# The role of chromosome rearrangements in reproductive isolation and speciation

## (chromosomal speciation, particularly in plants)

*Coghlan et al. (2005)*

Are chromosomal rearrangements merely a problem for the genome, or do they have functional significance in the short term (e.g. by enabling a species to adapt to changing environmental conditions) or in the long term (e.g. by facilitating speciation)?

# Problems

Both morphologically distinct species that lack chromosomal differences (e.g. translocations and inversions) and morphologically cryptic species with chromosomal differences can be found

⇩

The amount of phenotypic evolution is not a good predictor of the amount of karyotypic evolution

⇩

YES!

M.J.D. White

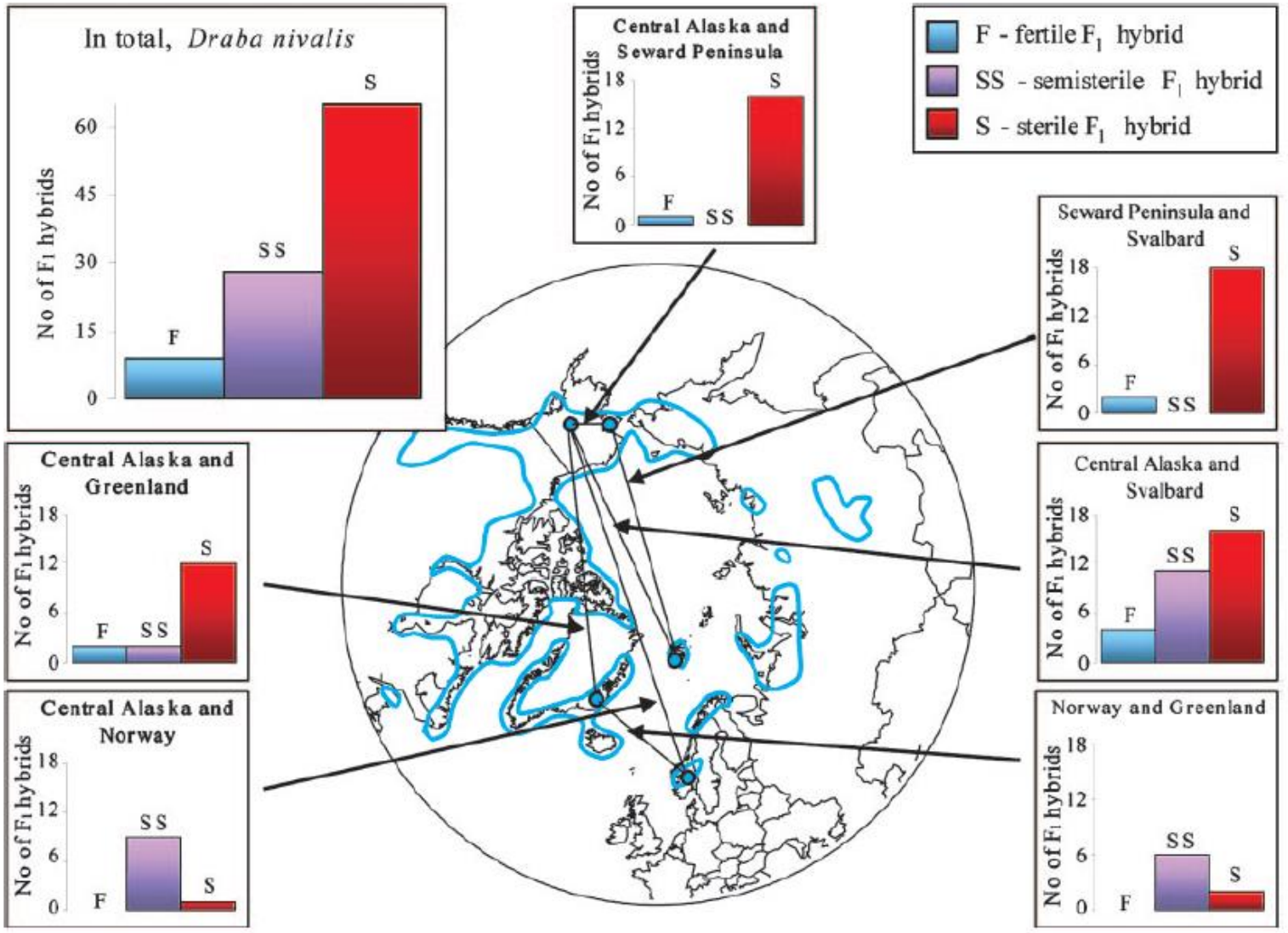
Are chromosome rearrangements important for creating reproductive isolation barriers and speciation?

Or is the accumulation of chromosomal differences between populations largely incidental to speciation?

(e.g., = speciation → chromosome rearrangements)

# Cryptic species with population-specific chromosome rearrangements?

*Draba nivalis*




Grund et al. 2006, PNAS 103

# Cryptic species with population-specific chromosome rearrangements?

*Draba nivalis*

Grund et al. 2006, PNAS 103:

Although 99% of parental individuals were fully fertile, the fertility of intraspecific crosses was surprisingly low. Hybrids from crosses within populations were mostly fertile (63%), but only 8% of the hybrids from crosses within and among geographic regions (Alaska, Greenland, Svalbard, and Norway) were fertile.

The frequent occurrence of intraspecific crossing barriers is not accompanied by significant morphological or ecological differentiation, indicating that numerous cryptic biological species have arisen within each taxonomic species despite their recent (Pleistocene) origin.

See also:
Gustafsson ALS, Skrede I, Rowe HC, Gussarova G, Borgen L, et al. (2014) Genetics of Cryptic Speciation within an Arctic Mustard, Draba nivalis. PLoS ONE 9(4): e93834. doi:10.1371/journal.pone.0093834

# Models of chromosomal speciation (Rieseberg 2001)

- Chain or Cascade models
- Chromosomal transilience model
- Monobrachial fusion model
- Recombinational model
- Quantum speciation model
- Stasipatric model
- Saltational model

**References**


White, M.J.D. (1978) *Modes of Speciation.*

Templeton, A.R. (1981) Mechanisms of speciation – a population genetic approach. *Annu. Rev. Ecol. Syst.* 12, 23–48.

Baker, R.J. and Bickham J.W. (1986) Speciation by monobrachial centric fusions. *Proc. Natl. Acad. Sci. U. S. A.* 83, 8245–8248.

Grant, V. (1981) *Plant Speciation*.

Lewis, H. (1966) Speciation in flowering plants. *Science* 152, 167–172.

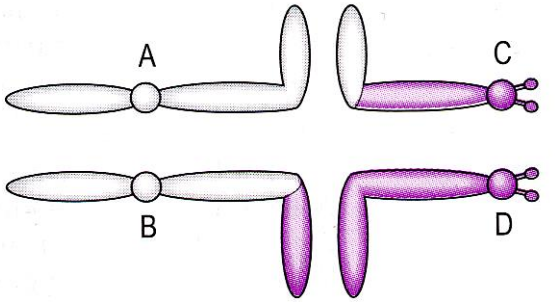

Fundamental feature of the models: **chromosomal differences that have accumulated between the neospecies and its progenitor(s) are assumed to impair the fertility or viability of interspecific hybrids, thereby reducing gene flow**

Deviating features of the models:

- geographical isolation is (not) required for speciation
- the means by which chromosomal rearrangements arise and become fixed
- effects of rearrangements on the fitness of chromosomally heterozygous individuals

# Chromosomal speciation: problems

- newly arisen chromosomal rearrangements will exist in the population almost exclusively as heterozygotes (inversion or translocation heterozygotes)



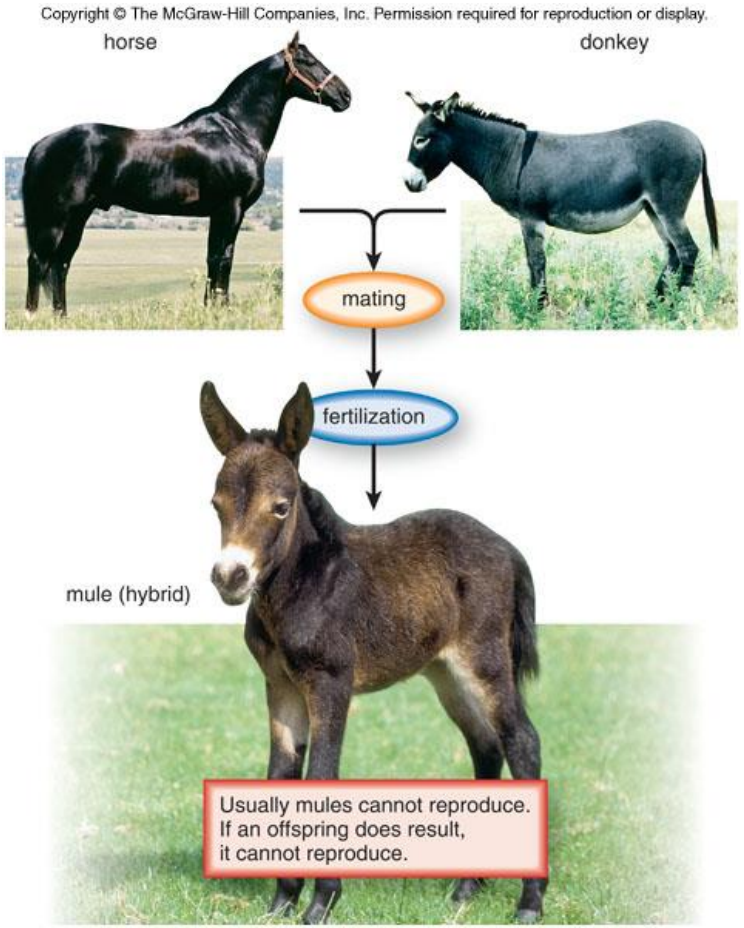

- many chromosomal rearrangements have little effect on fertility (ineffectiveness of chromosomal differences as barriers to gene flow)

