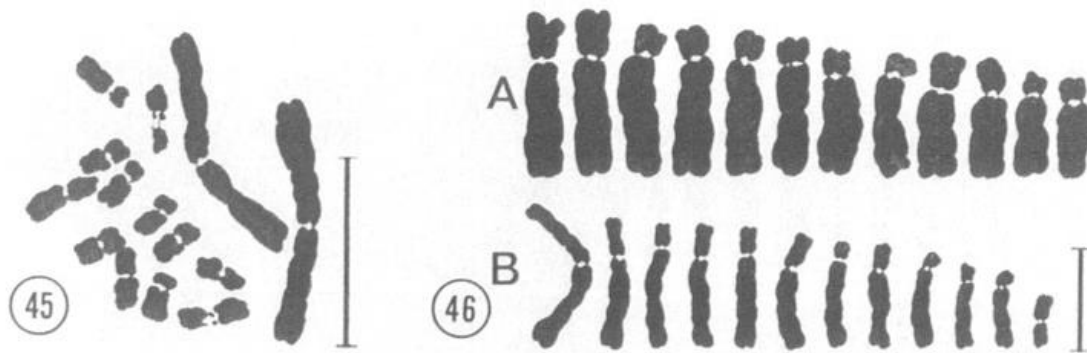


Evolutionary trends and mechanisms of chromosome number variation



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

Chromosome number variation

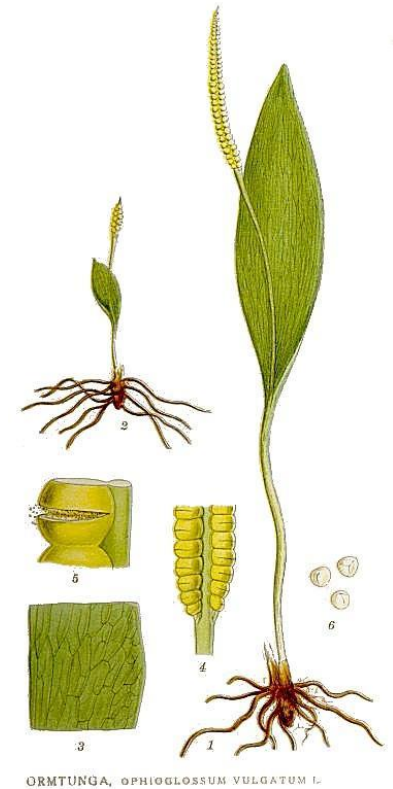
Chromosome numbers of plants vary enormously over a 360-fold range.

$n = 2$ in five angiosperm species

$n = 630$ in the fern *Ophioglossum reticulatum*



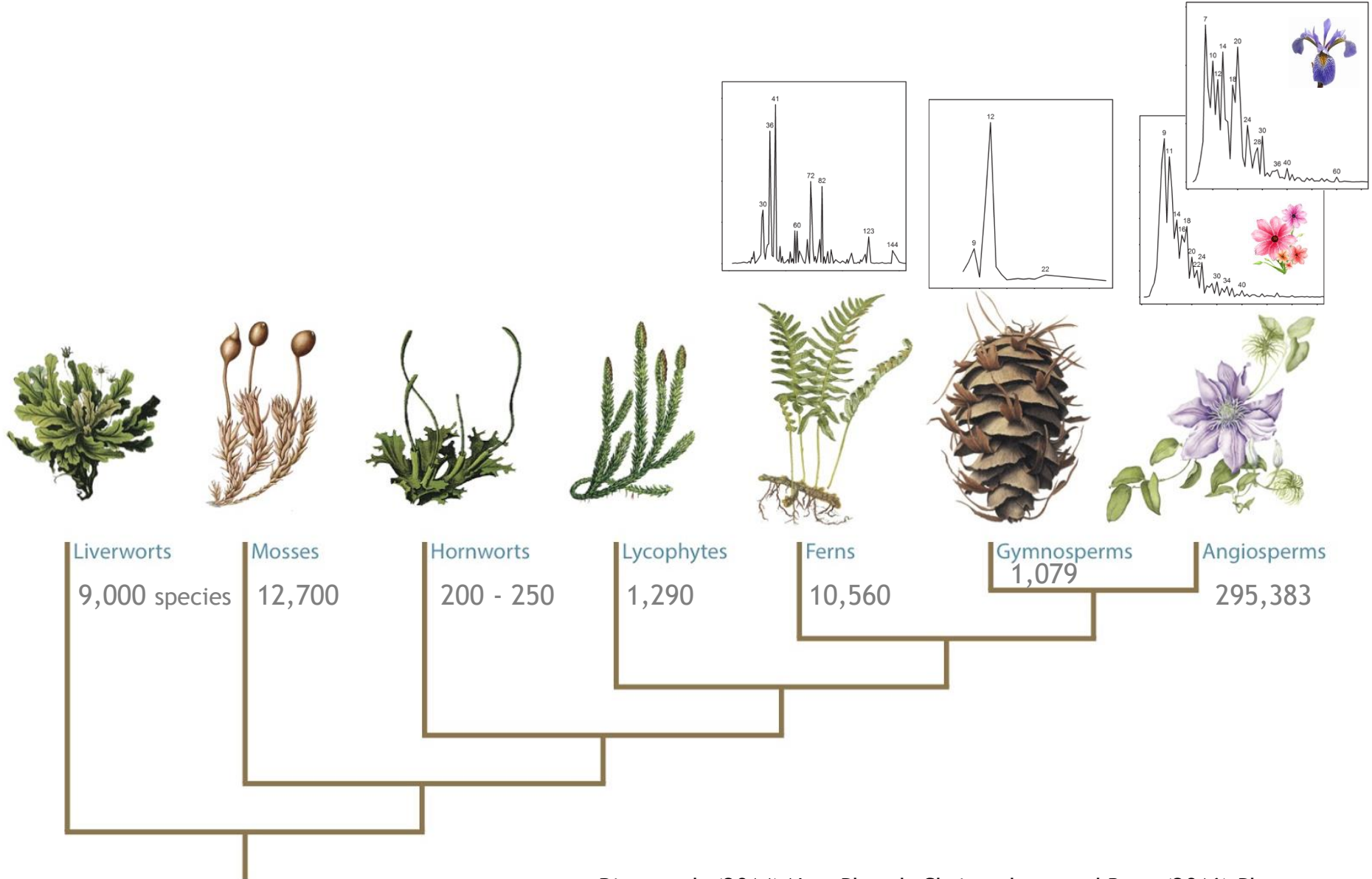
Haplopappus gracilis



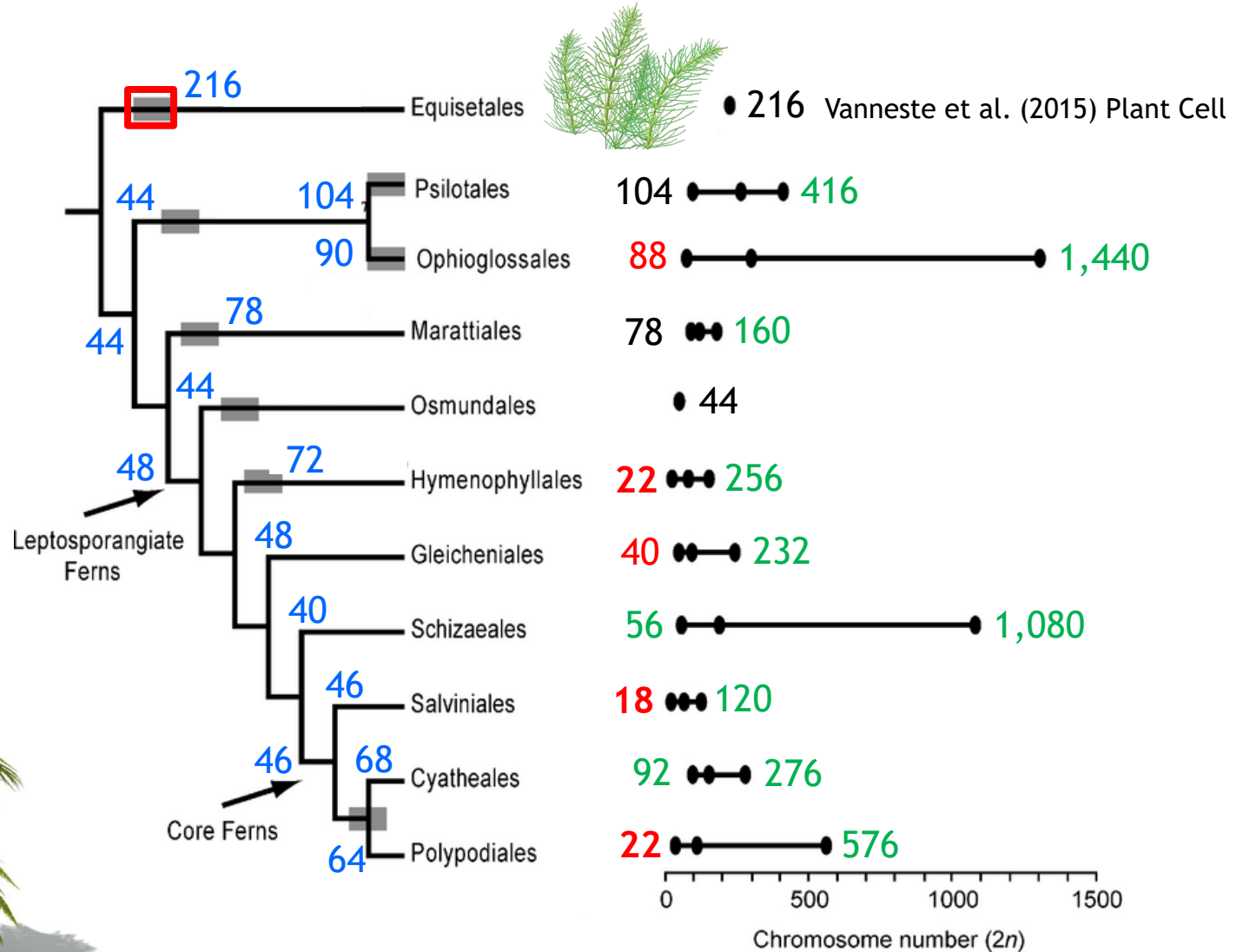
Chromosome number variation

- Stasis
- Decrease (descending dysploidy)
- Increase (polyploidy and ascending dysploidy)

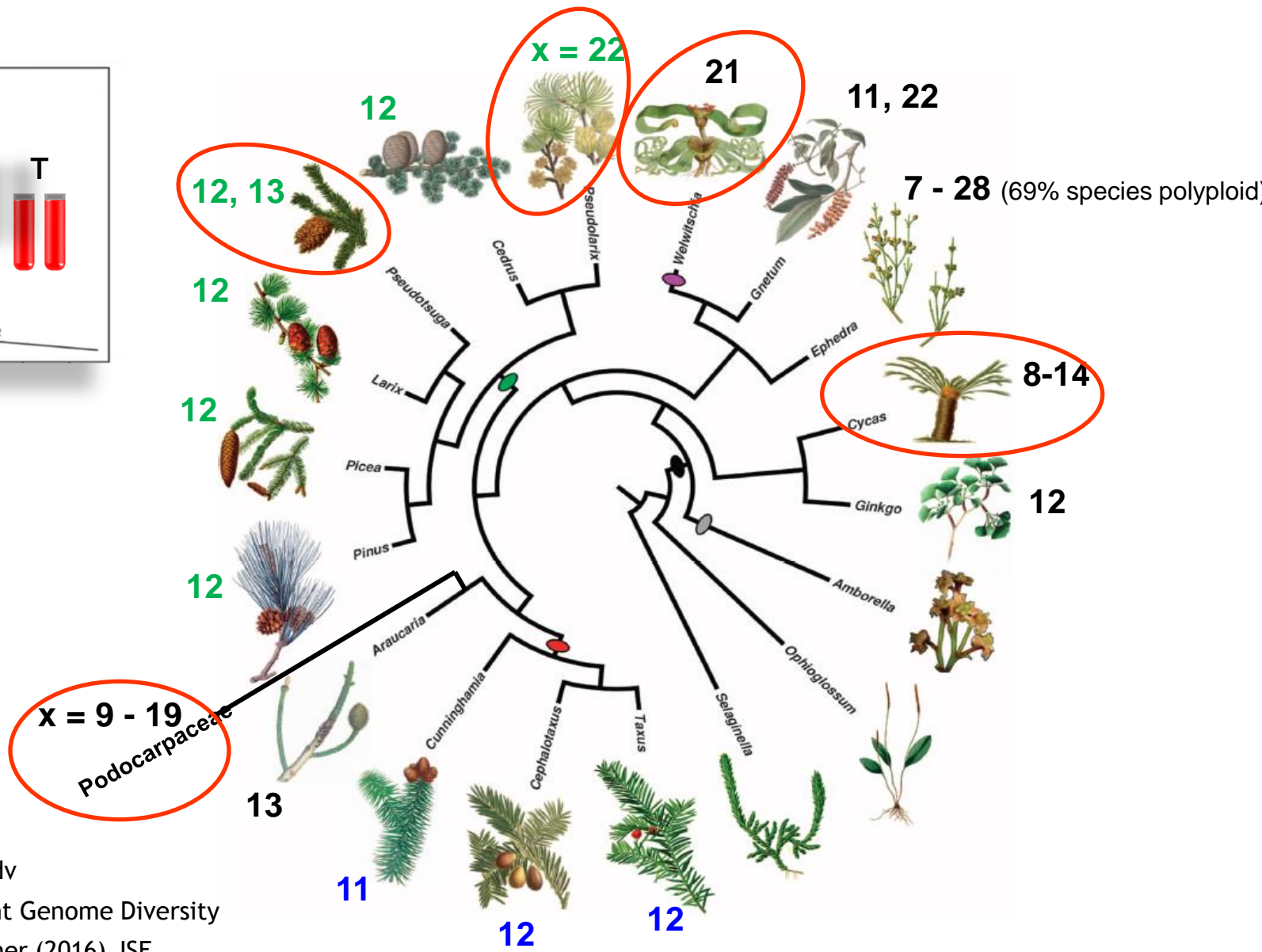
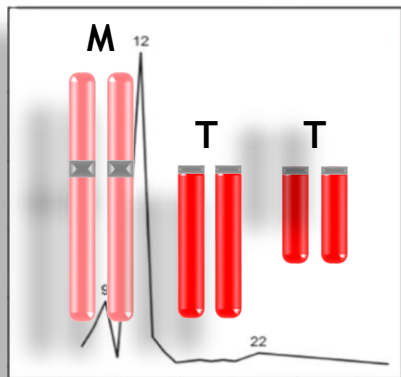
Chromosome Number Diversity Across Land Plants



Fern „Polyploidy Paradox“: high chromosome nos., but no clear evidence. Multiple WGDs and Dysploidies ?



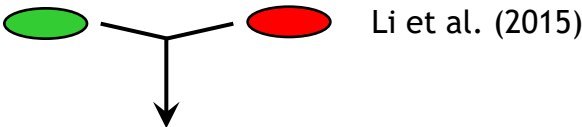
Reduced Chromosome Number Diversity and Rare Neopolyploids in Gymnosperms



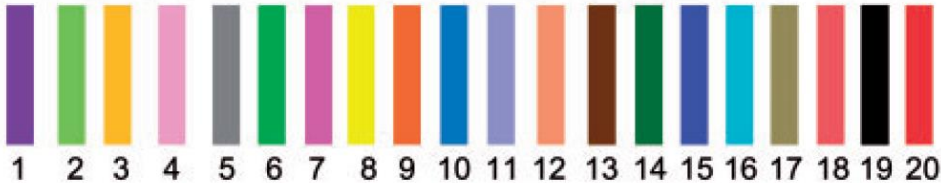
Li et al. (2015) Sci Adv
 Murray (2013) in Plant Genome Diversity
 Ickert-Bond and Renner (2016) JSE

Post-Polyploidy Chromosomal Schuffling in Coniferales ?

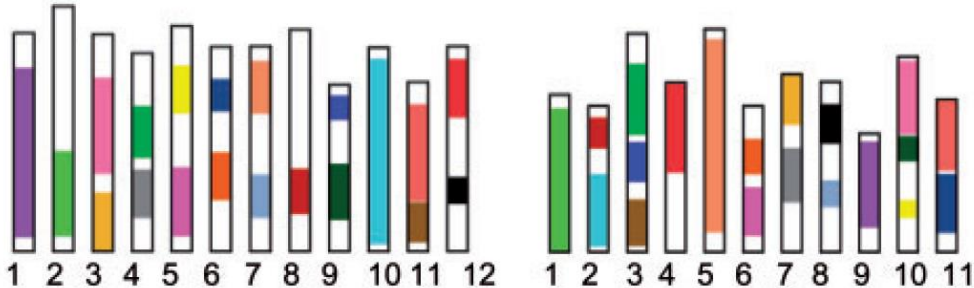
2 independent WGDs ?



