Evolutionary trends and mechanisms of chromosome number variation

㊺

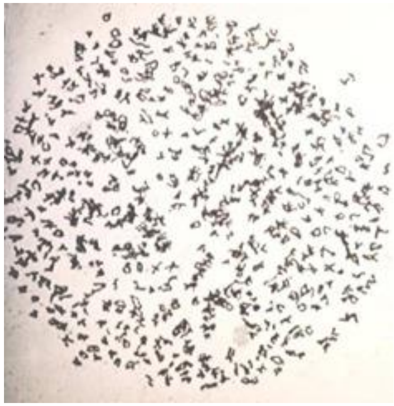
㊻

Chromosome number variation

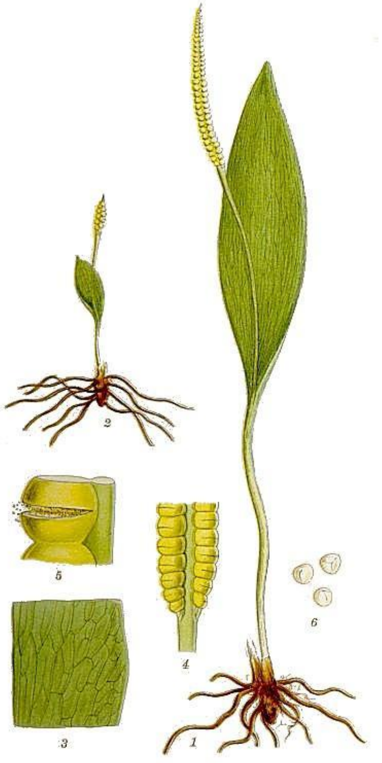
Chromosome numbers of plants vary enormously over a 360-fold range.

n = 2 in five angiosperm species

n = 630 in the fern *Ophioglossum reticulatum*



Haplopappus gracilis



ORMTUNGA, OPHIOGLOSSUM VULGATUM L.

Basic chromosome number (x)

- a relative concept [x has to be related to a certain taxonomic unit, e.g. genus or (sub)family]
- monobasic taxa (single x number), dibasic taxa (two x nos.) and polybasic taxa (>2 x nos.)
- are there any evolutionary trends in chromosome number changes?
- are the same chromosome number and similar karyotype structure indicative of close phylogenetic relationship?
- can polybasic taxa be regarded as monophyletic?
- is the most common basic chromosome number automatically the ancestral one?

Asteraceae – example of a polybasic family

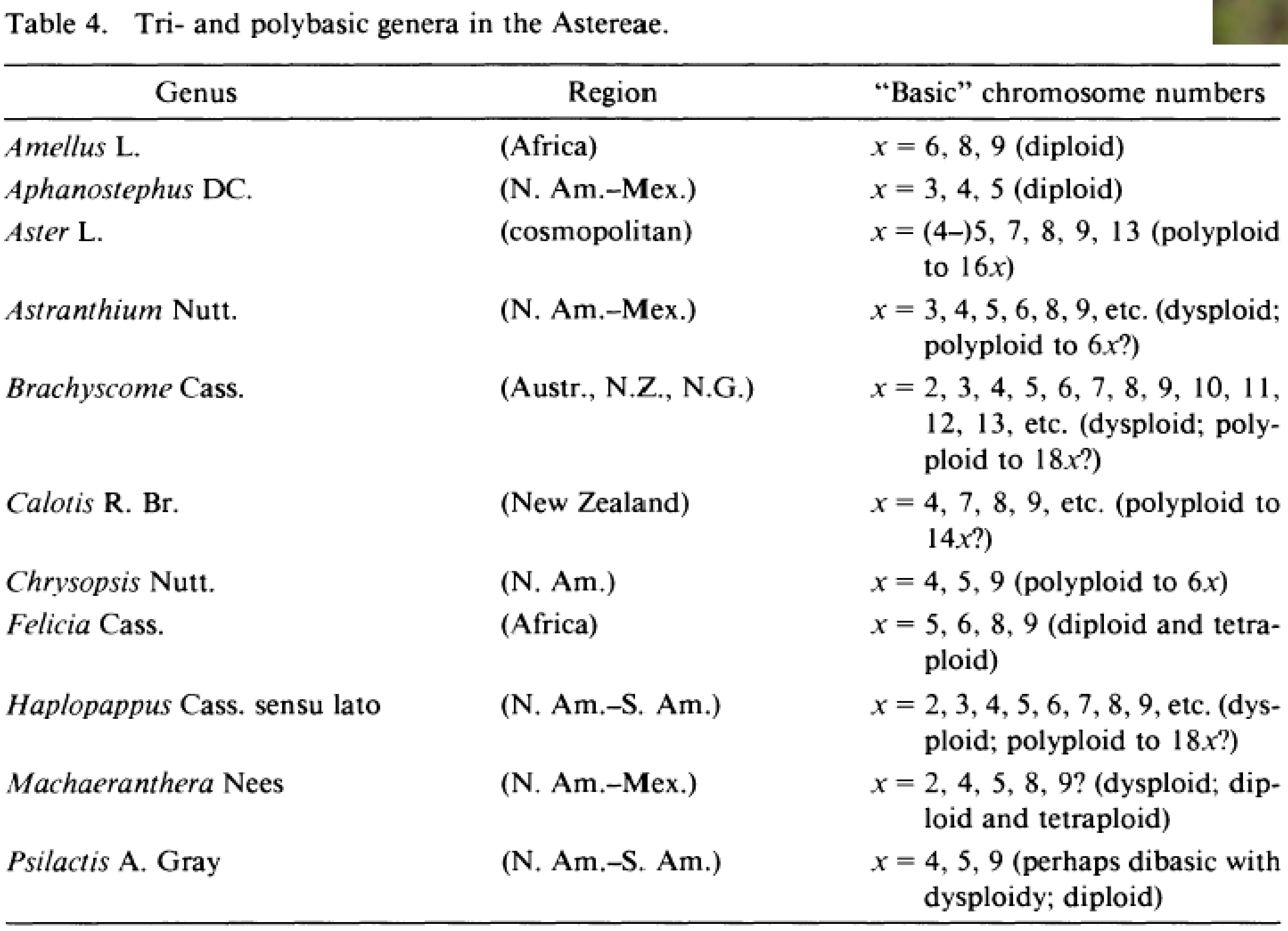
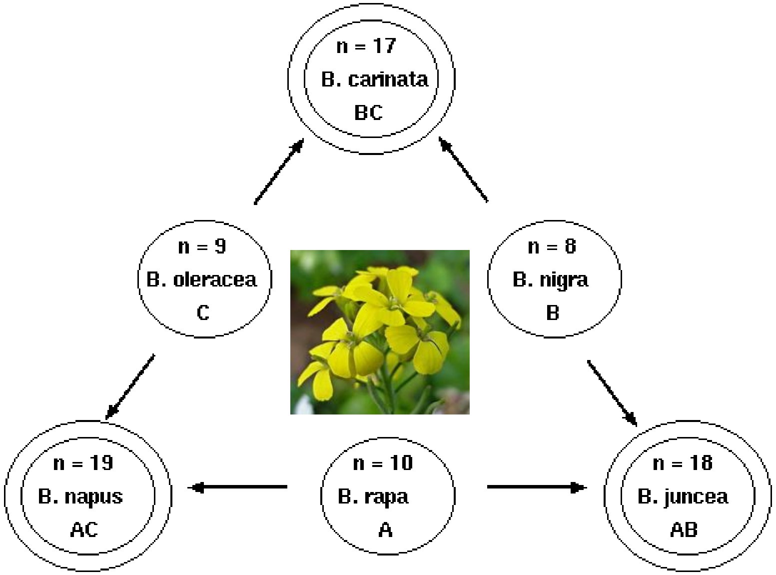


Table 4. Tri- and polybasic genera in the Astereae.

Genus	Region	"Basic" chromosome numbers
Amellus L.	(Africa)	$x = 6, 8, 9$ (diploid)
Aphanostephus DC.	(N. Am.–Mex.)	$x = 3, 4, 5$ (diploid)
Aster L.	(cosmopolitan)	$x = (4–)5, 7, 8, 9, 13$ (polyploid to $16x$)
Astranthium Nutt.	(N. Am.–Mex.)	$x = 3, 4, 5, 6, 8, 9$, etc. (dysploid; polyploid to $6x$?)
Brachyscome Cass.	(Austr., N.Z., N.G.)	$x = 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13$, etc. (dysploid; polyploid to $18x$?)
Calotis R. Br.	(New Zealand)	$x = 4, 7, 8, 9$, etc. (polyploid to $14x$?)
Chrysopsis Nutt.	(N. Am.)	$x = 4, 5, 9$ (polyploid to $6x$)
Felicia Cass.	(Africa)	$x = 5, 6, 8, 9$ (diploid and tetraploid)
Haplopappus Cass. sensu lato	(N. Am.–S. Am.)	$x = 2, 3, 4, 5, 6, 7, 8, 9$, etc. (dysploid; polyploid to $18x$?)
Machaeranthera Nees	(N. Am.–Mex.)	$x = 2, 4, 5, 8, 9$? (dysploid; diploid and tetraploid)
Psilactis A. Gray	(N. Am.–S. Am.)	$x = 4, 5, 9$ (perhaps dibasic with dysploidy; diploid)

Evolutionary changes of basic chromosome number

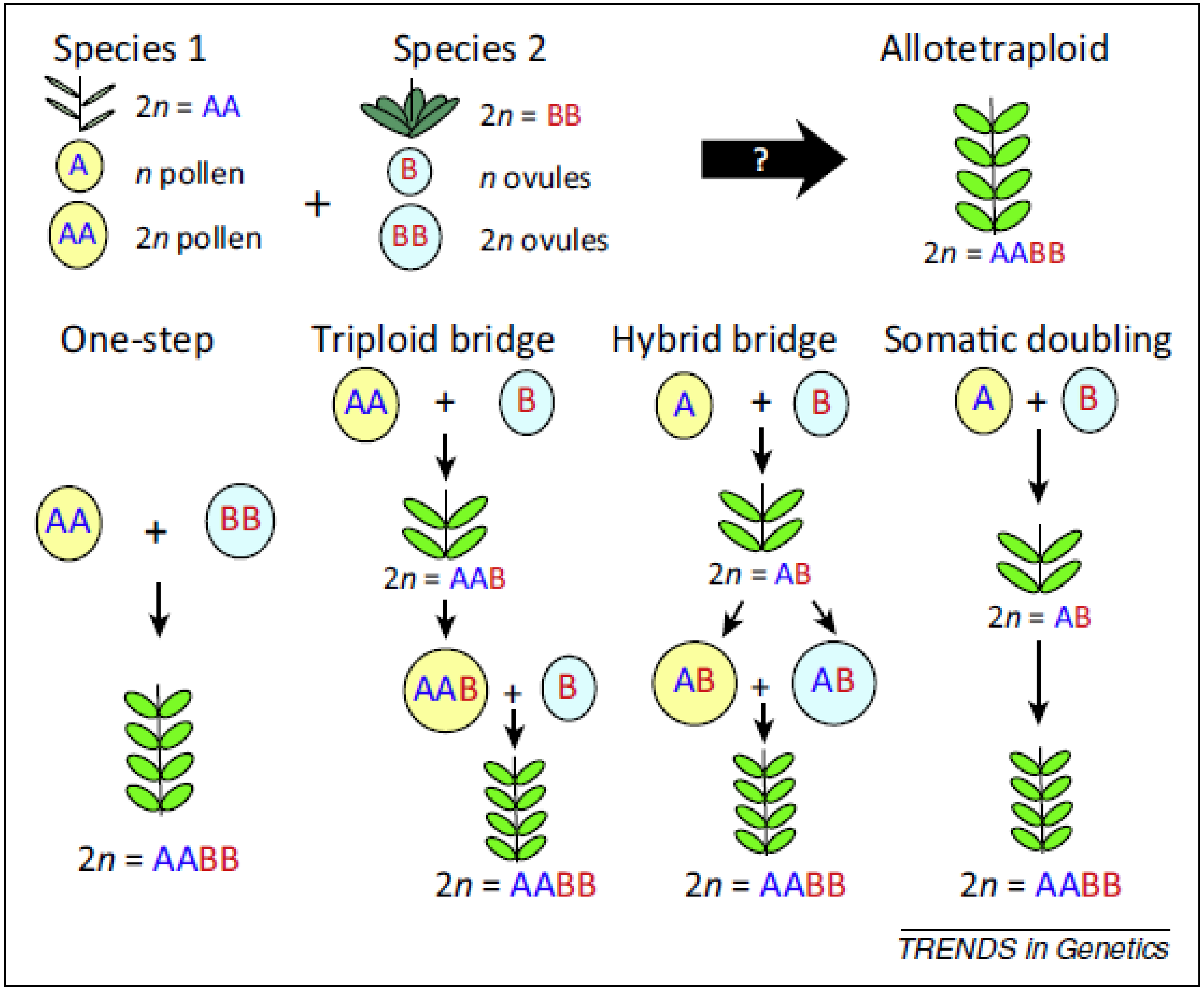
I. allopolyploidy



II. dysploidy (aneuploidy)

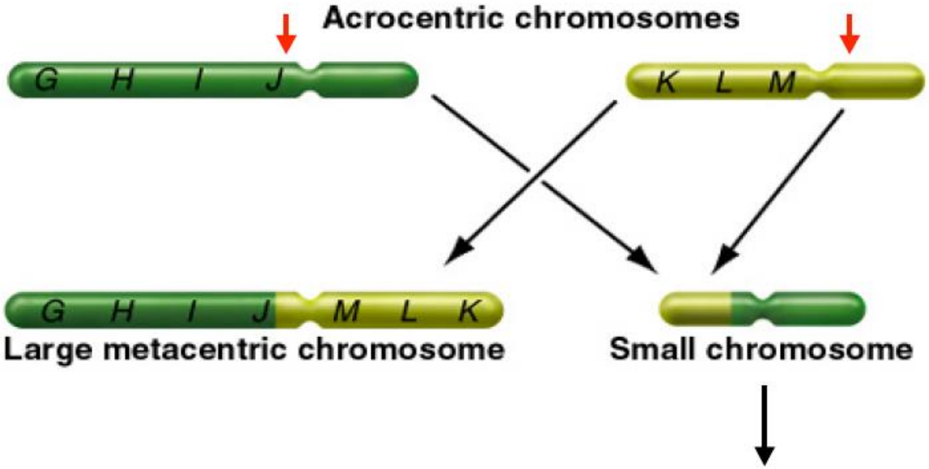
- descending
- ascending (aneuploidy) dysploidy

