The role of chromosome rearrangements in reproductive isolation and speciation: 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)?

Chromosome mutations

are variations in:

1. Chromosome structure (chromosomal rearrangements)
   - deletions
   - duplications
   - translocations
   - inversions
   - transpositions

   } mostly due to errors or mispairings in crossing over

2. Chromosome number
   - aneuploidy
   - abnormal euploidy

When chromosome rearrangements occur they are rare in a population:

How do they spread?

Problems

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

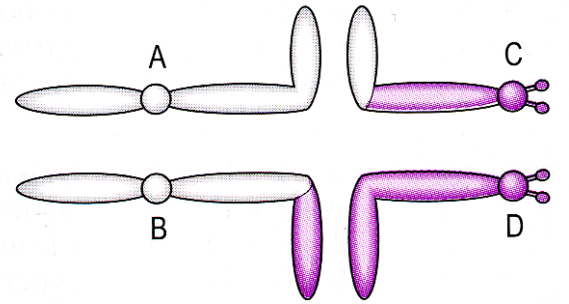
The amount of phenotypic evolution is not a good predictor of the amount of karyotypic evolution

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

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

More 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)

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

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

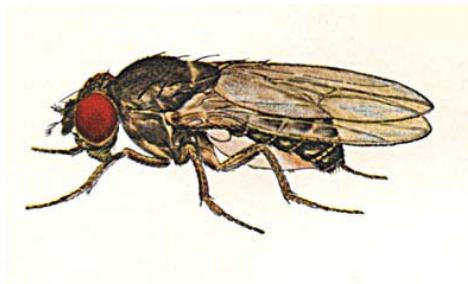
Chromosome speciation in *Drosophila*

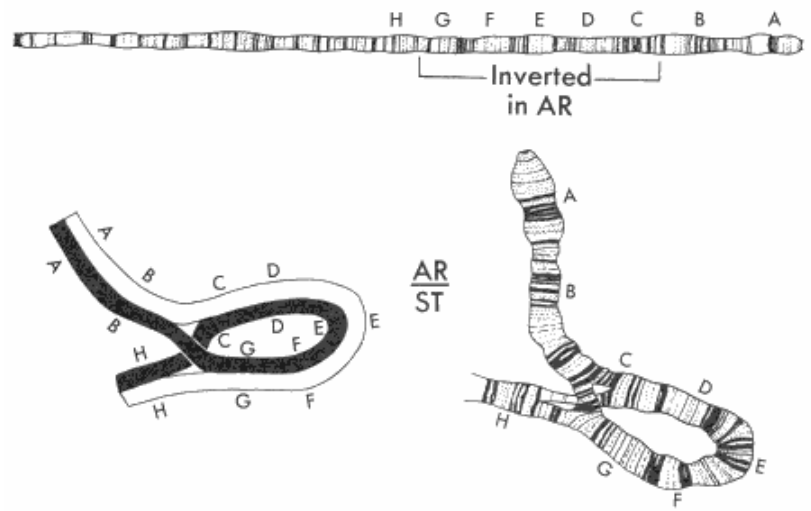
- gross chromosomal rearrangement in *Drosophila* are well characterized as they are easily detected in the chromosomes of their giant salivary glands