20 CARs de Miguel et al. (2015)

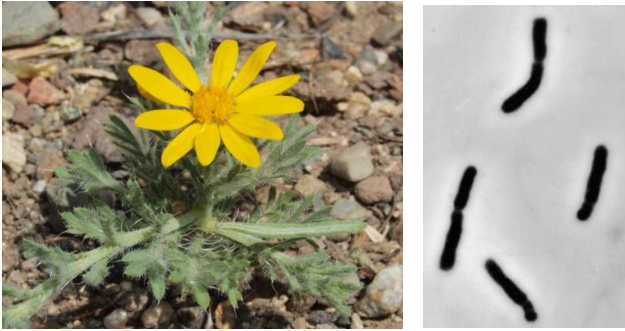


LG



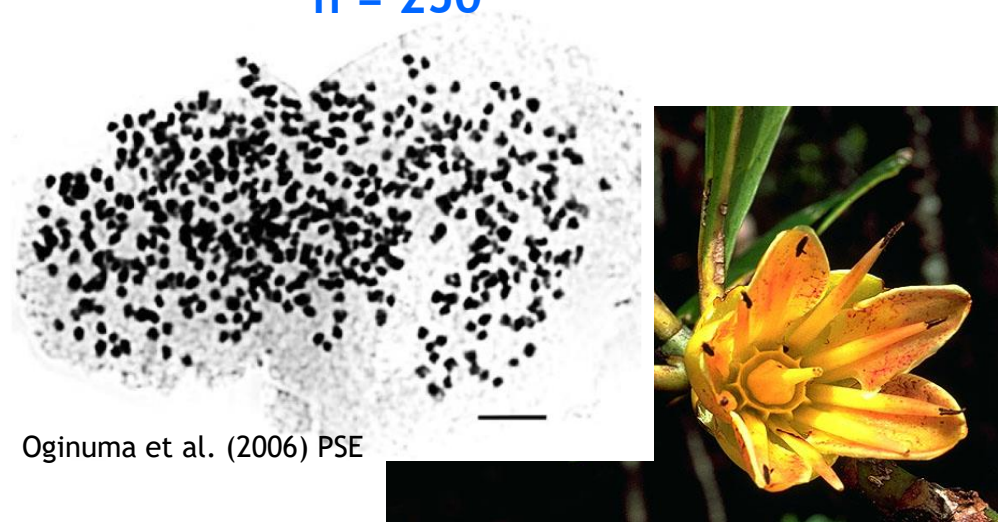
A 160-Fold Variation of Chromosome Numbers in Angiosperms

Haplopappus gracilis
n = 2



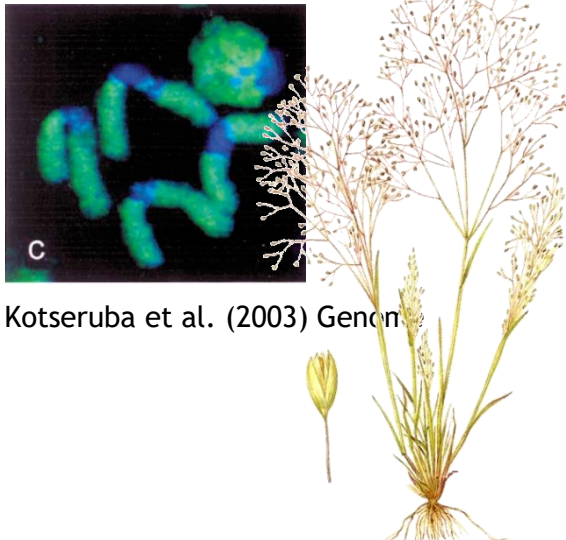
Jackson et al. (2002) Am J Bot

Strasburgeria robusta
n = 250



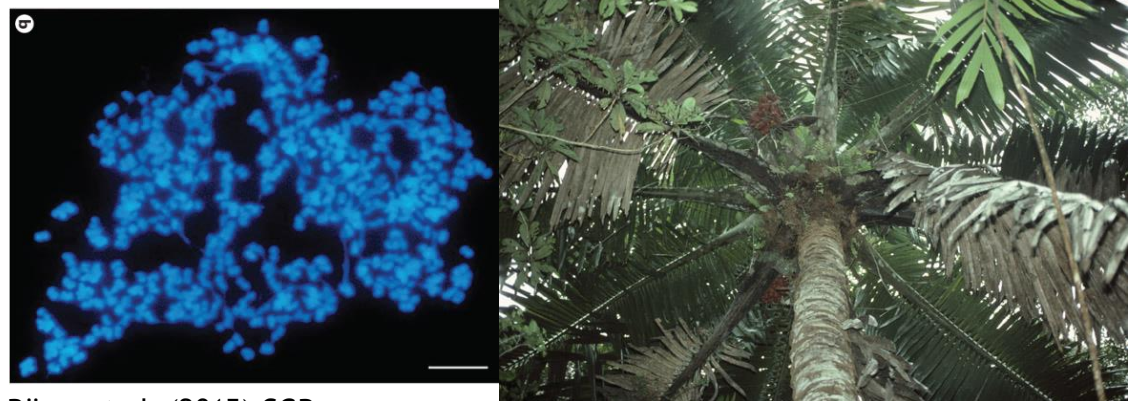
Oginuma et al. (2006) PSE

Zingeria biebersteiniana
n = 2



Kotseruba et al. (2003) Genom

Voanioala gerardii
n = 303



Röser et al. (2015) CGR

Distribution of Haploid Chromosome Numbers in Angiosperms Was Shaped by Polyploidy and Subsequent Dysploidy

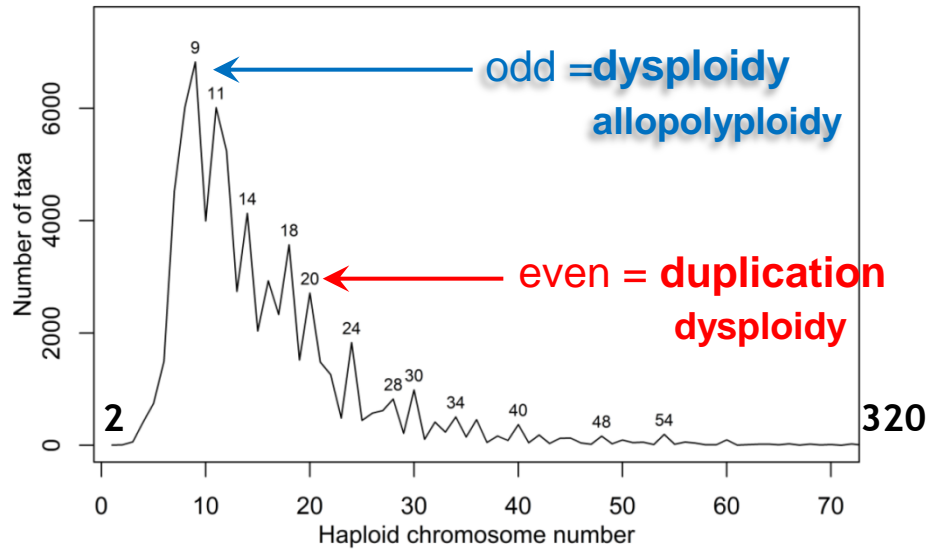
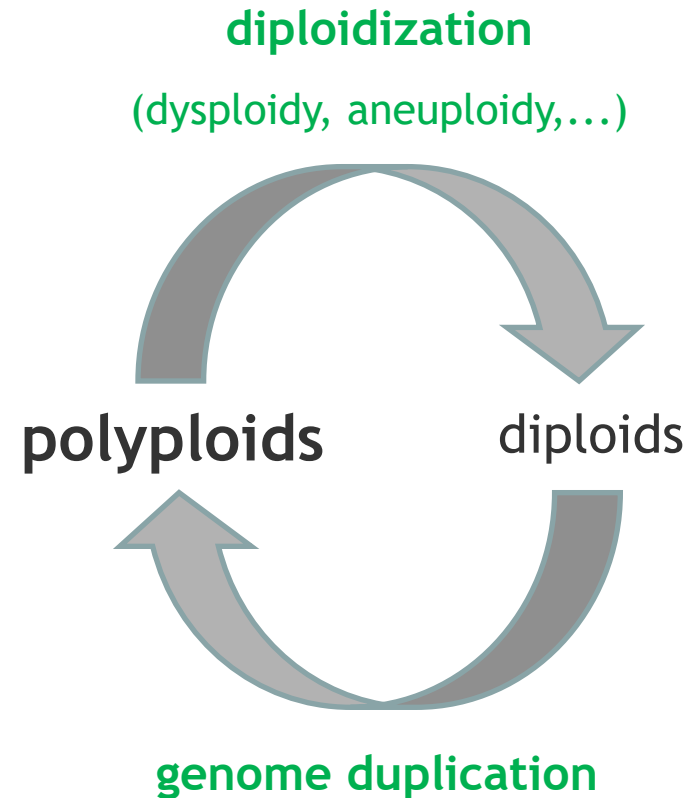


Table 3 Major groups even vs odd chromosome counts^a

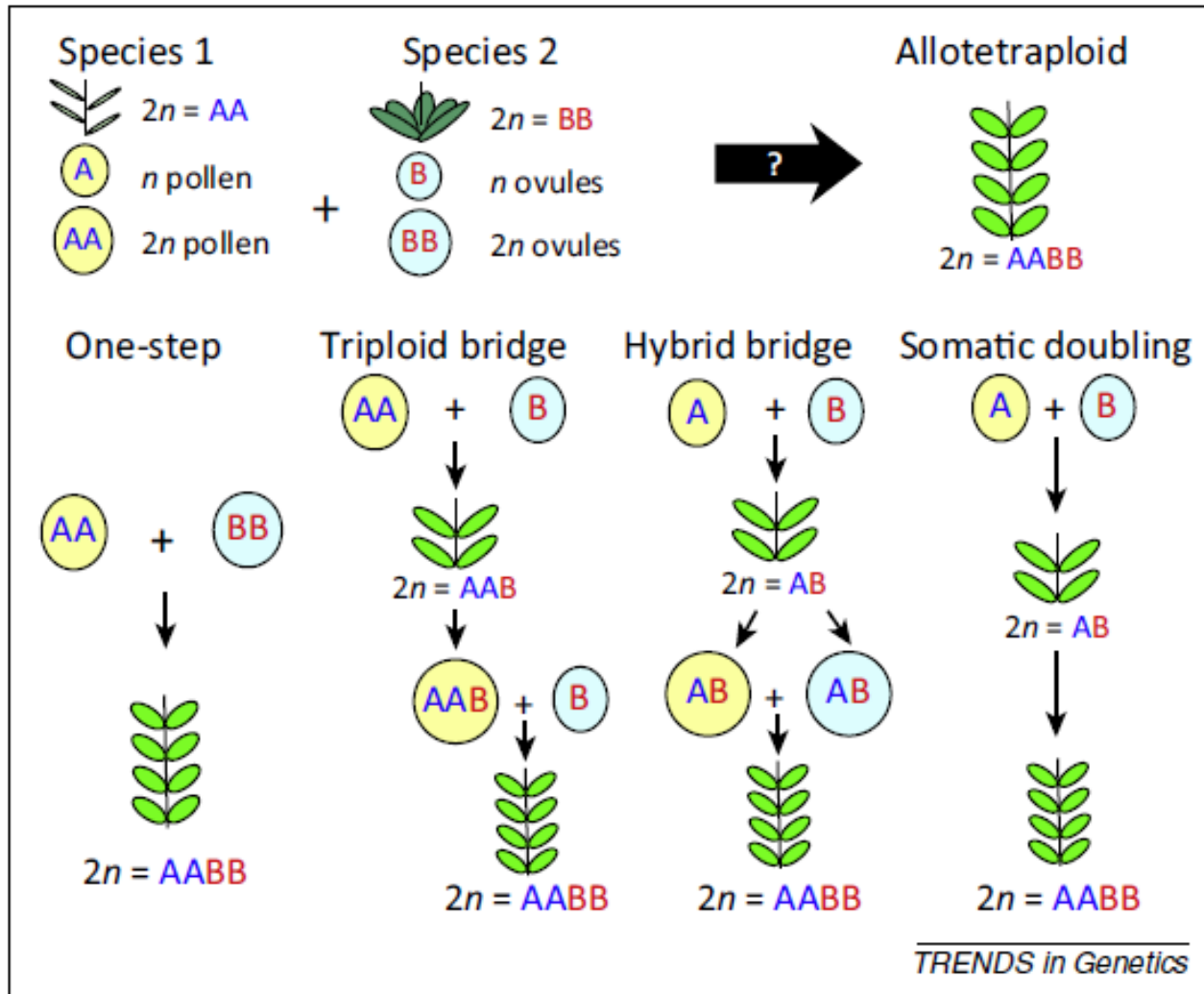
Clade	Number of total counts	Percentage of even counts
Angiosperm	70 338	56%
Monocots	15 528	58%
Eudicots	53 492	55%
Monilophytes	2986	63%
Lycophytes	220	53%
Gymnosperms	488	59%
Bryophytes	1446	48%



Chromosome number variation

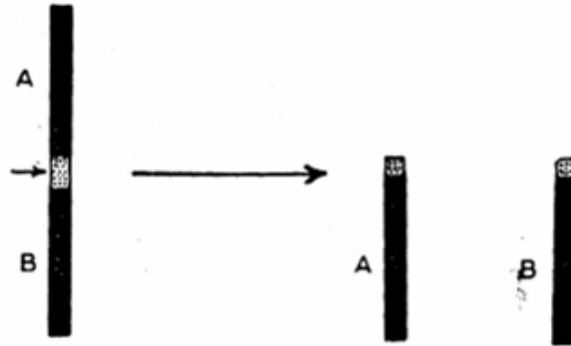
- Stasis
- Decrease (descending dysploidy)
- Increase (polyploidy and ascending dysploidy)

Allopolyploidy (and of course autopolyploidy)



Ascending dysploidy

1. Centric fission (1 metacentric chromosome \rightarrow 2 telocentrics)

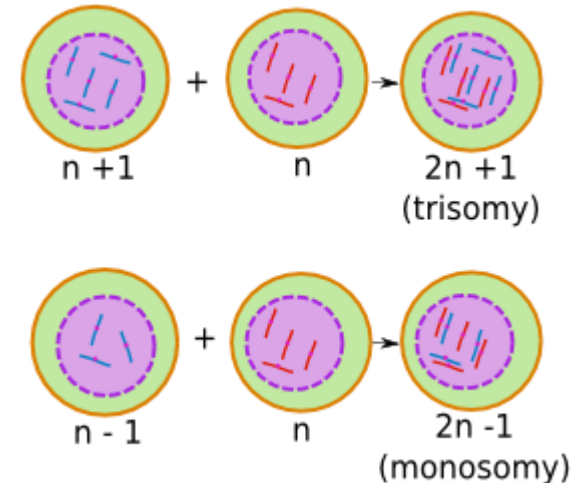


probably in orchids,
cycads...

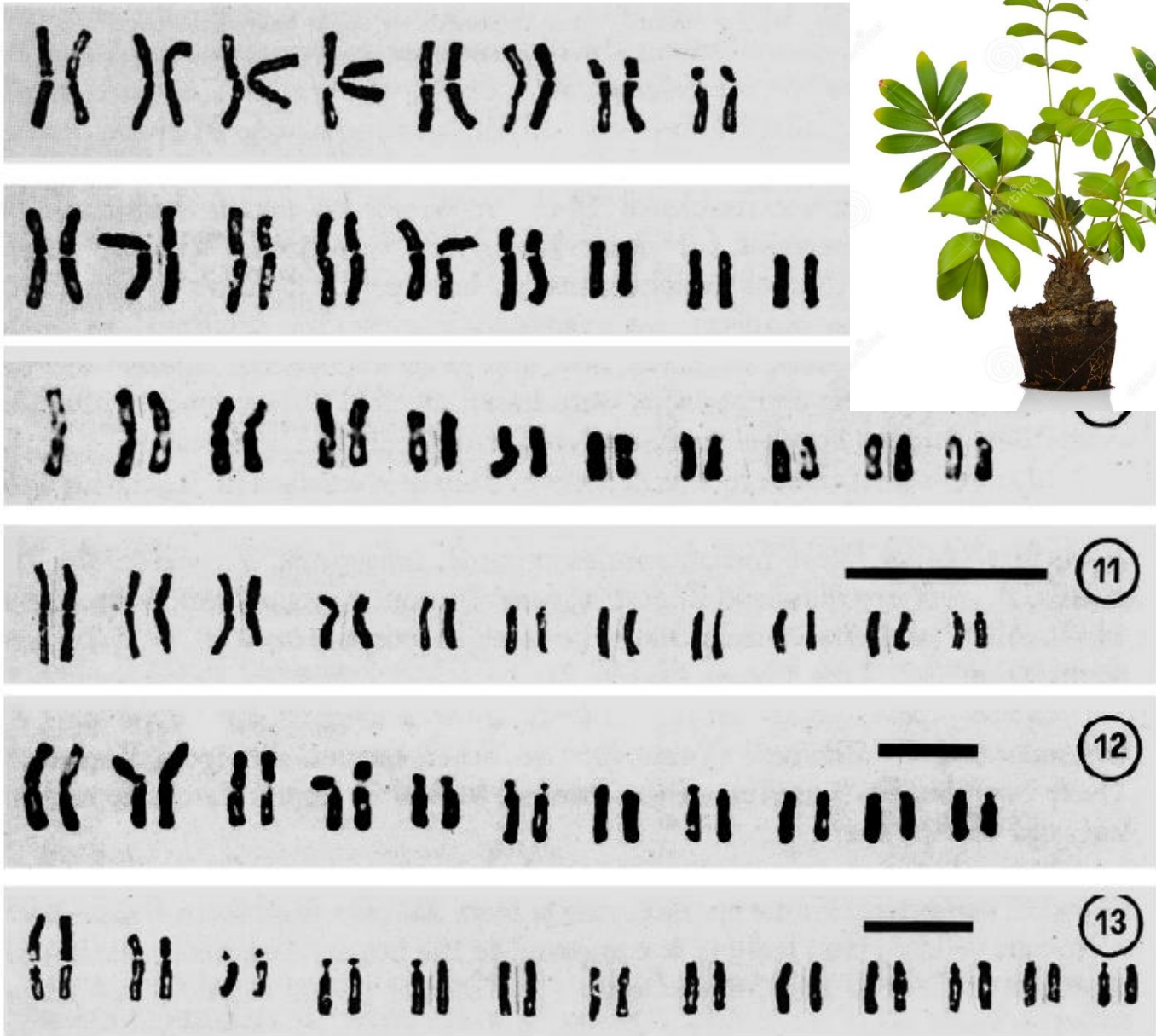
2. Meiotic misdivision (non-disjunction)