Allopolyploidy

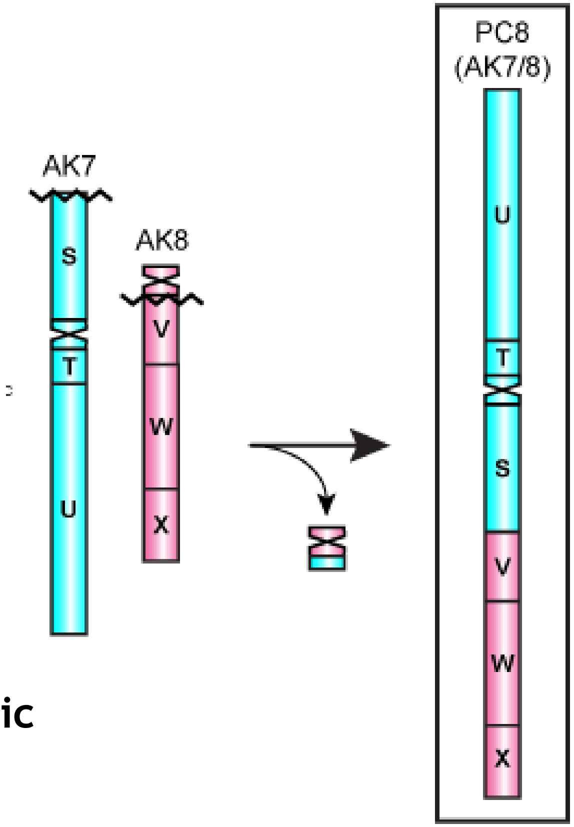


TRENDS in Genetics

Descending dysploidy due to Robertsonian translocation (centric „fusion“)



lost



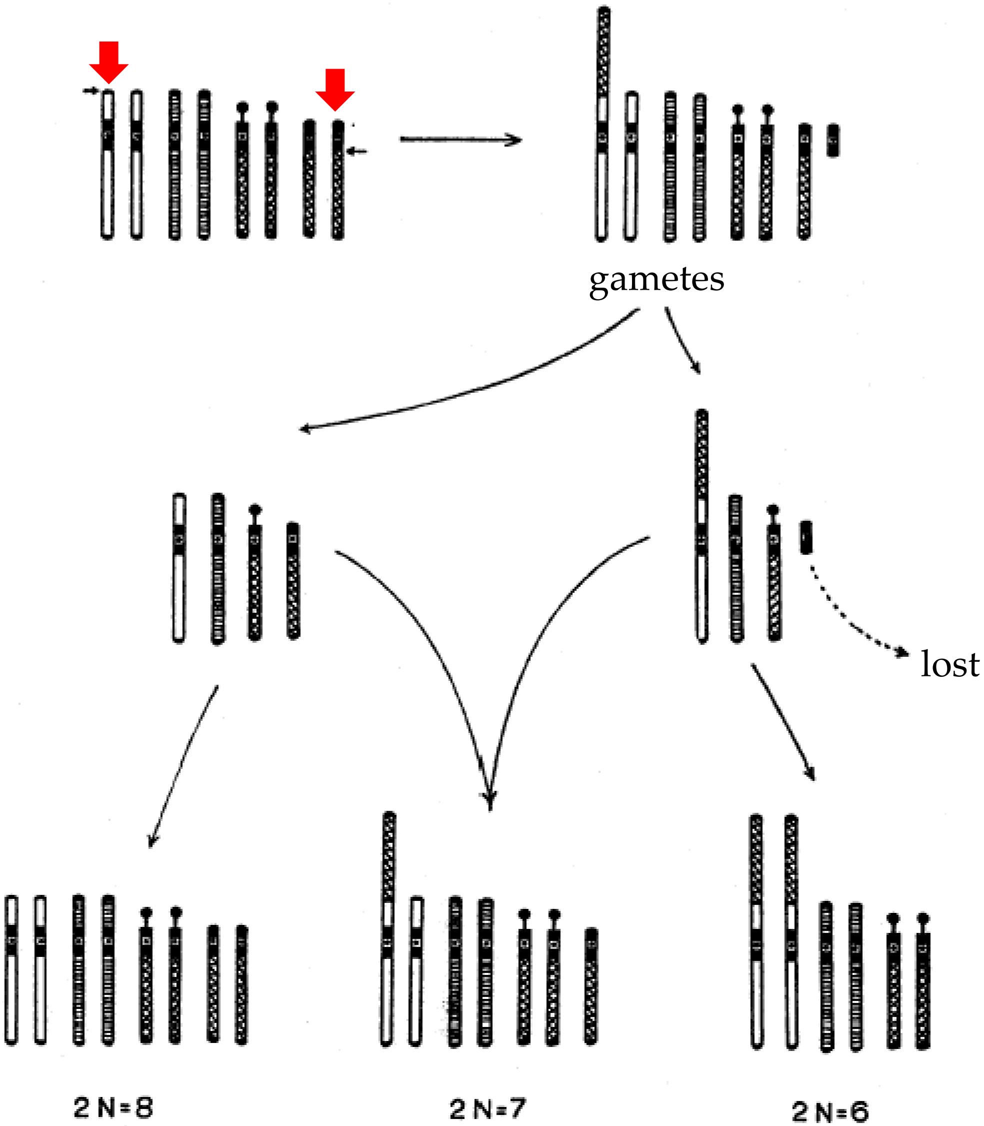
acrocentric
and metacentric
chromosome

Robertsonian (unequal reciprocal) translocation and meiotic seqregation

2n = 8

chromosome number reduction

2n = 8 or 7 or 6



Nested Chromosome Fusion (NCF) in grasses

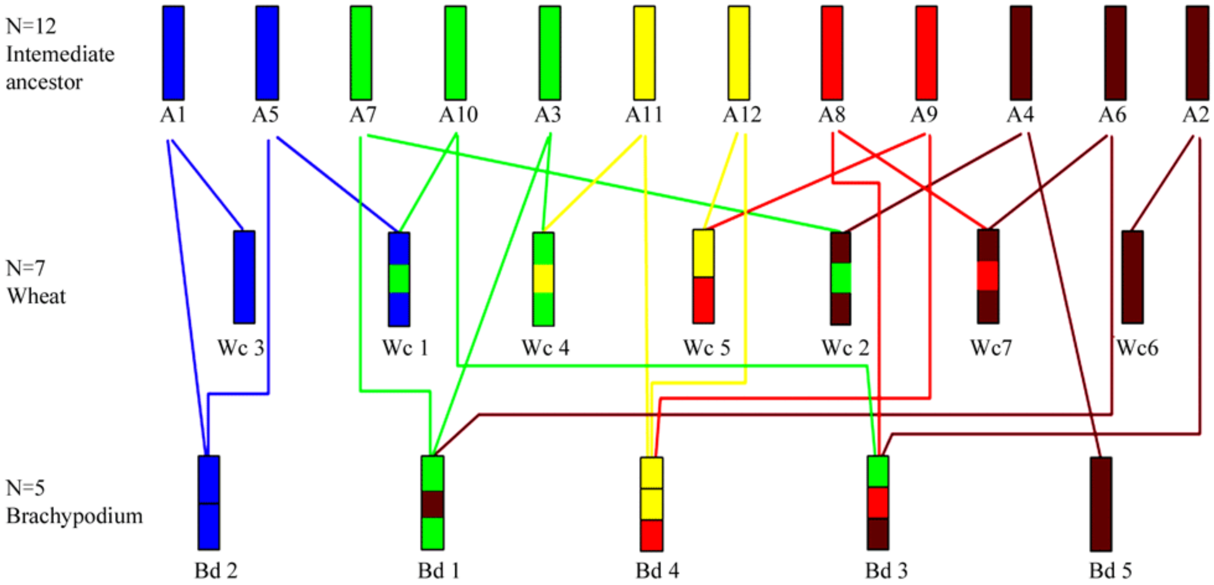
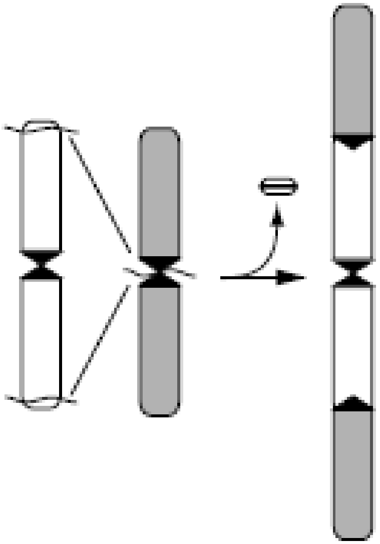
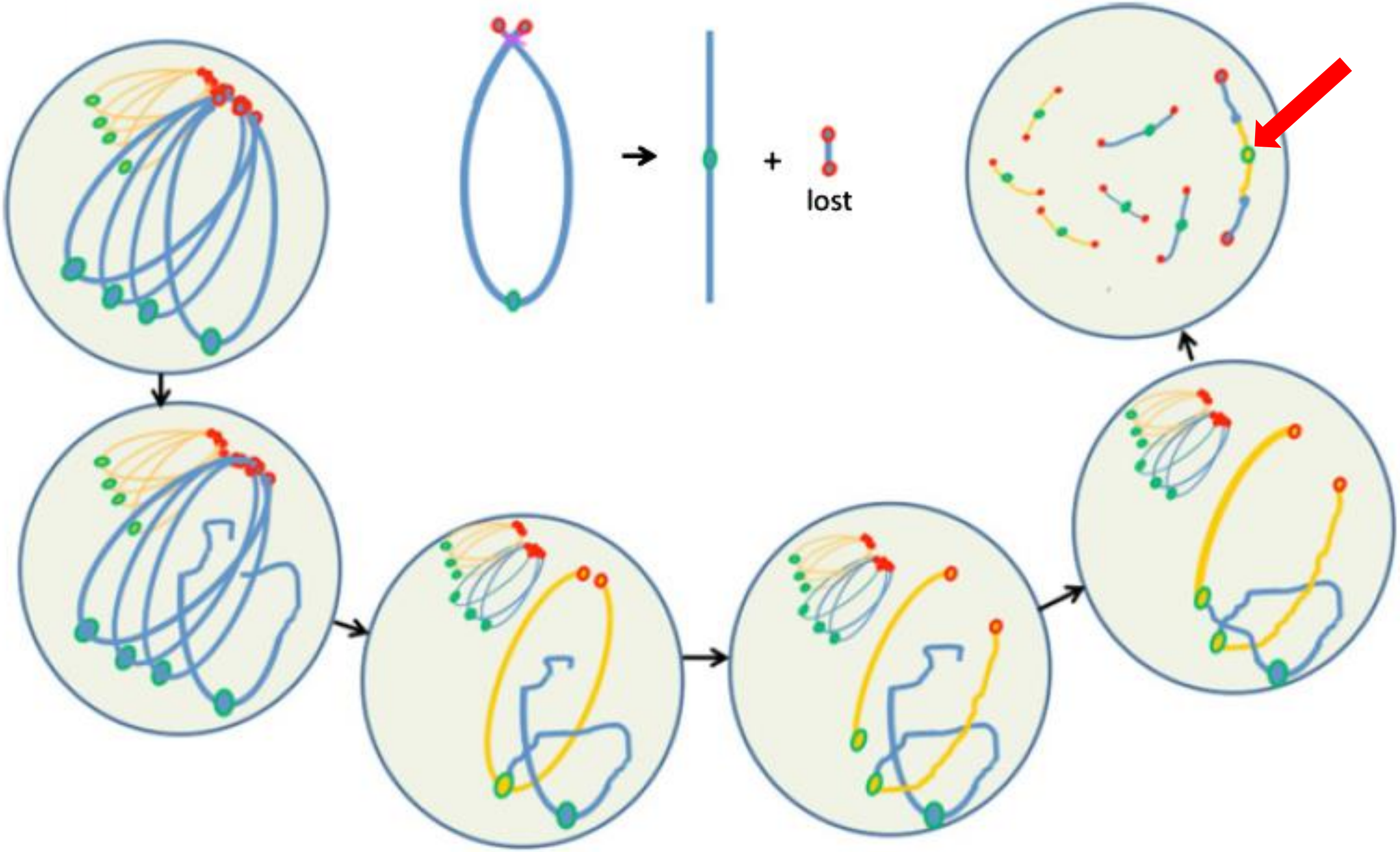


Figure 3 Evolution of the wheat ($n = 7$) and brachypodium ($n = 5$) chromosomes from a common intermediate ancestral genome with $n = 12$



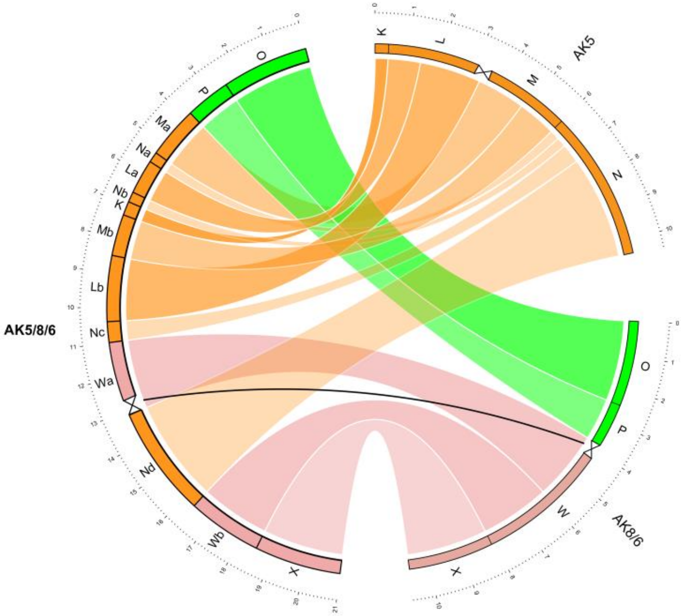
Nested Chromosome Fusion (NCF) in grasses

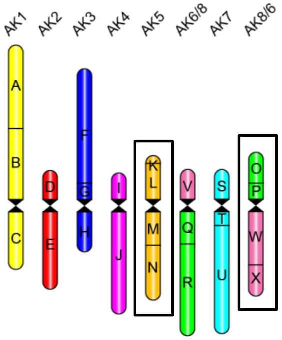


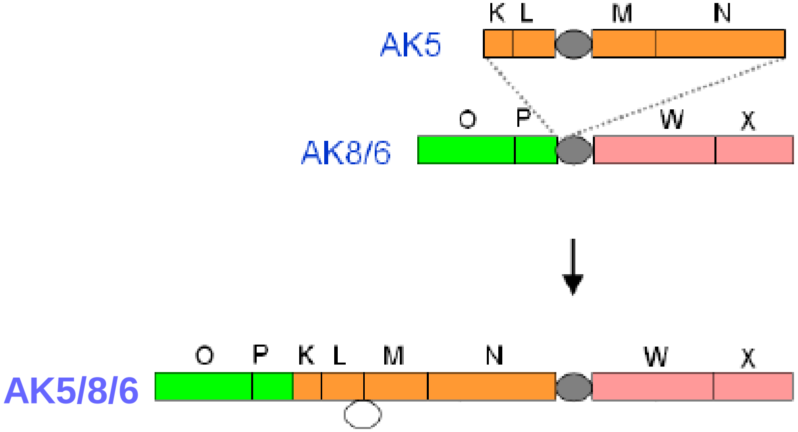
Wang et al. (2014) New Phytol

Nested Chromosome Fusion (NCF) in *Cardamine*

Lady‘s Smock (*Cardamine pratensis*)