- novel chromosomal arrangements have a selective disadvantage when they first appear in a population:
the problem of underdominance: difficulties associated with fixing chromosomal rearrangements that are strongly underdominant (i.e. reduce the fitness of heterozygotes)

# Chromosomal speciation

Two models:

- **the hybrid-sterility model**
- **recombination-suppression model**



# **Suppressed-recombination model** (Rieseberg 2001, Noor et al. 2001)

## Experimental data

### Drosophila

- inversions have contributed to speciation between the close relatives *D. pseudoobscura* and *D. persimilis*: inversions are found within the genomic regions associated with hybrid sterility

- chromosomal rearrangements reduce recombination between the genomes of the species, thereby enabling genetic differences to accumulate within the rearranged regions

- inversions are more common between *Drosophila* species that are sympatric compared to allopatric pairs (also true for butterflies)

### Plants

- the exact relationship between chromosomal rearrangement and speciation remains unclear in plants! ....but it is expected and probable

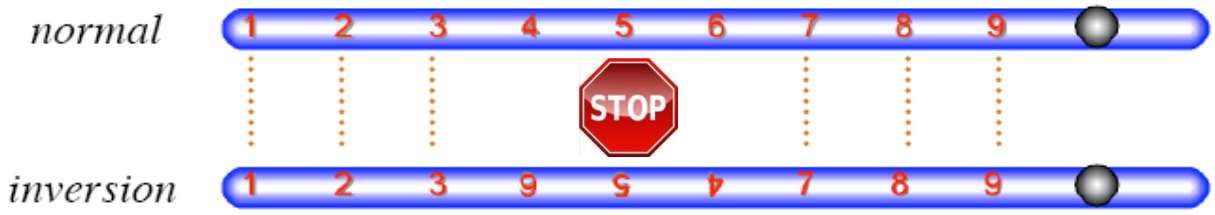
- sunflowers (*Helianthus*), *Mimulus*

# Suppressed-recombination model

(Rieseberg 2001, Noor *et al.* 2001, Faria and Navarro 2010)

**Chromosome rearrangements provide large regions of the genome protected from gene flow where isolating genes may accumulate until complete reproductive barriers exist.**

**The model suggests that rearrangements may reduce gene flow by suppressing recombination. CRs allow genes located in these regions to differentiate, in contrast to genes in freely recombining collinear regions.**



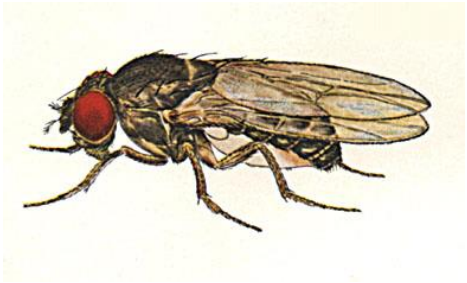
# Chromosome speciation in *Drosophila*

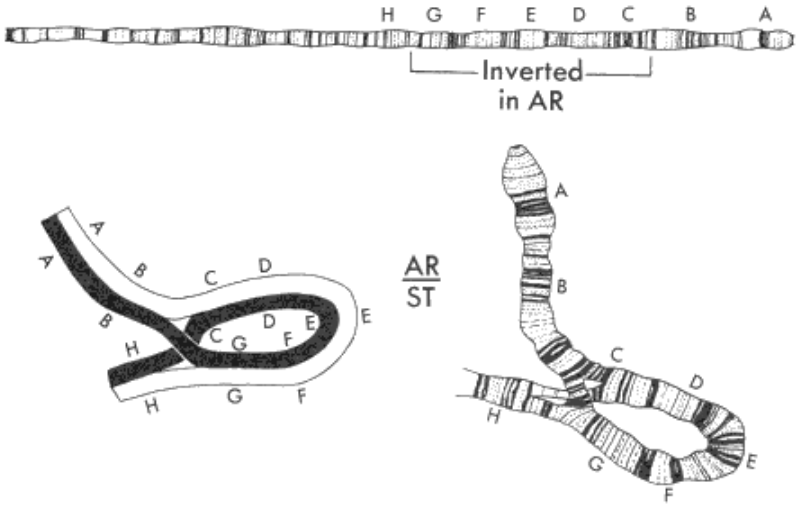
• gross chromosomal rearrangements in *Drosophila* are well characterized as rearrangements are easily detected in the chromosomes of their giant salivary glands

• the most common type of gross chromosomal rearrangement are paracentric inversions (do not span the centromere)

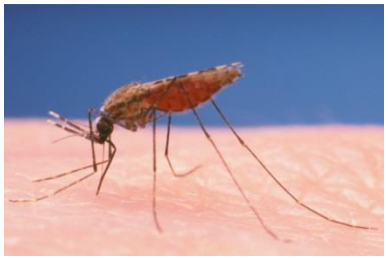
• paracentric inversions are common polymorphisms in drosophilas and other fly species (different populations of *D. melanogaster* harbor more than 500 inversion polymorphisms)

