

# Genome Evolution

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## Genome Size Evolution

<i>Saccharomyces cerevisiae</i>	12.05	6213
<i>Plasmodium falciparum</i>	22.85	5268
<i>Trypanosoma spp.</i>	39.2	10000
<i>Aspergillus nidulans</i>	30.07	9541
<i>Dictyostelium discoideum</i>	34	9000
<i>Arabidopsis thaliana</i>	125	25498
<i>Oryza sativa</i>	466	60256
<i>Lotus japonicus</i>	472	26000

Lynch (2006)

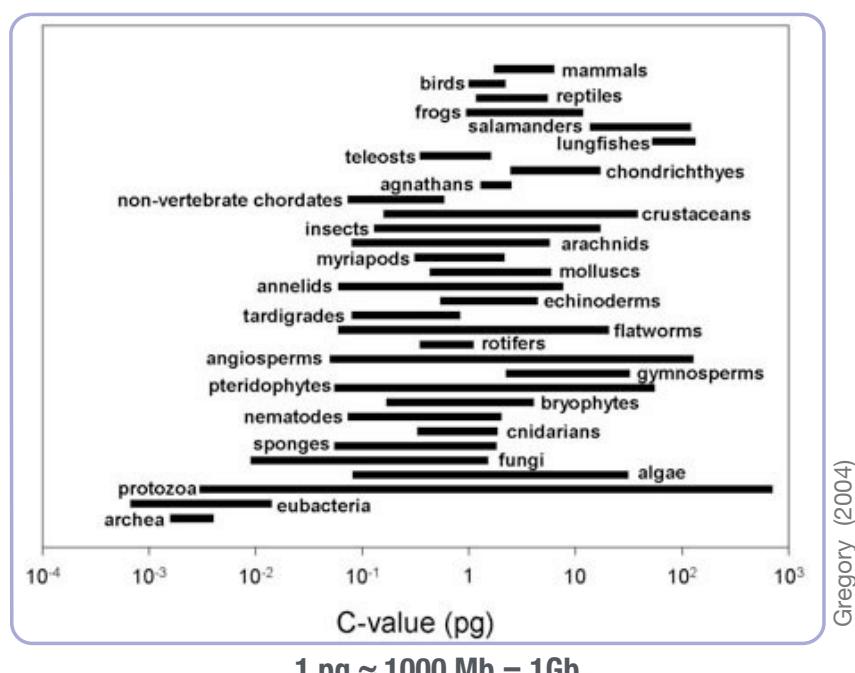
## Genome Size Evolution

<i>Caenorhabditis elegans</i>	100.26	21200
<i>Drosophila melanogaster</i>	137	16000
<i>Ciona intestinalis</i>	156	16000
<i>Anopheles gambiae</i>	278	13683
<i>Fugu rubripes</i>	365	38000
<i>Gallus gallus</i>	1050	21500
<i>Mus musculus</i>	2500	24000
<i>Homo sapiens</i>	2900	24000