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

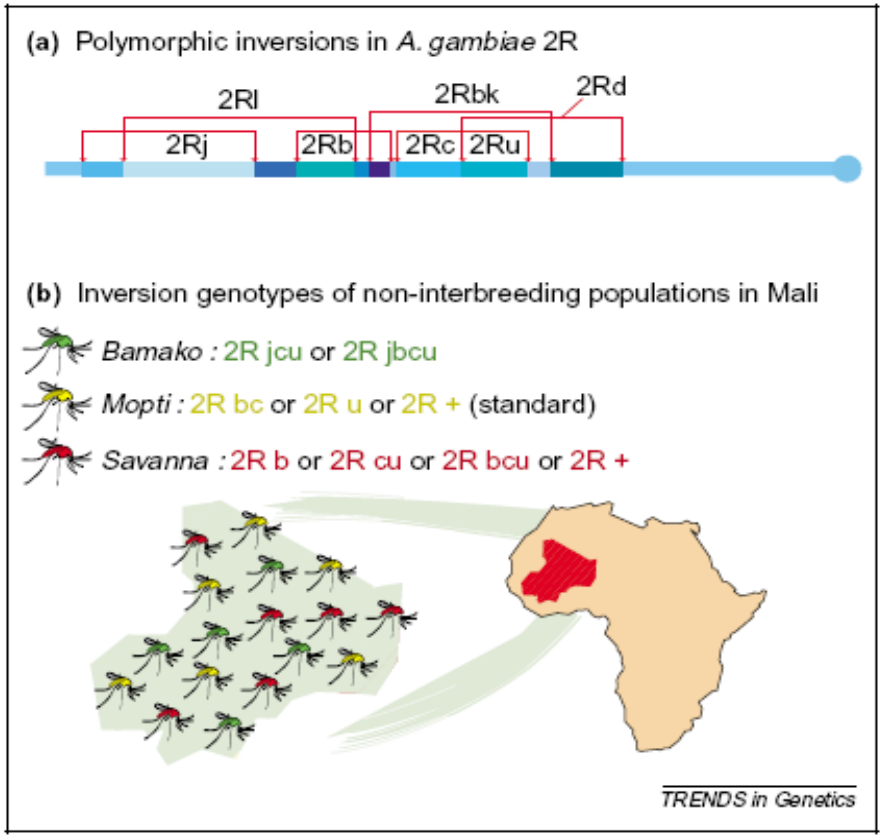
- paracentric inversions are common polymorphisms in drosophilas and other fly species (different populations of *D. melanogaster* harbor more than 500 inversion polymorphisms)

paracentric inversion in *D. pseudoobscura*





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



(a) The main 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 three populations differ by chromosomal inversions that might be contributing to speciation in *A. gambiae*.

For example, a chromosome with arrangement 2Rjcu has inversions j, c and u.

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.

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

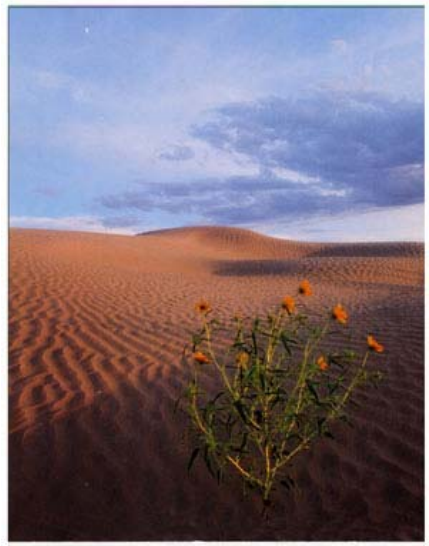
PLANTS

• the exact relationship between chromosomal rearrangement and speciation remains unclear in plants!

• seems that sunflowers (*Helianthus*) are only example: recombination between two divergent diploid species appears to have provoked speciation events in sunflowers (Loren Rieseberg's lab)

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

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



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 more by suppressing recombination than by directly reducing fitness.

Unlike other models of chromosomal speciation, the suppressed-recombination model does not suffer from the problem of underdominance.

A secondary contact scenario: chromosomal rearrangements favor speciation because they impede introgression across a large block of the genome, thus allowing certain chromosomal regions of two hybridizing populations to persist without fusing longer than regions where no chromosomal changes were present.

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

Homoploid hybrid speciation in sunflowers (*Helianthus*)