- 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)

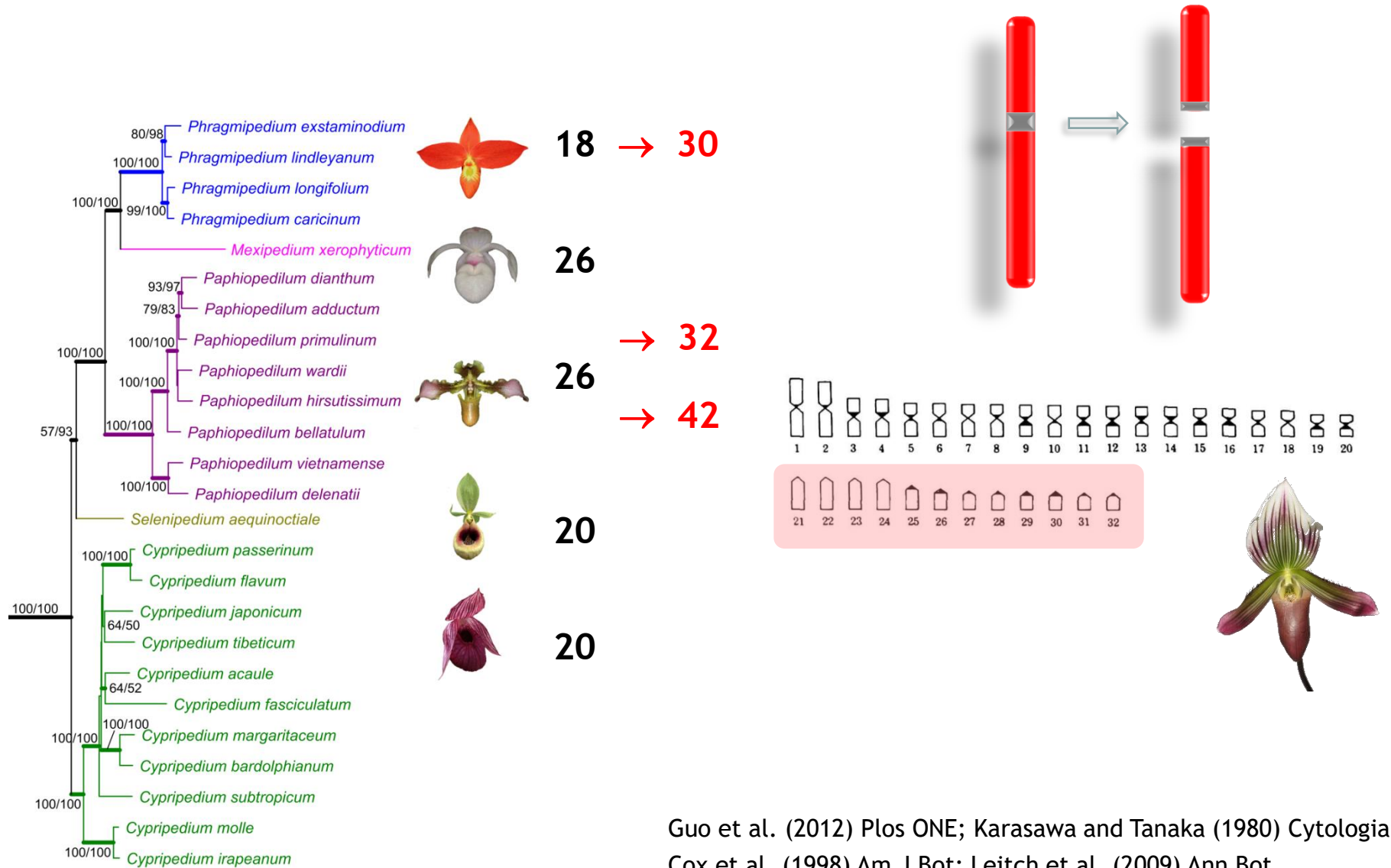
- the extra chromosome can diverge from their homologues through a translocation with non-homologous chromosomes



Centric fissions → telocentric chromosomes in cycads (Zamia)



Centric Fissions Increase Chromosome Number in Seed Plants



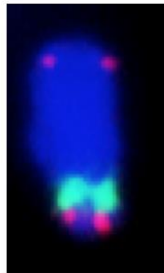
Guo et al. (2012) Plos ONE; Karasawa and Tanaka (1980) Cytologia
 Cox et al. (1998) Am J Bot; Leitch et al. (2009) Ann Bot

Centric Fissions Are Rare in Seed Plants Despite Evidence of Efficient Chromosome Healing



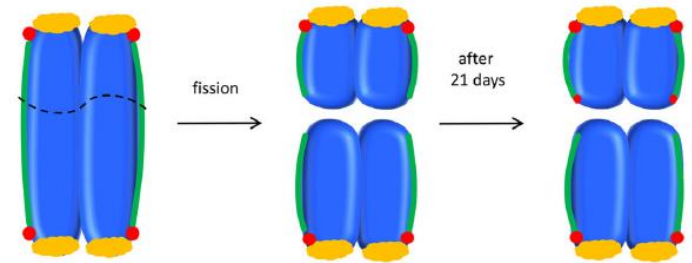
„Sticky“ chromosome ends can be stabilized by *de novo* telomere formation or „chromosome healing.“

CRW Telomere



Luzula elegans

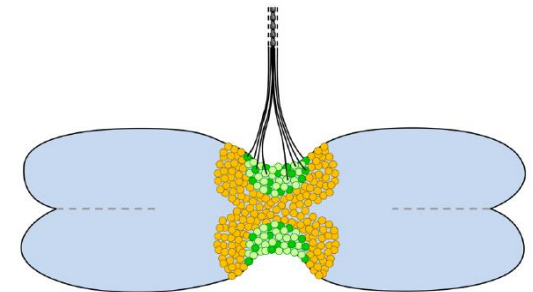
Jankowska et al. (2015) Chromosoma



bread wheat, ditelosomic Dt 3AS
courtesy of B. Friebe (Koo et al. 2015, PLoS ONE)

Telocentrics can be „unstable“...

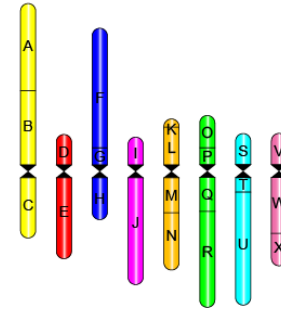
Wanner et al. (2015) Chromosoma



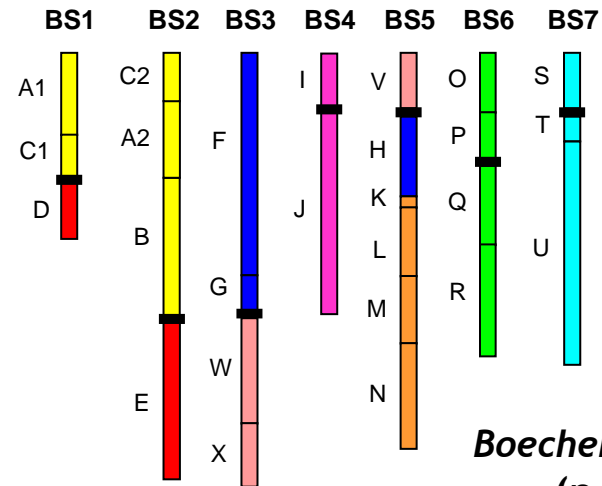
McClintock (1941), Marks (1957), Brighton (1978), Schubert et al. (1992), Slijepcevic and Bryant (1998), Tsujimoto et al. (1999), Jankowska et al. (2015), Koo et al. (2015), Wanner et al. (2015), Rocha et al. (2016)



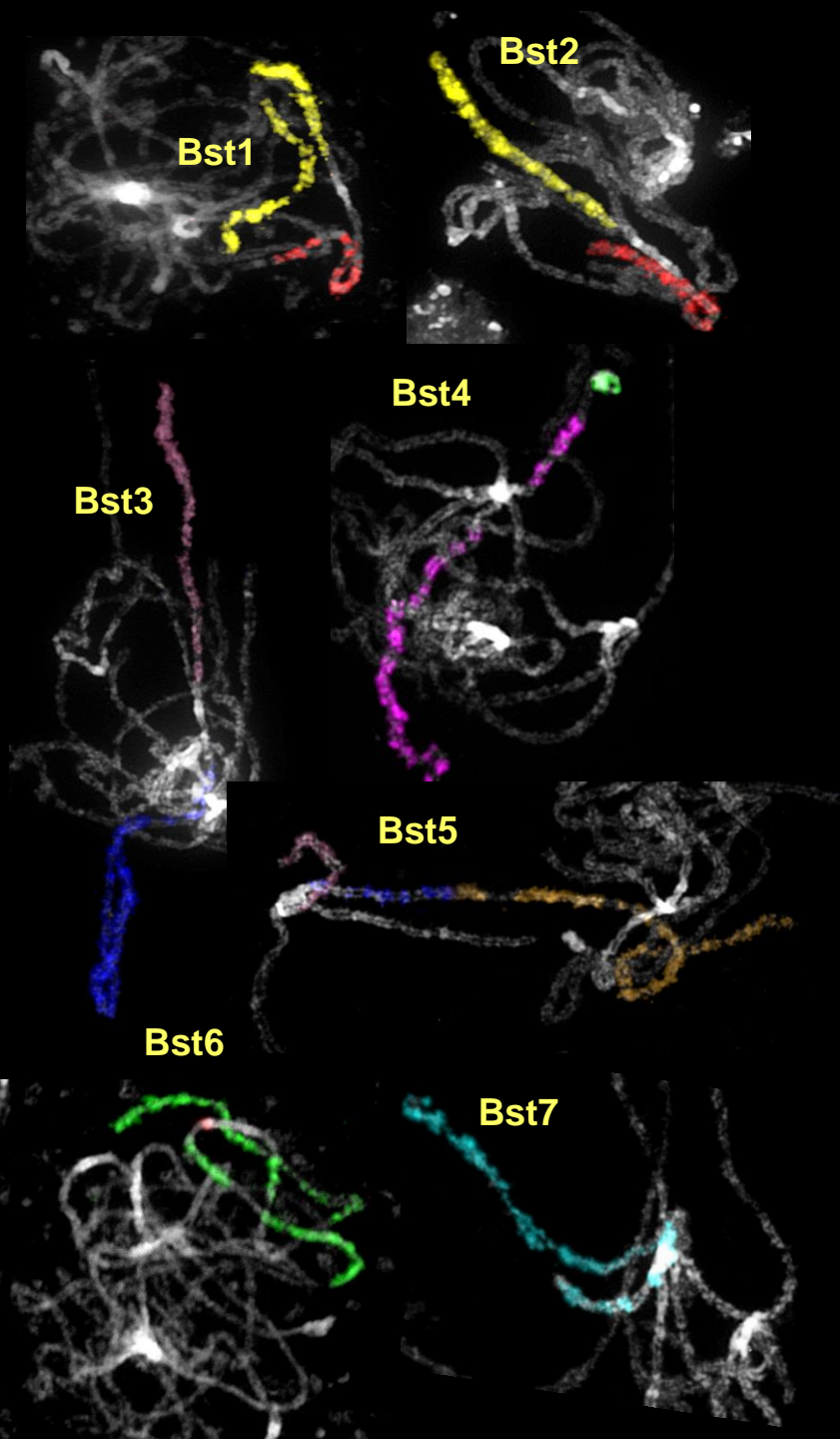
Boecheera genomes originated from eight ancestral chromosomes



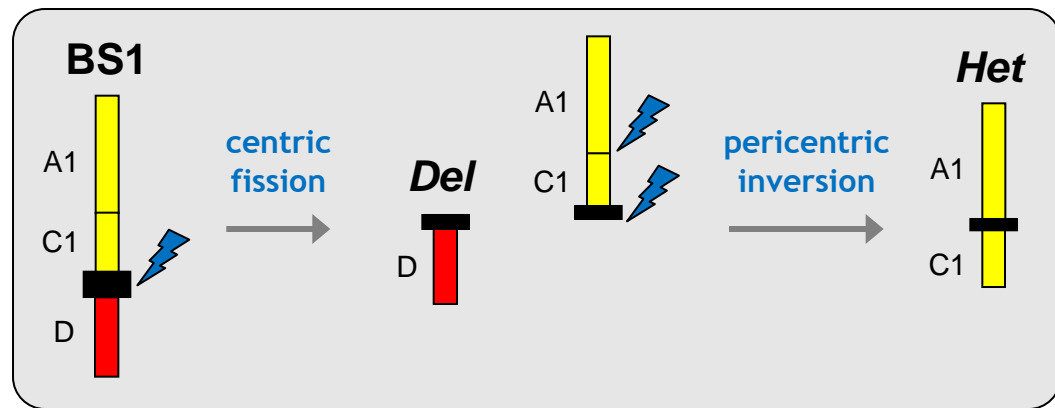
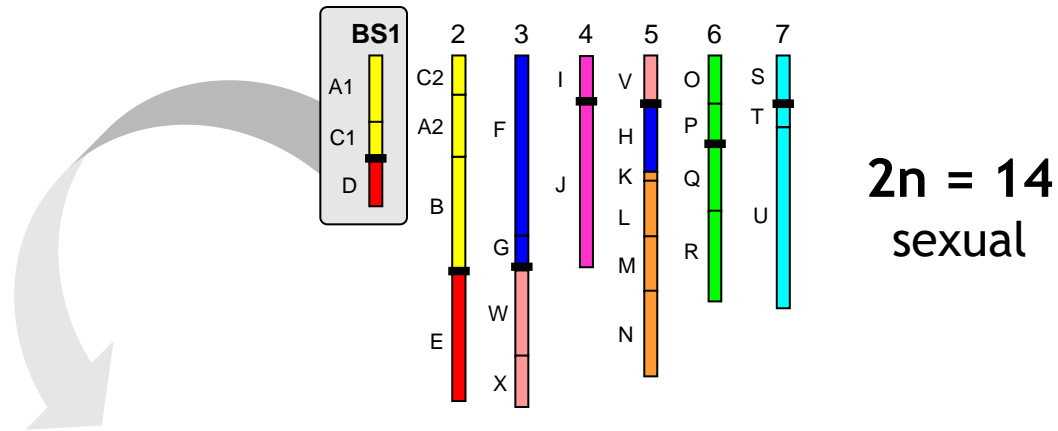
descending dysploidy
 $n = 8 \rightarrow n = 7$



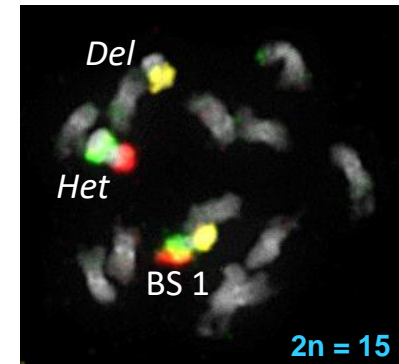
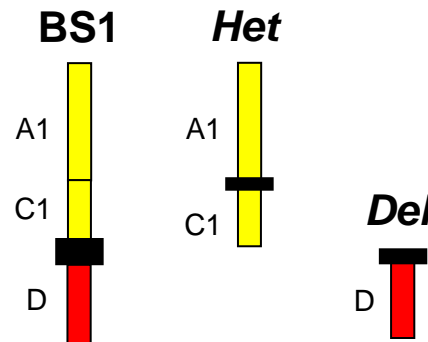
Boecheera stricta
($n = 7$)