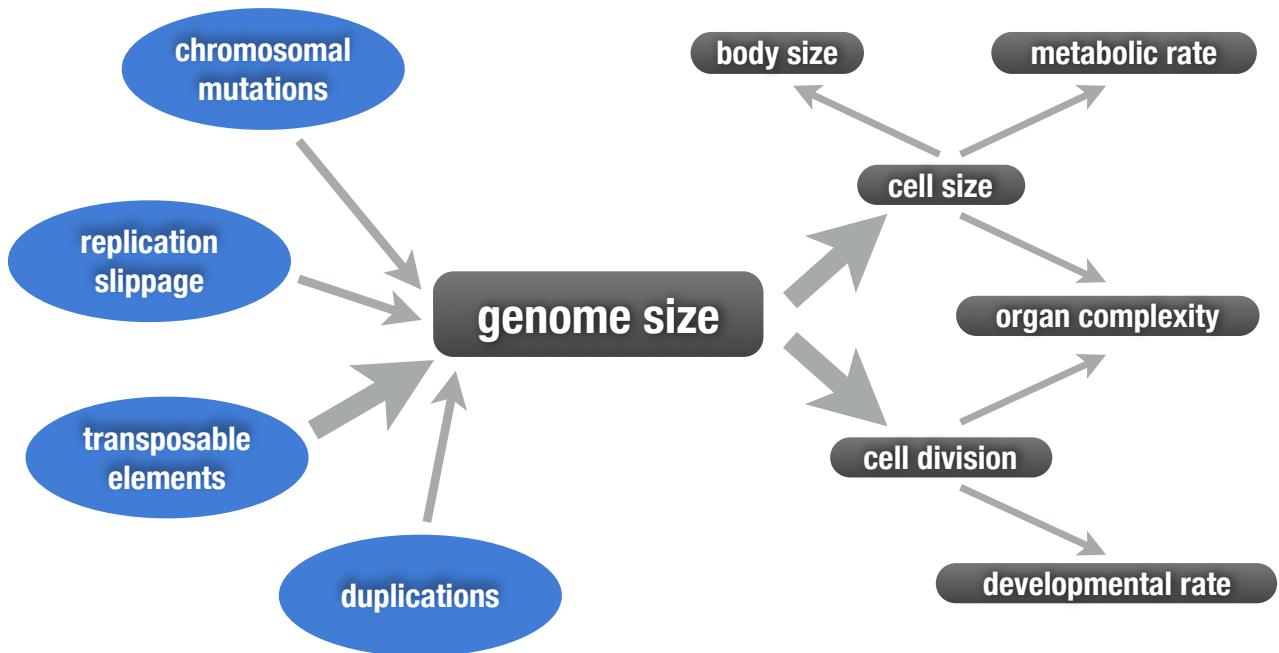
Lynch (2006)

## Genome Size Evolution

### “C-value enigma”



## Genome Size Evolution



Lynch (2007)

## Chromosomal Rearrangements

### Chromosomal mutations

- ▶ Mutations that affect entire chromosomes or large parts thereof are called **chromosomal mutations**
- ▶ The phenotypic effects of chromosome mutations are **difficult to generalize**
- ▶ Phenotypic consequences also arise, as gene expression is at least partly regulated by the relations between neighboring genes