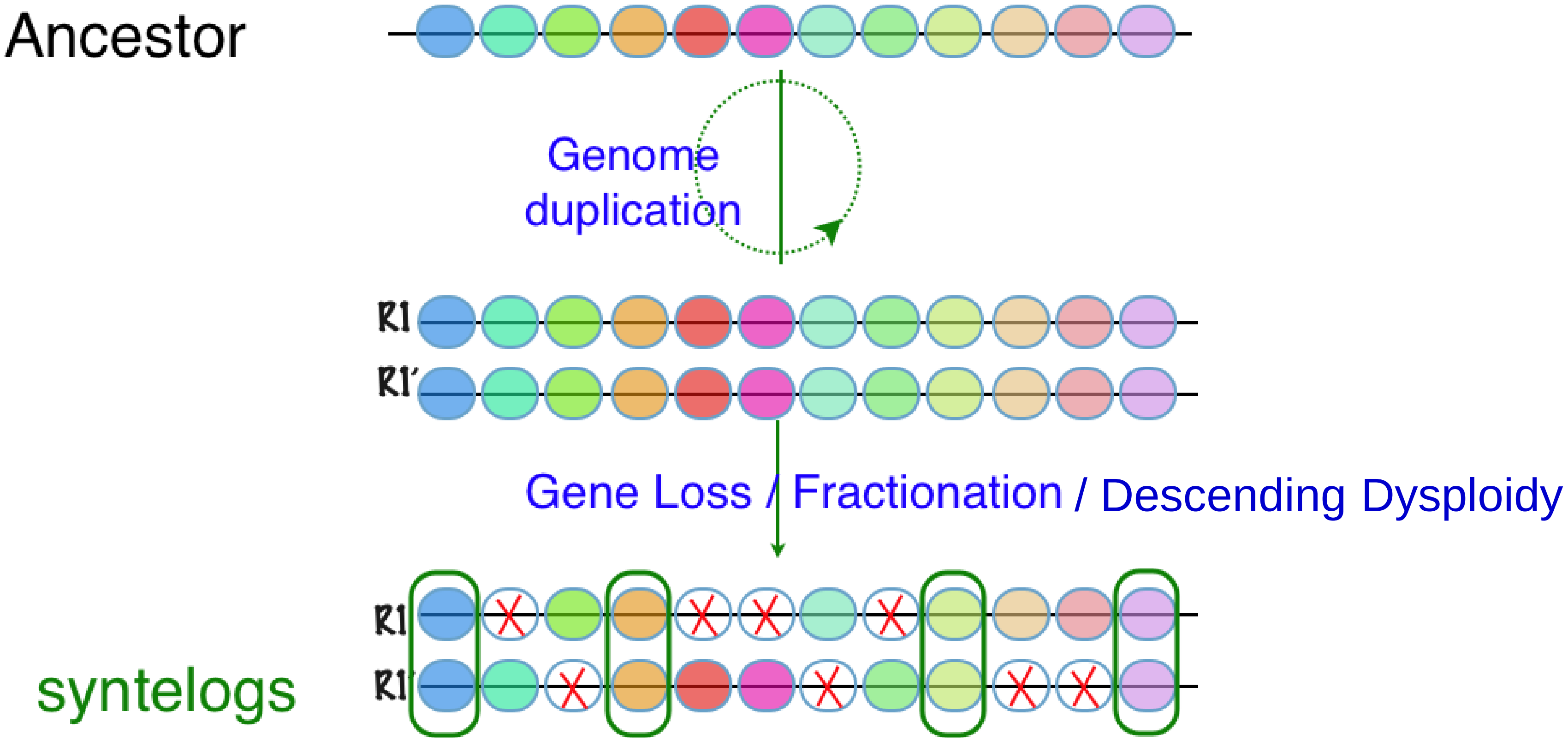
Nested Chromosome Fusion (NCF)

2n = 32 ⟶ 2n = 30

~~aneuploidy~~

Mandakova et al. (2013) Plant Cell

Whole-genome duplication are frequently followed by genome fractionation (diploidization) and accompanied by descending dysploidy



Ancestor

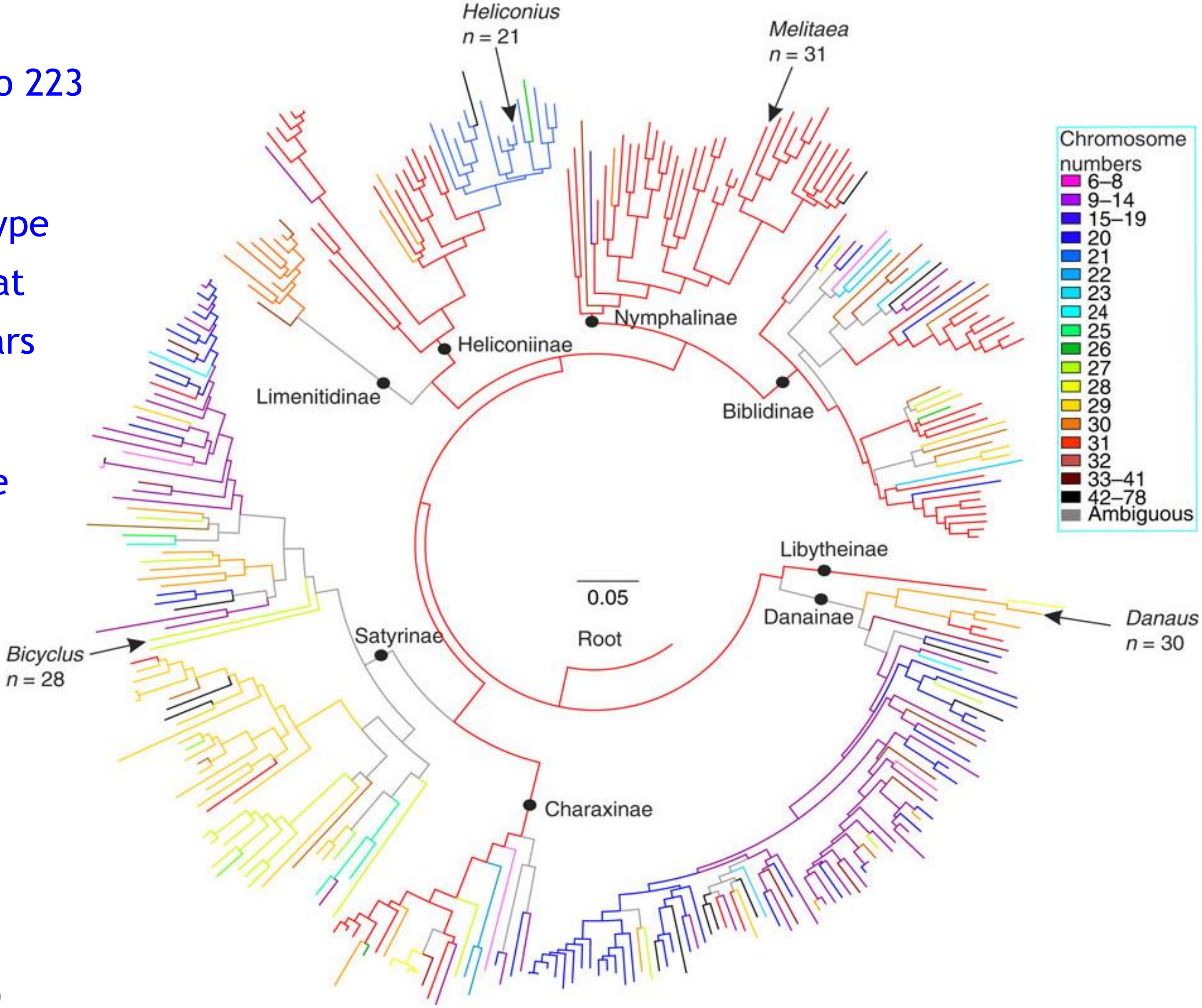
Genome duplication

Gene Loss / Fractionation / Descending Dysploidy

syntelogs

The Glanville fritillary genome retains an ancient karyotype and reveals selective chromosomal „fusions“ in Lepidoptera

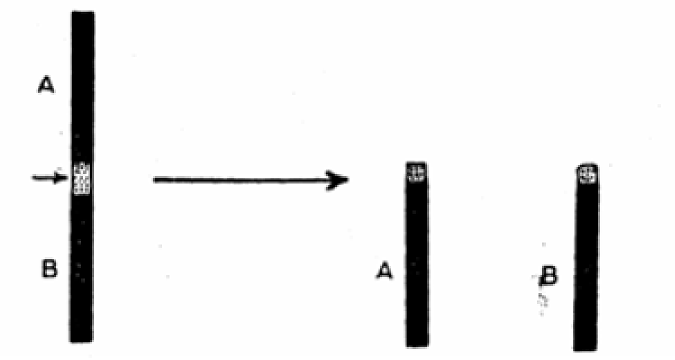
- Lepidoptera: n = 5 to 223
- the ancestral lepidopteran karyotype has been n = 31 for at least 140 million years
- karyotype evolution through chromosome „fusions“



Ahola et al., Nat Comm (2014)

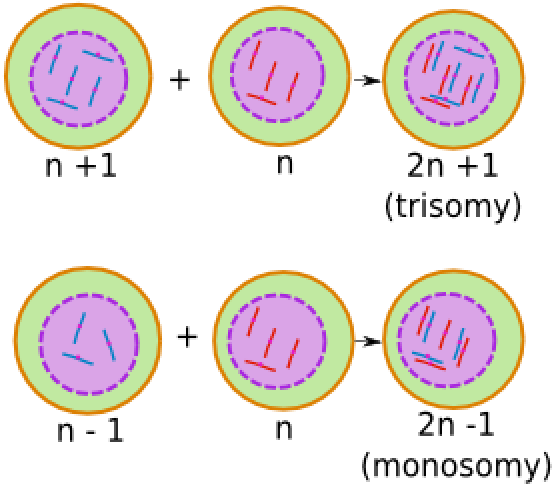
Ascending dysploidy

1. Centric fission (1 metacentric chromosome → 2 telocentrics)



probably in orchids, cycads...

2. Meiotic misdivision (non-disjunction)



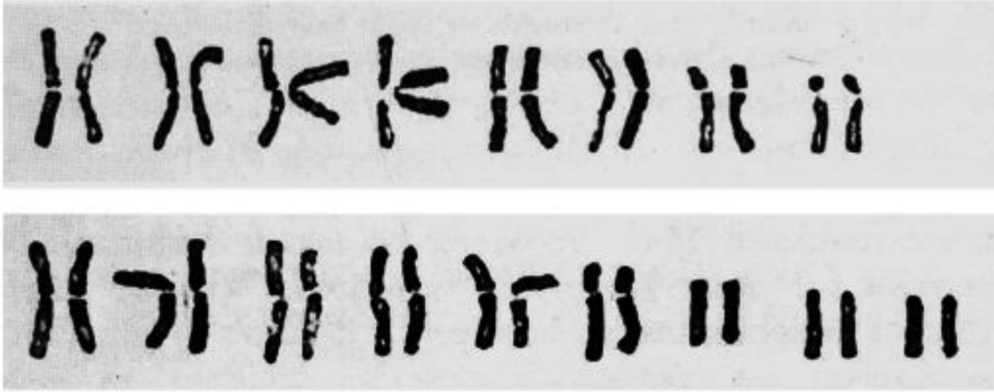
- misdivision resulting in a tetrasomic plant (2n+2)

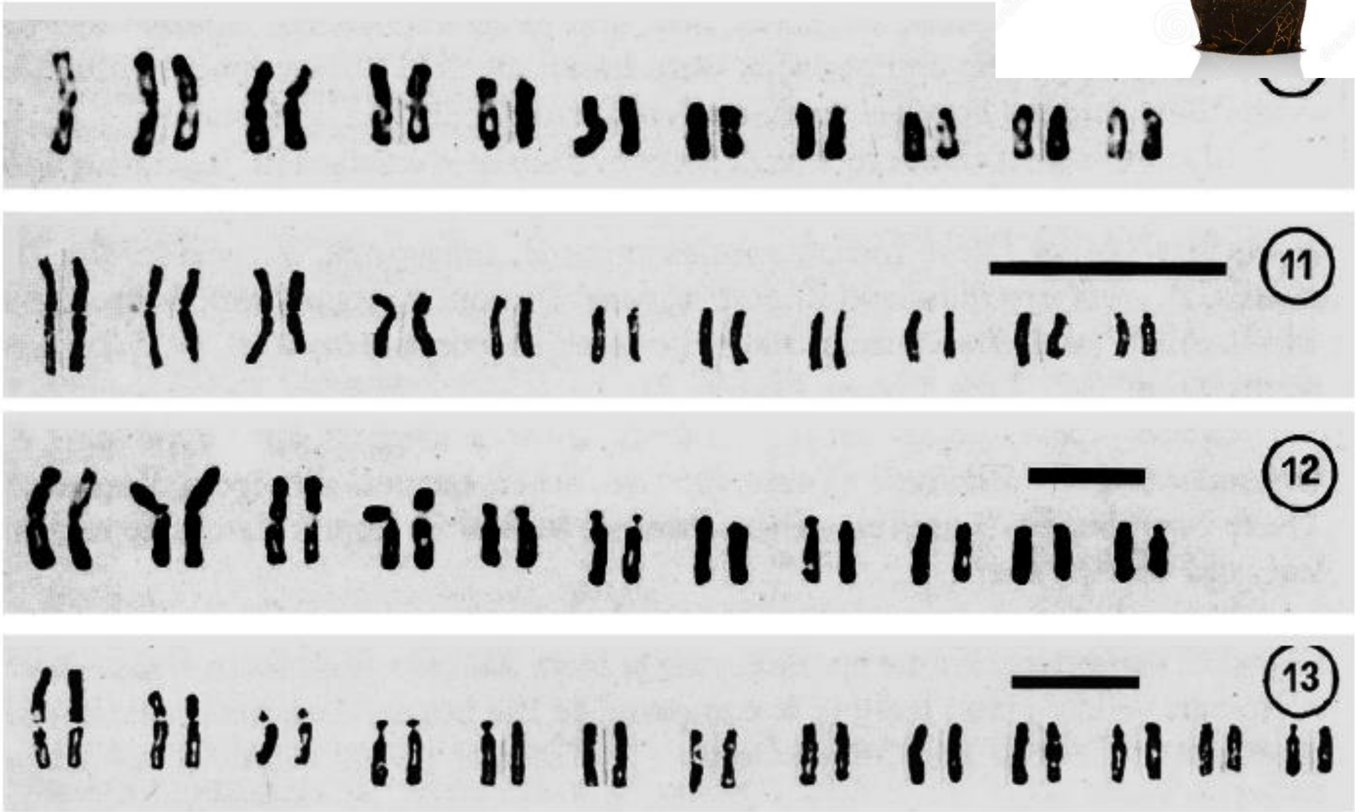
(or first trisomy: 2n+1 followed by tetrasomy, 2n+2)

or monosomic plant (2n-1, this is descending dysploidy)

- the extra chromosome can diverge from their homologues through a translocation with non-homologous chromosomes

Centric fissions → telocentric chromosomes in cycads (*Zamia*)





Chromosome number pattern congruent with phylogenetic relationships: *Ranunculaceae*

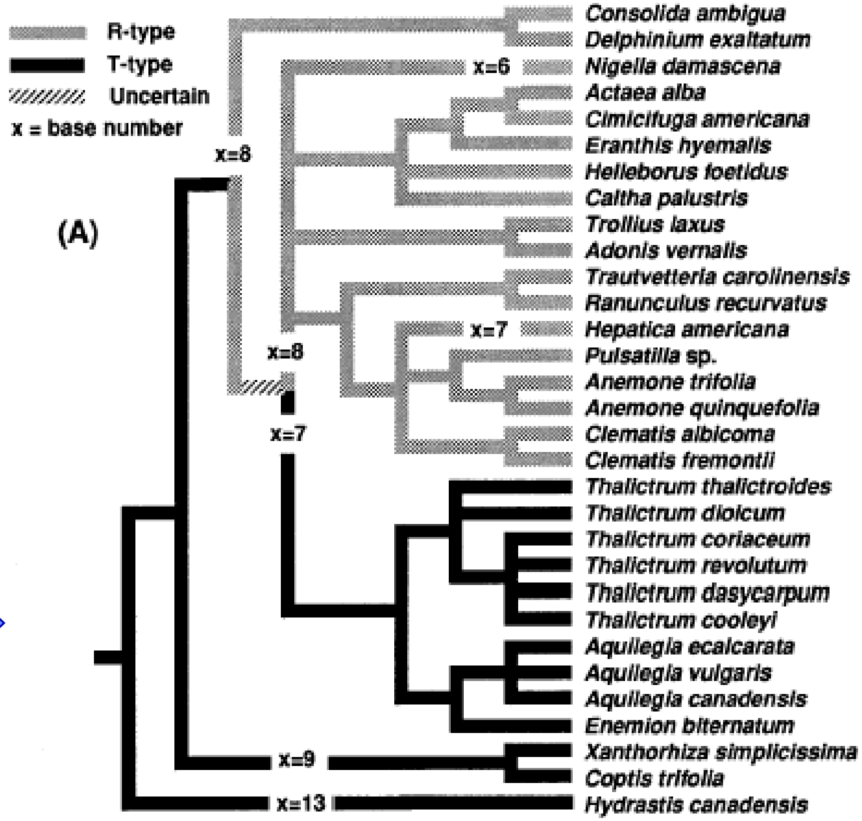
- Langlet (1927, 1932) recognized two subfamilies of *Ranunculaceae* (*Ranunculoideae* and *Thalictroideae*) on the basis of cytological characters, including chromosome size and basic number

- the *Ranunculus* group of genera (R-chromosome group) has large and long chromosomes with a basic number of 8