Ascending dysploidy by centric fission fixed due to apomixis



2n = 15
apomict

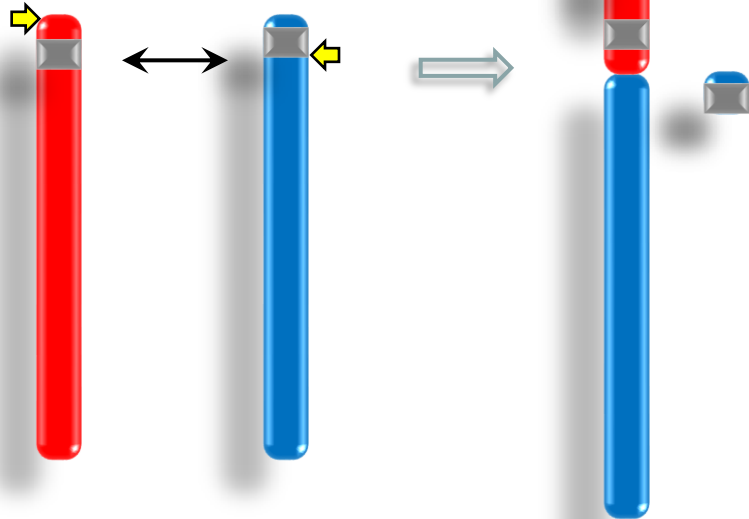


Chromosome number variation

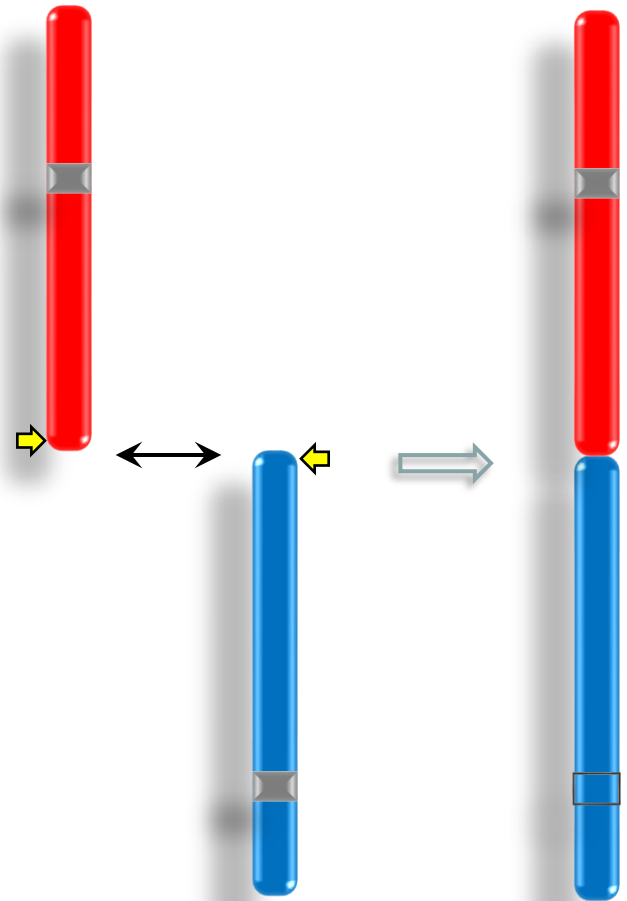
- Stasis
- Decrease (descending dysploidy)
- Increase (polyploidy and ascending dysploidy)

Descending Dysploidy via Terminal Chromosome Translocations

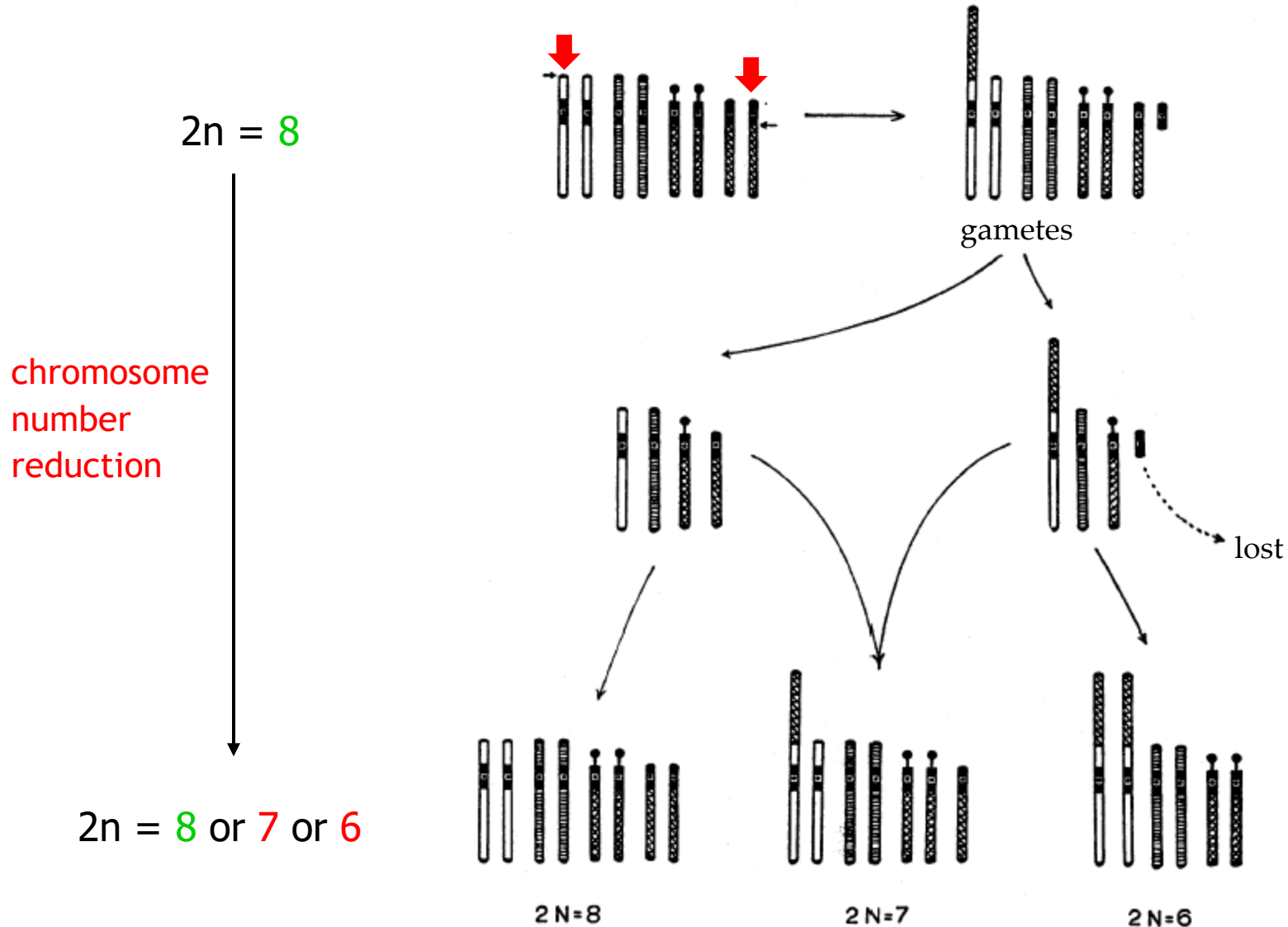
Roberstonian(-like)
translocations /
centric „fusions“



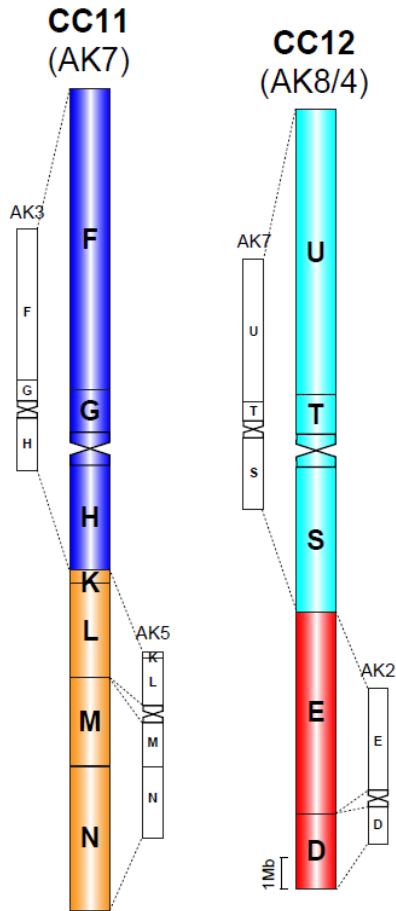
End-to-end
translocations / „fusions“



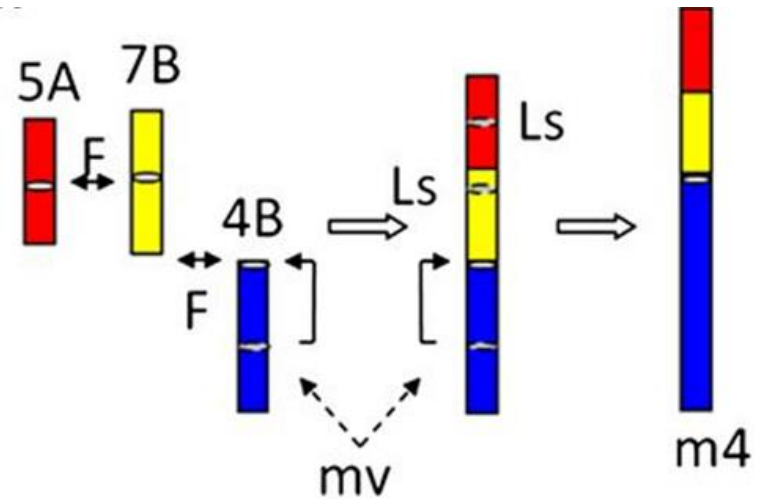
Robertsonian (unequal reciprocal) translocation and meiotic segregation



End-to-End Translocations in Plants Are Probably More Common Than Previously Thought

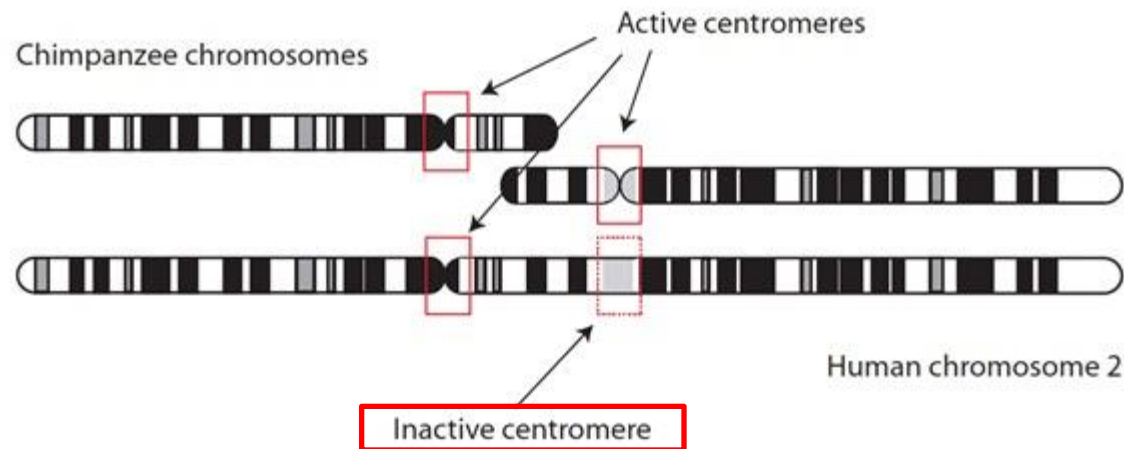


Mandáková et al. (2016) Am J Bot



Wang and Bennetzen (2012) PNAS

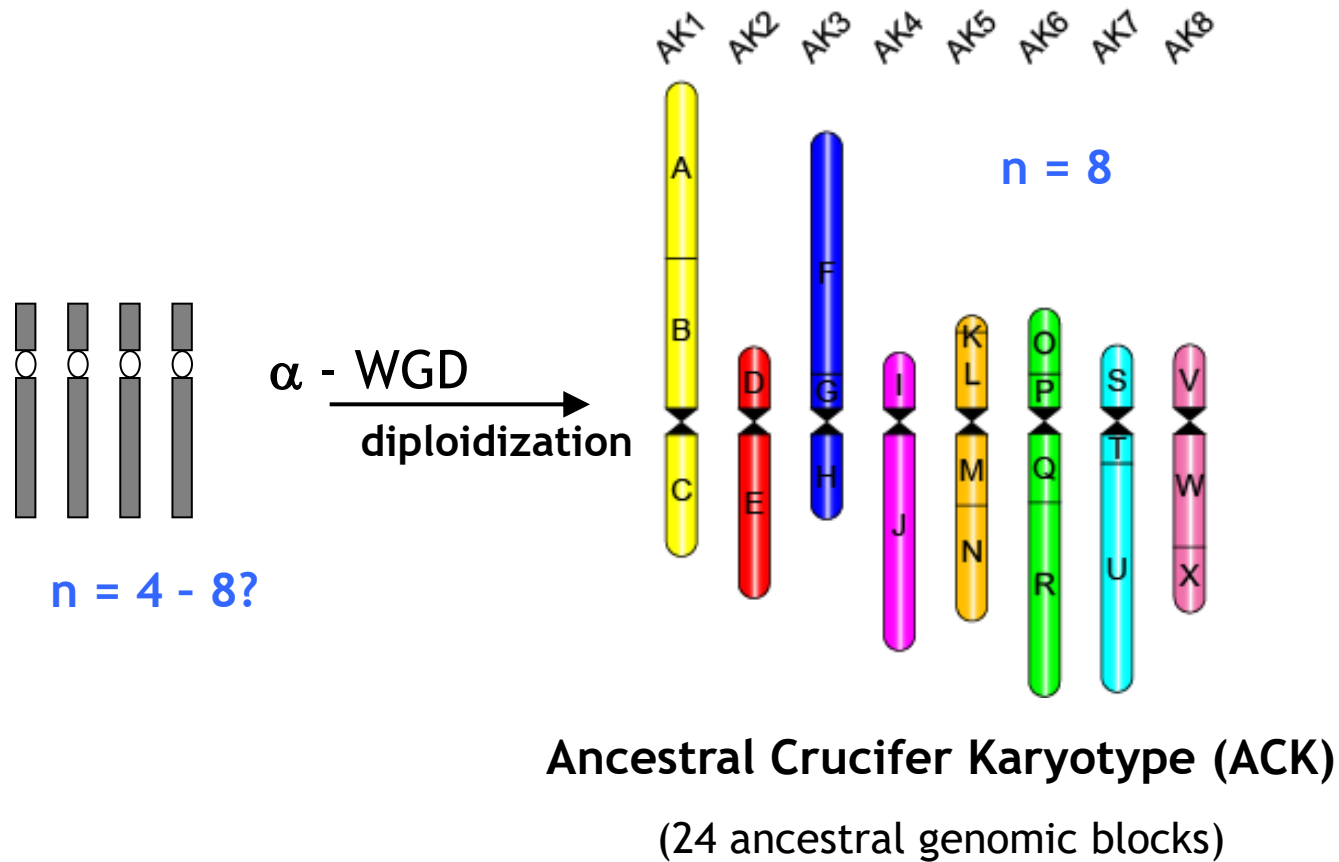
Chromosome „fusion“ - the origin of the human (dicentric) chromosome 2



Two options how the „fusion“ chromosome 2 was stabilized

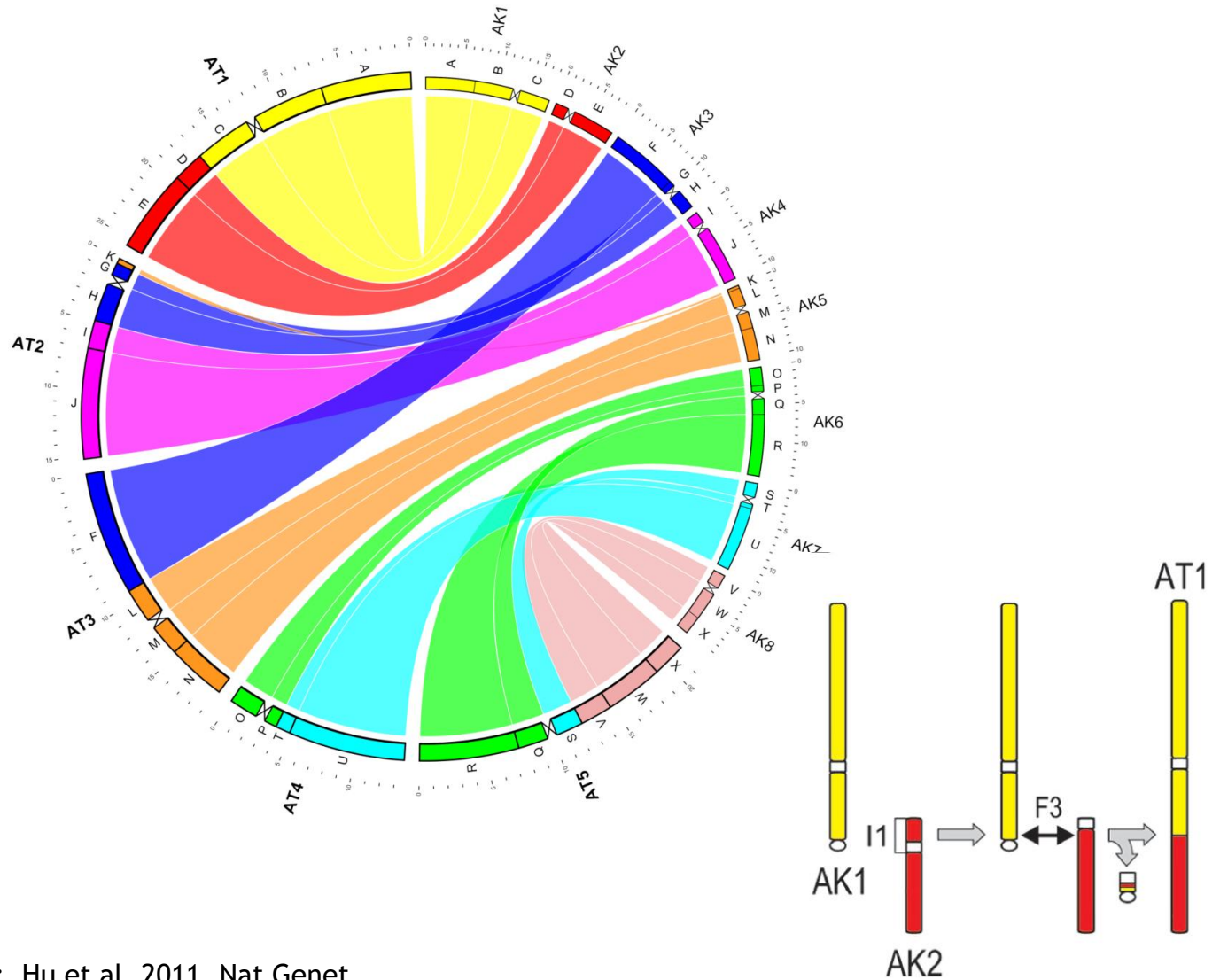
- the ancestral centromere (AC) was either epigenetically inactivated or centromere-determining sequences were excised
- the excision is more probable - what mechanism?
- recombination-based excision, most likely in one step (similar human clinical cases...)