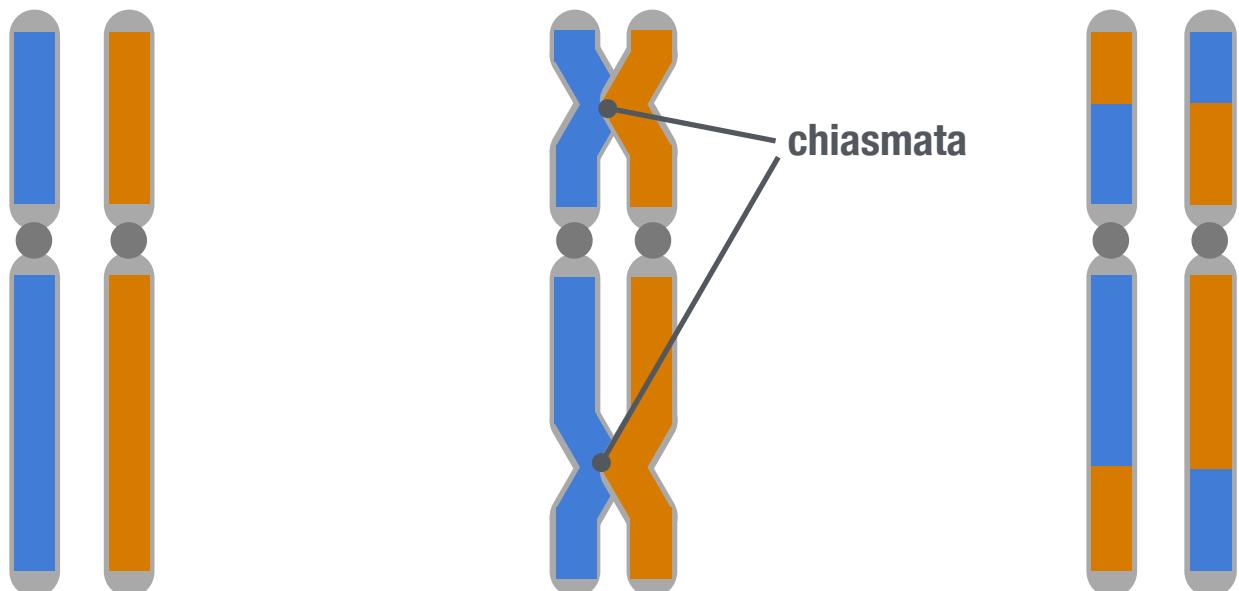
## Chromosomal Rearrangements

### Recombination

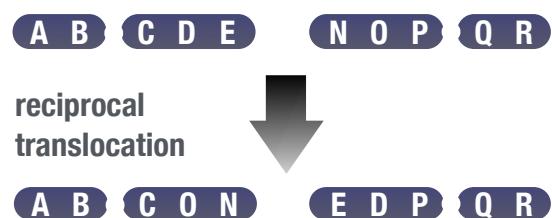
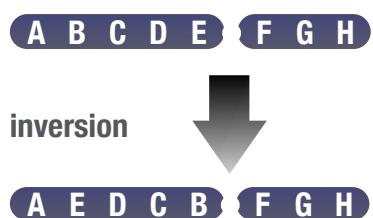
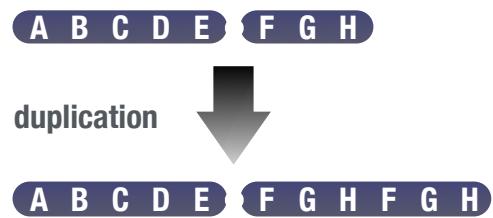
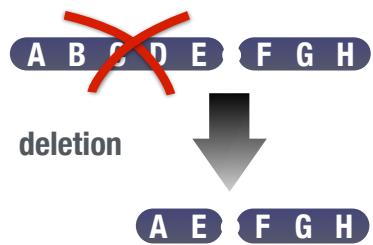
- ▶ ...occurs by the **crossing-over** of homologous chromosomes during **meiosis**
- ▶ ...leads to the **exchange of DNA** between a pair of chromosomes
- ▶ As a consequence, two previously unlinked genes may become linked or *vice versa*
- ▶ ...may lead to rearrangements