all diploid (n=17), annual and self-incompatible

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

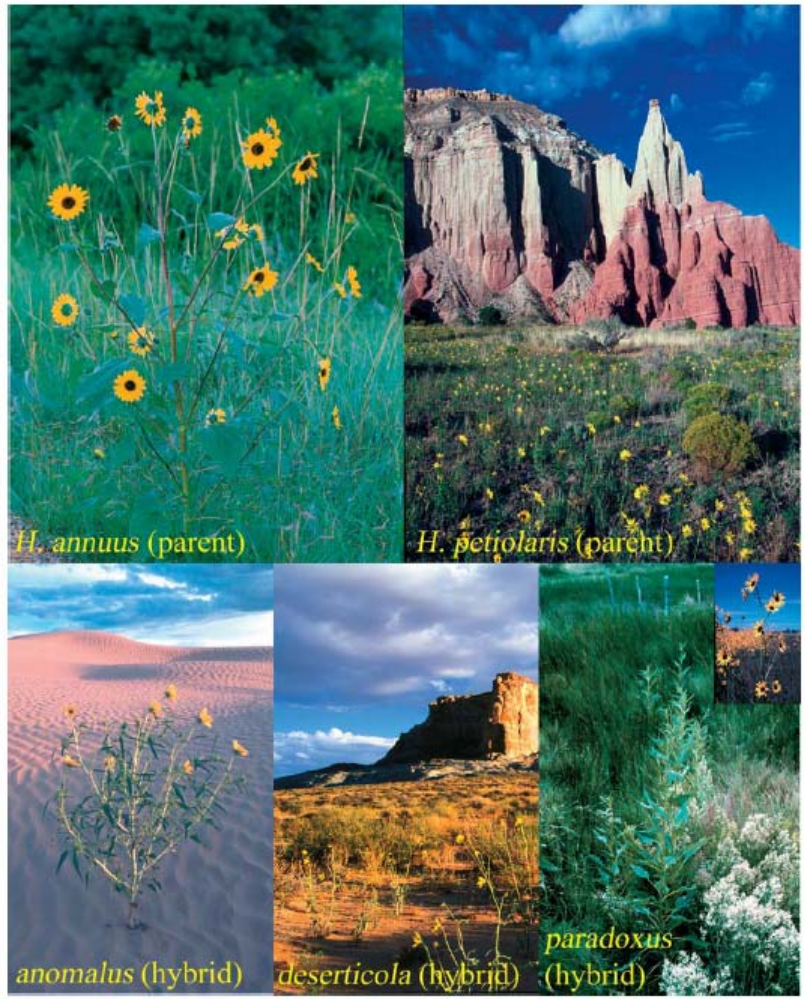
⇩

H. anomalus

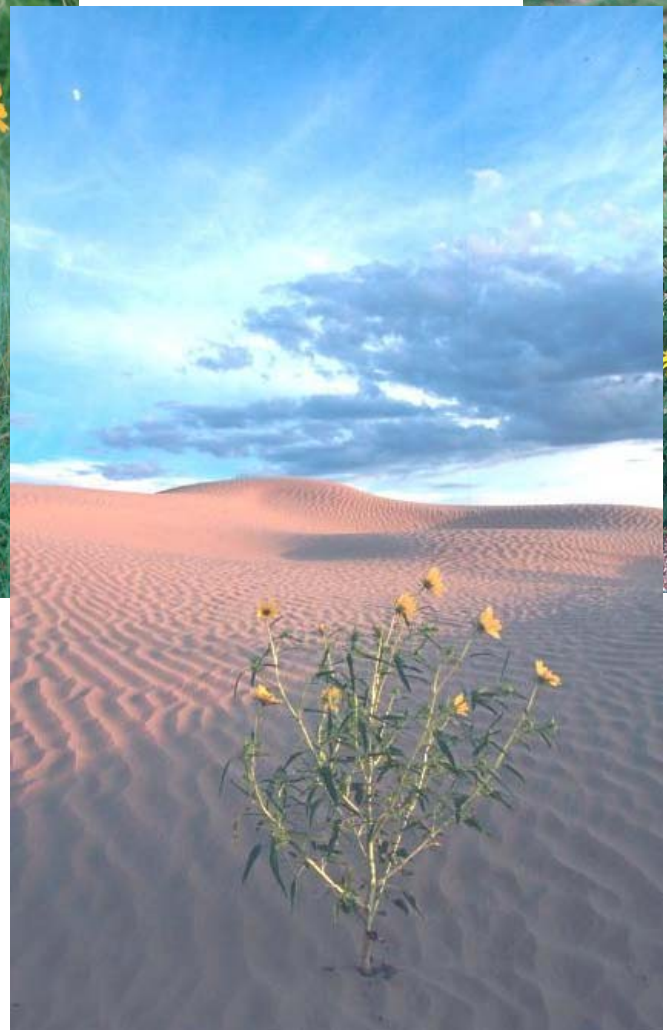
H. deserticola

H. paradoxus

three different hybrid species occupying different habitats



Helianthus annuus



Helianthus anomalus

Helianthus petiolaris

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)

