The role of chromosome rearrangements in reproductive isolation and 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

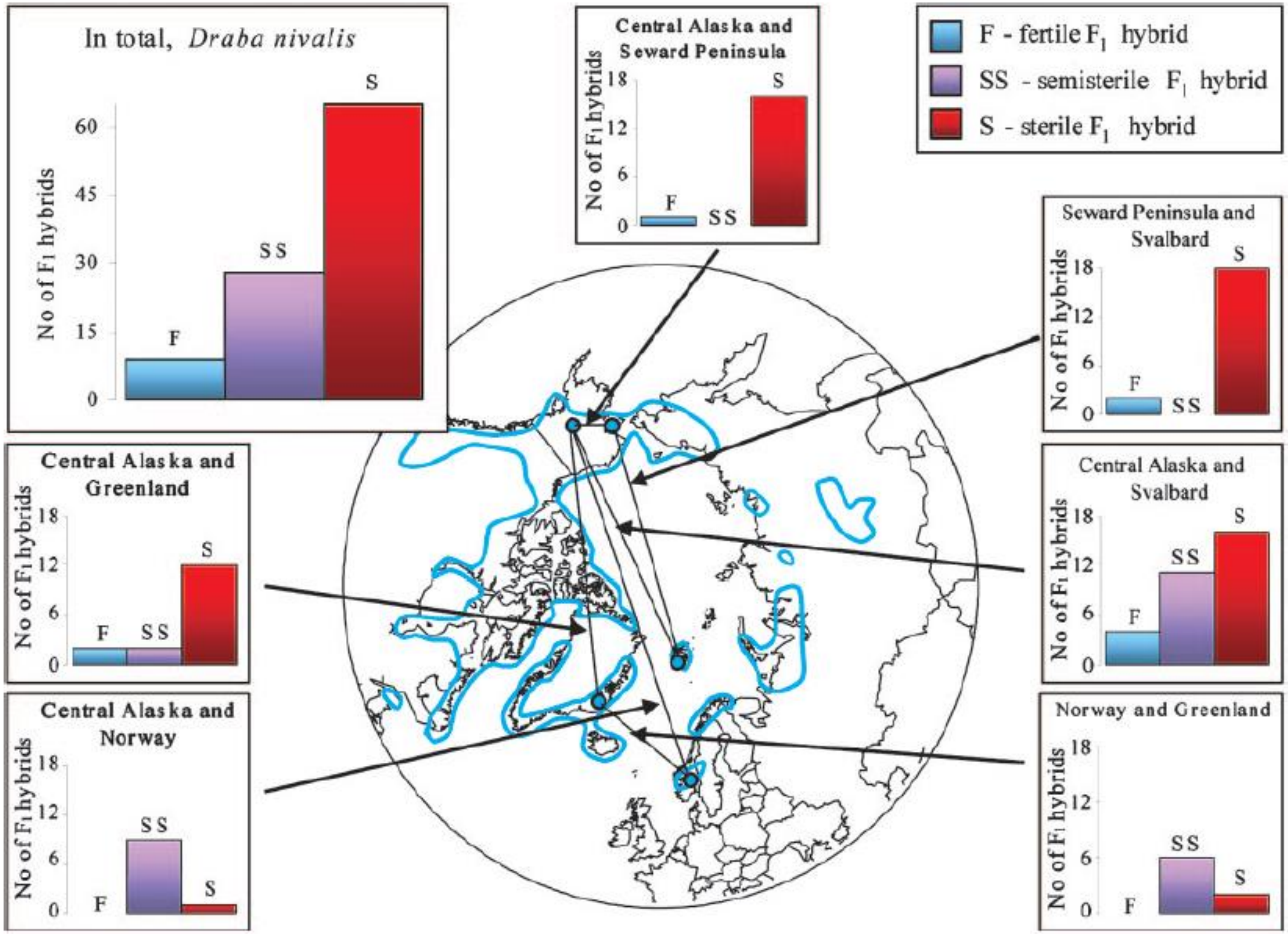
Are chromosome rearrangements important for creating reproductive isolation barriers and speciation?

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

M.J.D. White

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.