A paracentric inversion in *D. pseudoobscura*

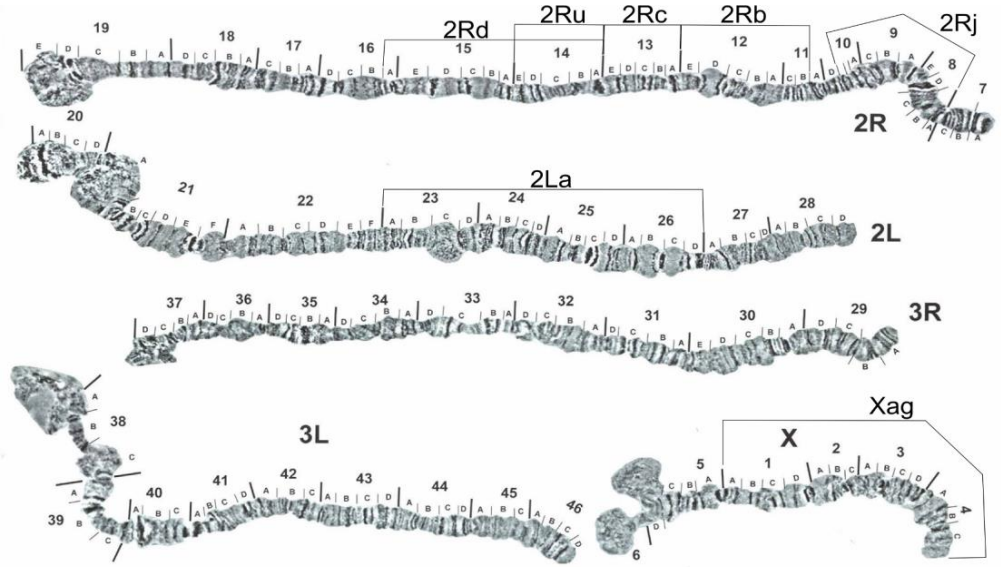


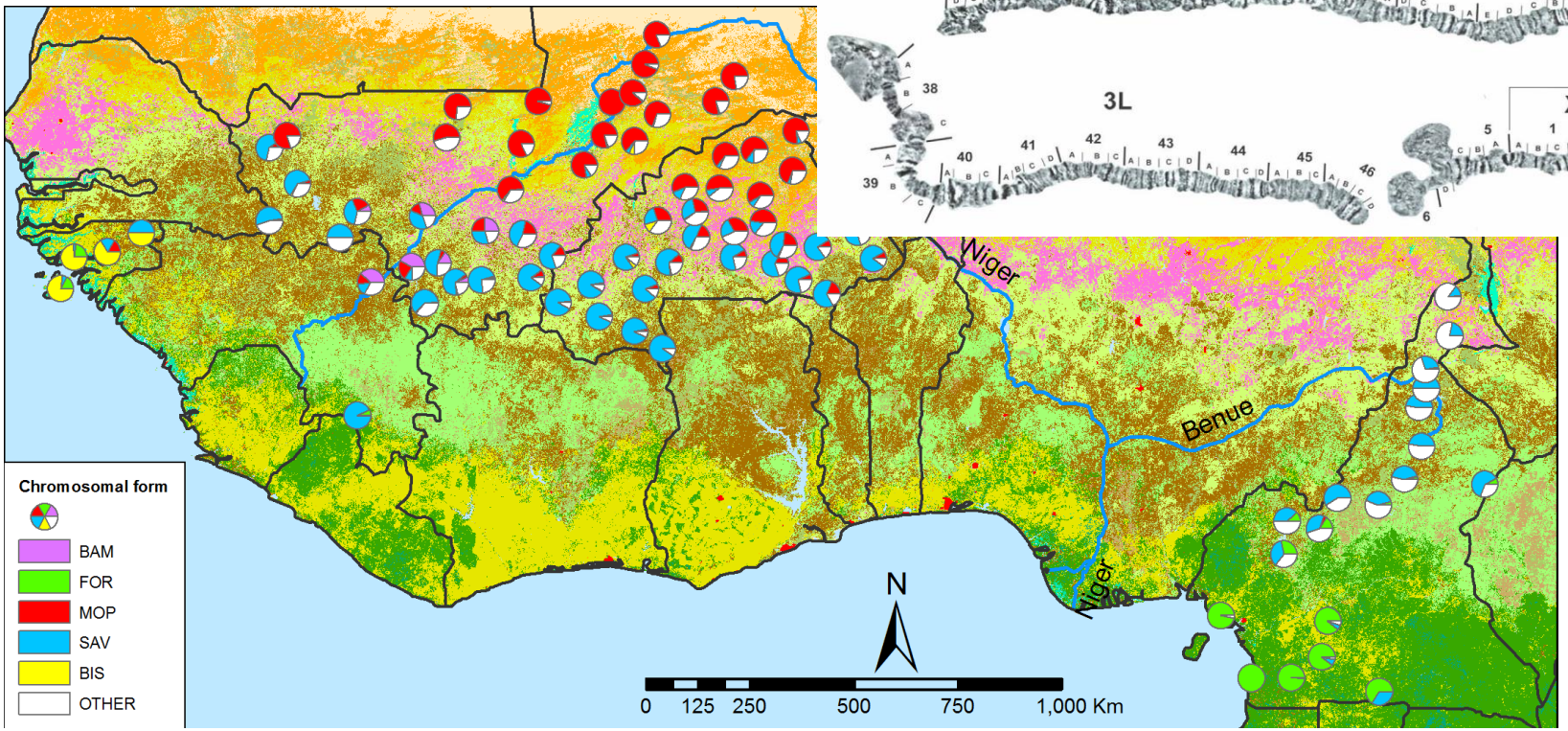


# Inversions are crossover suppresors – evolutionary consequences (speciation)

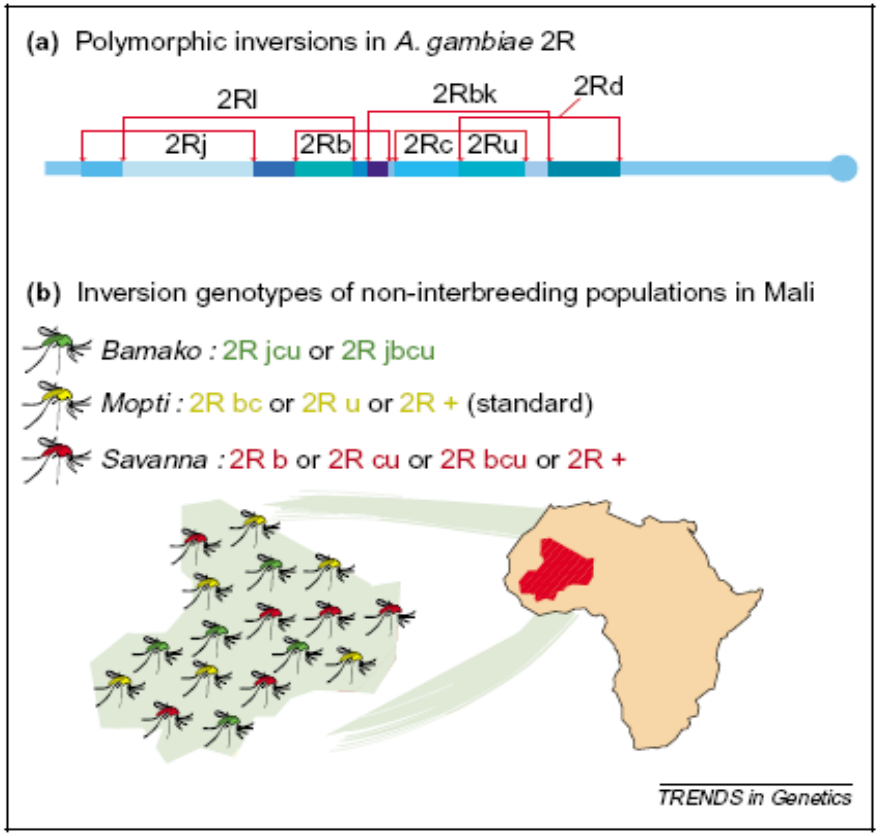


*Anopheles gambiae*



# Do chromosomal rearrangements contribute to speciation in *Anopheles gambiae*?

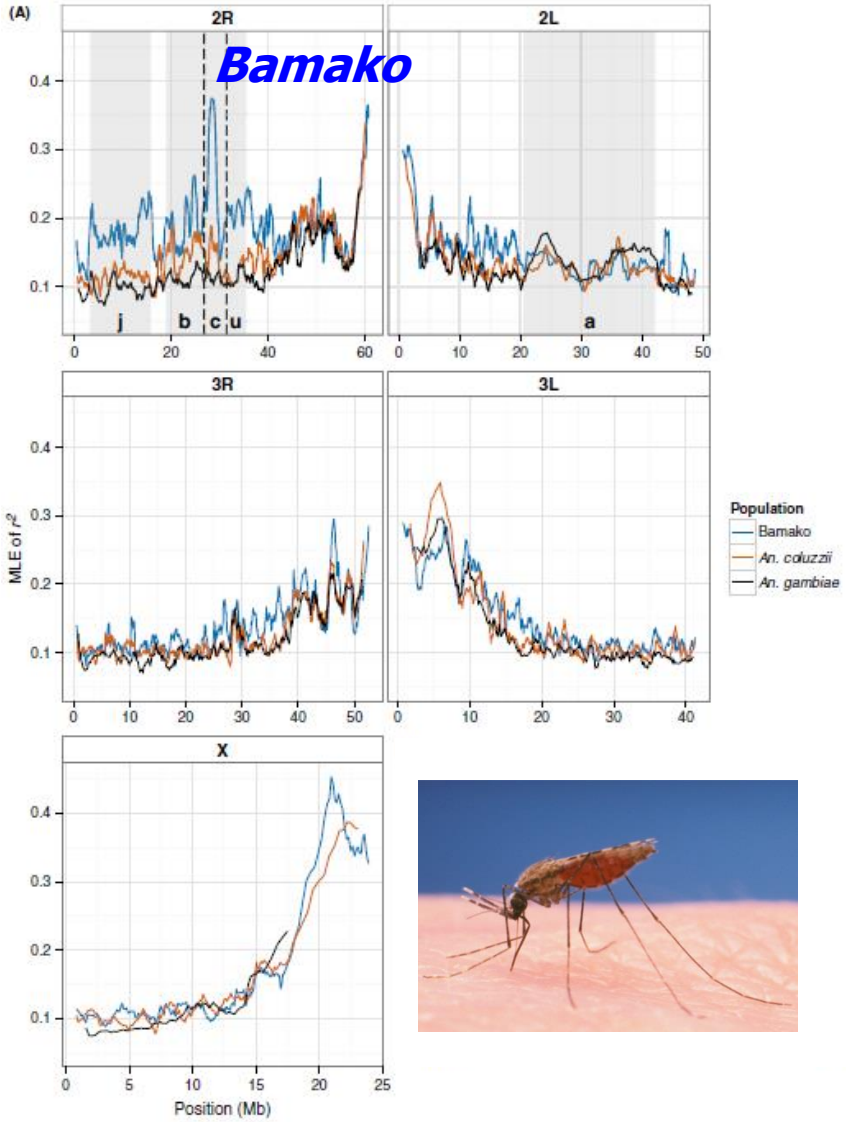



(a) Polymorphic paracentric inversions in *A. gambiae* chromosome arm 2R.

(b) Three non-interbreeding populations of *A. gambiae* (named *Bamako*, *Savanna* and *Mopti*) that live in the same region of Mali. The 3 populations differ by chromosomal inversions that might be contributing to speciation in *A. gambiae*. (For example, a chromosome with arrangement 2R jcu has inversions j, c and u on chrosmosome arm 2R.)