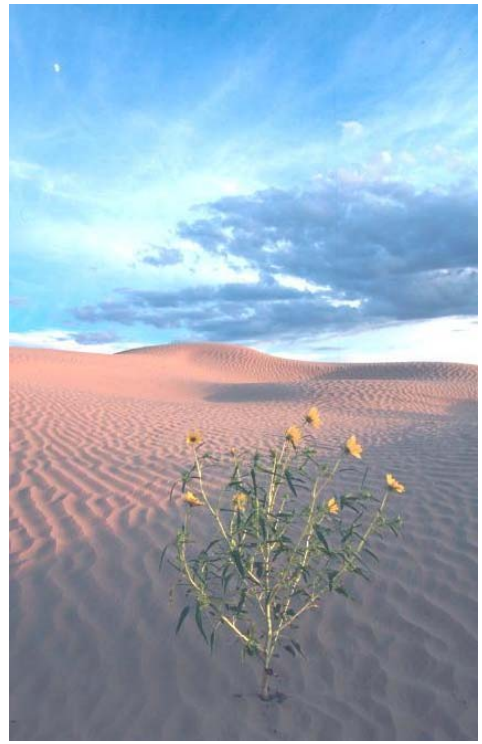
- the three synthetic lineages were cross-compatible with each other and with *H. anomalus* (Rieseberg 2000)

- *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



Reccurent origin of *H. anomalus* ?

• documentation of the multiple origins of diploid hybrid species is difficult

• introgression following a hybrid origin may produce patterns that are almost indistinguishable from those produced by recurrent hybrid speciation

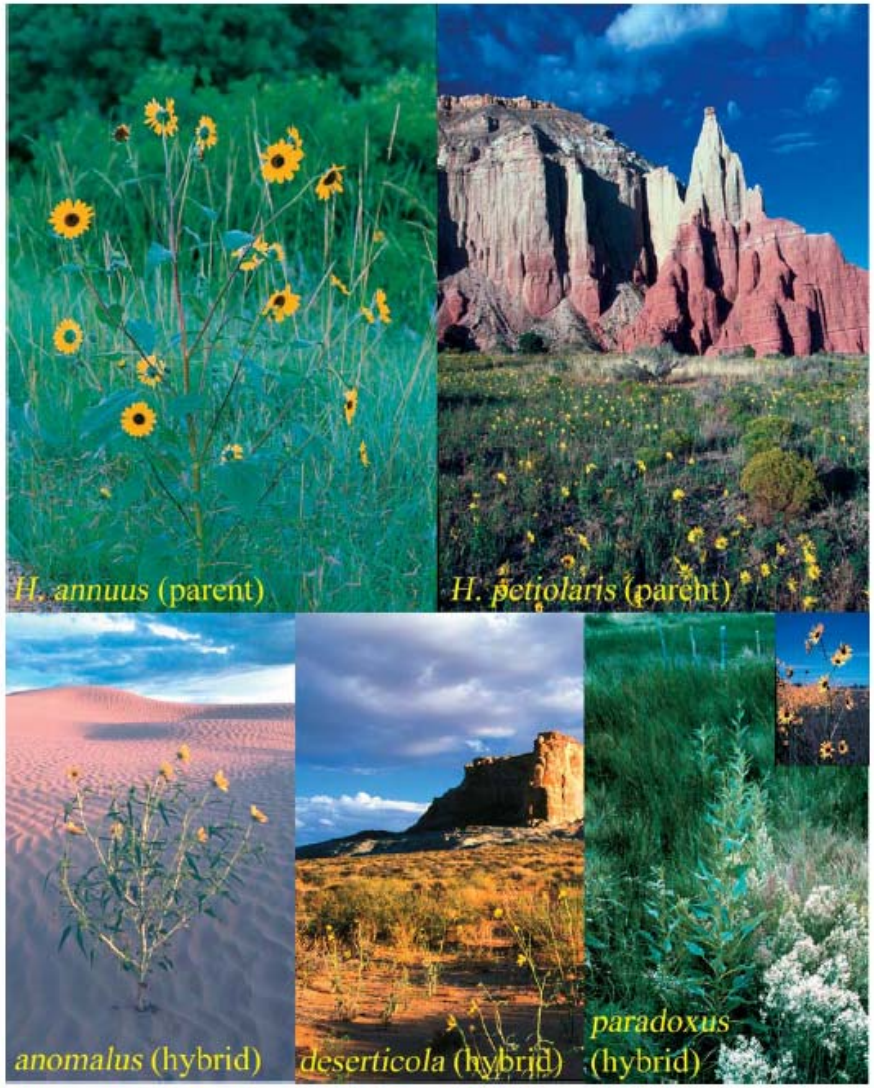
✓ greenhouse experiments showed that the genomic composition of hybrid lineages is shaped in part by deterministic forces