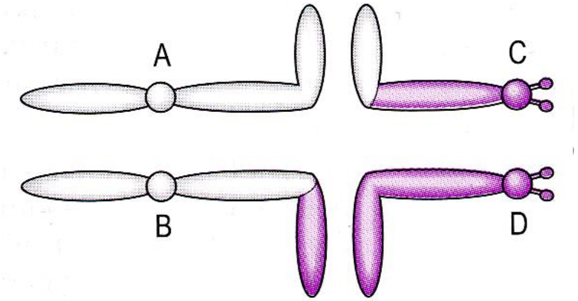
Fundamental feature of the models: **chromosomal differences that have accumulated between the neospecies and its progenitor(s) are assumed to impair the fertility or viability of interspecific hybrids, thereby reducing gene flow**

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)

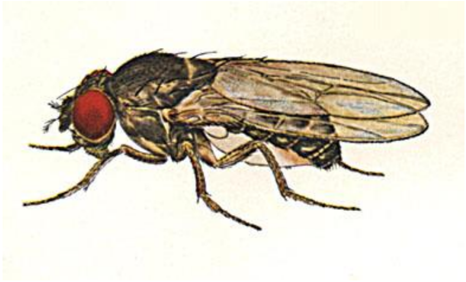
Chromosome speciation in *Drosophila*

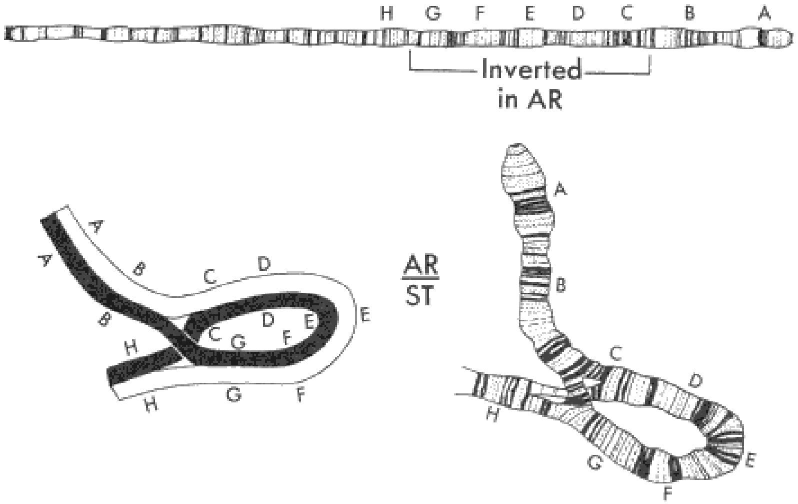
- gross chromosomal rearrangements in *Drosophila* are well characterized as rearrangements are easily detected in the chromosomes of their giant salivary glands

- 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)

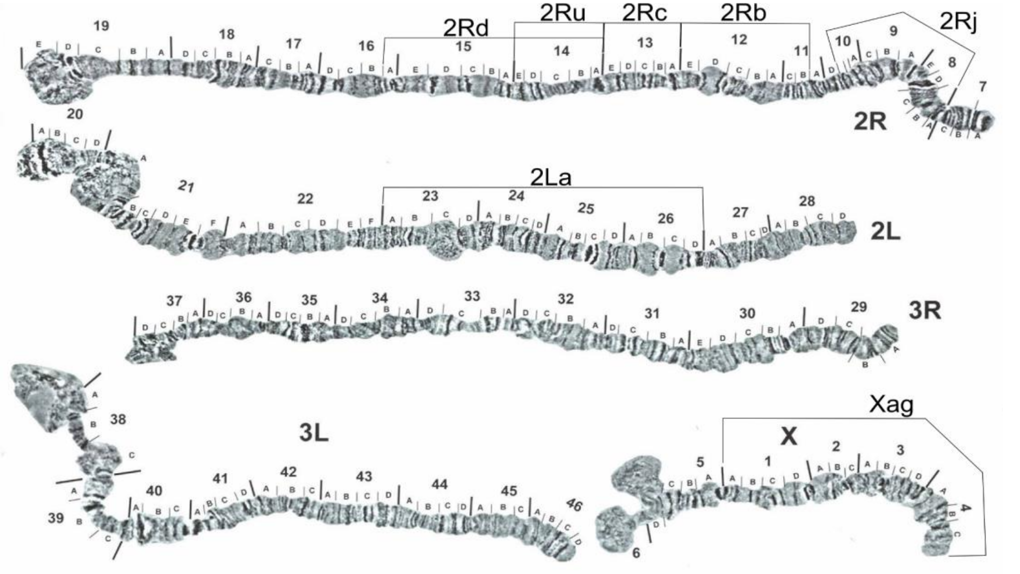
A paracentric inversion in *D. pseudoobscura*