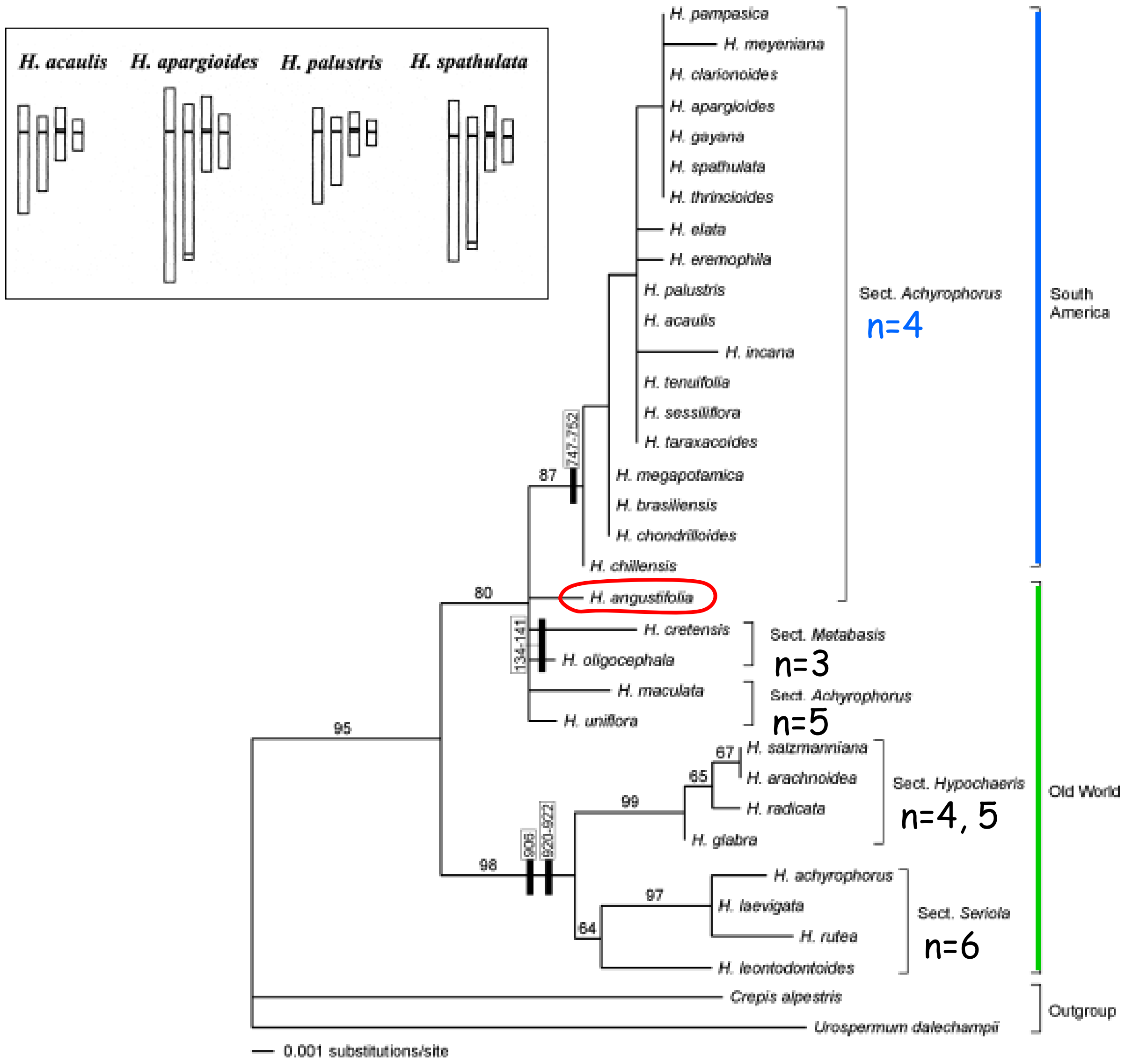
- the *Thalictrum* group (T-chromosome group) has short and small chromosomes with a basic number of 7 or 9

- Ro et al. (1997): chromosome type and base number are congruent with the inferred molecular (rDNA) phylogeny

- fruit type (often used for the higher classification) was not congruent with karyological data and phylogenetic patterns



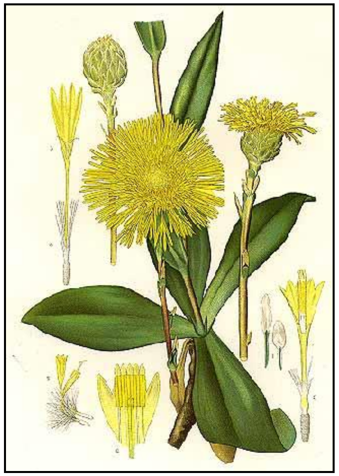
Descending dysploidy in *Hypochaeris* (*Asteraceae*)



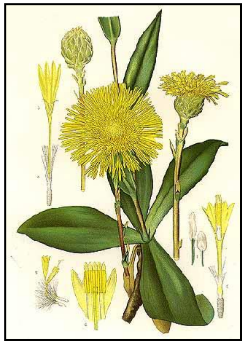
Descending dysploidy in *Podolepis* (*Asteraceae*)

- the extraordinary series of chromosome numbers, n = 12, 11, 10, 9, 8, 7 and 3 (dysploidy)

- chromosome number of n = 10 is the most common in the genus, and thus, x = 10 was regarded as the ancestral chromosome base number for the genus



Descending dysploidy in *Podolepis* (*Asteraceae*)



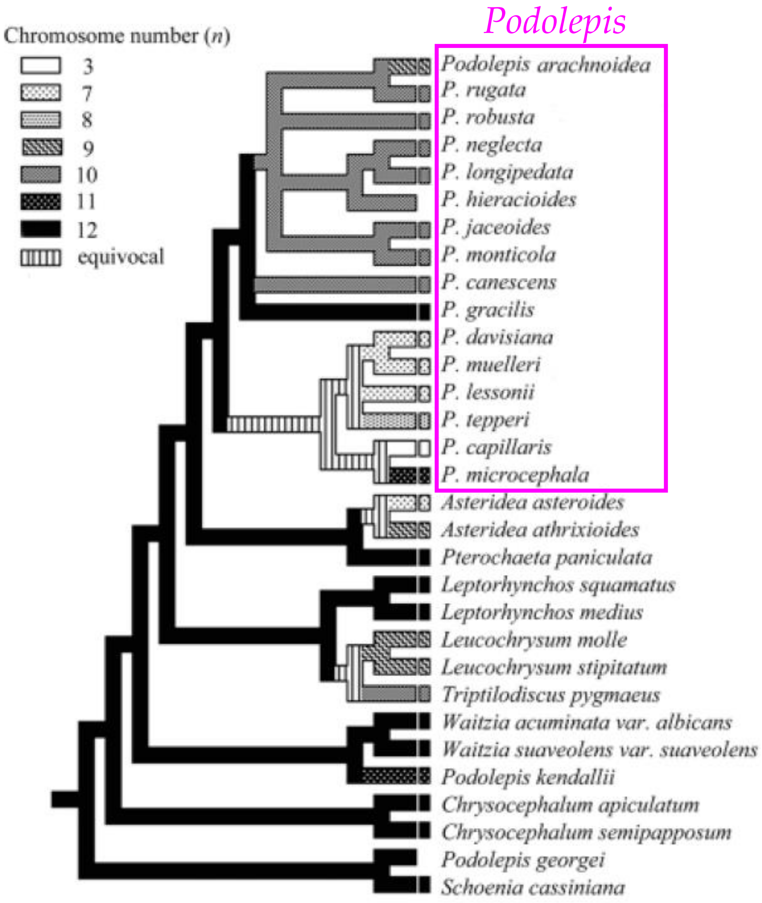
- the haploid chromosome number of n = 12 is the most common in the related genera (*Chrysocephalum*, *Waitzia*, *Leptorhynchos*, *Pterochaeta*)

- **according to the phylogenetic analysis, the ancestral chromosome base number in the genus *Podolepis* may be x = 12**

- chromosome number reduction has occurred in three lineages:
  - from n = 12 to n = 10 and 9 in the subclade A
  - from n = 12 to n = 8 and 7 in the subclade B1
  - from n = 12 to n = 11 and 3 in the subclade B2

- the low chromosome numbers of n = 8, 7 and 3 were found only in annual species which were distributed in semi-arid regions

- comparing the karyotypes between the taxa with n = 12 (in *Waitzia* and *Chrysocephalum*) and n = 10 (perennial *Podolepis*), **the increase in the number of large chromosomes accompanies the decrease in the number of medium-sized chromosomes** in *Podolepis* → **the reduction in chromosome number has been achieved by the unequal reciprocal translocations, followed by the loss of the short translocation product**

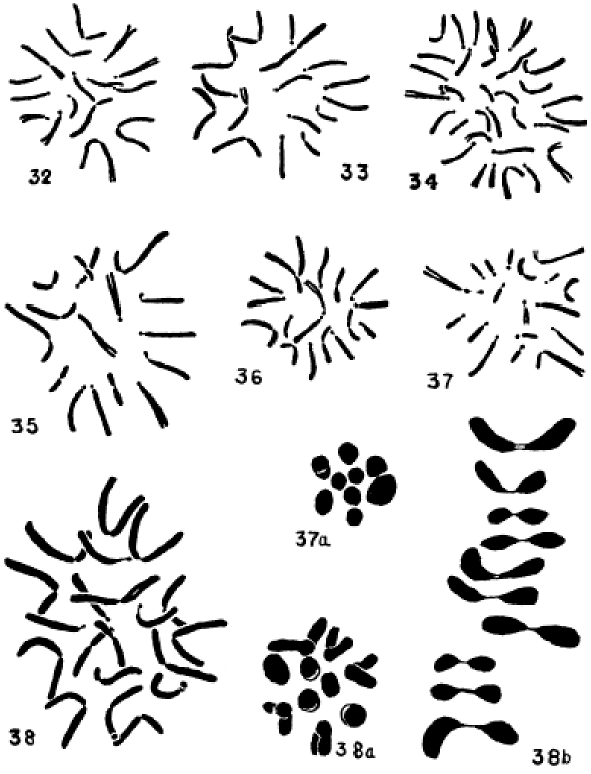


Descending and ascending dysploidy in *Calochortus* (*Liliaceae*)

- c. 67 spp.

- chromosome numbers n = 6, 7, 8, 9, and 10

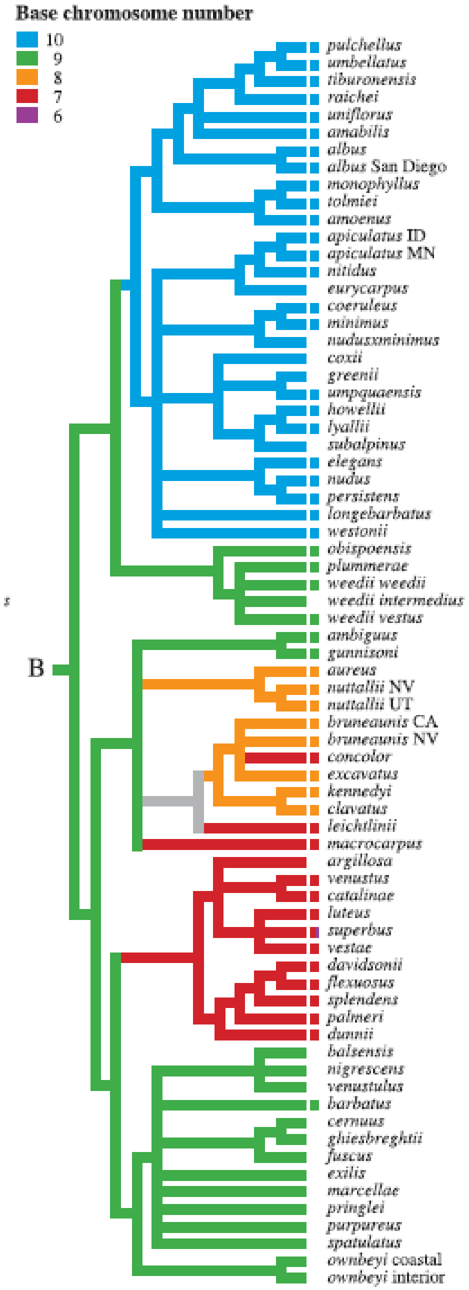
- molecular phylogenetic study carried out to test the monophyly of the three sections and 12 subsections erected by Ownbey (1940) based on morphology and chromosome number



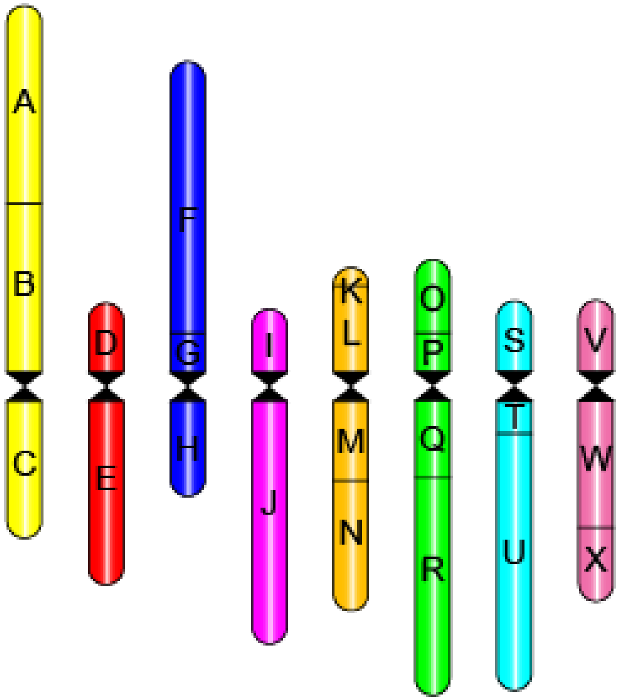
Descending and ascending dysploidy in *Calochortus* (*Liliaceae*)

- the ancestral chromosome number of *Calochortus* is $x = 9$
- descending aneuploidy (9 → 8, 7, 6)
- ascending aneuploidy (9 → 10) BUT is this true or the phylogeny is wrong?