The role of chromosomal rearrangements in speciation in the *A. gambiae* species complex is difficult to prove: even a highly significant coincidence in time between chromosomal rearrangements and speciation does not prove a causal relationship.

# Chromosomal inversions and ecotypic differentiation in *Anopheles gambiae*: the perspective from whole-genome sequencing






- initial genomic and ecological differentiation – sympatric speciation
- the majority of differentiated regions between Bamako and typical *An. gambiae* are located inside inversions
- differentiated genomic regions were enriched for genes implicated in nervous system development and signalling

Love et al. (2016), Mol Ecol

# Suppressed-recombination model

## PLANTS

- the exact relationship between chromosomal rearrangement and speciation remains unclear in plants! ...but more evidence is emerging (new methods available)

- seems that sunflowers (*Helianthus*) are only example: hybridization between two divergent diploid species appears to have provoked speciation events in sunflowers (Loren Rieseberg's lab) ....and recently *Mimulus guttatus* (Lowry and Willis 2010)

HYBRID SUNFLOWER SPECIES, H. ANOMALUS
(PHOTO BY J. RICK)

**Hybrid**
**Homoploid**
**Chromosomal**
**Recombinational**

} **speciation in sunflowers (*Helianthus*)**

The rate of introgression is lower within rearranged chromosomes /chromosome regions (vs. collinear regions). The strongest difference close to the breakpoints - consistent with suppressed-recombination models (the strongest reduction in recombination).

**Hybrid**
**Homoploid**
**Chromosomal**
**Recombinational**

} **speciation in sunflowers (*Helianthus*)**

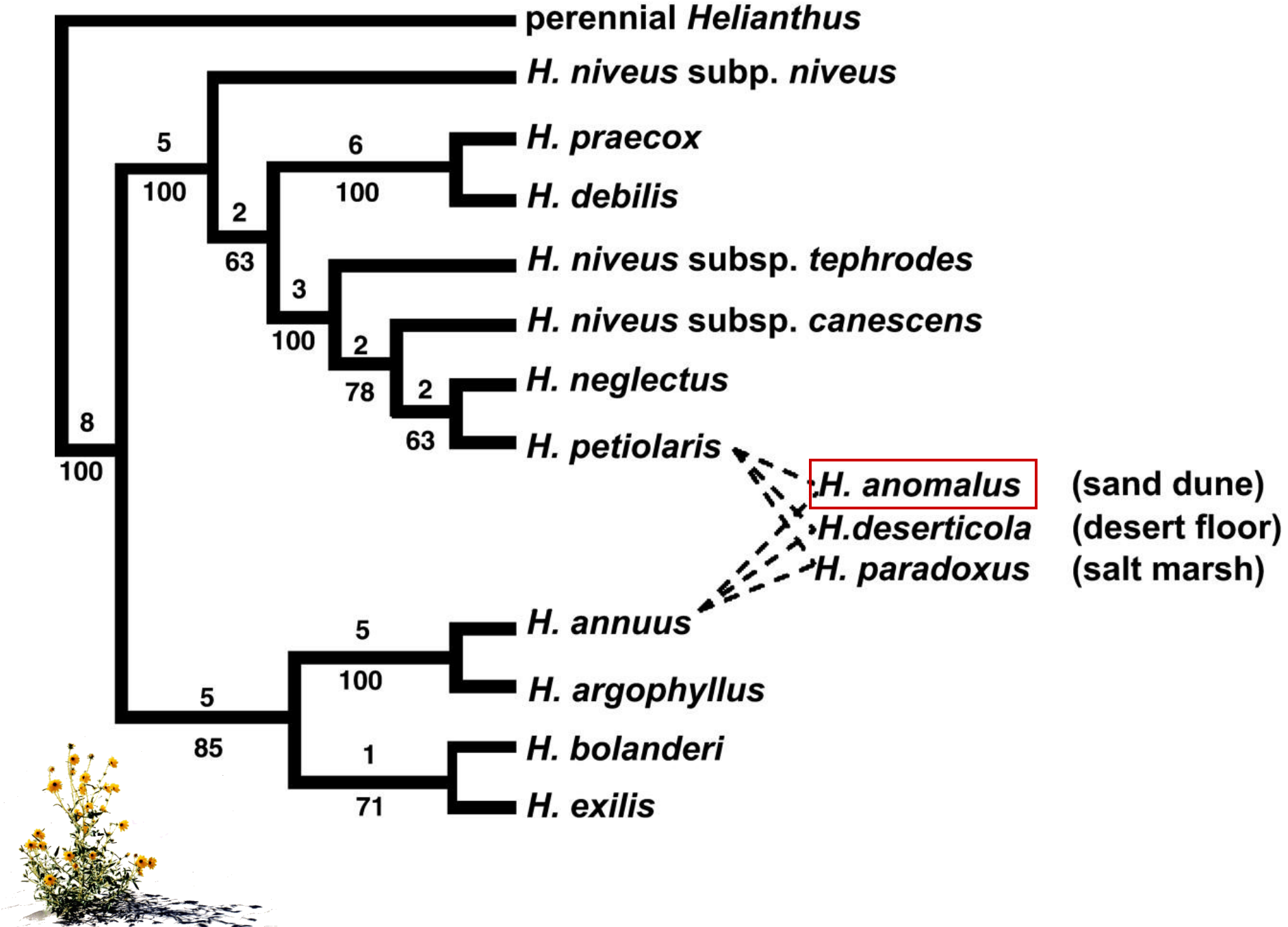
- hybrid or recombinational speciation refers to the origin of a new homoploid species *via* hybridization between chromosomally or genetically divergent parental species

- homoploid hybrid speciation is theoretically difficult because it requires the development of reproductive barriers in sympatry or parapatry (the possibility of backcrossing with their parental species)

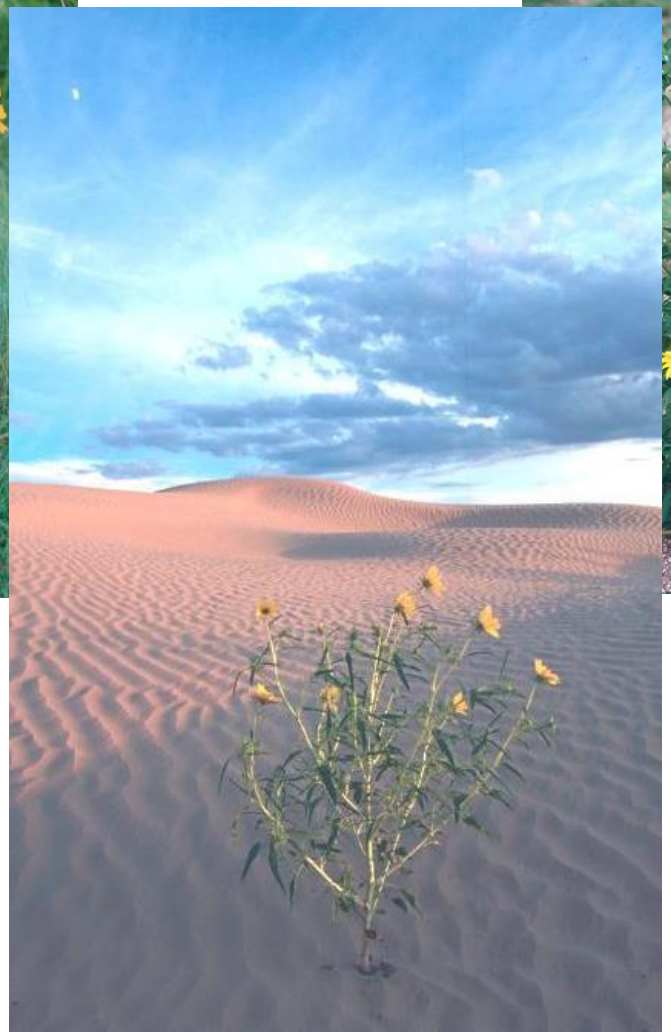
- theory suggests that isolation may arise through rapid karyotypic evolution and/or ecological and spatial divergence of hybrid neospecies

- it is assumed that new hybrid lineage diverge karyotypically from its parental species through the chromosomal rearrangements that differentiate the parental species and/or by new chromosomal rearrangements induced by recombination

- three *Helianthus* species are probably the best documented examples of homoploid hybrid speciation in either animals or plants


perennial Helianthus
H. niveus subp. niveus
H. praecox
H. debilis
H. niveus subsp. tephrodes
H. niveus subsp. canescens
H. neglectus
H. petiolaris
H. anomalus
H.deserticola
H. paradoxus
H. annuus
H. argophyllus
H. bolanderi
H. exilis
(sand dune)
(desert floor)
(salt marsh)
5
100
6
100
2
63
3
100
2
78
2
63
8
100
5
100
5
85
1
71

*Helianthus annuus*



*Helianthus petiolaris*

*Helianthus anomalus*

# Homoploid hybrid speciation: *H. anomalus*

• a sand dune endemic, central Utah and northern Arizona

• both parental species are widespread; hybridize but retain their genetic integrity because of the synergistic action of several reproductive barriers