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

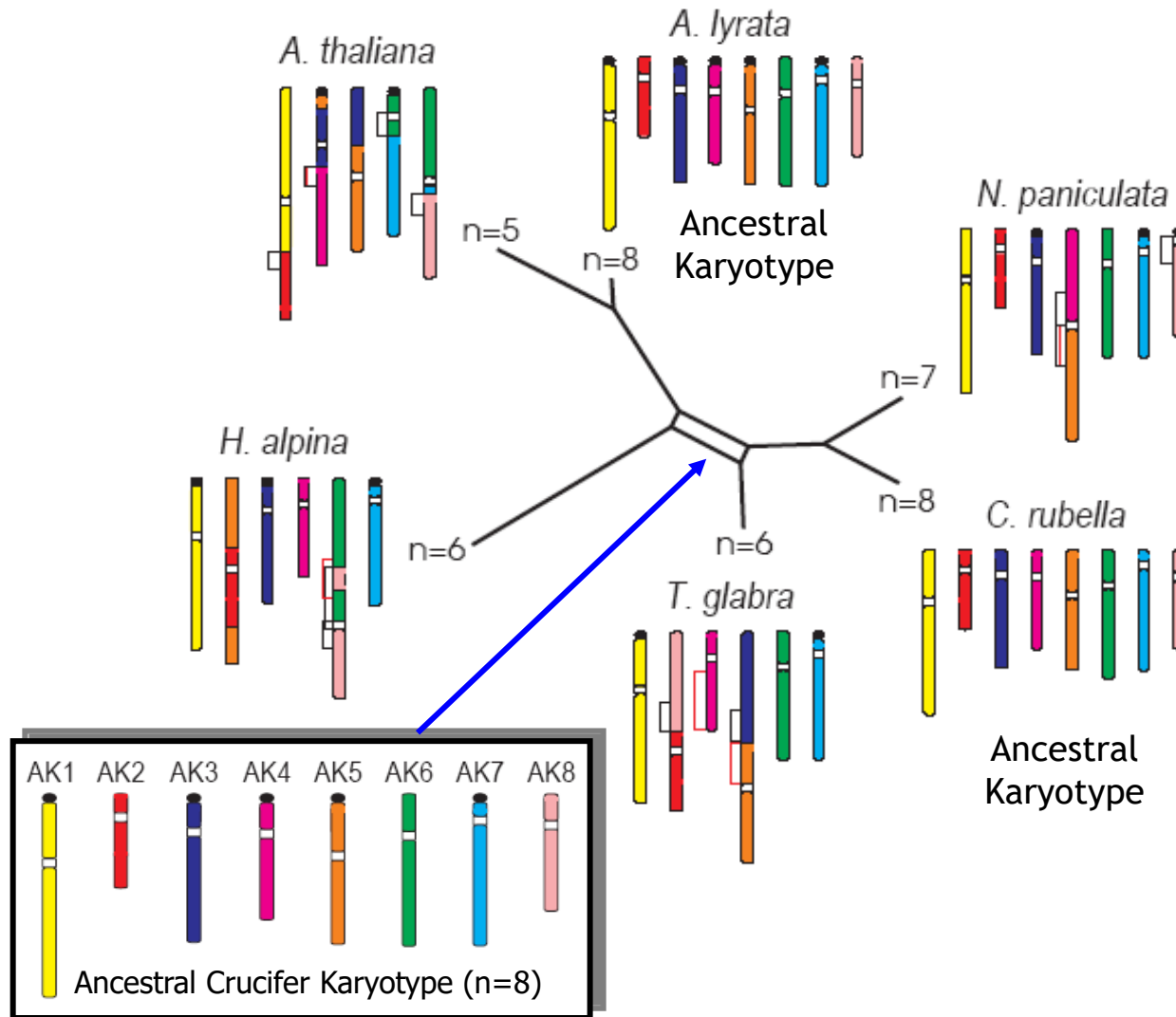


WGD = whole-genome duplication (polyploidization)

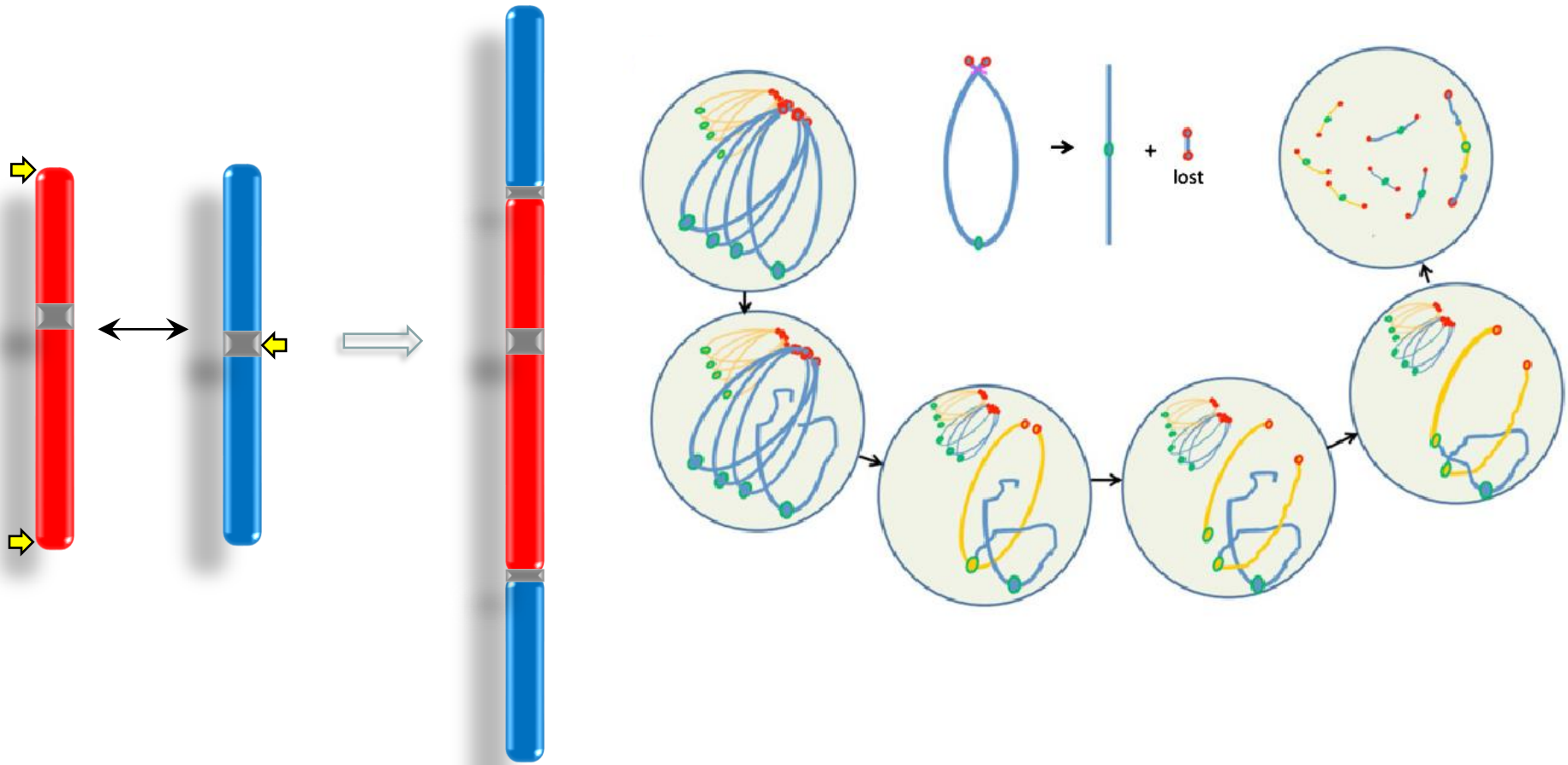
Arabidopsis thaliana - extensive chromosome reshuffling linked with chromosome number reduction from $n=8$ to $n=5$



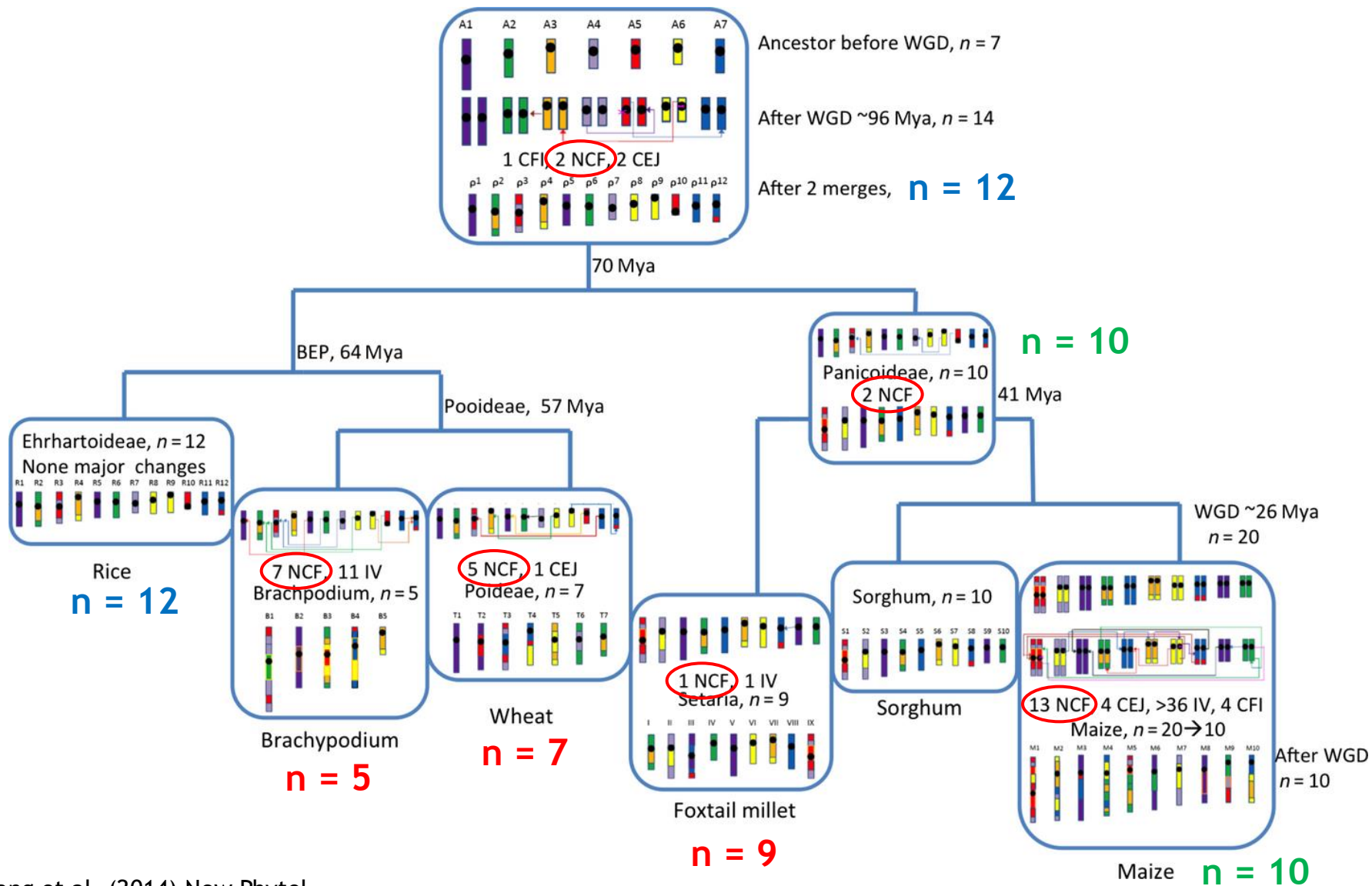
Reductions of Chromosome Numbers Were Independent and Used Different Chromosome Breakpoints



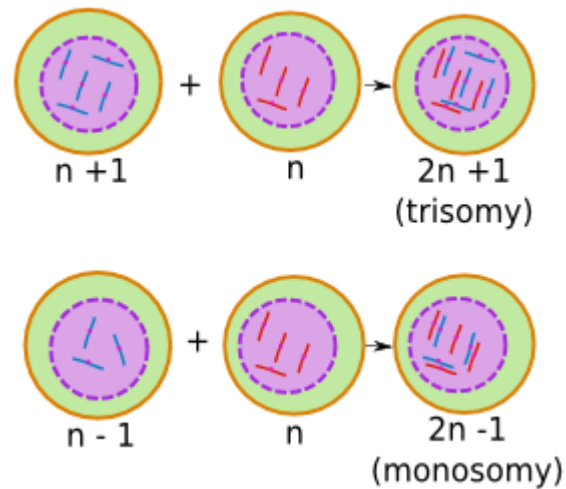
Descending Dysploidy in Grasses is Mediated by Nested Chromosome Insertions (NCIs)



Nested Chromosome Insertions Repeatedly Mediated Descending Dysploidy in Grasses



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

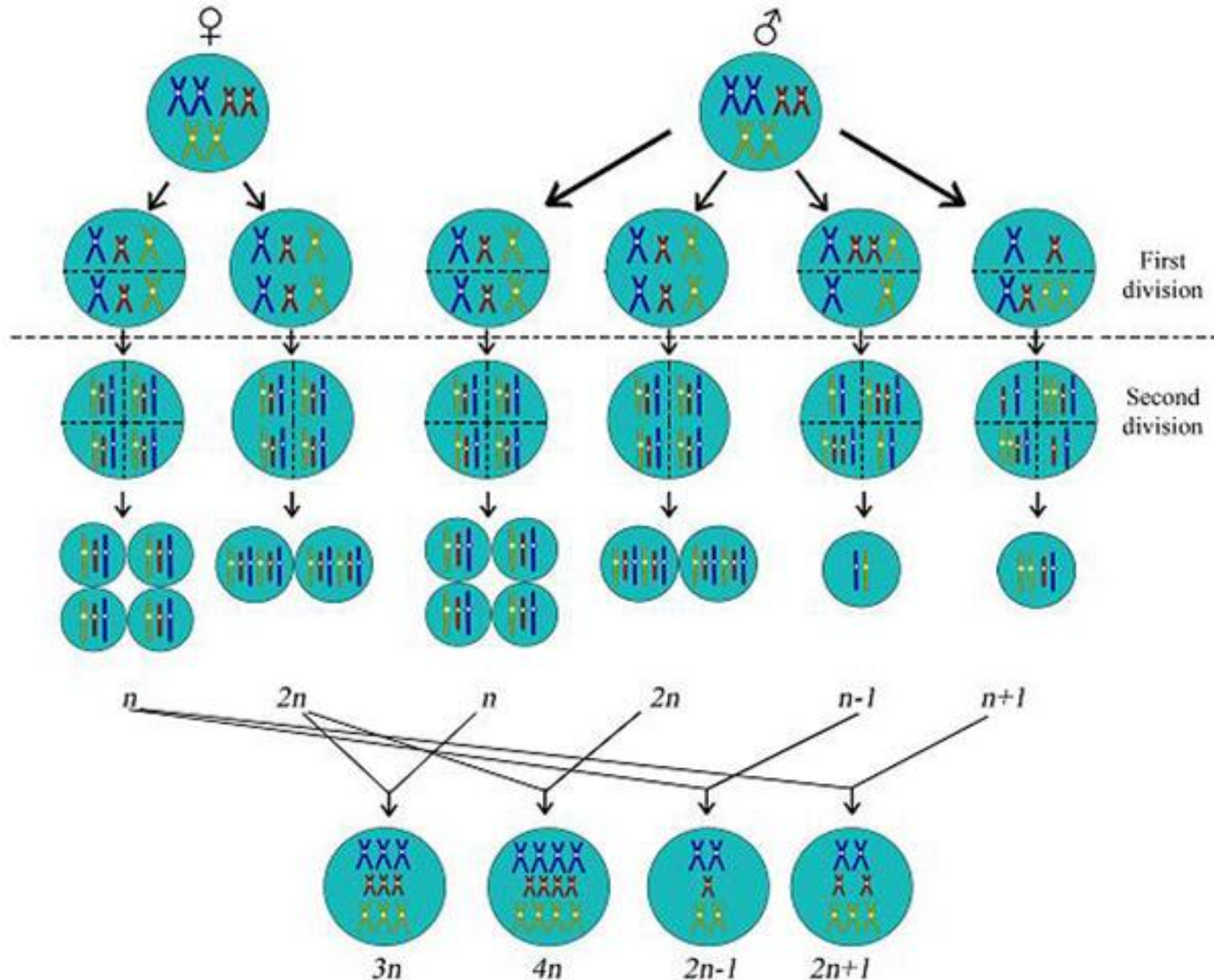


the diploid apple tree - *Malus* (Considine et al.)

- all tetraploid seedlings were derived from $2n$ ova fertilized with $2n$ spermatozoa
- all triploids from $2n$ 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*.