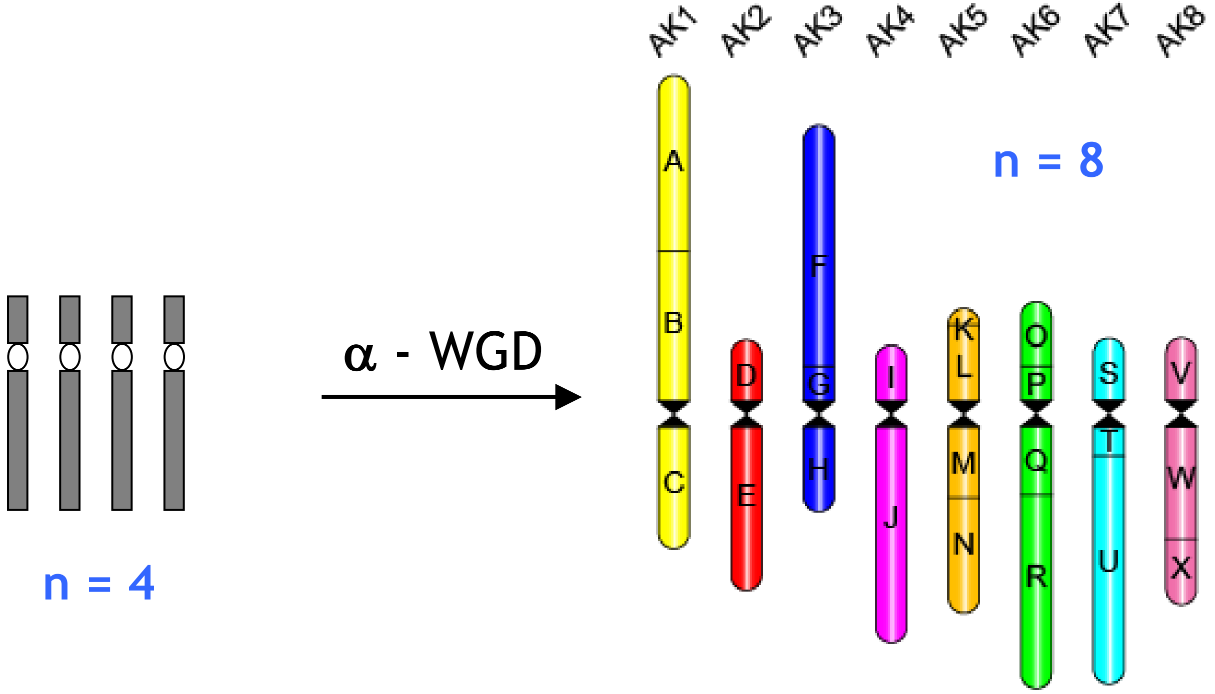
Patterson and Givnish (2003)



Crucifers (Brassicaceae): evolution of an ancestral genome



Crucifers (Brassicaceae): the origin of the common ancestral genome



n = 4

α - WGD

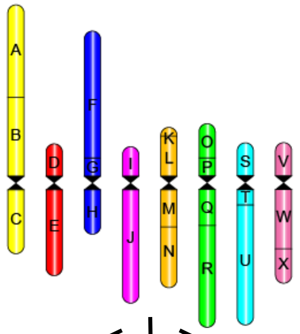
n = 8

Ancestral Crucifer Karyotype (ACK)

(24 ancestral genomic blocks)

WGD = whole-genome duplication (polyploidization)

Evolution of the Ancestral Crucifer Genome – DIPLOIDS

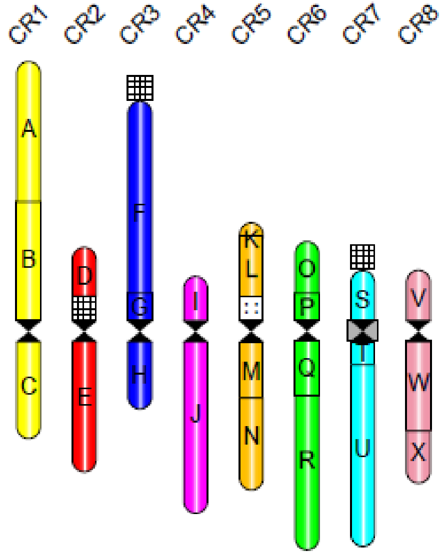


unchanged

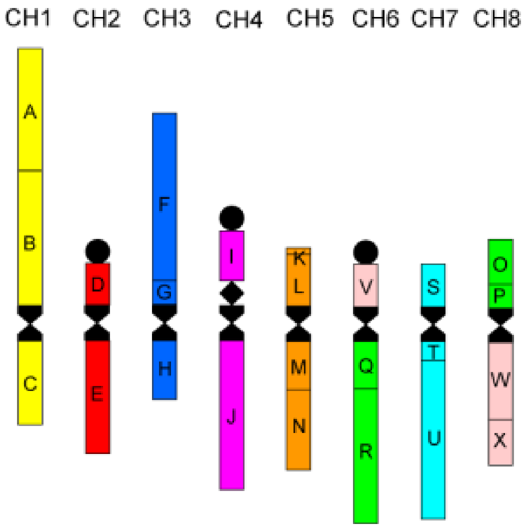
eight but rearranged

descending dysploidy

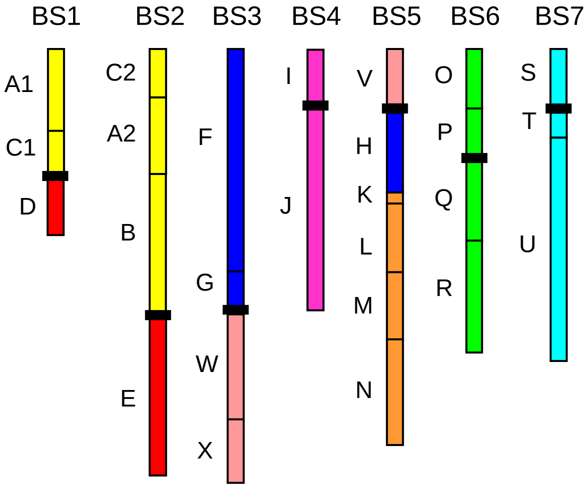
n = 8 → n = 7, 6 & 5



Capsella rubella
(n = 8)

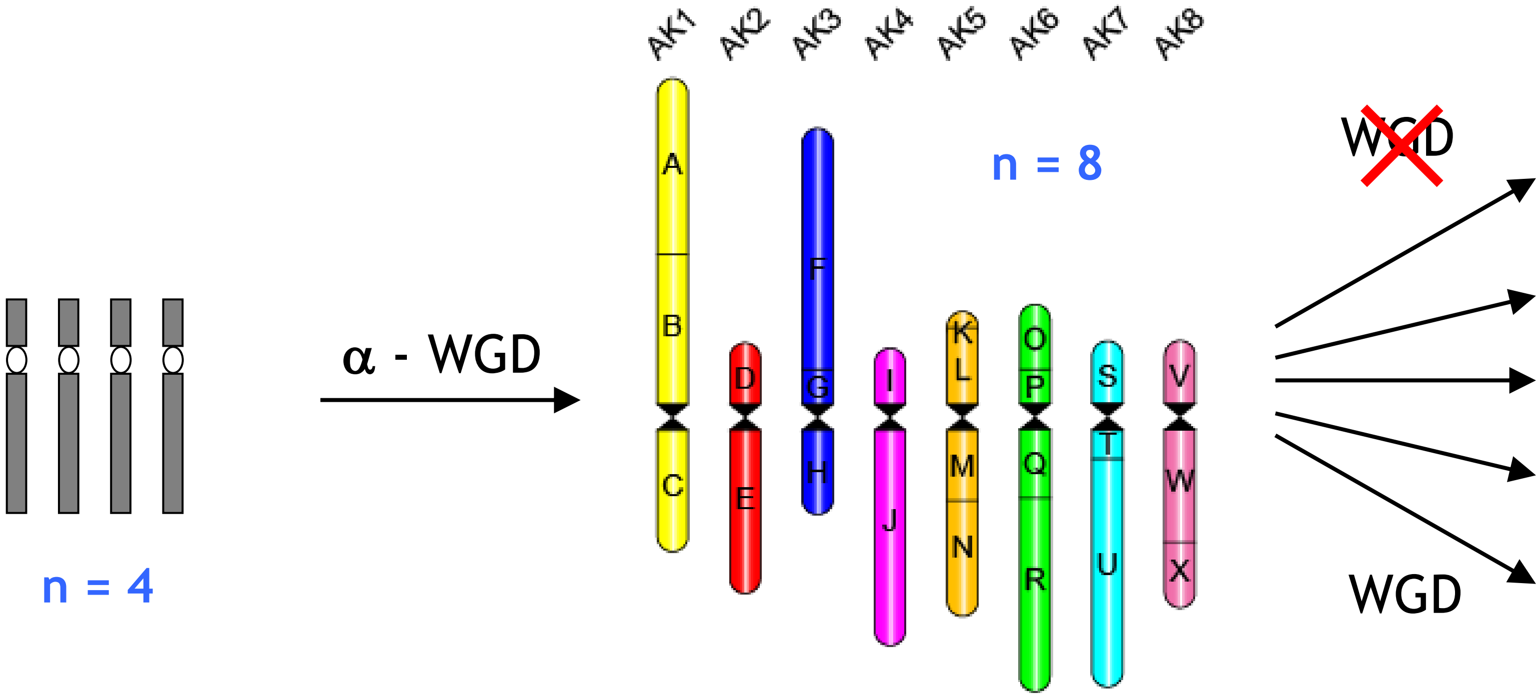


Cardamine hirsuta
(n = 8)



Boechera stricta
(n = 7)

Different fates of the Ancestral Crucifer Karyotype in „diploids“ and polyploids


n = 4

α - WGD

n = 8

~~WGD~~

WGD

Ancestral Crucifer Karyotype (ACK)

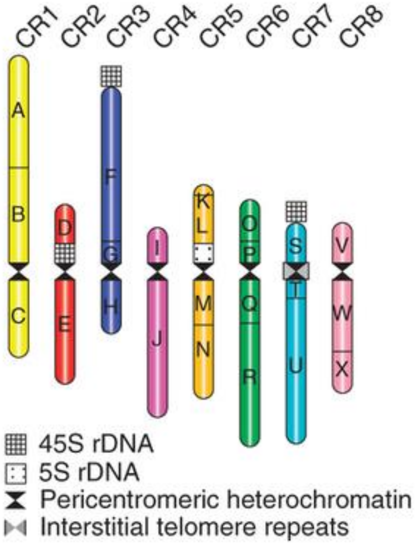
(24 ancestral genomic blocks)

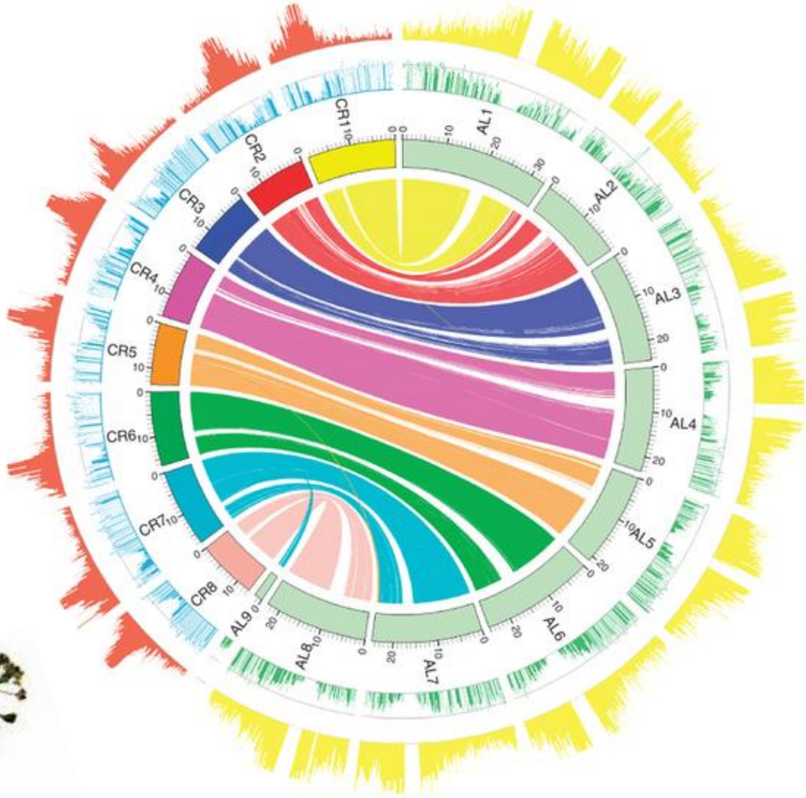
WGD = whole-genome duplication (polyploidization)

Lysak et al. 2006, PNAS; Schranz, Lysak & Mitchell-Olds 2006, TiPS

Ancestral Crucifer Karyotype remained conserved in some taxa of Lineage I

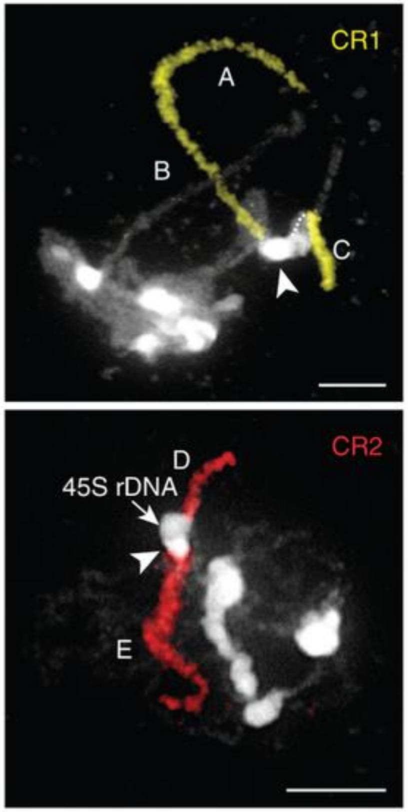
Capsella rubella
(n = 8)





Capsella rubella vs. *Arabidopsis lyrata*

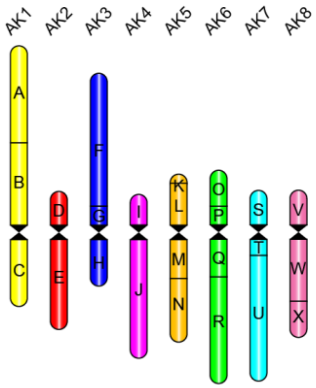
(both have ACK-like genome)

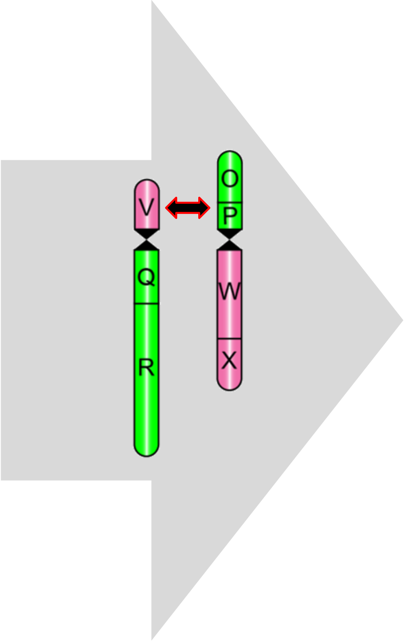


Slotte et al. 2013, Nat Genet

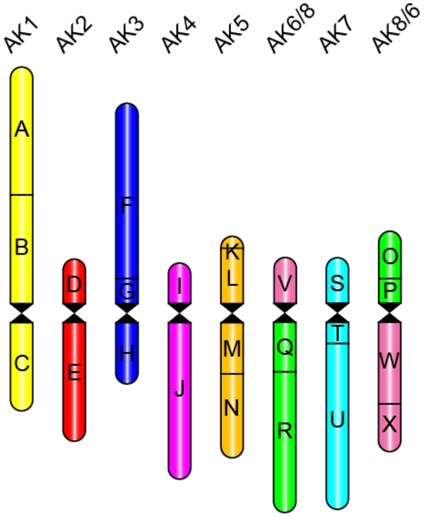
Diversification without large-scale chromosome rearrangements: karyotype stasis in the Cardamineae

ACK
(n = 8)

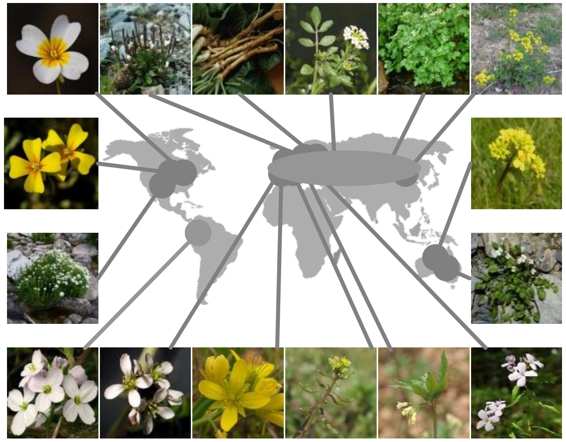




ancestral karyotype of
Cardamineae (n = 8)

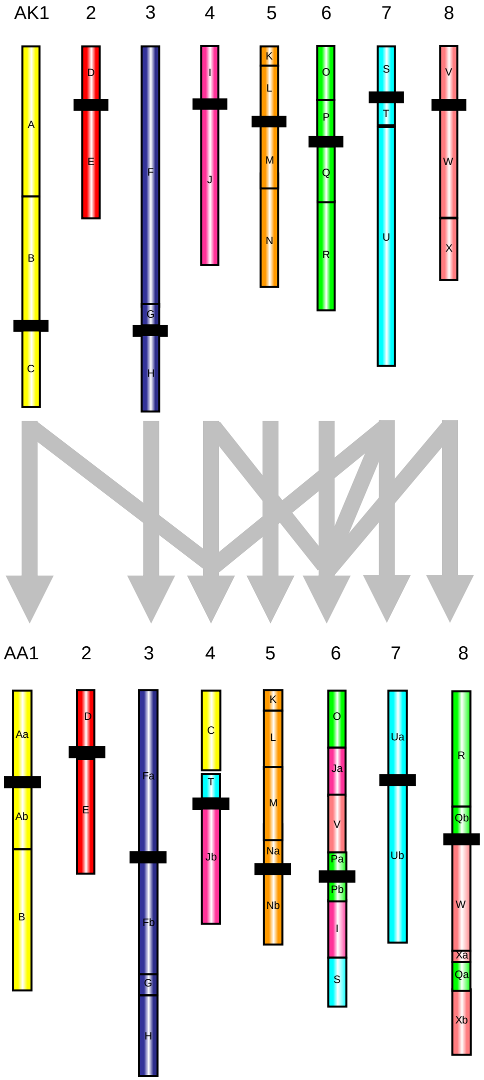


Cardamineae:
12 genera (337 spp.) worldwide



Mandáková and Lysak, in prep.