• three experimentally generated hybrid lineages (*H. annuus* x *H. petiolaris*) showed a combination of chromosomal blocks similar to that found in *H. anomalus* (Rieseberg et al. 1996)

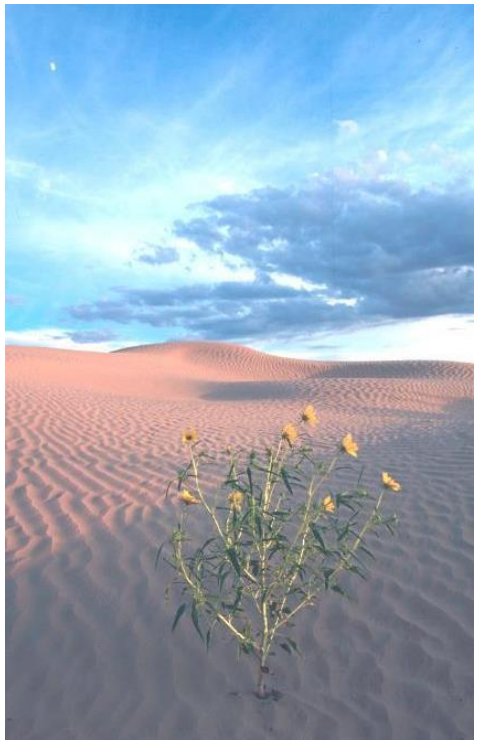
• the three synthetic lineages were cross-compatible with each other and with *H. anomalus* (Rieseberg 2000)

**• *H. anomalus* has diverged considerably from its parents in both karyotype and ecological preference due to the sorting of chromosomal rearrangements that differentiate the parental species. *H. anomalus* also possesses several unique arrangements, possibly induced by recombination. As a result, *H. anomalus* is almost completely intersterile with its parental species**

***H. annuus* x *H. petiolaris***

⇩

***H. anomalus***



# Homoploid hybrid speciation in sunflowers (*Helianthus*)

➢ it is assumed that new hybrid lineage diverge karyotypically from its parental species through the chromosomal rearrangements that differentiate the parental species and/or by new chromosomal rearrangements induced by recombination

***H. annuus* x *H. petiolaris***

⇩

*H. anomalus*

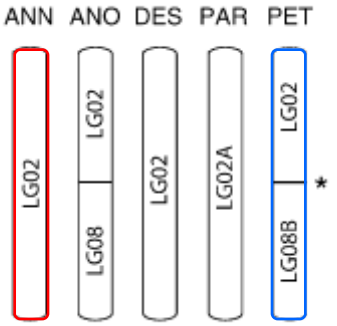
*H. deserticola*

*H. paradoxus*

• karyotypes of the three hybrid species are massively divergent from their parental species

• about one-third of the karyoypic differences arose through the sorting of parental chromosomal rearrangements

• the remainder of karyotypic differences appear to have arisen *de novo* (6 breakages/6 fusions in *H. anomalus*, 4 breakages/3 fusions in *H. deserticola*, and 5 breakages/5 fusions in *H. paradoxus*)




**• karyotypic differences contribute to reproductive isolation: 9 of 11 pollen viability QTLs occur on rearranged chromosomes and all but one map close to a rearrangement breakpoint**

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# A Widespread Chromosomal Inversion Polymorphism Contributes to a Major Life-History Transition, Local Adaptation, and Reproductive Isolation

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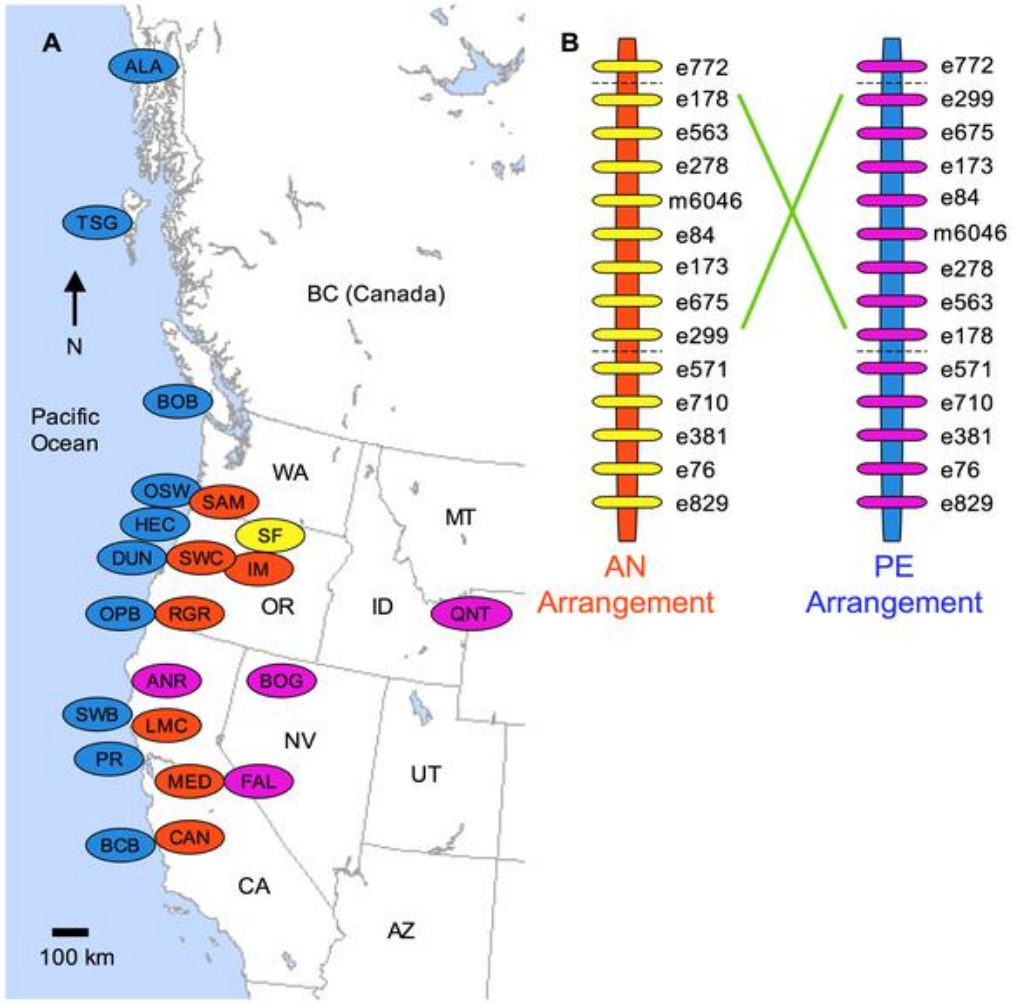
*yellow monkeyflower* (*Mimulus guttatus*)

perennial and annual plant

# *M. guttatus*: geographic distribution of the chromosomal inversion




**(A)** Map of western North America with the locations of populations of coastal perennials (blue), inland annuals (orange), and inland perennials (purple), as well as obligate self-fertilizing species *M. nasutus (yellow)*. **(B)** Marker order of the AN and PE inversion arrangements along linkage group eight. Inland annuals and *M. nasutus* had the AN arrangement while coastal and inland perennials all had the PE arrangement.

# Inversion polymorphism and adaptation in *Mimulus*

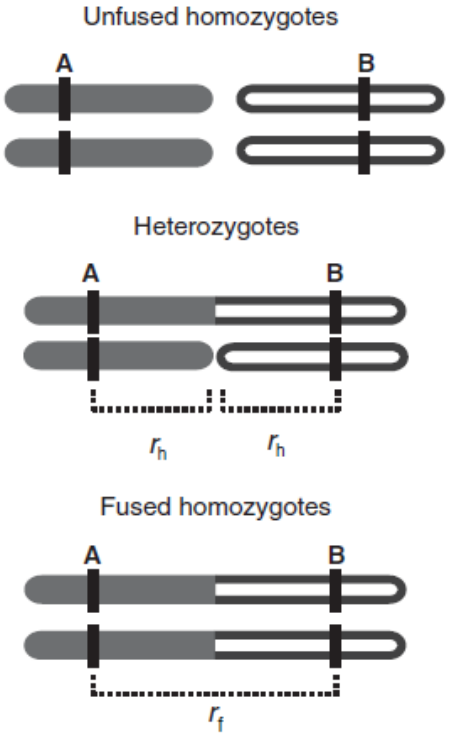
- a geographically widespread adaptive inversion polymorphism in the yellow monkeyflower (*Mimulus guttatus*)
- the inversion is involved in a classic life-history shift in plants - an adaptive response to differences in the seasonal availability of water resources:

  - one arrangement of the inverted region is found in an annual ecotype that lives in Mediterranean habitats characterized by reduced soil water availability in the summer;

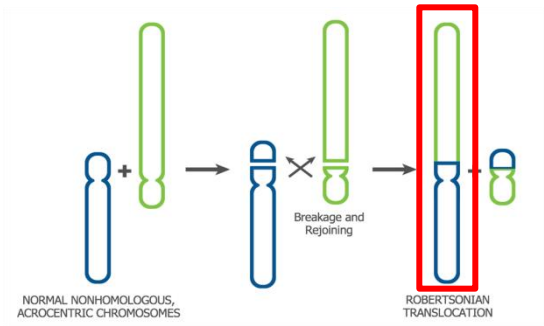
  - the other arrangement appears in a perennial ecotype that lives in habitats with high year-round soil moisture.