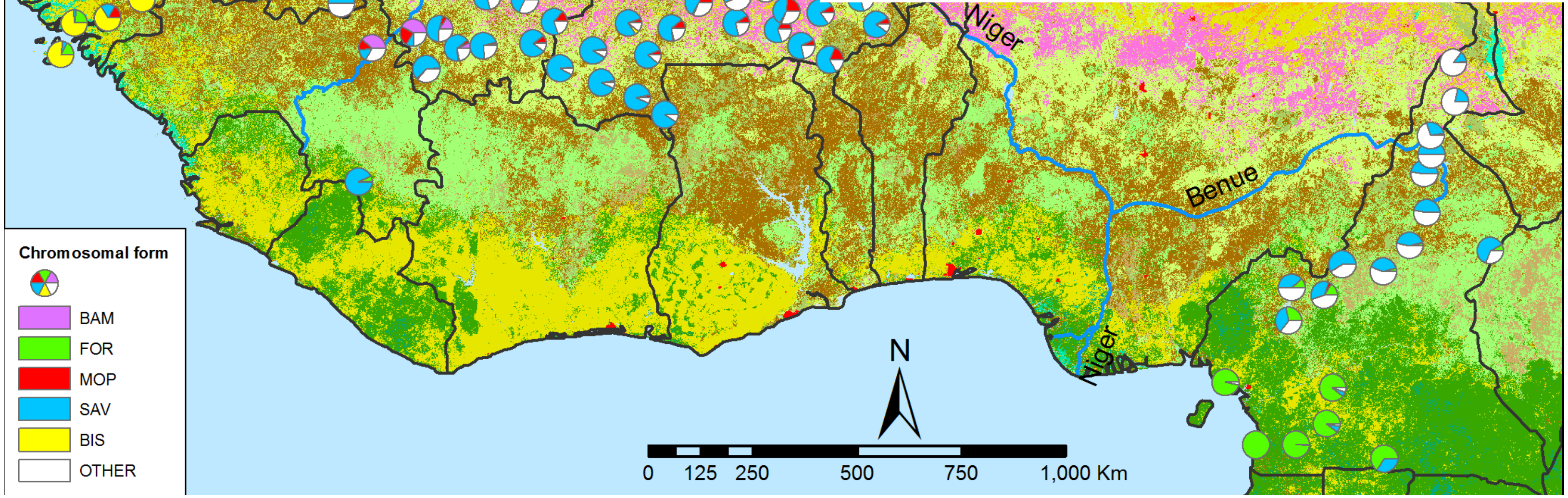




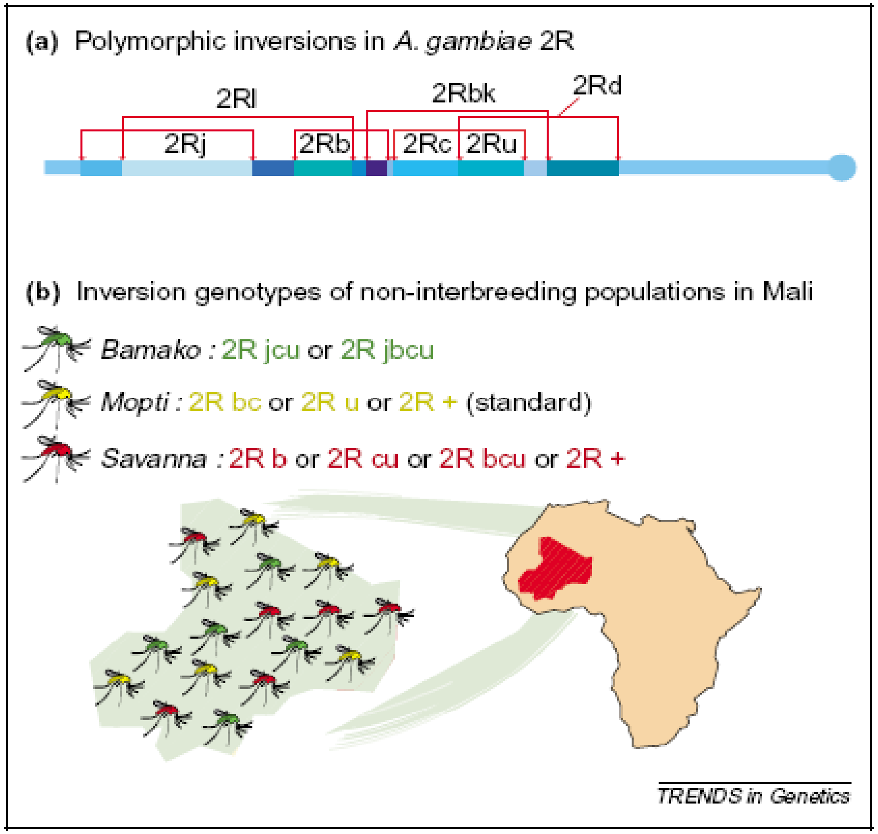
Inversions are crossover suppresors – evolutionary consequences (speciation)

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.