## Chromosomal Rearrangements

### Recombination

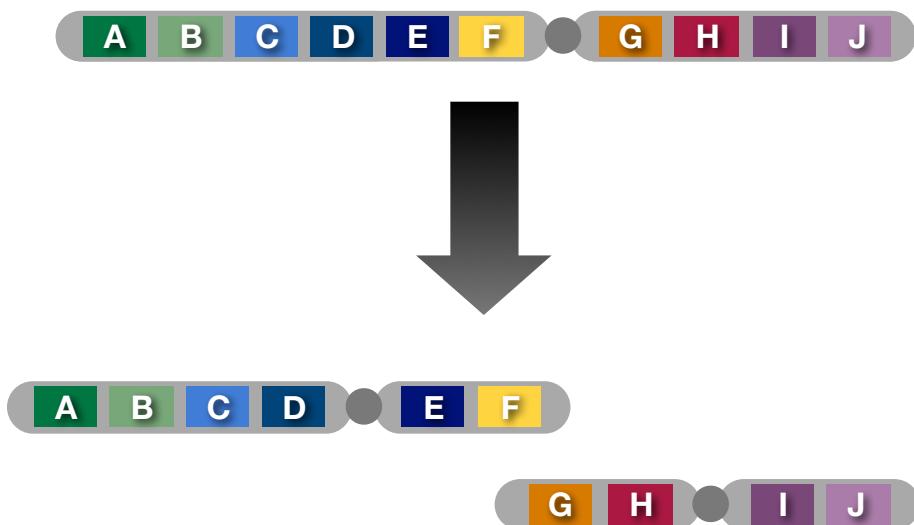


## Chromosomal Rearrangements



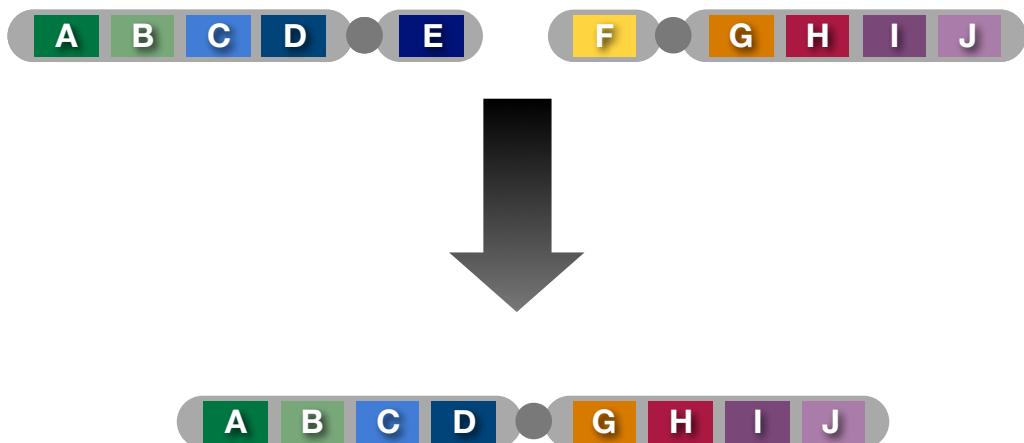
## Chromosomal Rearrangements

### Chromosomal Fission



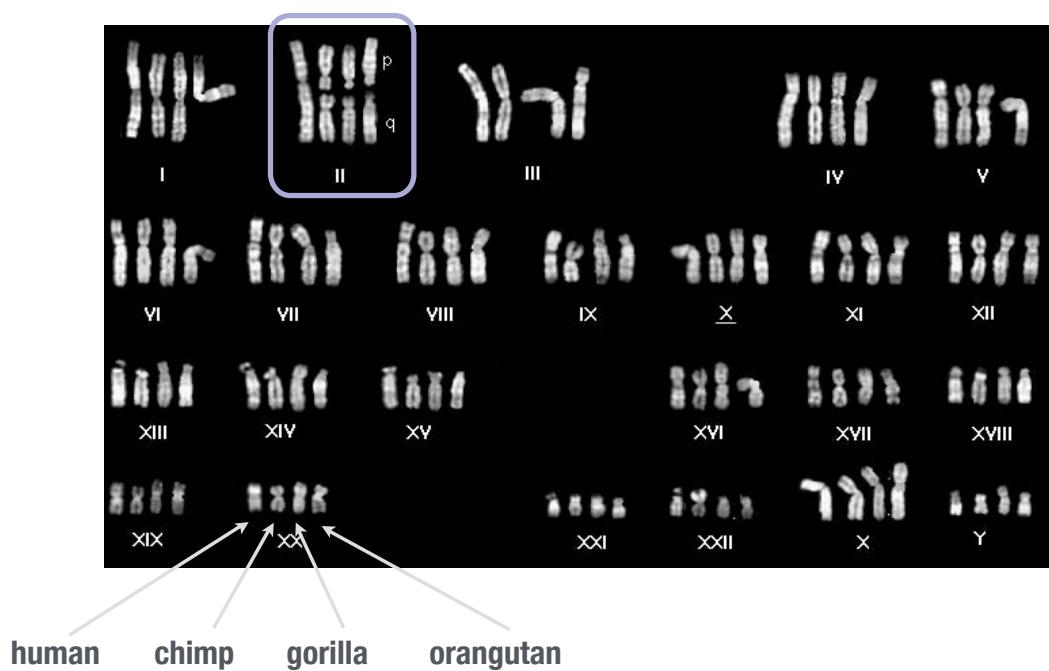
## Chromosomal Rearrangements

### Chromosomal Fusion



## Chromosomal Rearrangements

### Chromosomal fusion: human chromosome 2



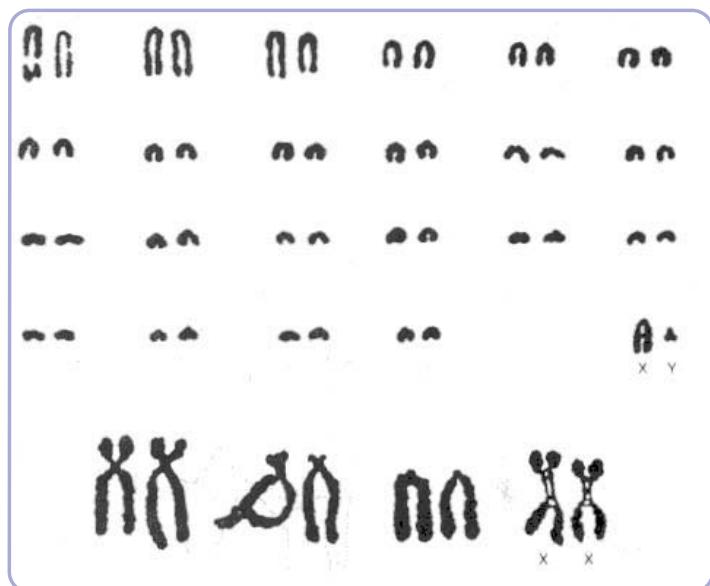