- inversion polymorphism influences morphological and flowering time differences between the two ecotypes = reproductive isolating barriers
- observation is consistent with the theory that adaptation to local environments can drive the spread of chromosomal inversions and promote speciation.
- for the first time in nature was shown the contribution of an inversion to adaptation, an annual/perennial life-history shift, and multiple reproductive isolating barriers

# Chromosome „fusions“ (CF) as a speciation agent ?

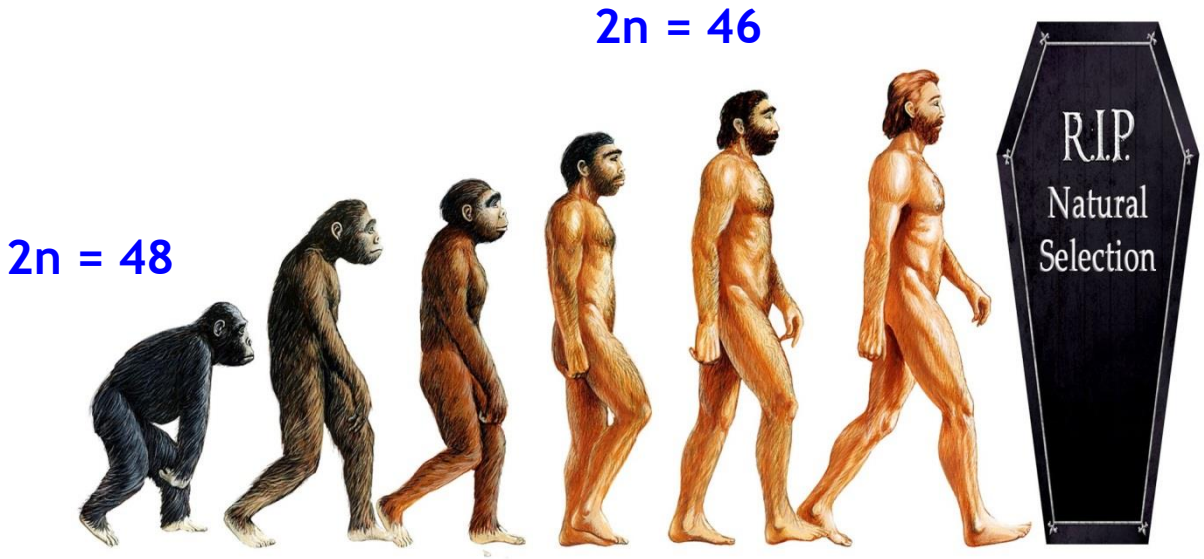


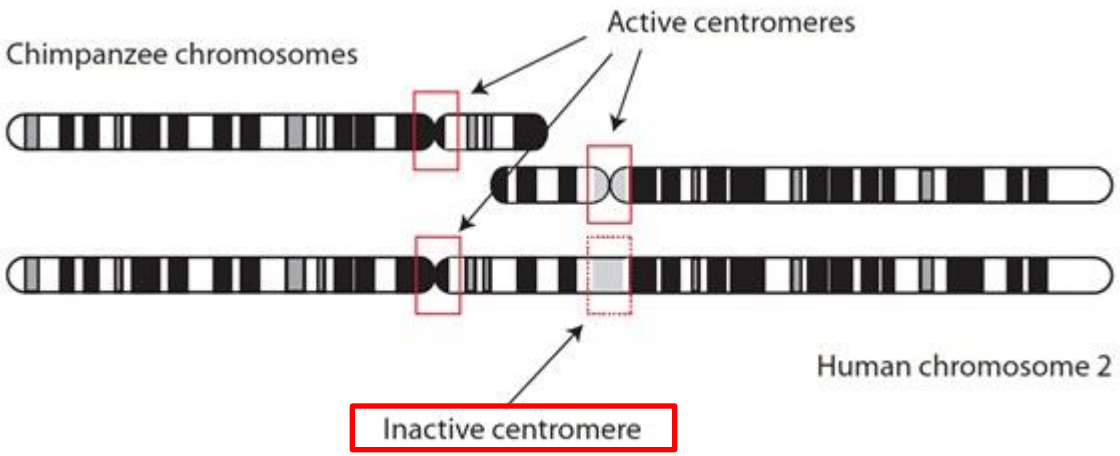

Loci A and B segregate independently in unfused homozygotes, recombination has different rates and segregation is different in heterozygotes and fused homozygotes




- CF can lead to tight linkage of genes („super-gene“)
- ... can avoid recombination between locally adapted alleles – adaptation / divergence / speciation
- ... can alter gene expression (silencing or higher expression)
- ...can confer mechanistic advantage (decreased no. of chromosomes = faster processes = possible adaptive advantage)

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



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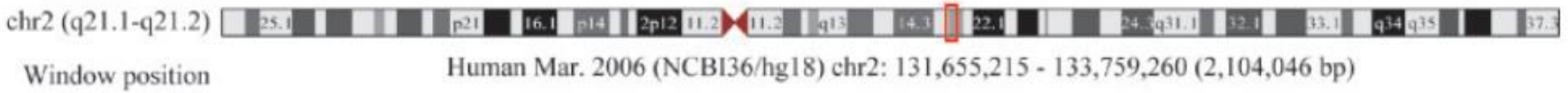
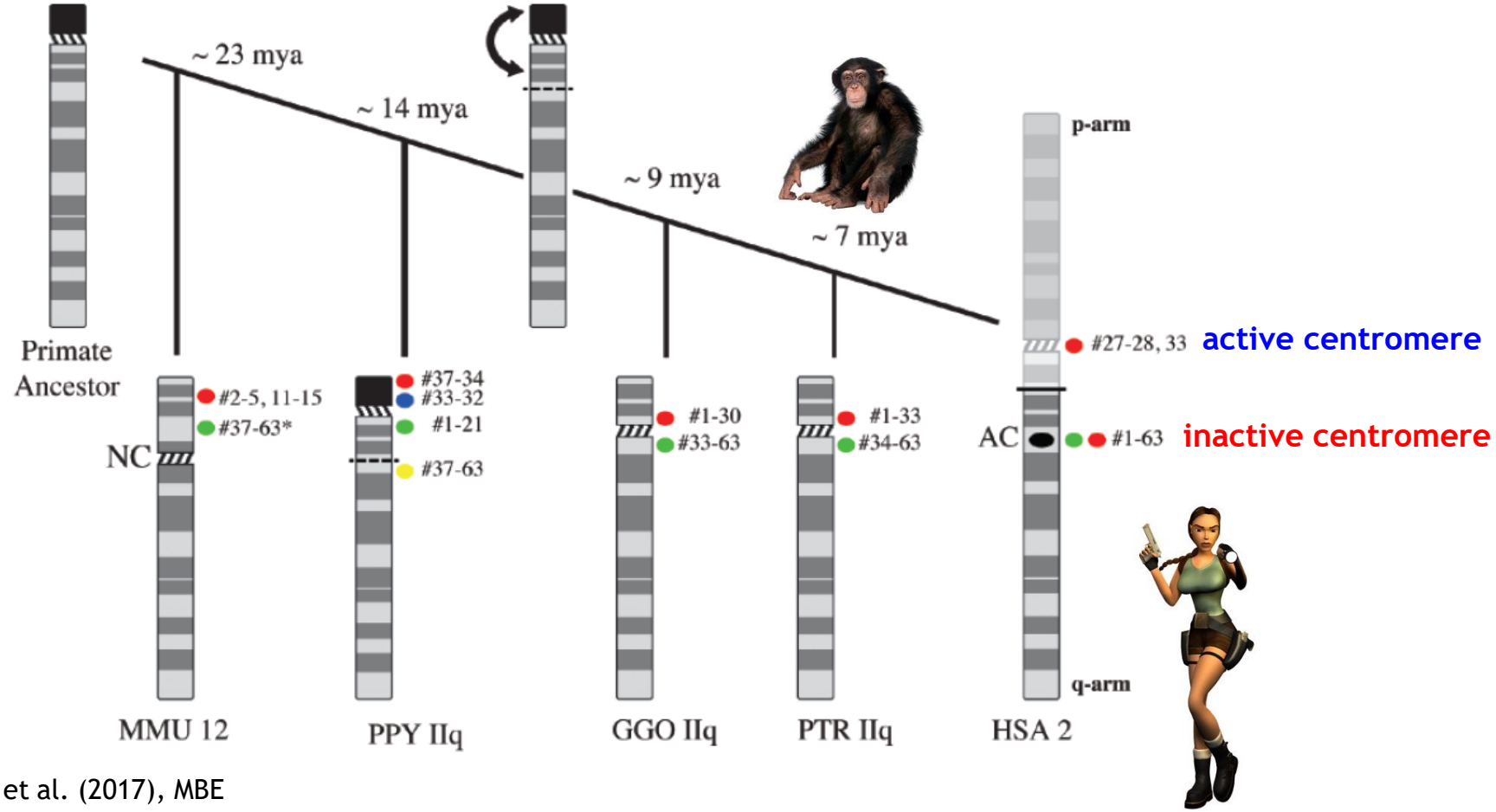
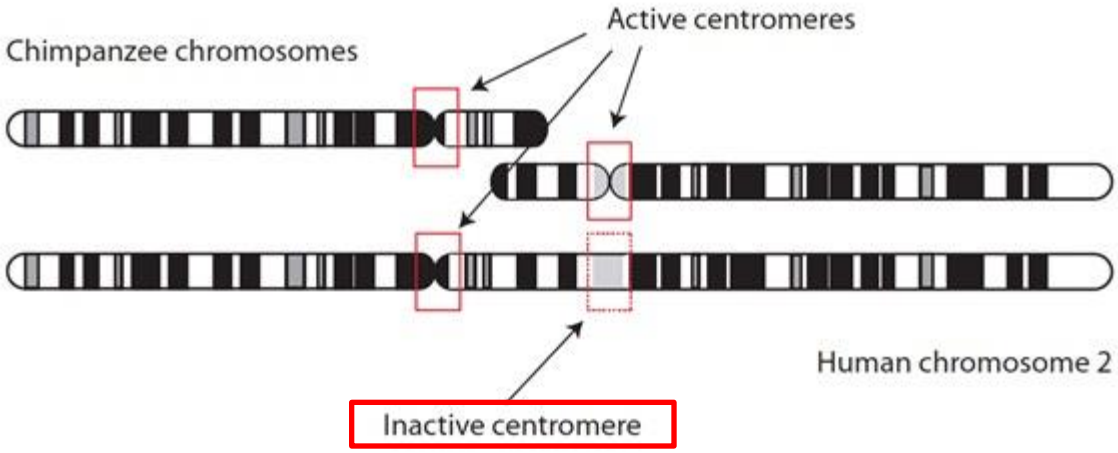