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



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

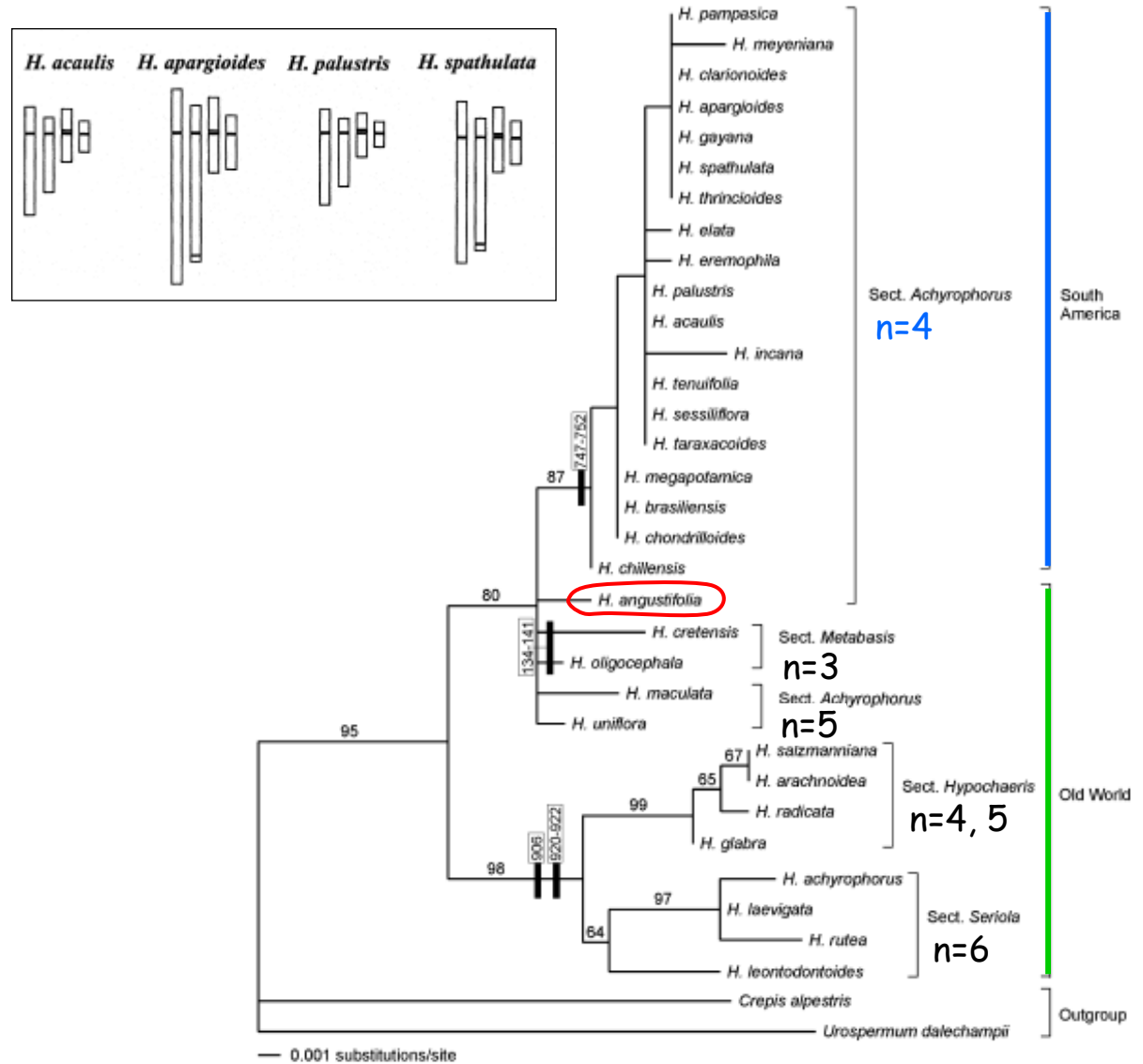
Three-step scenario to the odd basic chromosome number in *Malus*:
(aneuploidization - eupolyploidization - diploidization)

- ❖ 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.

Chromosome number evolution in phylogenetic contexts

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*)

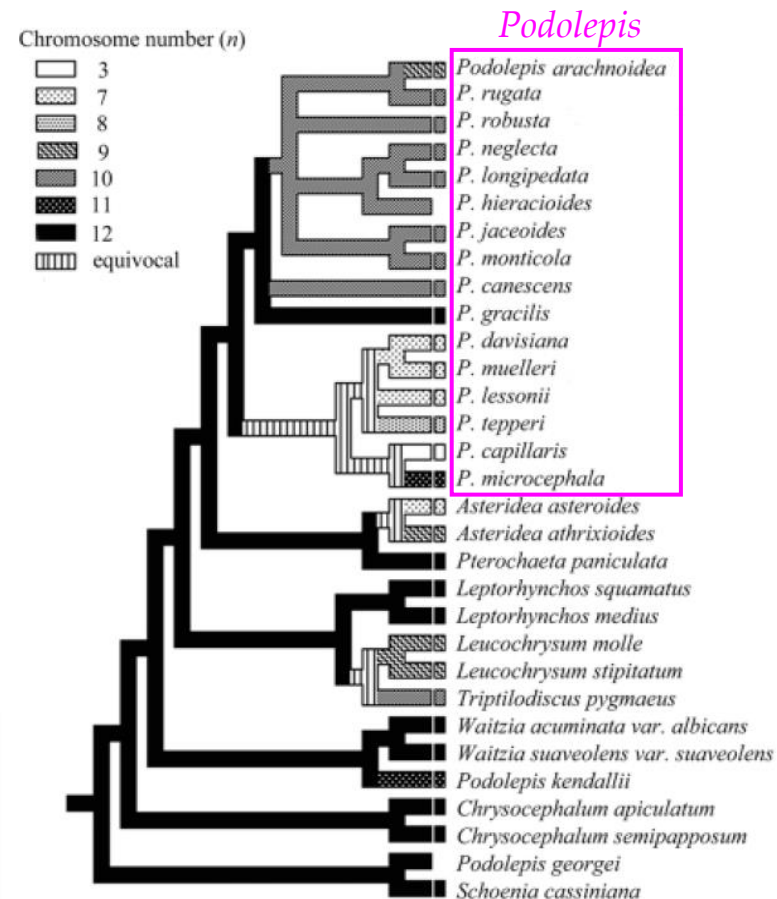
- according to the phylogenetic analysis, the ancestral chromosome base number in the genus *Podolepis* may be $x = 12$

- 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



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

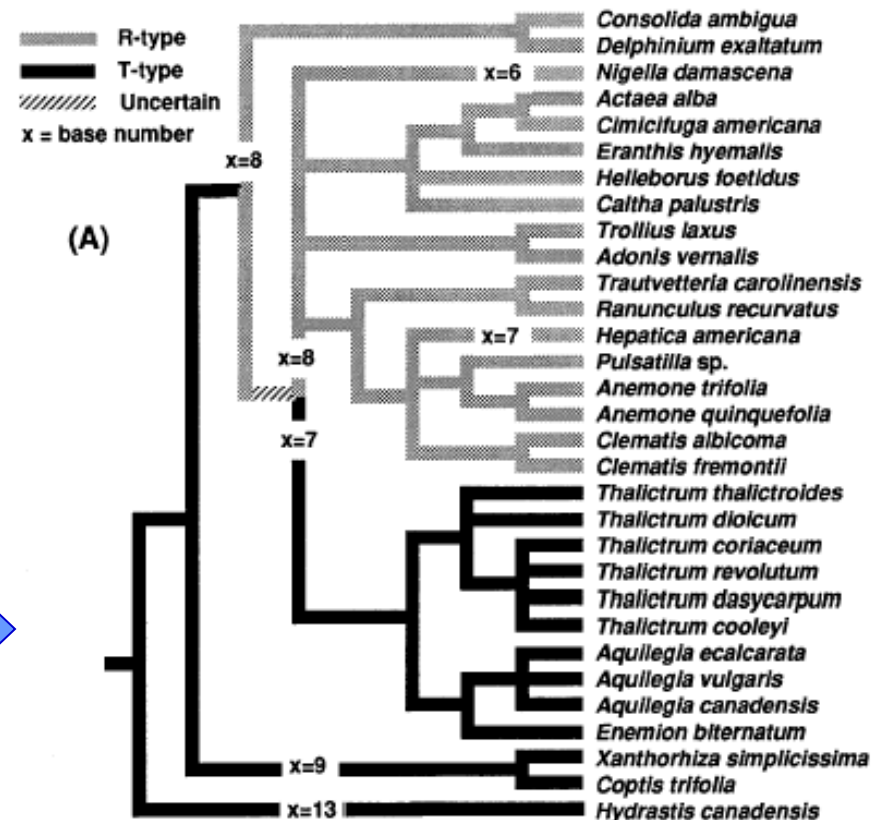
- the *Ranunculus* group of genera (R-chromosome group) has large and long chromosomes with a basic number of 8

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



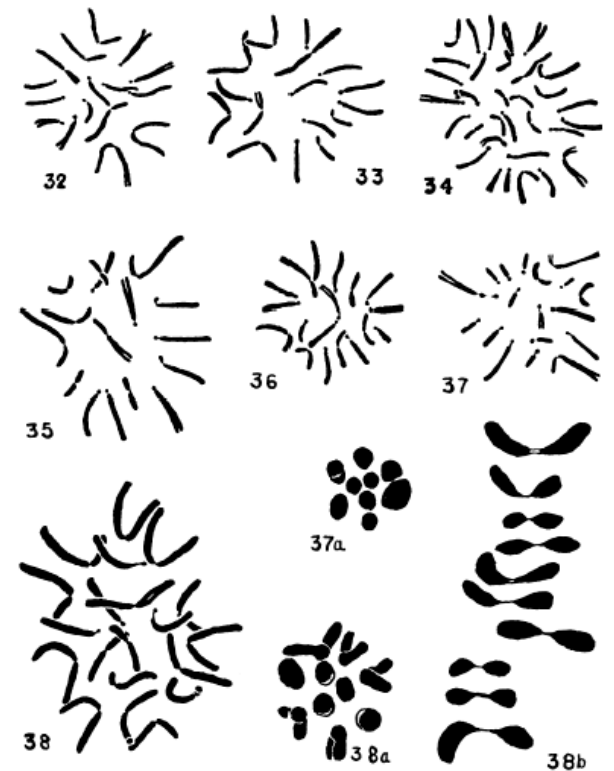
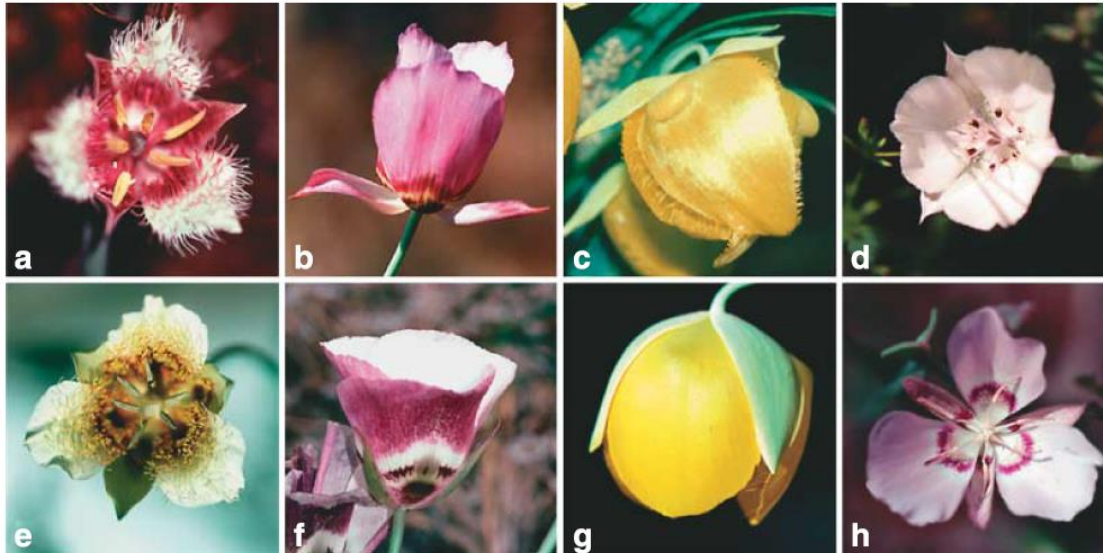
- Ro et al. (1997): chromosome type and base number are congruent with the inferred molecular (rDNA) phylogeny

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



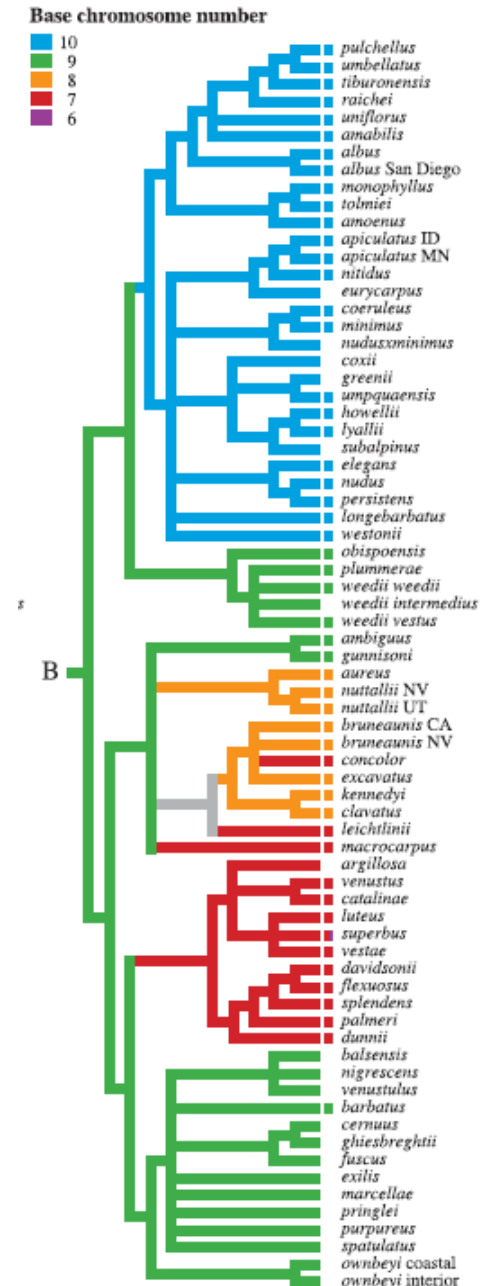
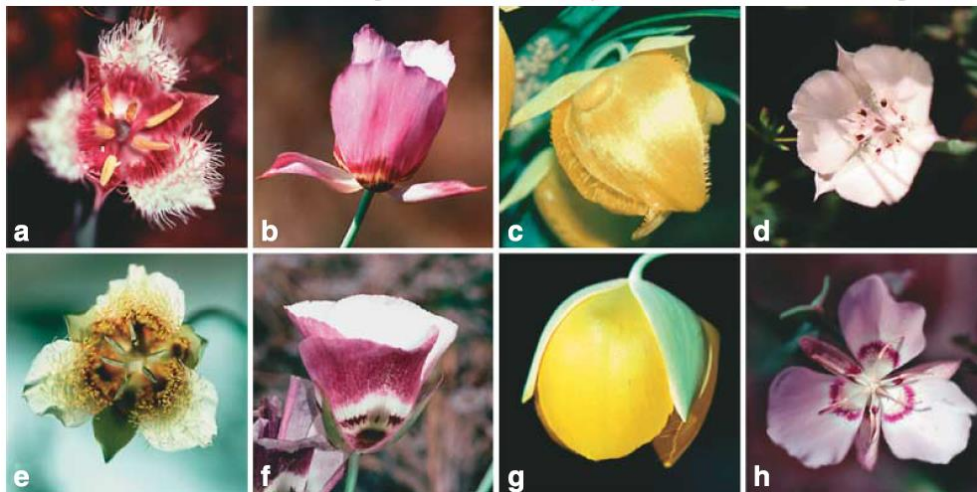
Descending and ascending (?) dysploidy in *Calochortus* (Liliaceae)

- 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



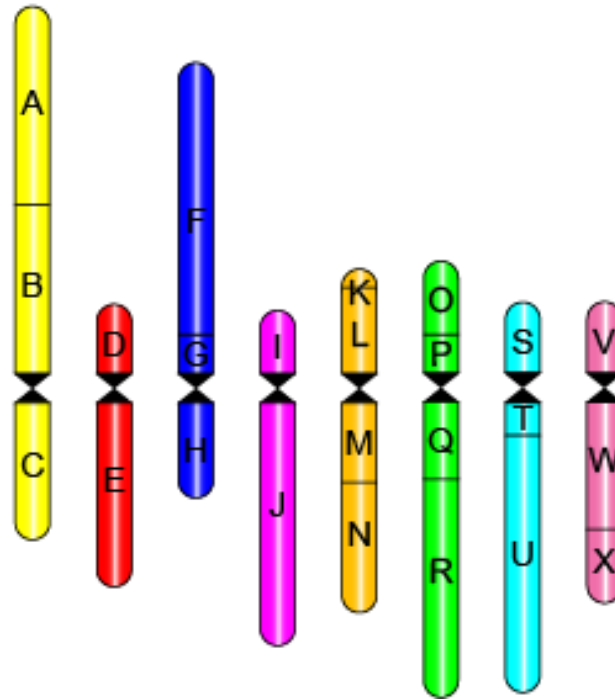
Descending and ascending (?) dysploidy in *Calochortus* (Liliaceae)