Arabis alpina: seven out of the eight ancestral chromosomes reshuffled



5 reciprocal translocations

4 pericentric inversions

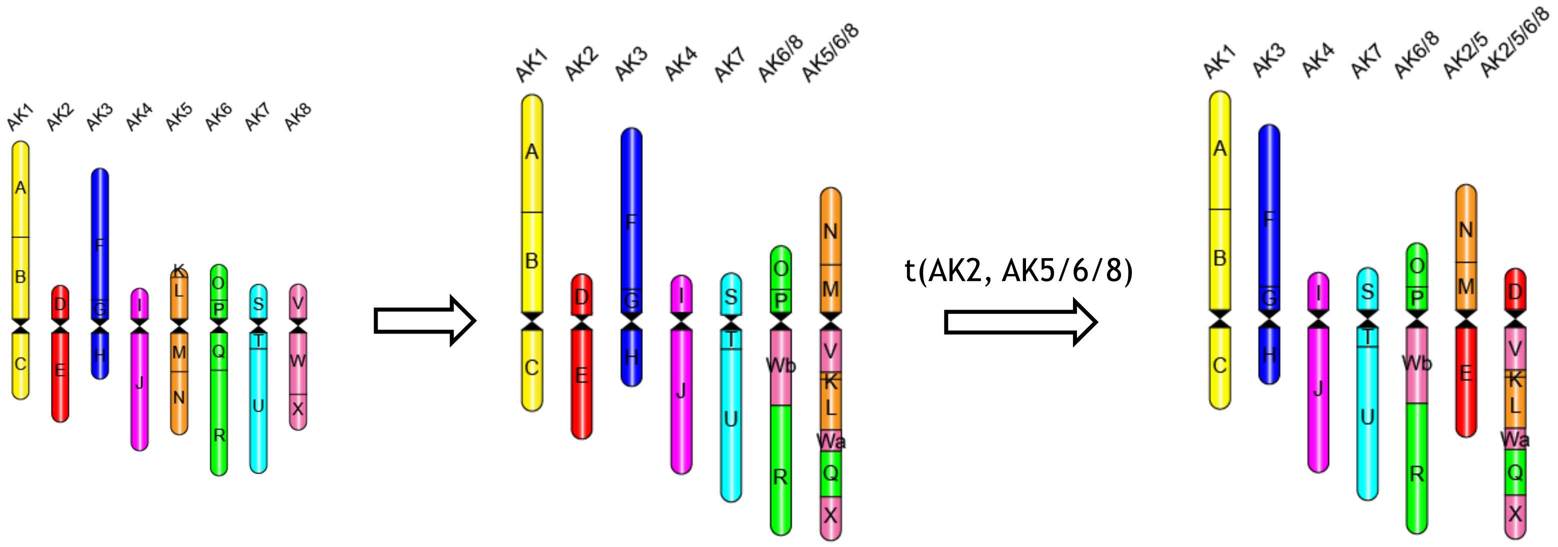
3 centromere repositions

1 centromere loss

1 new centromere emergence (?)

Willing et al., submitted to Nature Plants (collaboration G. Coupland, K. Schneeberger, MPI Cologne)

Descending dysploidy and genome stasis across Lineage II



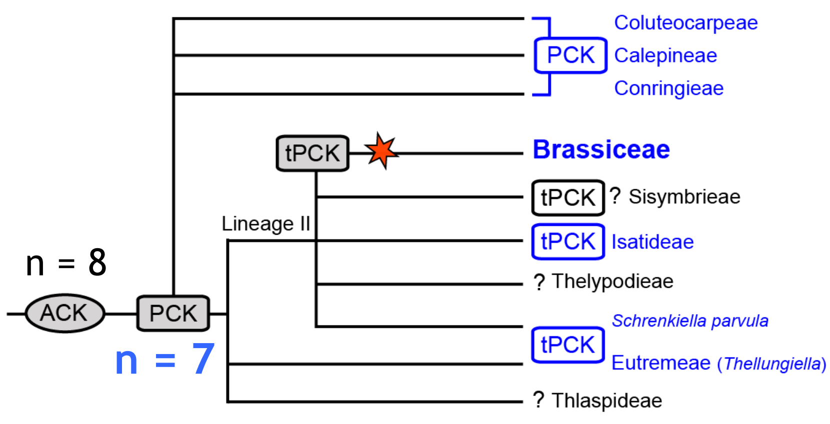
ACK (n = 8)

Ancestral Crucifer
Karyotype

PCK (n = 7)

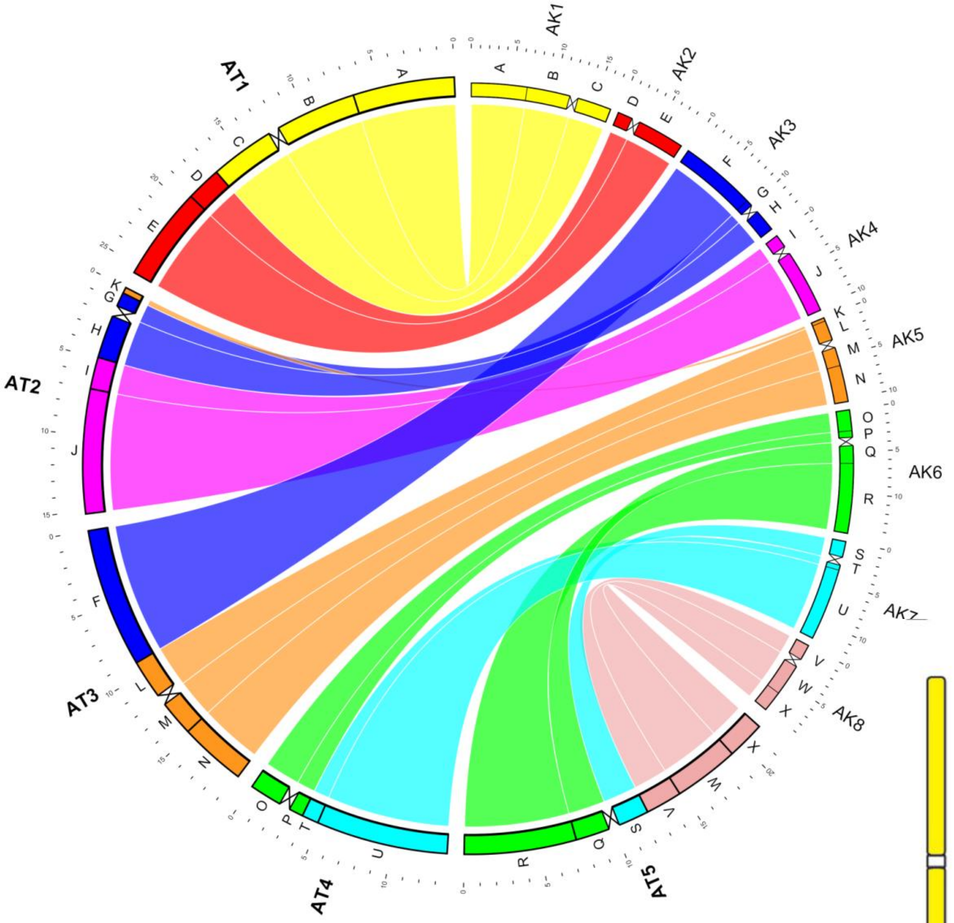
Proto-Calepineae Karyotype

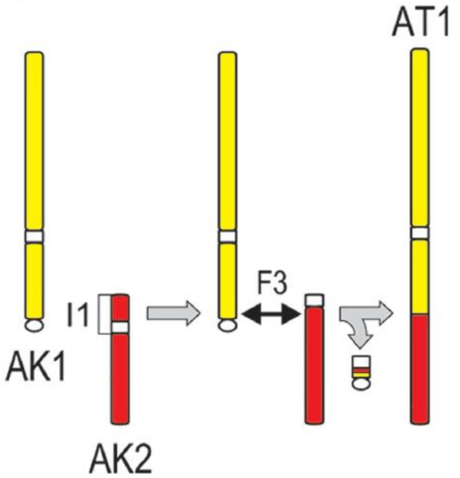
tPCK (n = 7)



Mandáková and Lysak 2008, Plant Cell; Cheng et al. 2013, Plant Cell

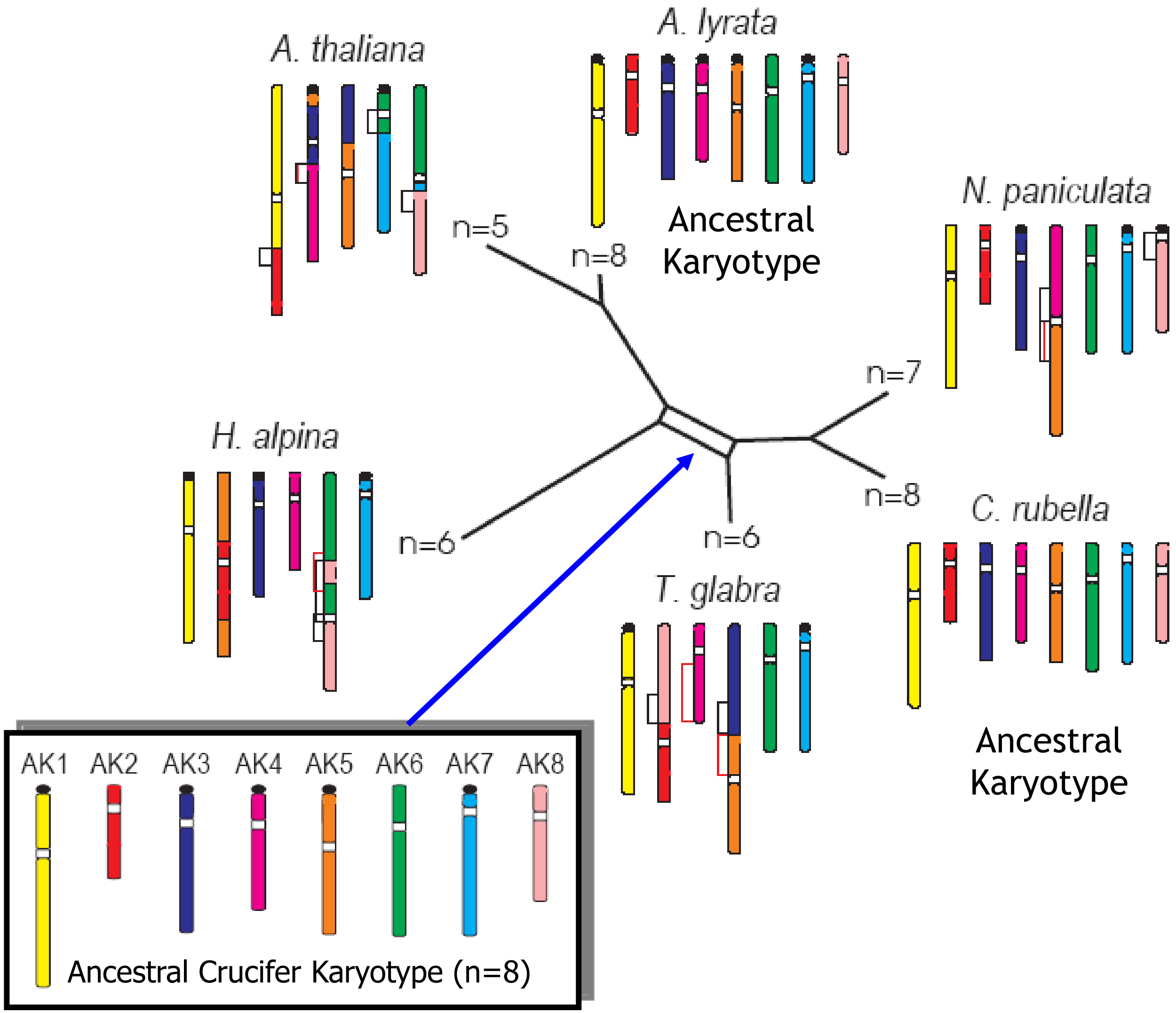
Arabidopsis thaliana - extensive chromosome reshuffling linked with chromosome number reduction from n=8 to n=5



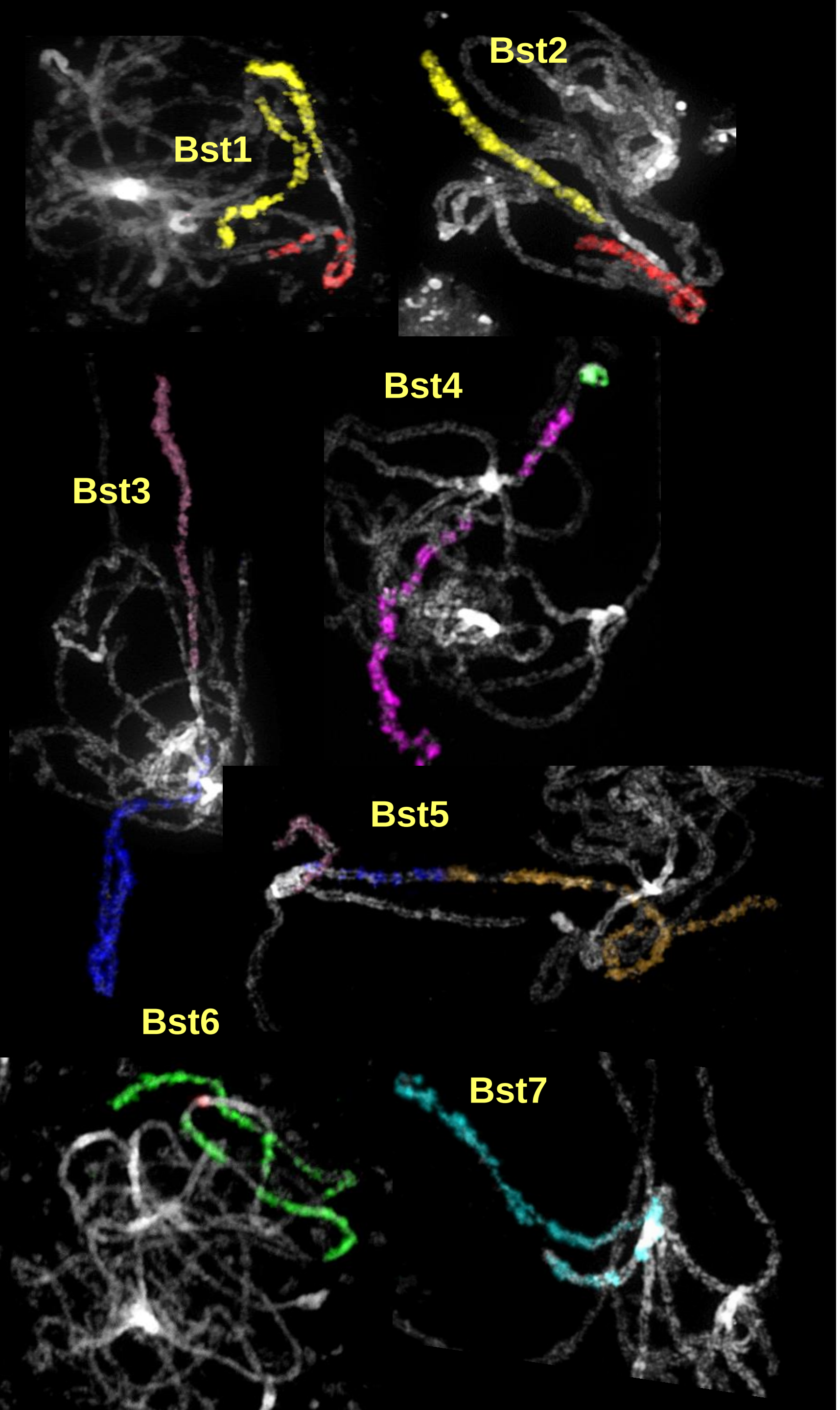


Lysak et al. 2006, PNAS; Hu et al. 2011, Nat Genet

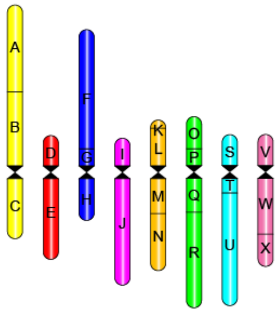
Reductions of Chromosome Numbers Were Independent and Used Different Chromosome Breakpoints