Chiatante et al. (2017), MBE

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

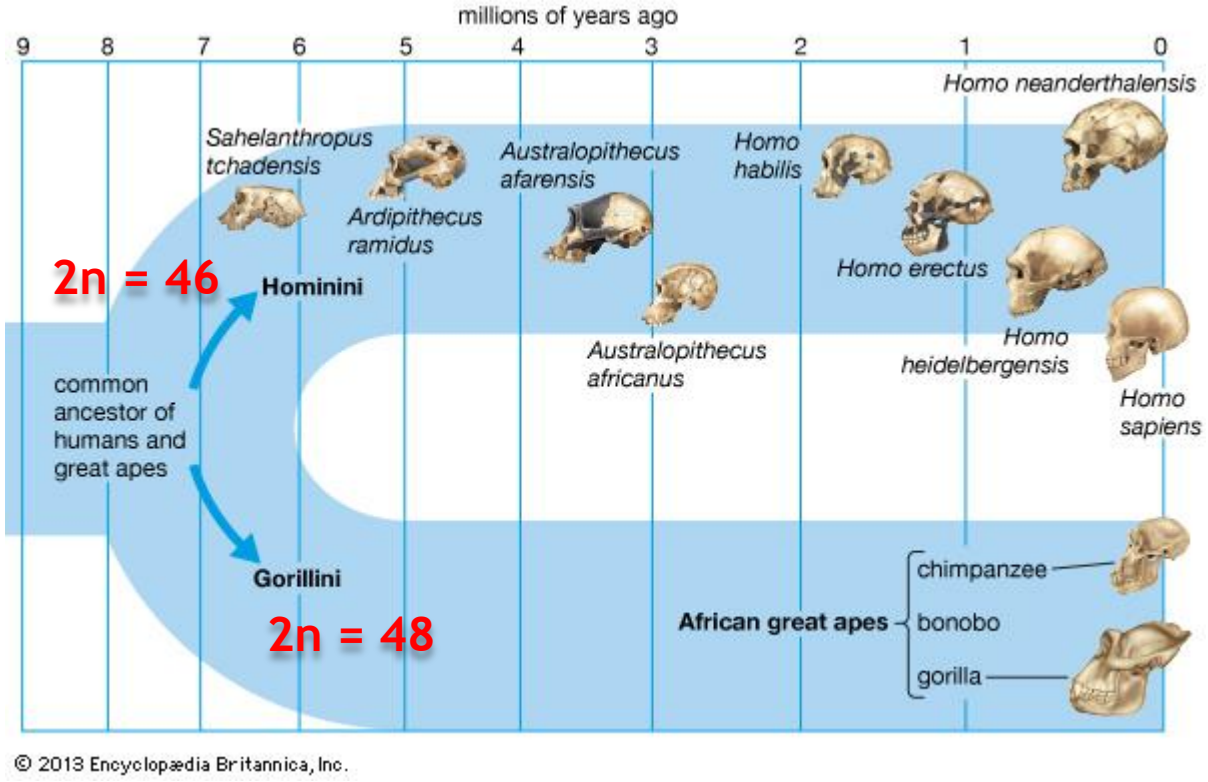



Two optionss how the „fusion“ chromosome 2 was stabilized

- 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...)

Chiatante et al. (2017), MBE

# Did the origin of „fusion“ chromosome 2 contributed to reproductive isolation of hominid species from great apes?





- different no. of chromosomes → reproductive isolation
- loss of gene(s) → adaptive advantage
- gene linkage? changed regulation of gene expression?