from Dr. M. Rocchi

## Chromosomal Rearrangements

### An extreme case: muntjac deers



www.wdr.de



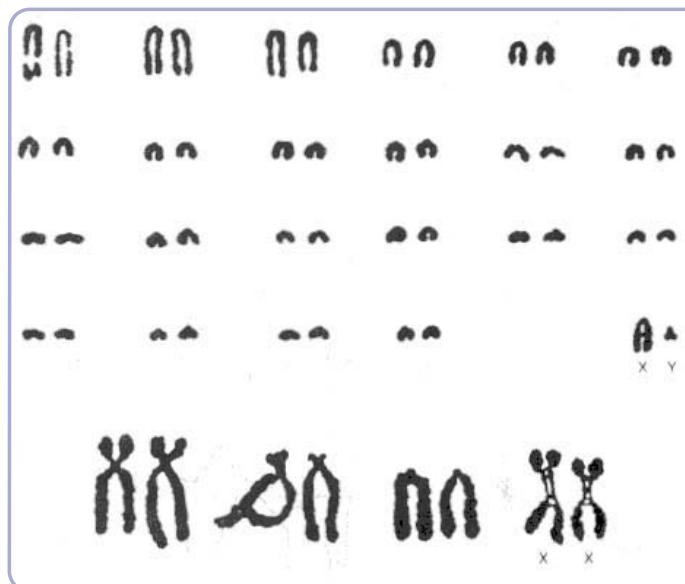
## Chromosomal Rearrangements

### An extreme case: muntjac deers

chinese muntjac deer

Translocation,  
chromosome fusion,  
and/or fission  
explain why these  
two very similar  
species of hoofed  
mammal have such  
different karyotypes.