✓ recurrent diploid hybrid speciation may be more feasible than previously believed

✓ chloroplast DNA (cpDNA) and crossability data were most consistent with a scenario in which *H. anomalus* arose three times: three different *H. anomalus* fertility groups were discovered, each with a unique cpDNA haplotype

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

Comparative genetics in five *Helianthus* species



Comparative genetic mapping in plants

Analyzing karyotypic changes

Cytology – chromosome structure/morphology and chromosome pairing in meiosis I

Limitations

- indistinguishable chromosomes
- disruption of meiotic pairing; ambigious interpretations of meiotic configurations

Comparative genetic (linkage) mapping (1988: first comparative studies published)

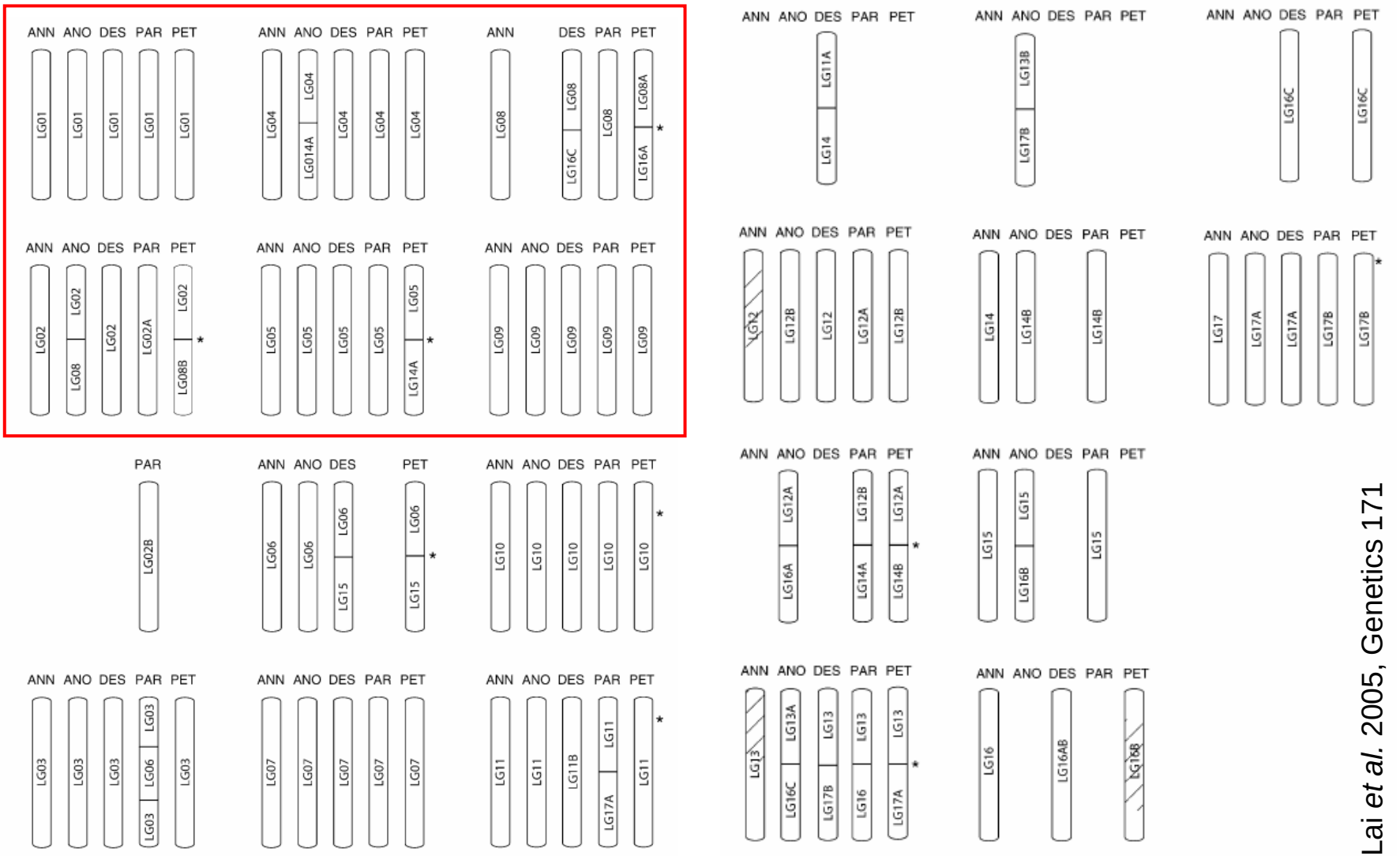
✓ based on inter-species mapping of known orthologous sequences