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

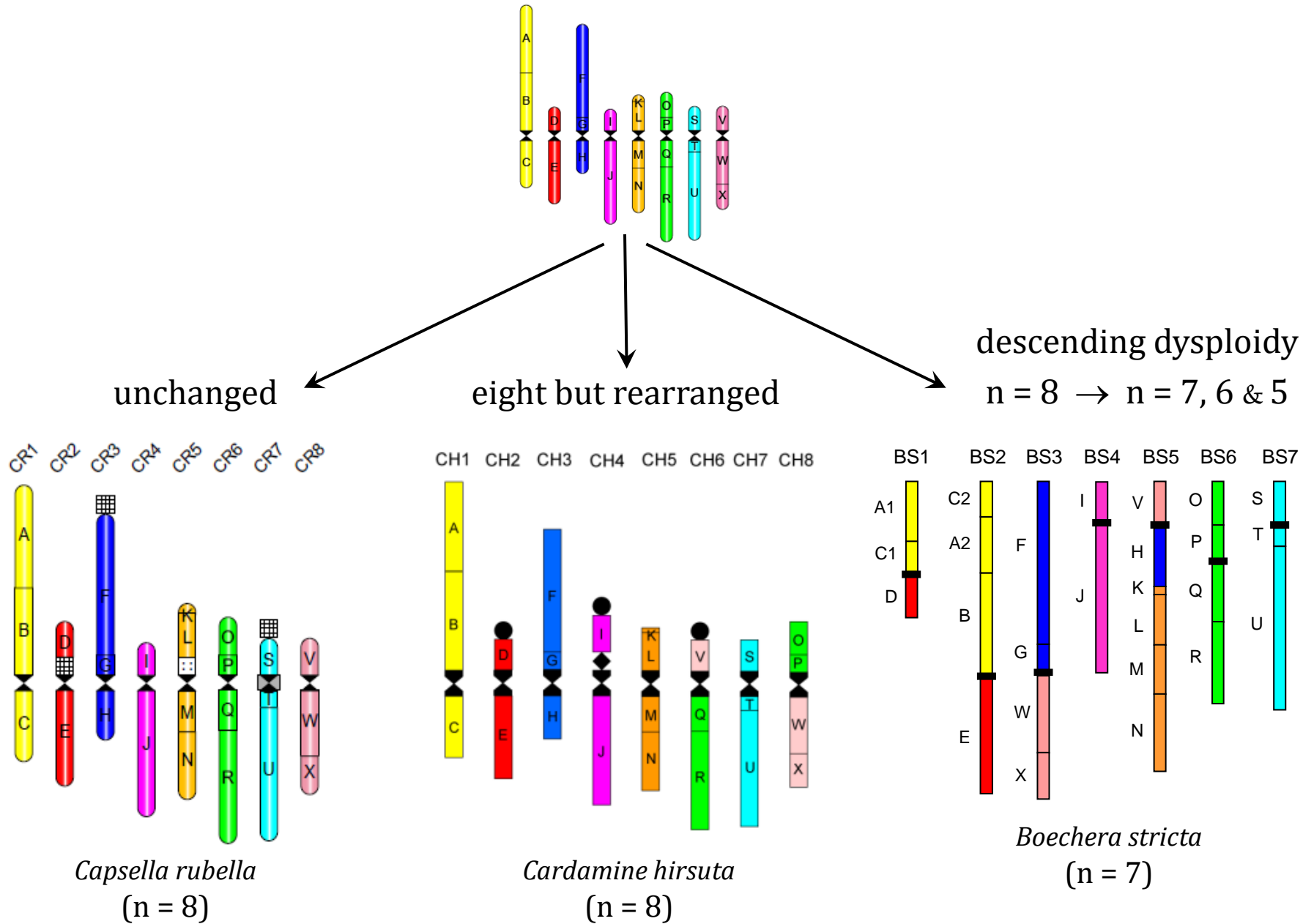


end

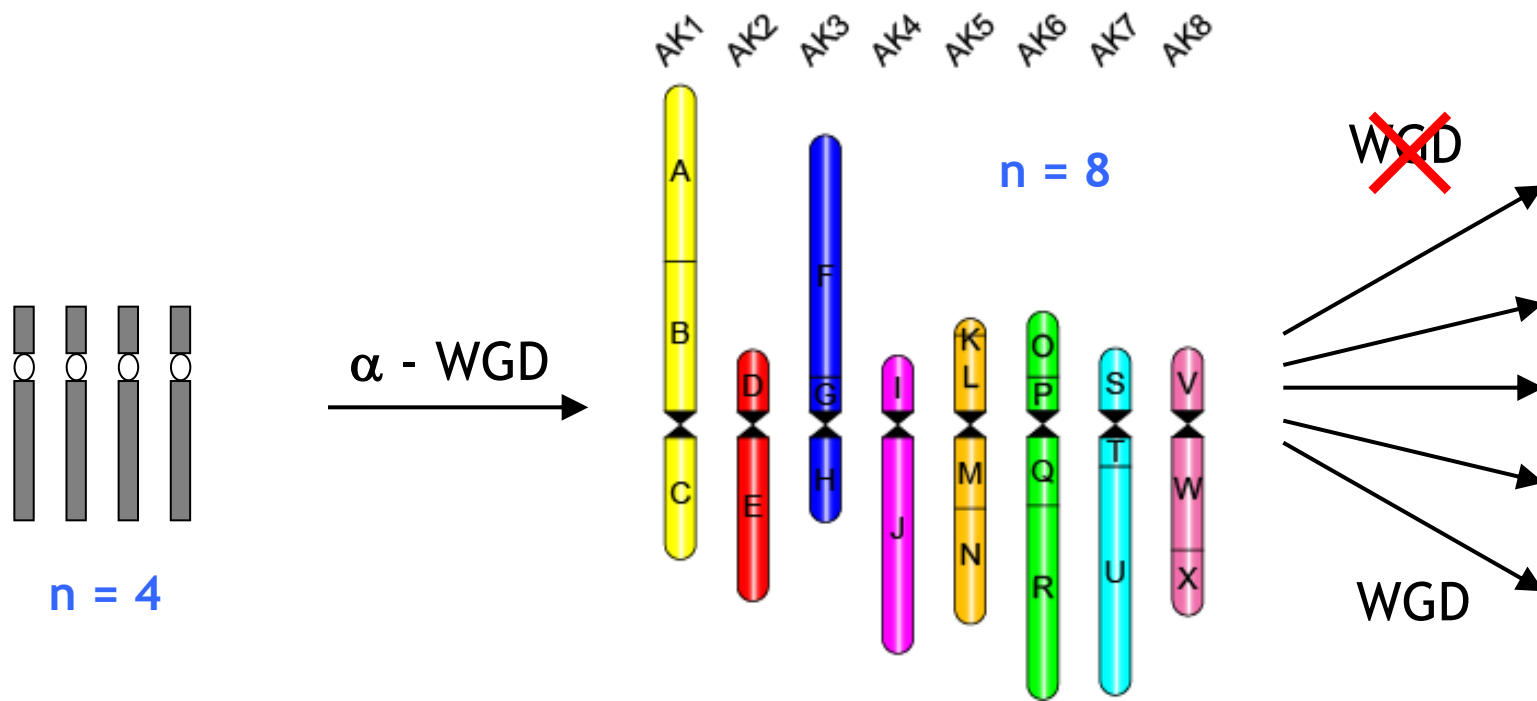
Crucifers (Brassicaceae): evolution of an ancestral genome



Evolution of the Ancestral Crucifer Genome – DIPLOIDS



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



Ancestral Crucifer Karyotype (ACK)

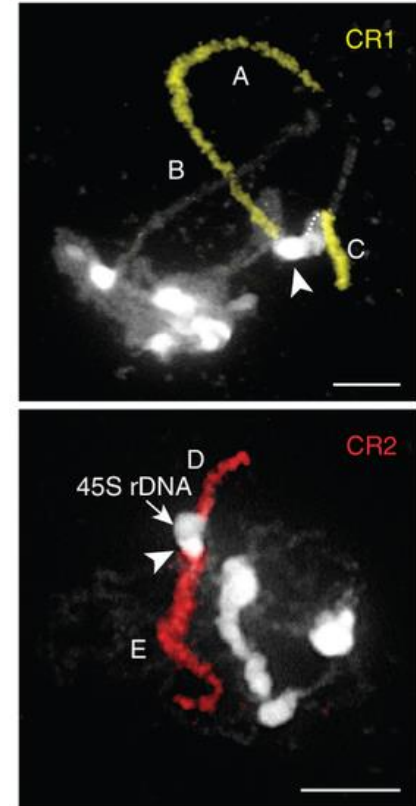
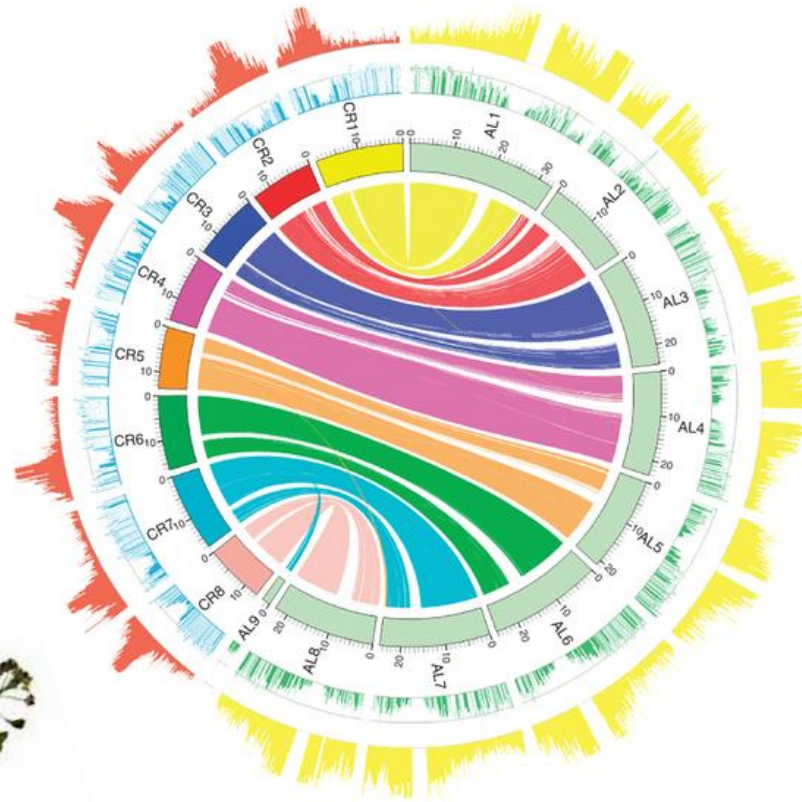
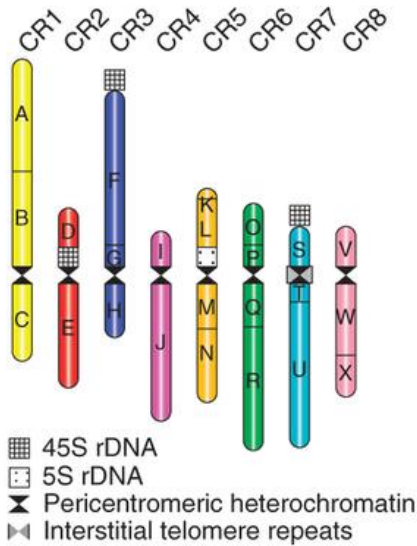
(24 ancestral genomic blocks)

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

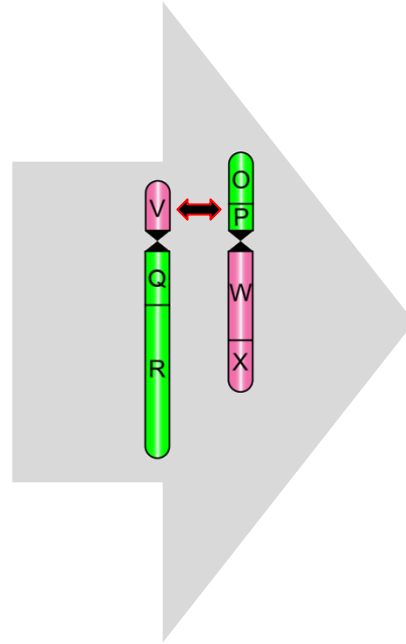
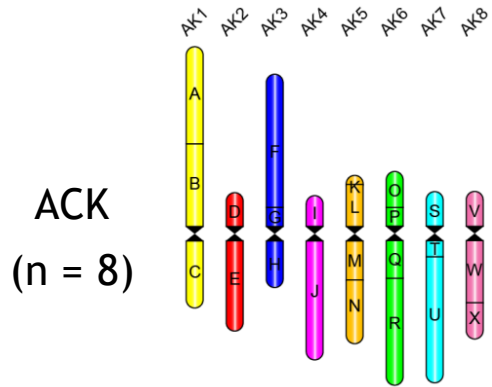
Capsella rubella
(n = 8)



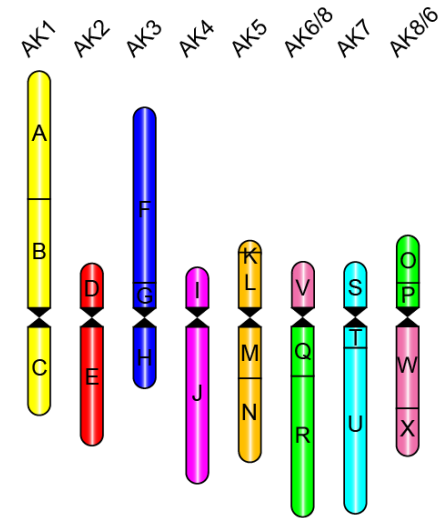
Capsella rubella vs. *Arabidopsis lyrata*

(both have ACK-like genome)

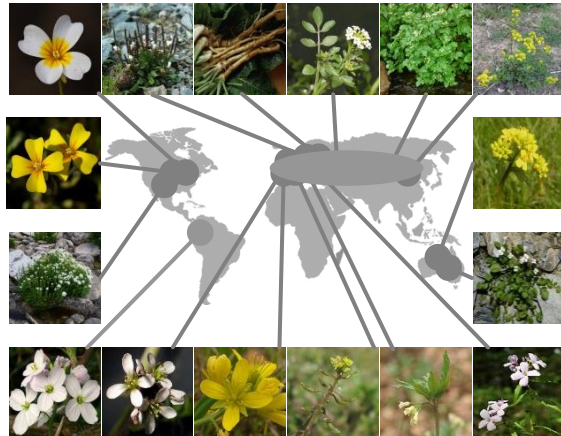
Diversification without large-scale chromosome rearrangements: karyotype stasis in the Cardamineae



ancestral karyotype of
Cardamineae (n = 8)

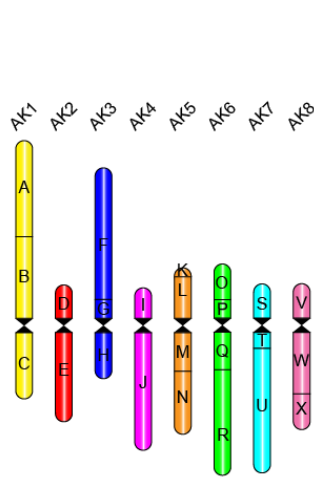


Cardamineae:
12 genera (337 spp.) worldwide



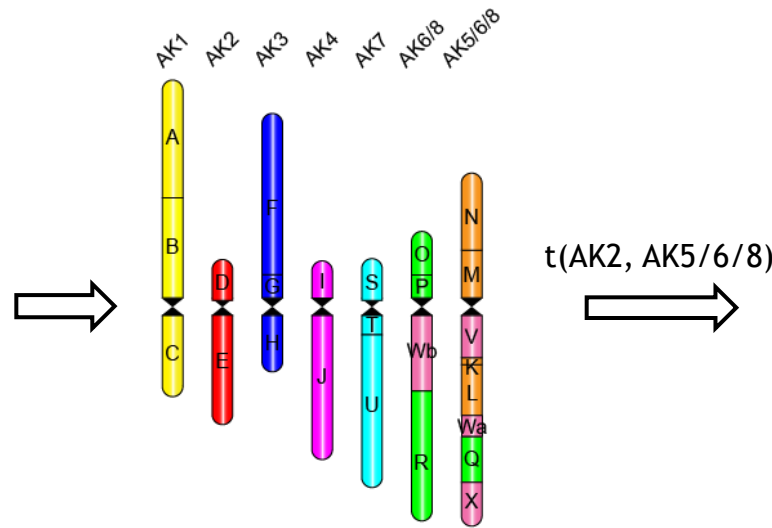
Mandáková and Lysak, in prep.