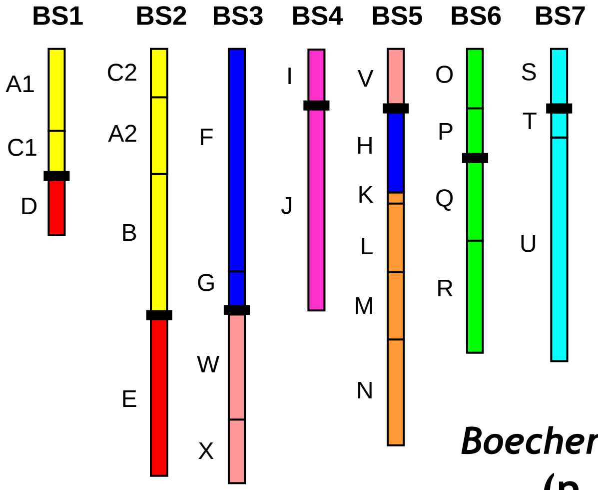
Lysak et al. (2006) PNAS



Boechera genomes originated from ACK



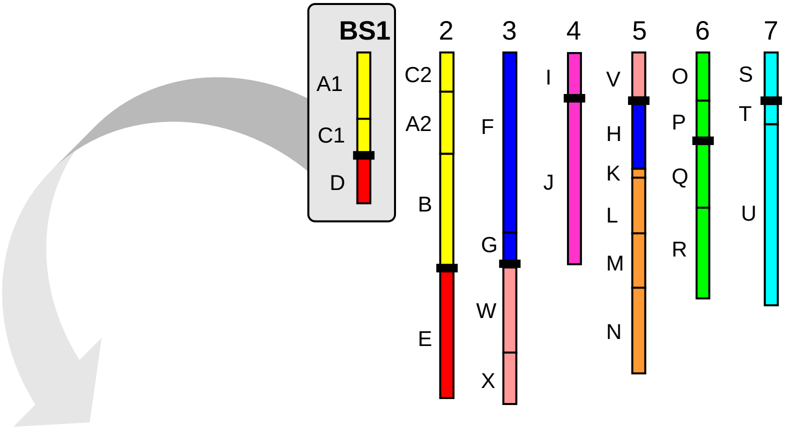
descending dysploidy
n = 8 → n = 7



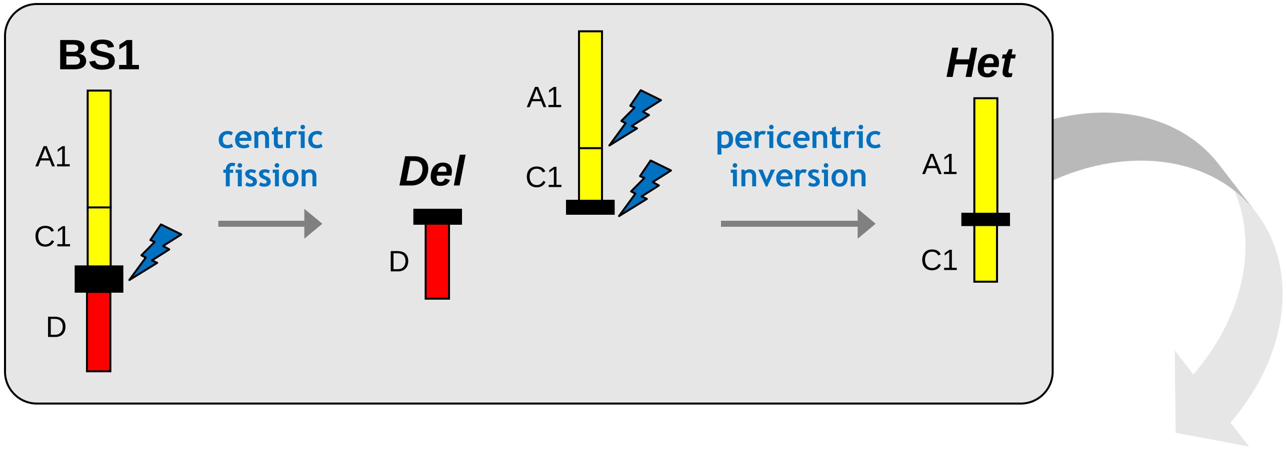
Boechera stricta
(n = 7)

Mandáková et al., unpublished

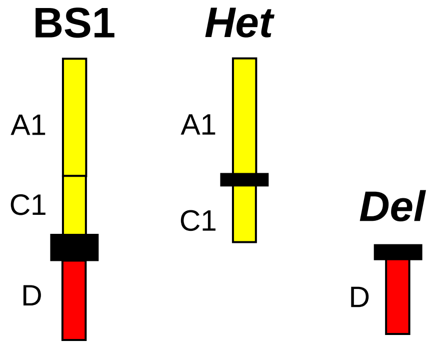
Ascending dysploidy by centric fission in *Boechera*

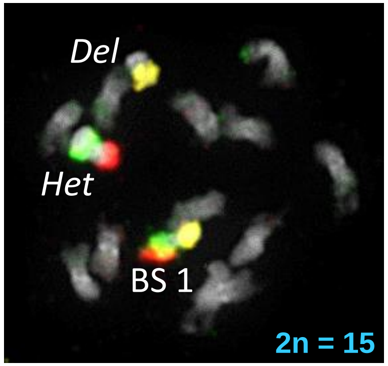


2n = 14
sexual



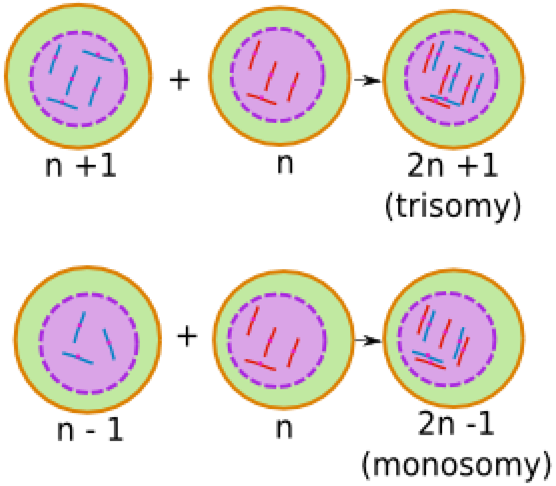
2n = 15
apomict





Mandáková et al., unpublished

Chromosome number change due to aneuploidy



Odd basic chromosome numbers in Rosaceae ($x=7, 8$ and 7; $x=17$ in the tribe Pyreae)

the Pyreae have long been considered an example of allopolyploidization between species related to extant Spiraeoideae ($x = 9$) and Amygdaleoideae ($x = 8$) taxa

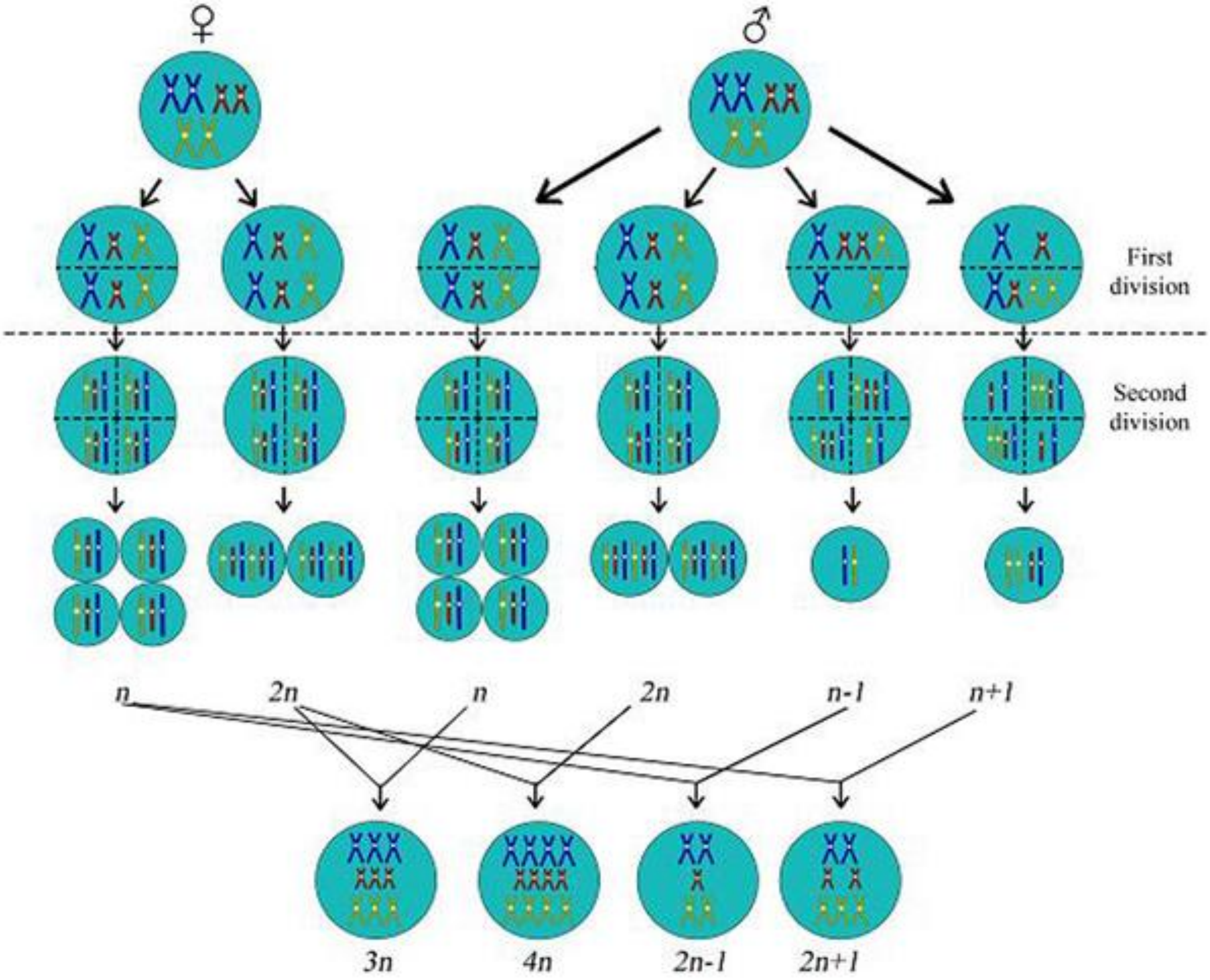
the diploid apple tree - *Malus* (Considine et al.)

- all tetraploid seedlings were derived from 2*n* ova fertilized with 2*n* spermatozoa
- all triploids from 2*n* ova fertilized with *n* spermatozoa
- **all aneuploids from *n* ova fertilized with aneuploid spermatozoa**

Thus ova only contributed euploidy while spermatozoa contributed a range of cytotypes, including aneuploidy, to non-diploid seedlings in the diploid *Malus*.

Considine MJ et al. (2012) Molecular Genetic Features of Polyploidization and Aneuploidization Reveal Unique Patterns for Genome Duplication in Diploid *Malus*. PLoS ONE 7(1): e29449.

Schematic Summary of the Features of Gametic Combinations for Apple Polyploidization in Diploid *Malus*



Odd basic chromosome numbers in the Pyreae (x=17)

Three-step scenario to the odd basic chromosome number in *Malus*:

(aneuploidization - eupolyploidization - diploidization)

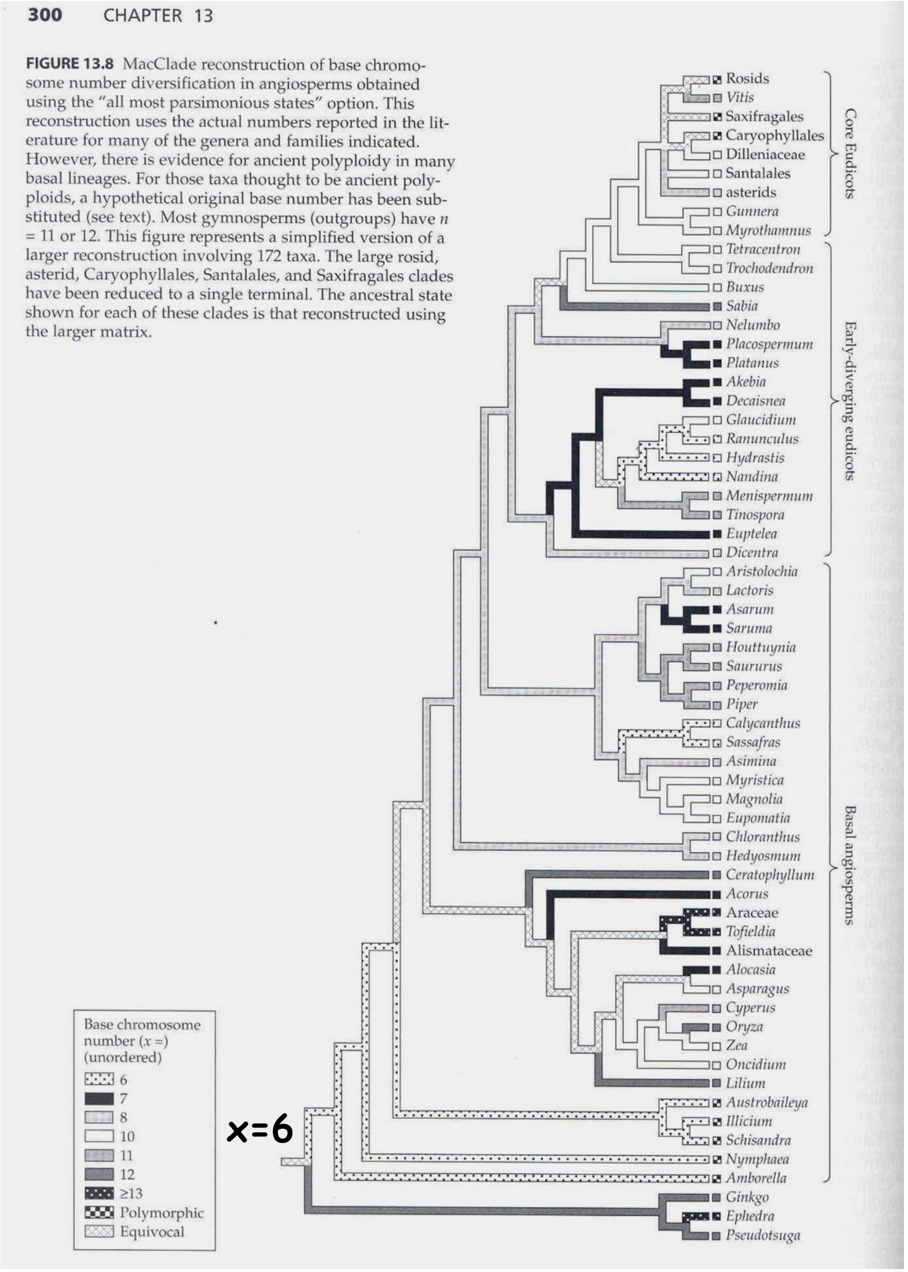
- ❖ aneuploidization of two sister taxa ($x = 9$, $2n = 18$) to $2n = 17$ ($x = 9$)
- ❖ whole-genome duplication in both ova and spermatozoa → tetraploids ($x = 9$, $4n = 34$)
- ❖ diploidization → the extant diploid state ($x = 17$, $2n = 34$) (diploid-like meiosis)

Aneuploidization can result in speciation with both odd and even basic chromosome numbers, while eupolyploidization can ONLY contribute to even basic chromosome numbers.