✓ allows comparisons to be made between species, genera, and even families (difficult by cytological analysis)

Limitations

- the quality of a genetic map depends on marker density
- large-scale rearrangements are readily detected, whereas those involving small chromosomal segments are likely to go undetected

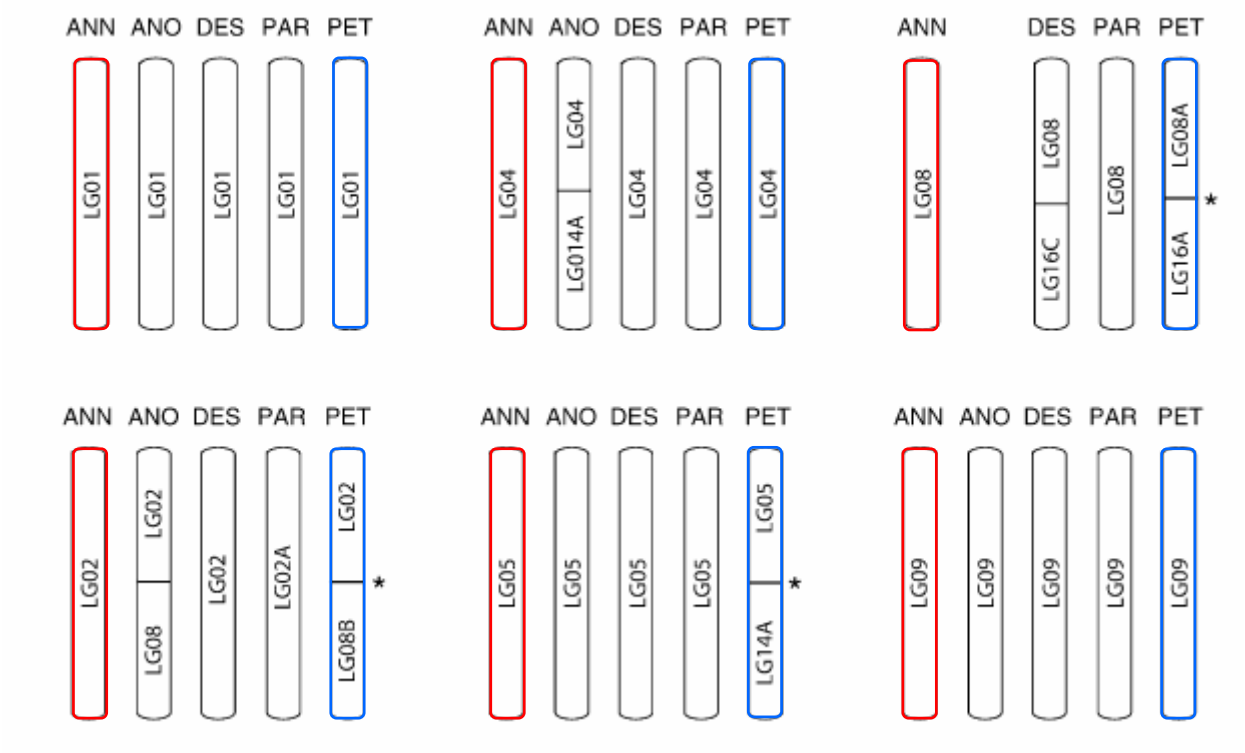
Inferred chromosomal structural relationships between the parental species, *H. annuus* and *H. petiolaris*, and their three diploid hybrid derivatives, *H. anomalus*, *H. deserticola*, and *H. paradoxus*



Lai *et al.* 2005, Genetics 171

Segments containing inversions are indicated by hatched lines.