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Chromosome Inversions, Genomic Differentiation and Speciation in the African Malaria Mosquito *Anopheles gambiae*

Yoosook Lee[1]*, Travis C. Collier[1], Michelle R. Sanford[1], Clare D. Marsden[1], Abdrahamane Fofana[3], Anthony J. Cornel[2], Gregory C. Lanzaro[1]

- data consistent with expectations that inversions are involved in the divergence process, elevated genomic divergence was found between *Bamako* and *Savanna* in the regions carrying inversions

- but divergence was not observed throughout the inversions, rather it is restricted to sites near the breakpoints

- surprisingly, the overall highest divergence was found on the X chromosome

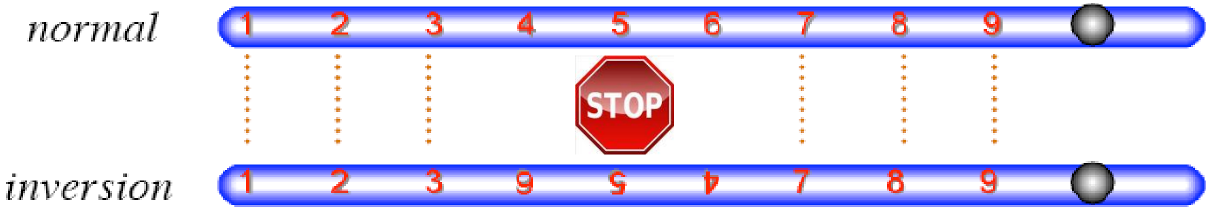
- genes located on the sex chromosome may be the major force driving speciation between the chromosomal forms of *A. gambiae*

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.



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

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

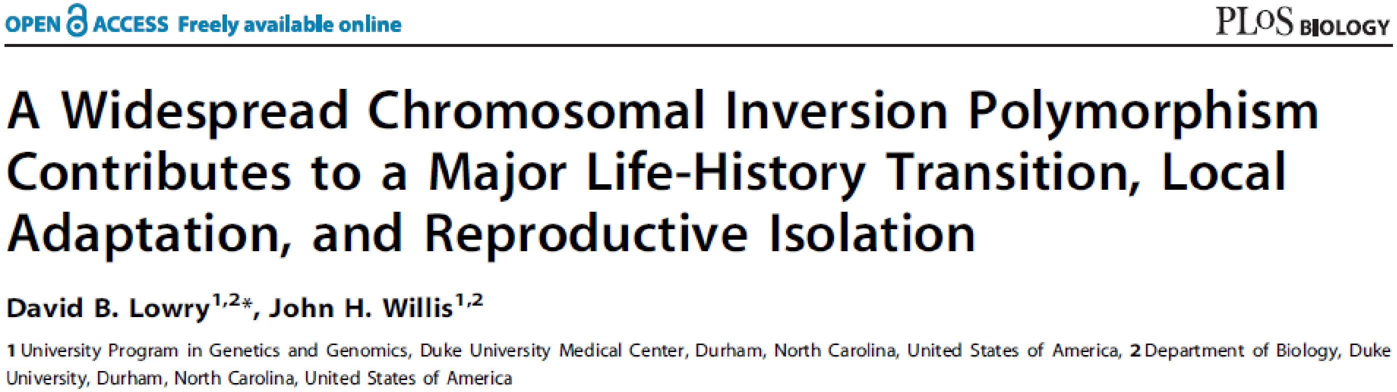
- chromosomal rearrangements reduce recombination between the genomes of the species, thereby enabling genetic differences to accumulate within the rearranged regions

- inversions are more common between *Drosophila* species that are sympatric compared to allopatric pairs

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)