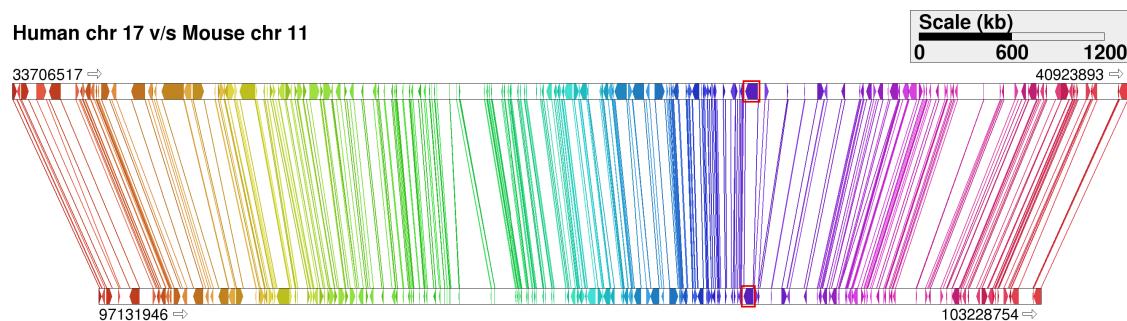
indian muntjac deer



# Chromosomal Rearrangements

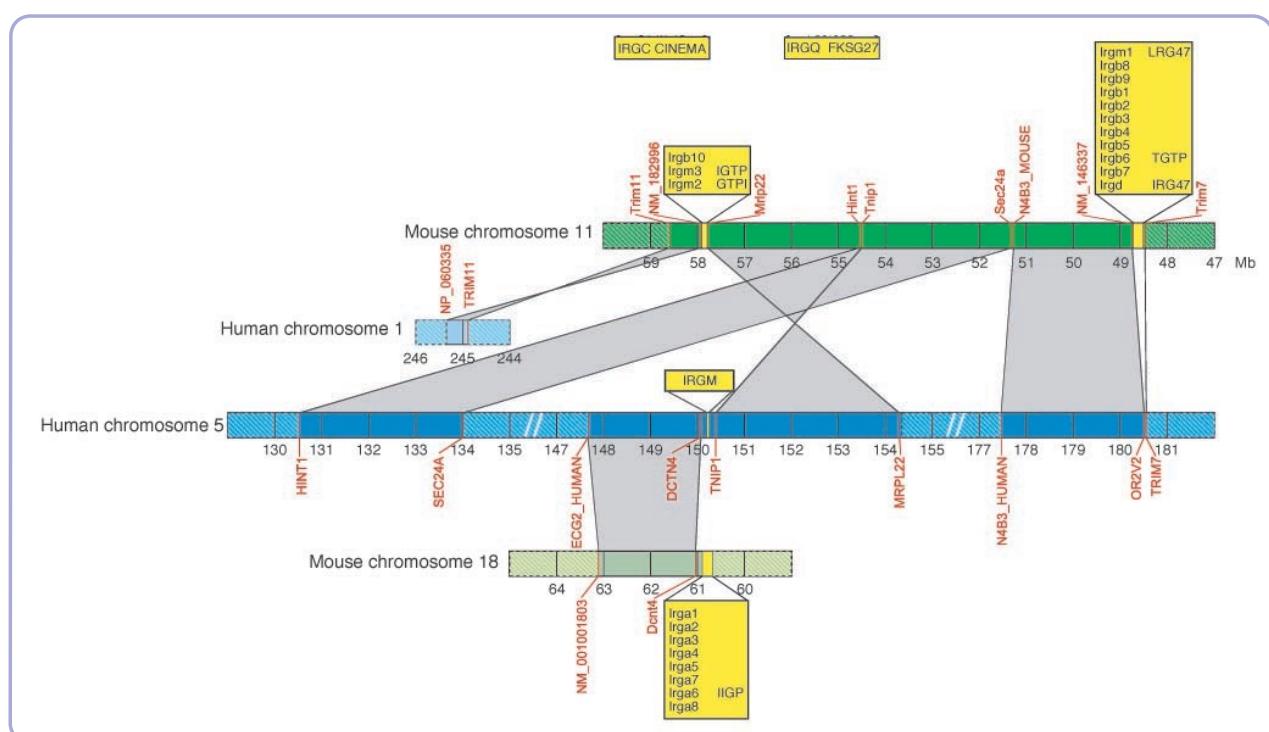
## Synteny

- In comparative genomic terms, synteny describes the **preserved order of genes** on chromosomes as a result of common ancestry



