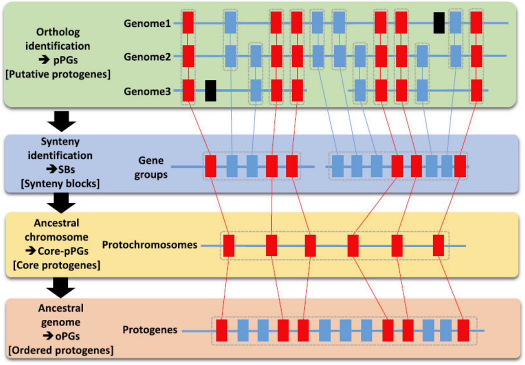


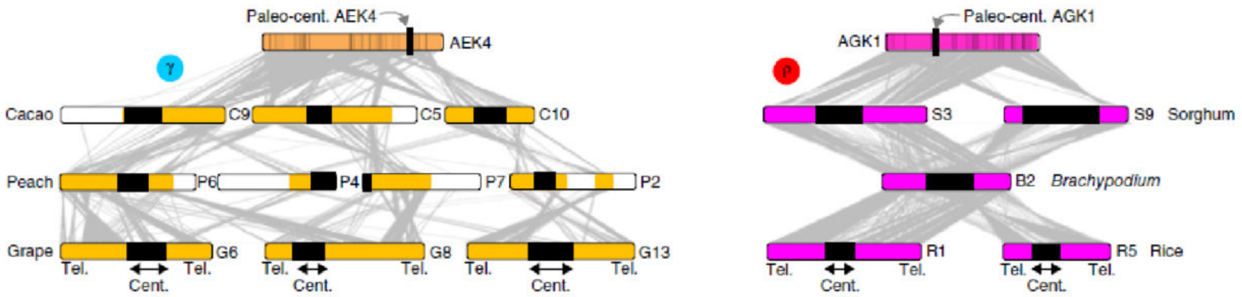
Paleogenomics

Murat et al., 2017; Pont et al., 2019

The reconstruction of ancestral genomes based on existing species

- Eudicots- n=7
- Monocots, n=5
- Note that angiosperms arose from a polyploid





Pont et al., 2019