Descending dysploidy and genome stasis across Lineage II



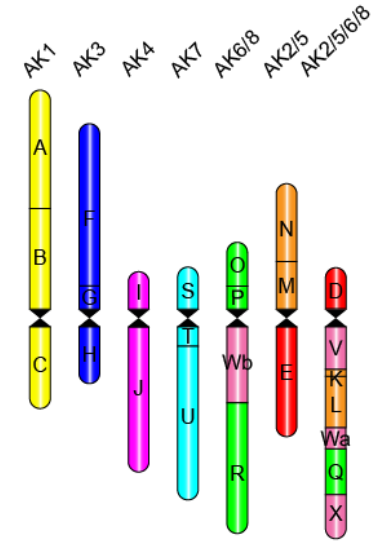
ACK (n = 8)

Ancestral Crucifer
Karyotype

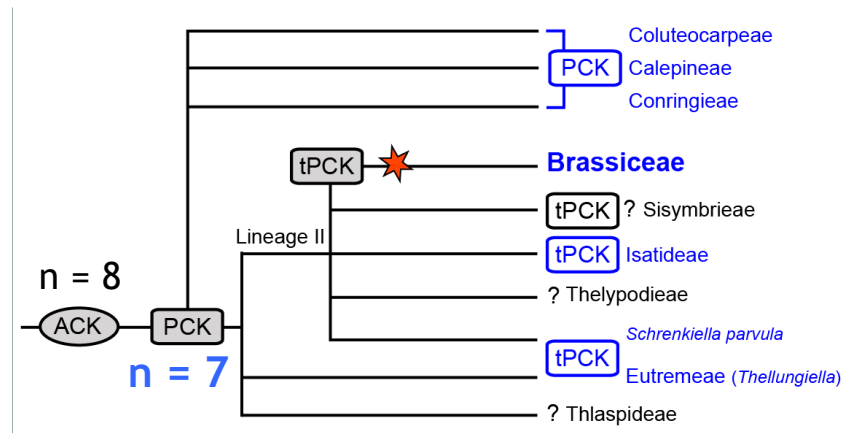


PCK (n = 7)

Proto-Calepineae Karyotype



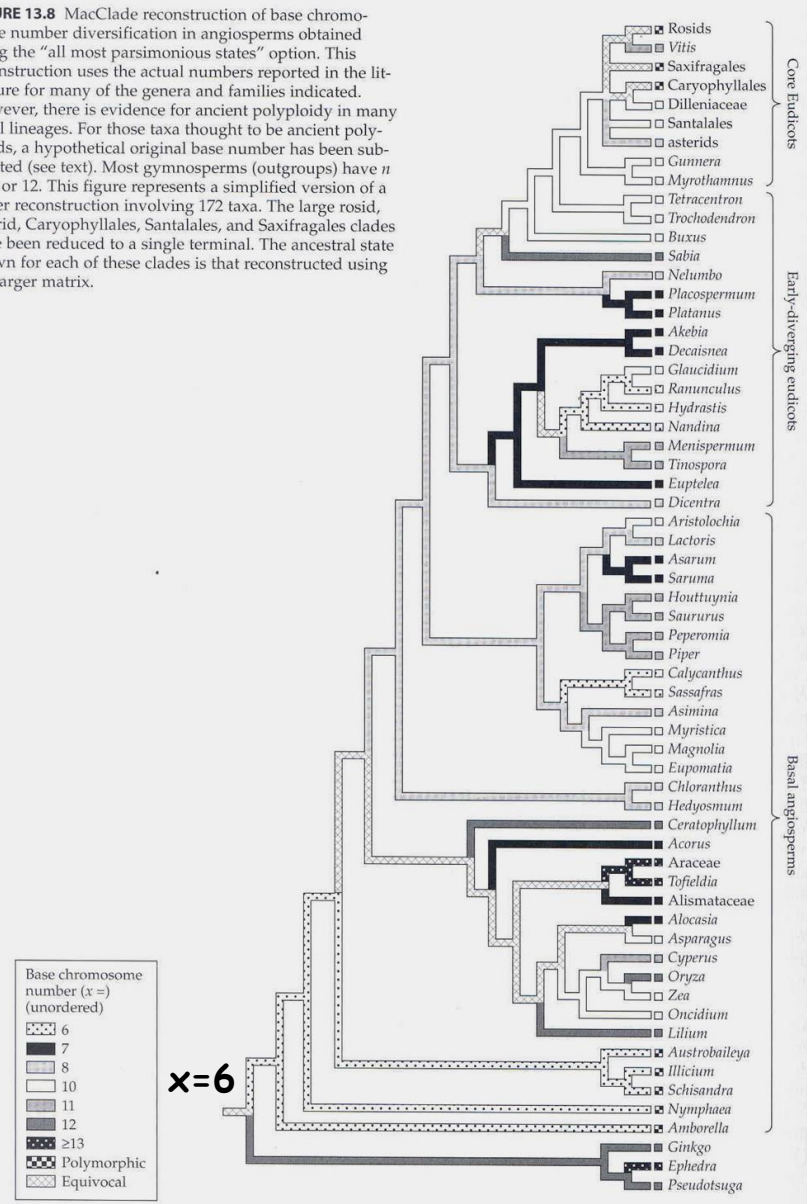
tPCK (n = 7)



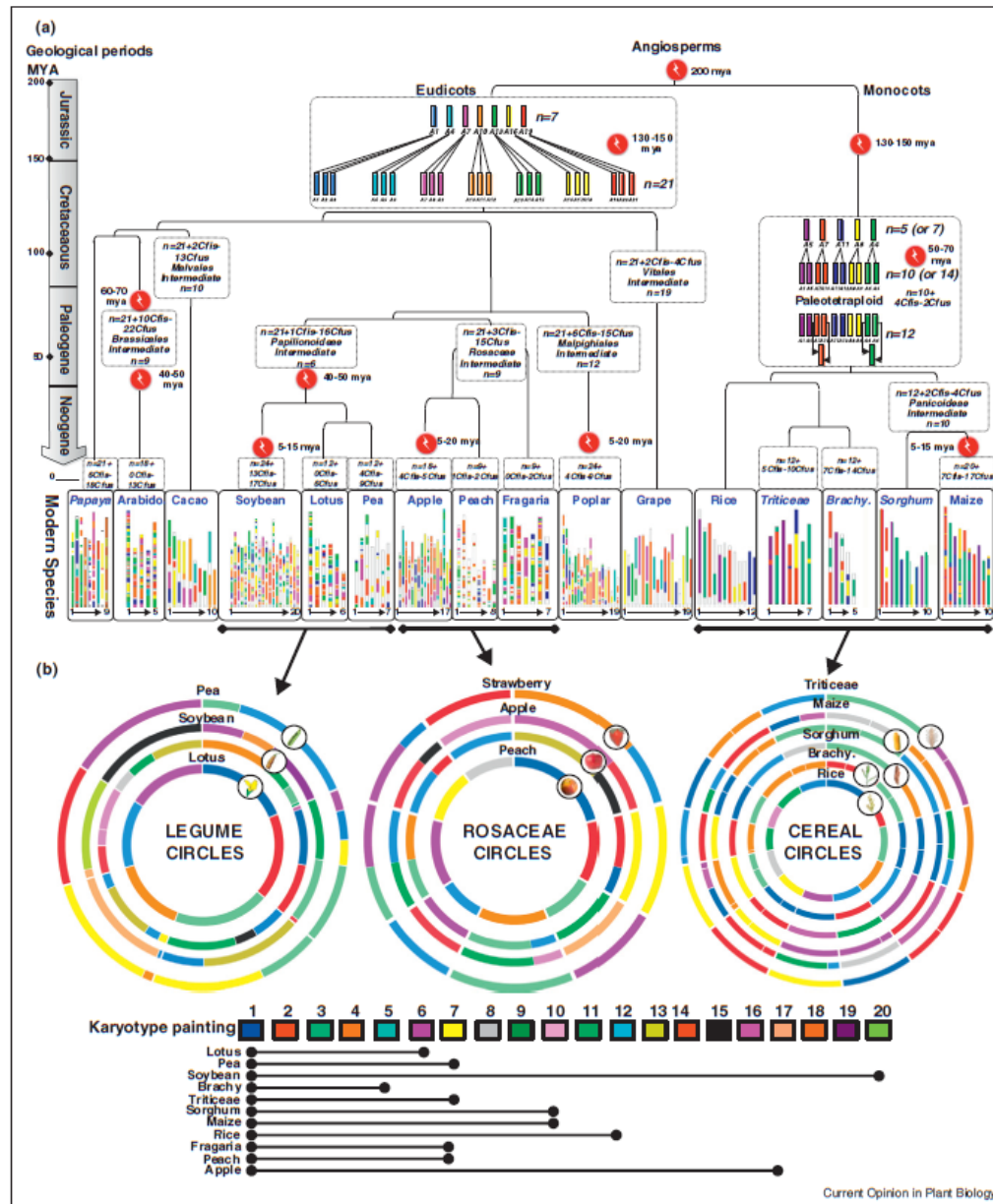
Reconstructing the ancestral base number for angiosperms

- 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

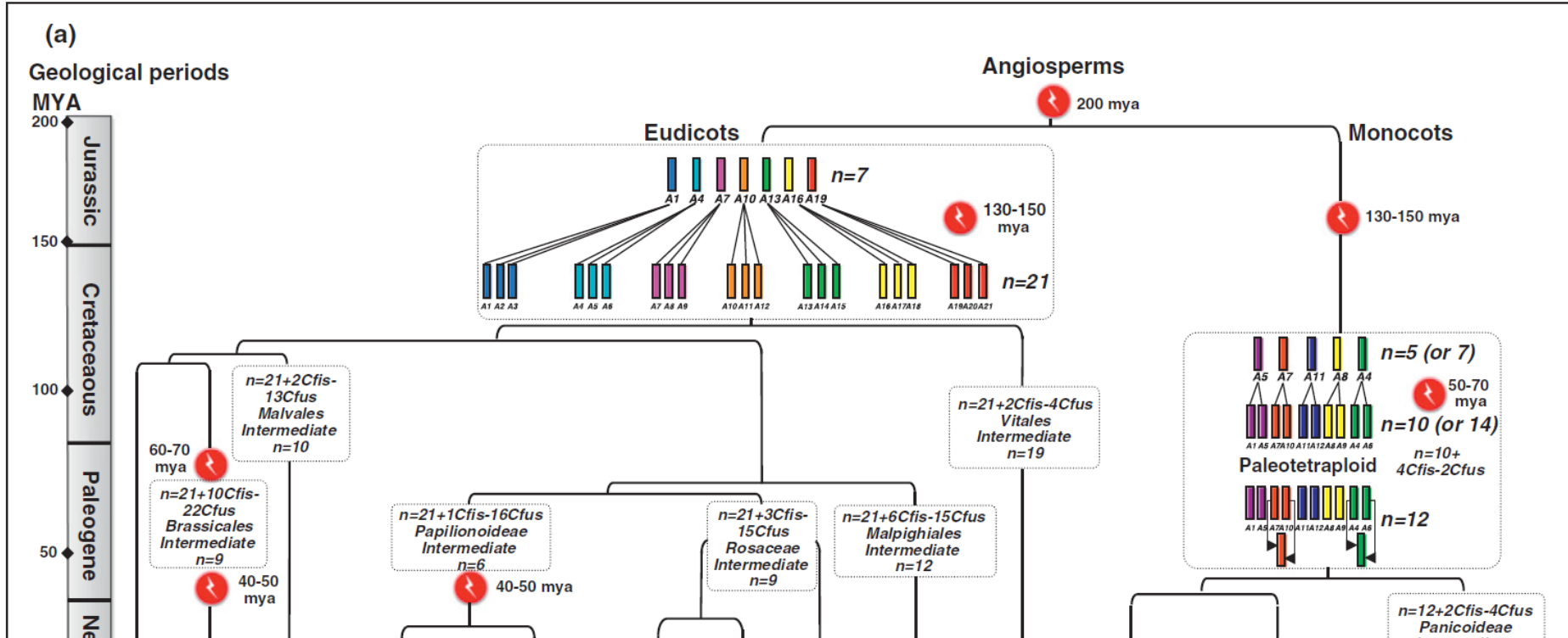
FIGURE 13.8 MacClade reconstruction of base chromosome number diversification in angiosperms obtained using the “all most parsimonious states” option. This reconstruction uses the actual numbers reported in the literature for many of the genera and families indicated. However, there is evidence for ancient 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.



Reconstructing the ancestral chromosome number and paleogenomes

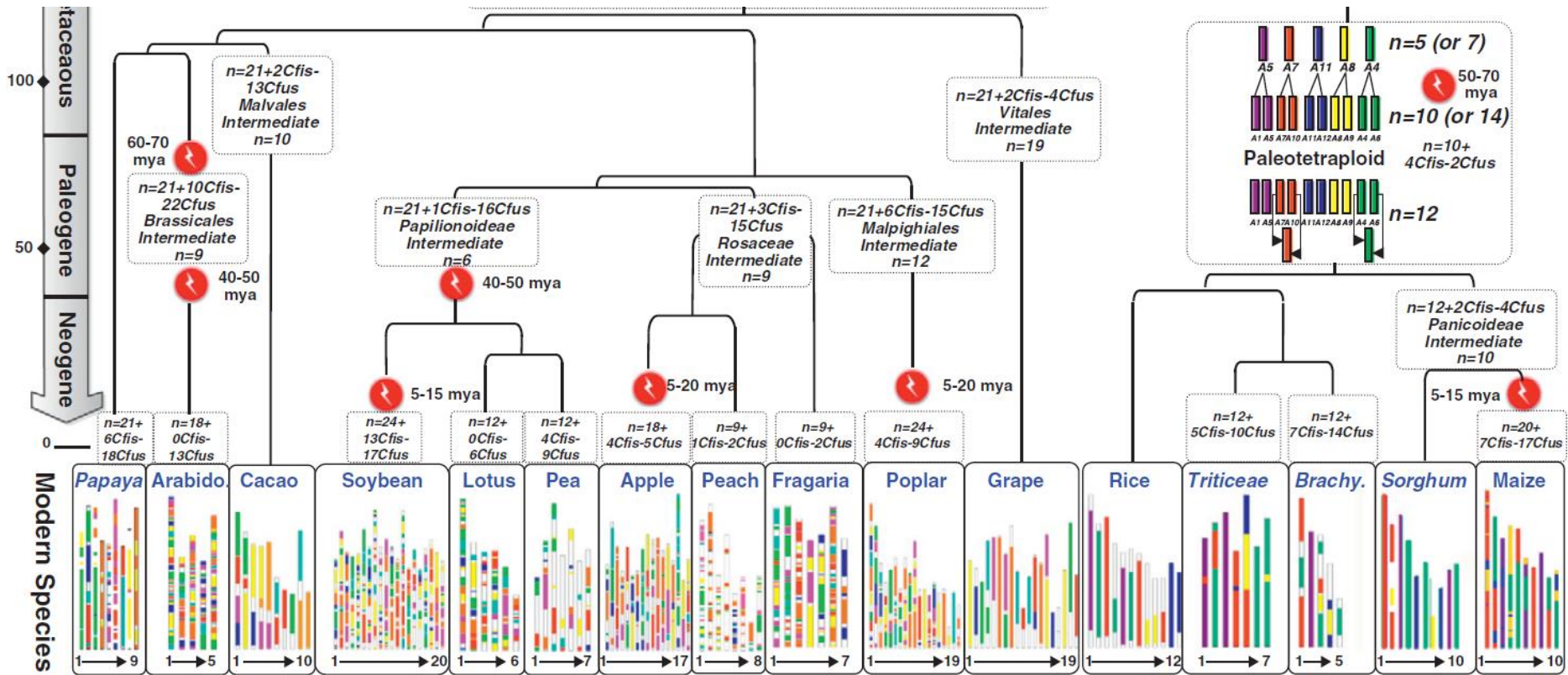


Reconstructing the ancestral chromosome number and paleogenomes



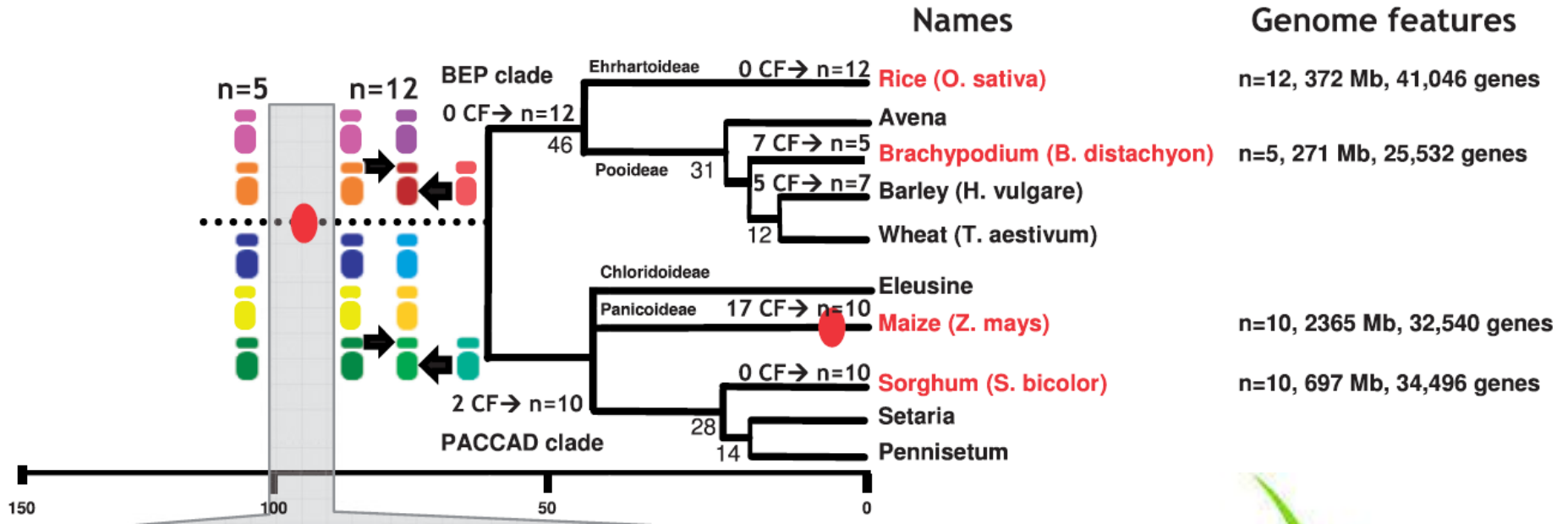
whole-genome duplication

Reconstructing the ancestral chromosome number and paleogenomes



whole-genome duplication

Reconstructed genome evolution in grasses



CF = „chromosome fusion“

● = whole-genome duplication (polyploidy)



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