Reconstructing the ancestral base number for angiosperms

- the reconstructed ancestral base chromosome number is x=6

- x=6 is a theoretical reconstructed base number, it can be said that the ancestral number was low - between x=6 nad 9

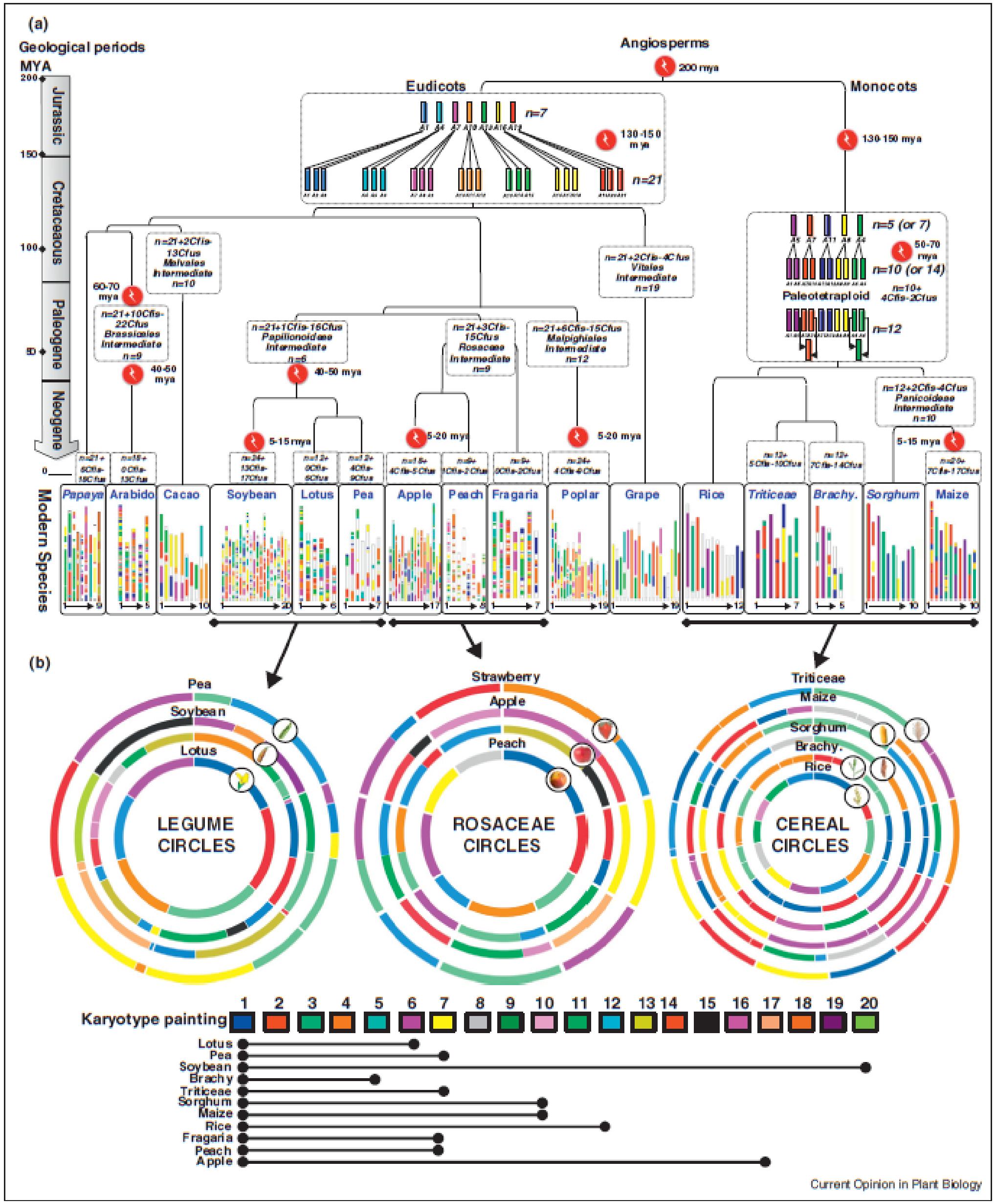


300 CHAPTER 13

FIGURE 13.8 MacClade reconstruction of base chromosome number diversification in angiosperms obtained using the "all most parsimonious states" option. This reconstruction uses the actual numbers reported in the literature for many of the genera and families indicated. However, there is evidence for ancient polyploidy in many basal lineages. For those taxa thought to be ancient polyploids, a hypothetical original base number has been substituted (see text). Most gymnosperms (outgroups) have *n* = 11 or 12. This figure represents a simplified version of a larger reconstruction involving 172 taxa. The large rosid, asterid, Caryophyllales, Santalales, and Saxifragales clades have been reduced to a single terminal. The ancestral state shown for each of these clades is that reconstructed using the larger matrix.

Soltis et al. 2005

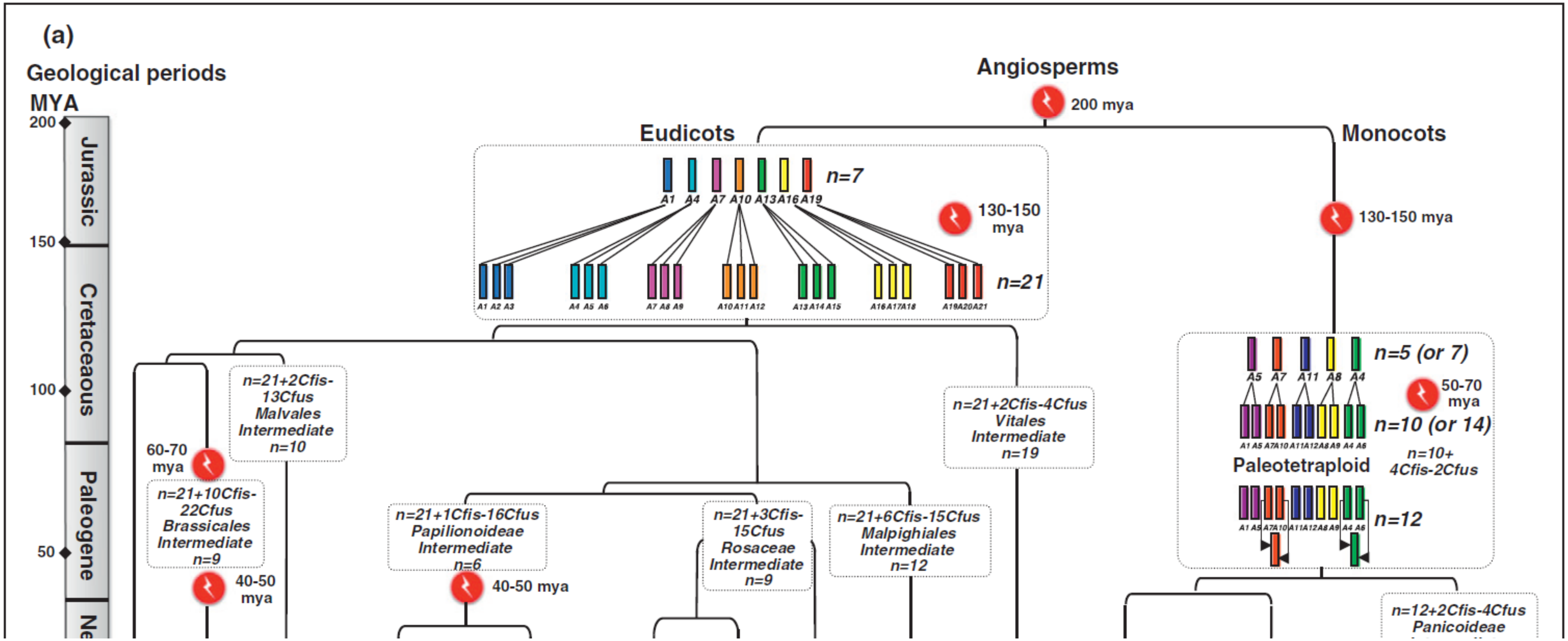
Reconstructing the ancestral chromosome number and paleogenomes



Current Opinion in Plant Biology

Salse 2012

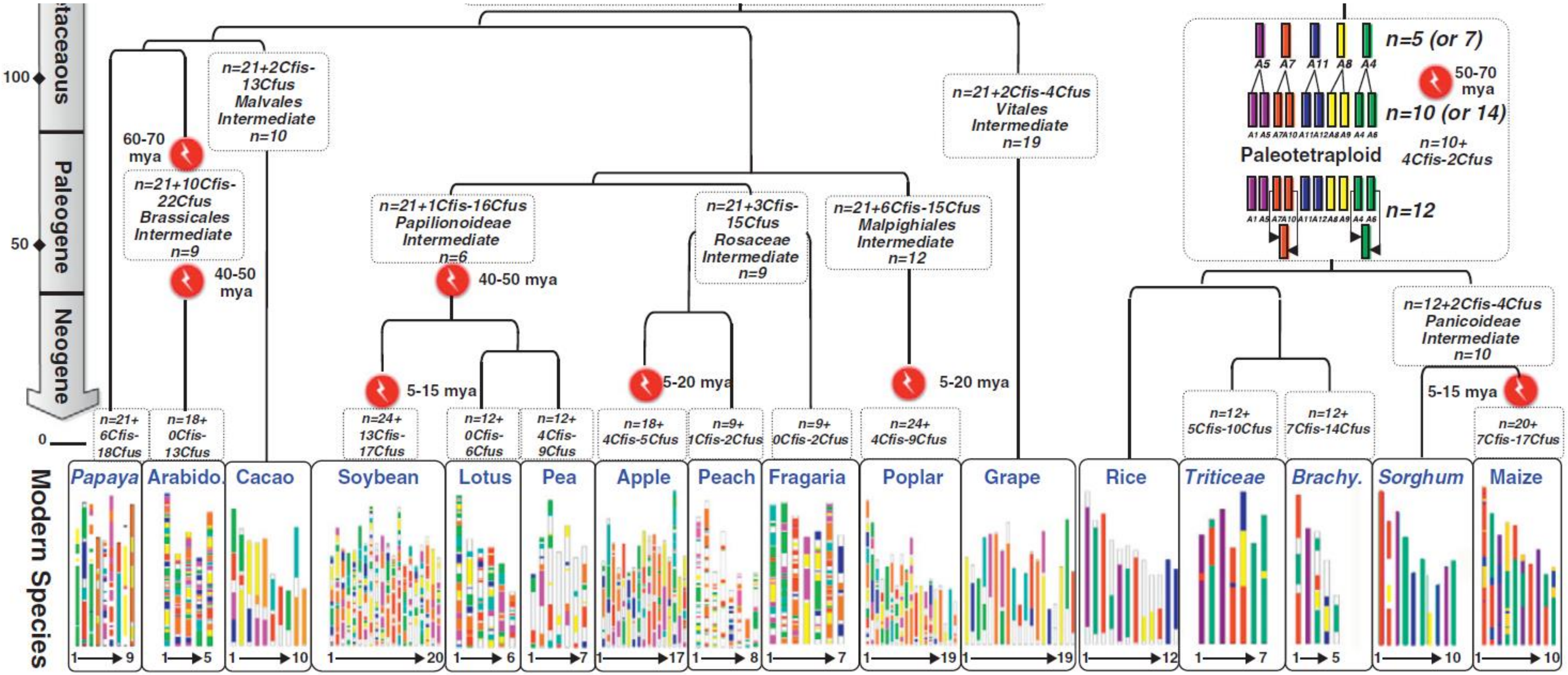
Reconstructing the ancestral chromosome number and paleogenomes



whole-genome duplication

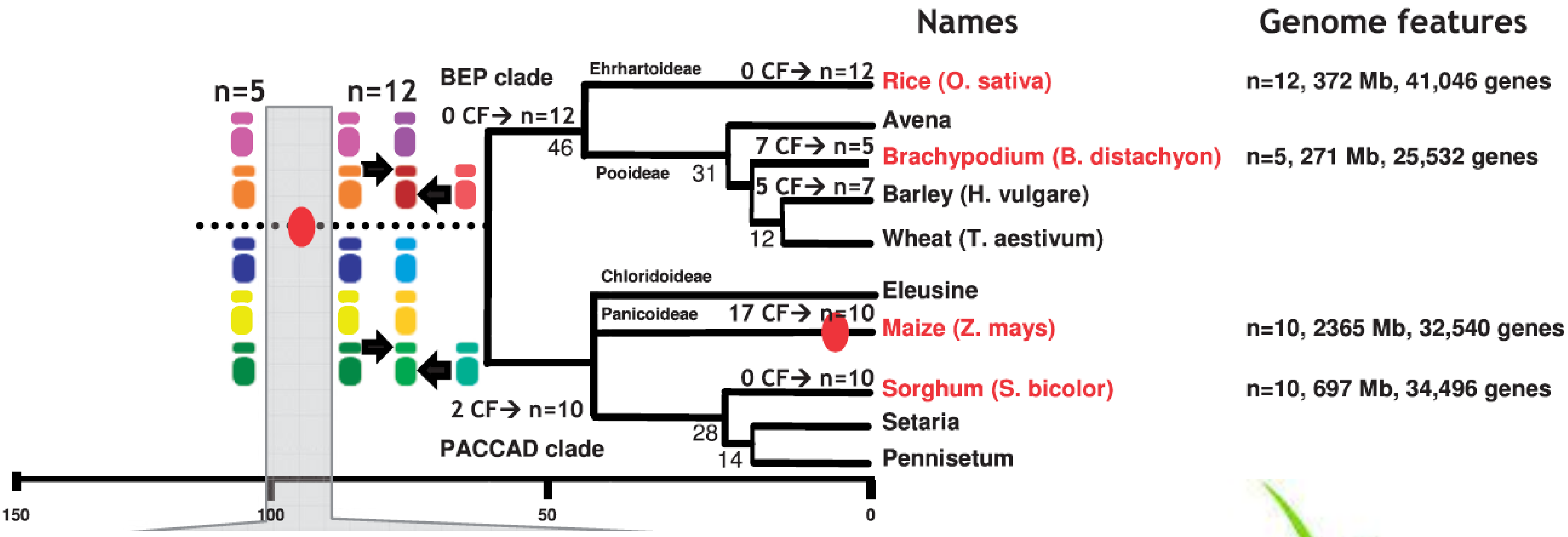
Salse 2012

Reconstructing the ancestral chromosome number and paleogenomes



whole-genome duplication

Reconstructed genome evolution in grasses



CF = „chromosome fusion“

● = whole-genome duplication (polyploidy)

Murat et al. (2014) GBE

Descending and ascending aneuploidy in *Sideritis* (*Lamiaceae*)

- bimodal pattern of chromosomal change

- Clade 1 shows decreasing aneuploid series, with 2n=44 being the ancestral number

- Clade 2 (with some ambiguity): 2n=36 is the ancestral number and ascending aneuploidy has occurred