# Chromosomal speciation – example of rock-wallabies

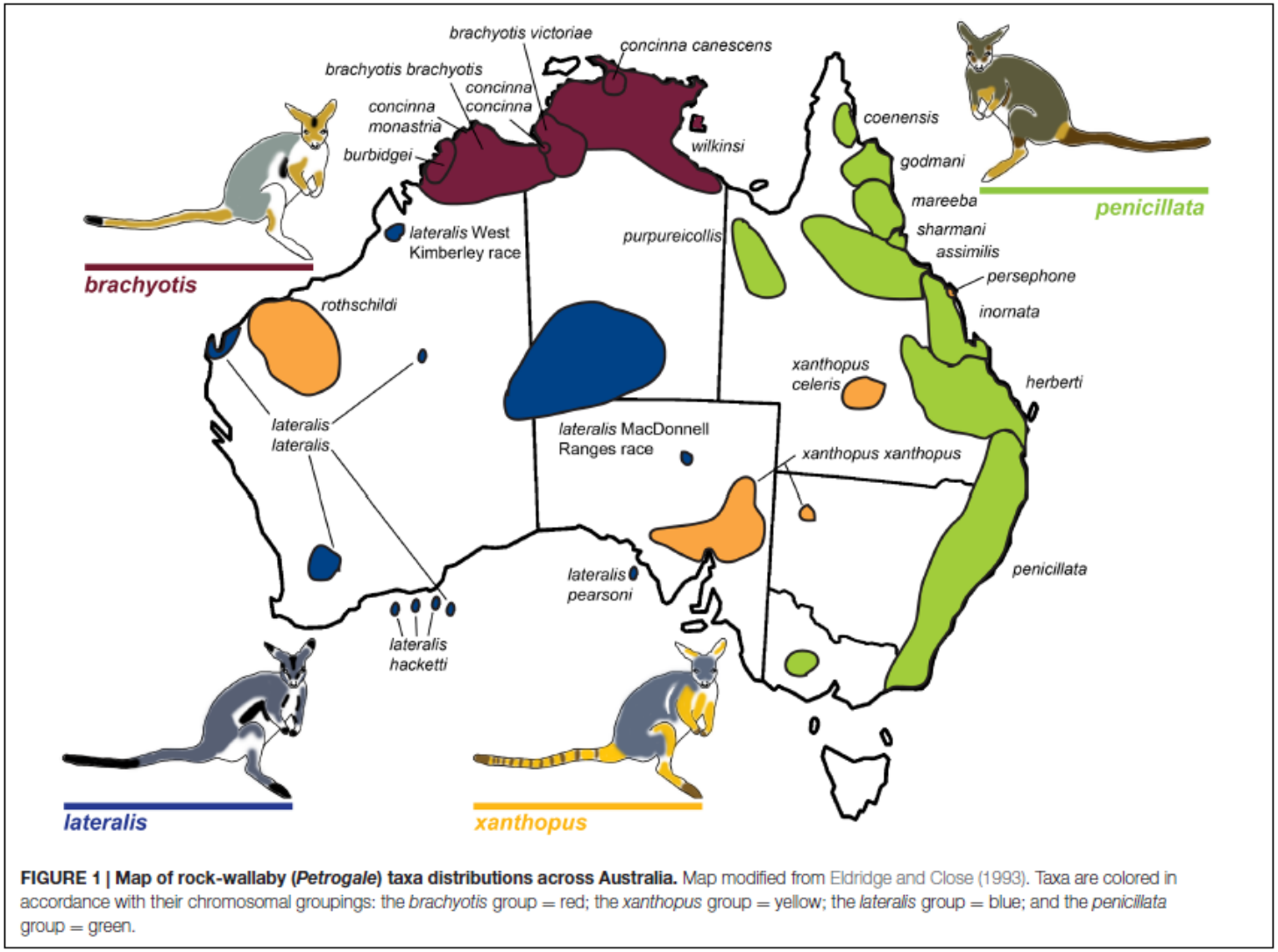
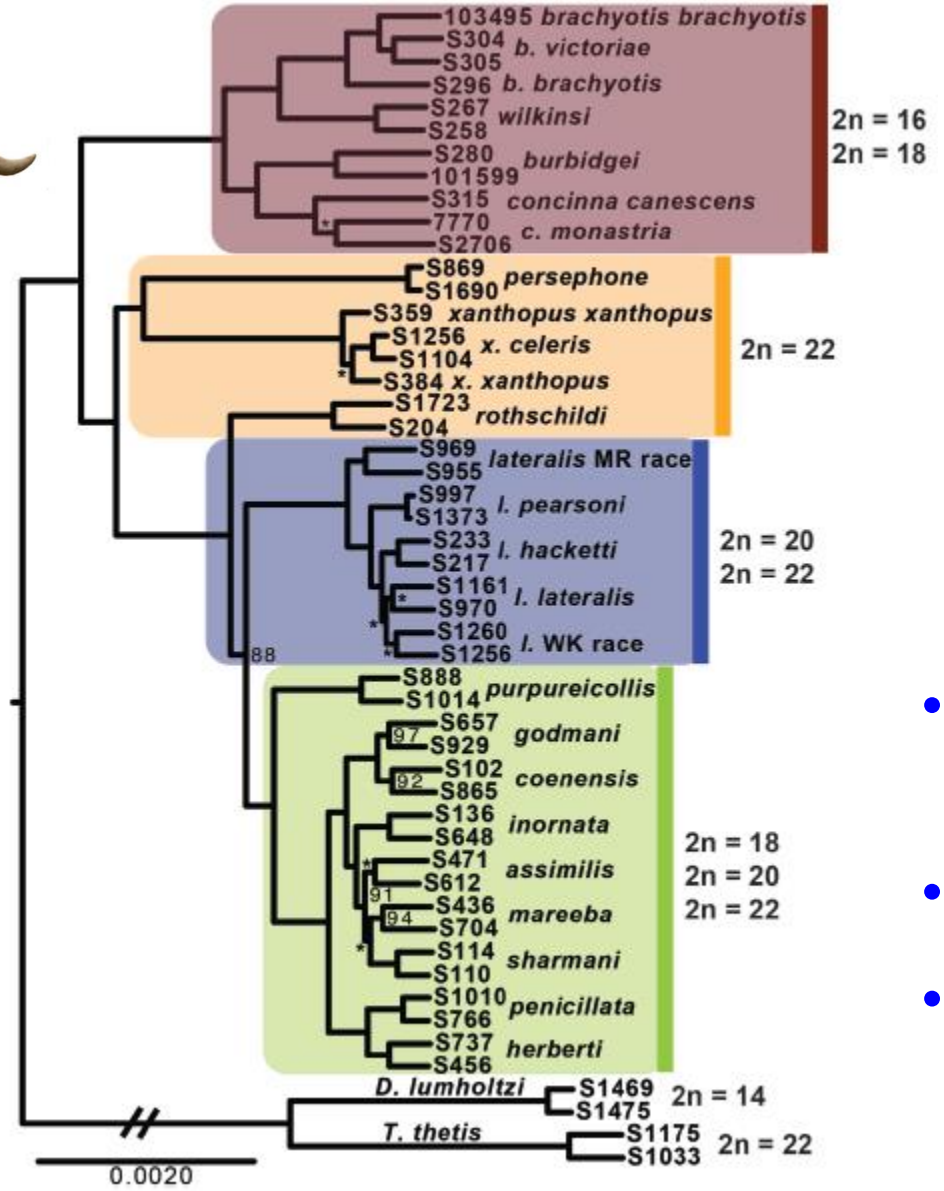



**FIGURE 1 | Map of rock-wallaby (*Petrogale*) taxa distributions across Australia.** Map modified from Eldridge and Close (1993). Taxa are colored in accordance with their chromosomal groupings: the *brachyotis* group = red; the *xanthopus* group = yellow; the *lateralis* group = blue; and the *penicillata* group = green.

Potter et al. (2017), Front Genet

# Chromosomal speciation – example of rock-wallabies




- chromosome „fusions“ via reciprocal translocations
- inversions
- centromere shifts

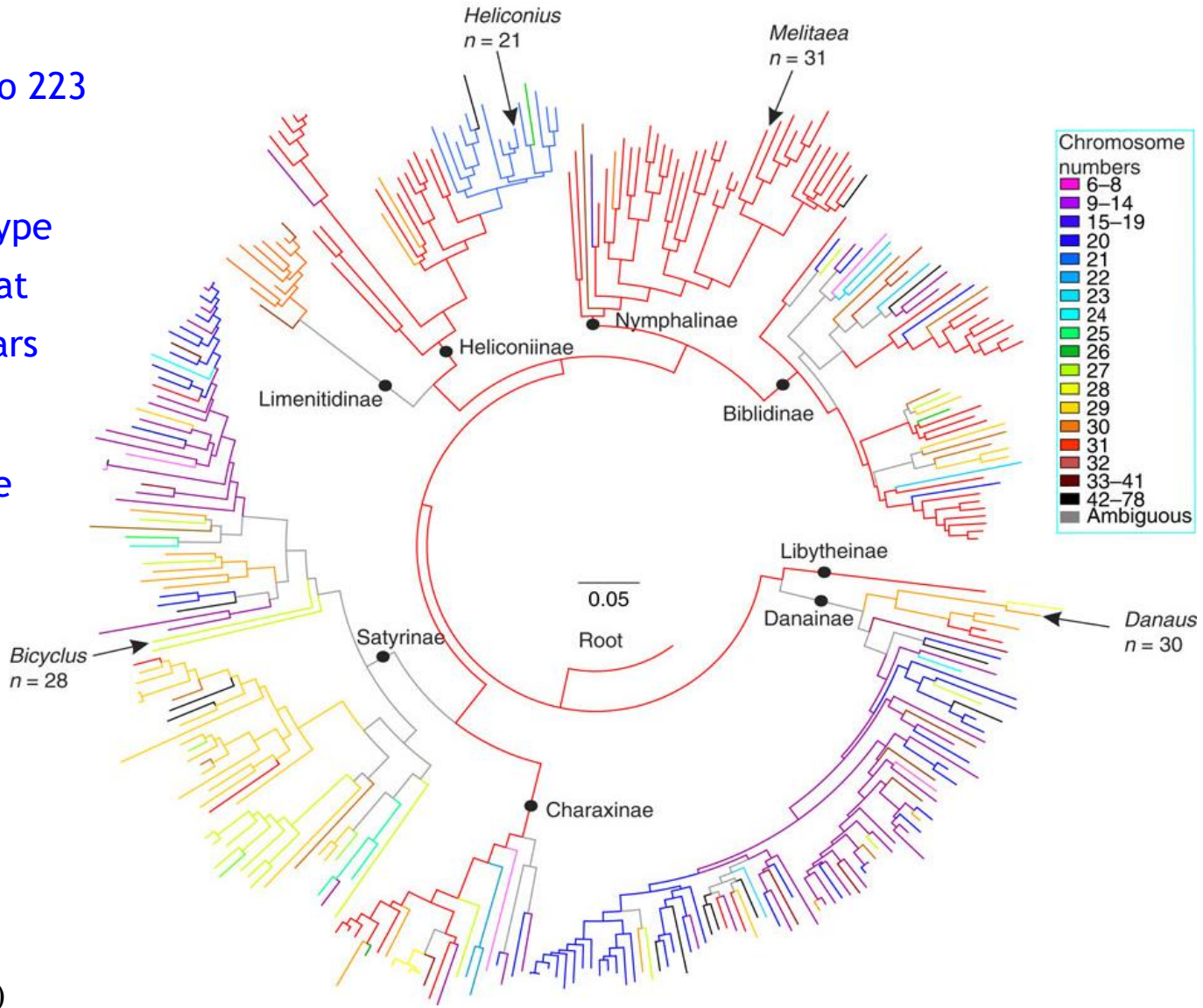
Potter et al. (2017), Front Genet

# Speciation by Robertsonia translocations („centric fusions“)

*Rhogeessa tumida*

# 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)