Inferred chromosomal structural relationships between the parental species, *H. annuus* and *H. petiolaris*, and their three diploid hybrid derivatives, *H. anomalus*, *H. deserticola*, and *H. paradoxus*



Lai *et al.* 2005, Genetics 171

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

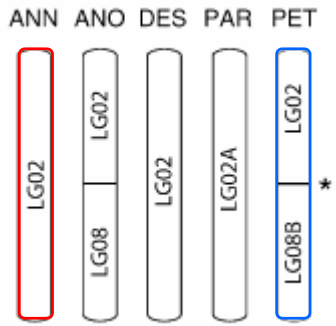
H. paradoxus

• 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

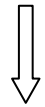


Homoploid hybrid speciation in sunflowers (*Helianthus*)

• approx. 50% of the reproductive barrier between *Helianthus* species is caused by chromosomal rearrangements

• the data in line with the theory (rapid karyotypic evolution in the hybrid sunflowers have facilitated the development of reproductive isolation with their parental species)

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



H. anomalus

H. deserticola

H. paradoxus

• the lengths of chromosomal segments around isolating genes that are prevented from introgressing across hybrid zones are longer in rearranged *vs.* collinear chromosomes

In Drosophila*, genes causing hybrid sterility and conditioning female species preferences between* D. pseudoobscura *and* D. persimilis *map predominantly to inversions that differentiate the species (Noor et al. 2001)*

Genome size increase in hybrid *Helianthus* species

• hybrid-derived species have 50% more nuclear DNA than the parental species (*H. annuus and H. petiolaris*)

• first- and sixth-generation synthetic hybrids and hybrid-zone plants did not show an increase from parental DNA content

⇩

• hybridization by itself does not lead to increased nuclear DNA content in *Helianthus*

• is amplification of repetitive sequences responsible for the increases in DNA content in the hybrid-derived species?

Current Biol 16 (2006)

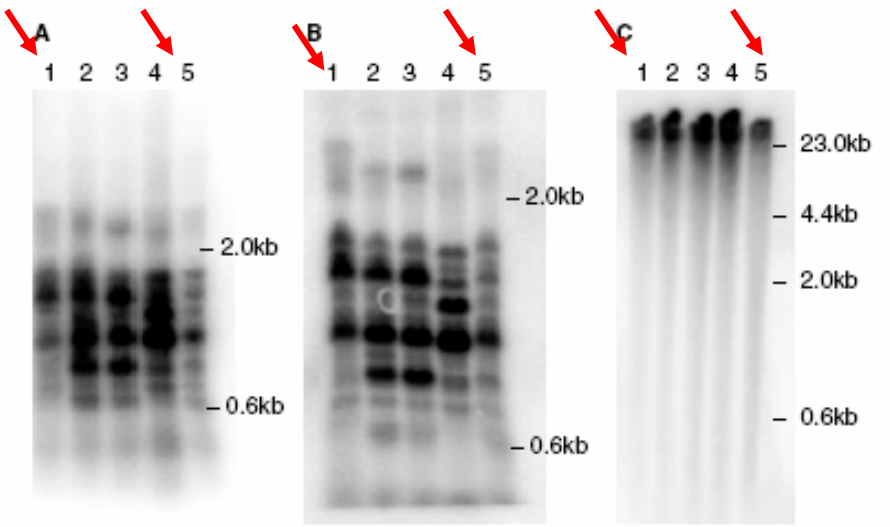
Genome expansion in three hybrid sunflower species is associated with retrotransposon proliferation

Mark C. Ungerer, Suzanne C. Strakosh and Ying Zhen

- 3 hybrid species exhibit species-specific karyotypic alterations
- all 3 species have at least 50% larger genomes than the parental species

- 5.6 to 23.6-fold increases in copy number of the *Ty3/gypsy* retrotransposon in all hybrid taxa

Southern blots probed with an 887 base pair region of the *Ty3/gypsy* integrase domain



Copy number estimates (per genome) of the *Ty3/gypsy* integrase domain based on quantitative PCR