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PLoS BIOLOGY

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

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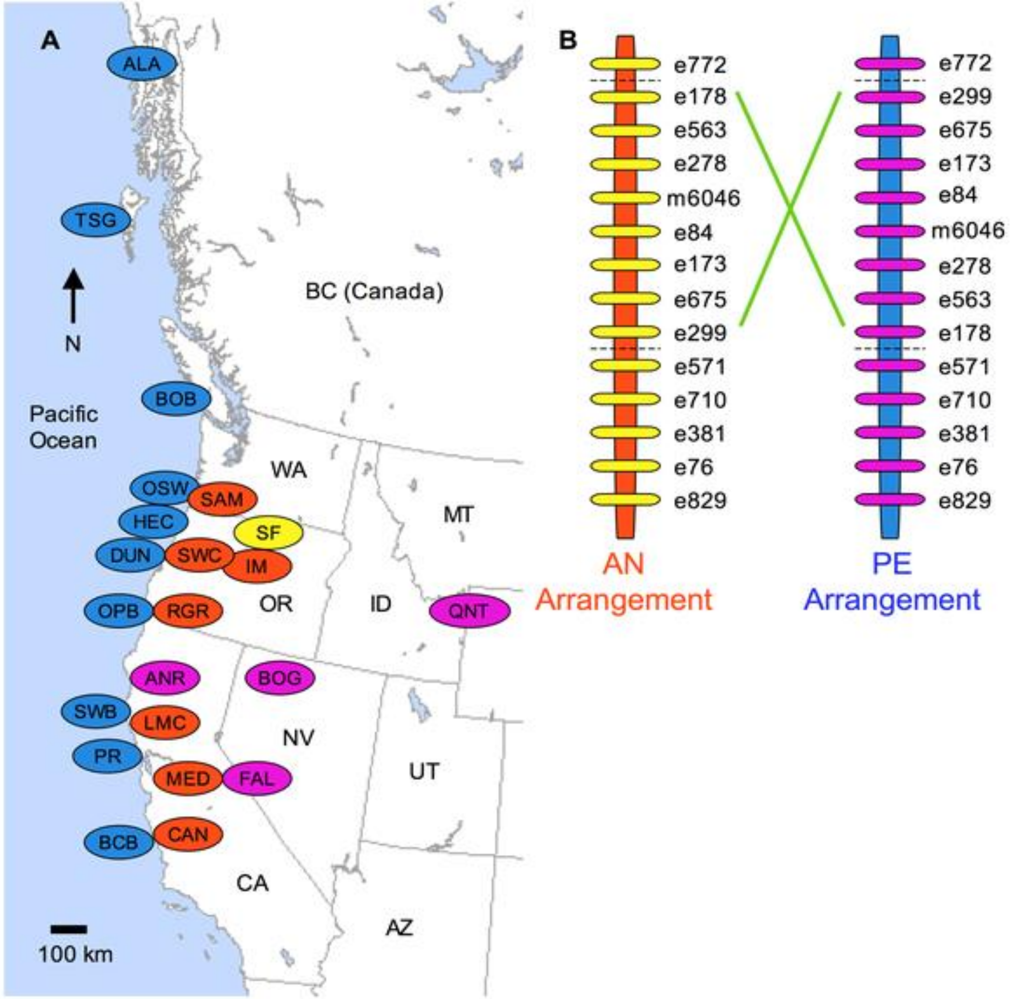
perennial and annual plant

yellow monkeyflower (*Mimulus guttatus*)

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

The question is, are CRs an important component of speciation?

When considering all the data, the emerging picture is that evidence for chromosomal speciation varies between lineages. For example, it is strong for *Drosophila*, but much weaker or absent in primates.

The observation that appears most consistently in many species is an increased level of divergence near rearrangement breakpoints. According to suppressed-recombination models, these regions constitute strong candidate regions to accumulate alleles involved in reproductive isolation.

Faria and Navarro (2010)

Chromosomal rearrangements as post-pollination barriers

- CRs can contribute to reproductive isolation through their effects on hybrid fertility, as well as by reducing interspecific gene flow
- CRs most commonly contributing to reproductive isolation in plants are inversions and translocations
- large translocations have the impact on the fertility of hybrids - 50% of gametes expected to carry deletions or duplications of whole chromosome arms (or portions of arms). Translocations also reduce recombination rates near centromeres or chromosomal breakpoints due to mechanical interference with pairing and crossing over.
- The fertility effects of inversions are less severe, although for large inversions up to 50% of gametes can be inviable due to recombination within the inversion resulting in unbalanced gametes. However, inversions have little or no effect on fertility if recombination is reduced (if the inversion is small, occurs in a low recombination region).
- Reduced recombination can contribute to the evolution of reproductive barriers by facilitating the accumulation of hybrid incompatibilities or other key reproductive barriers traits in the presence of gene flow.