# Chromosomal Rearrangements

## Synteny



# Gene and Genome Duplications

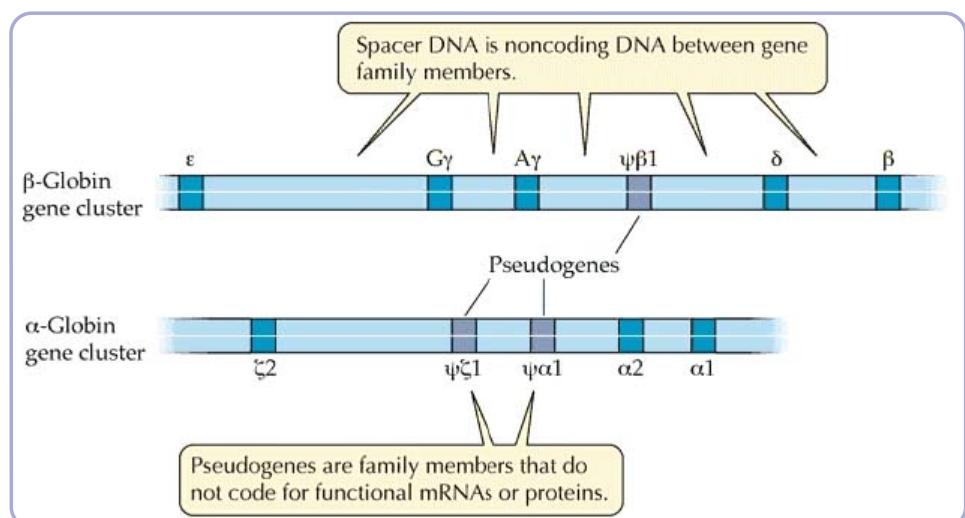
## Gene duplication

- ▶ A gene can be duplicated by **various kinds of mechanisms**, e.g., genome duplications, chromosome mutations, unequal crossing over, etc.
- ▶ Any such duplication will be rare initially but may increase its frequency by **natural selection or random drift**.
- ▶ Duplicated genes may undergo different **evolutionary fates** such as non-functionalization, neo-functionalization or sub-functionalization.

# Gene and Genome Duplications

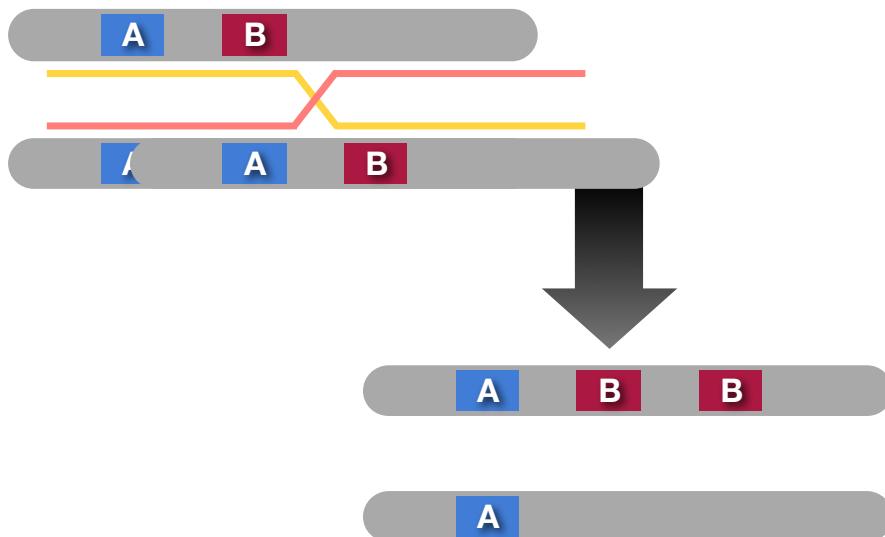
## Gene clusters

- ▶ ...is a set of two or more genes of common ancestry that encode similar products



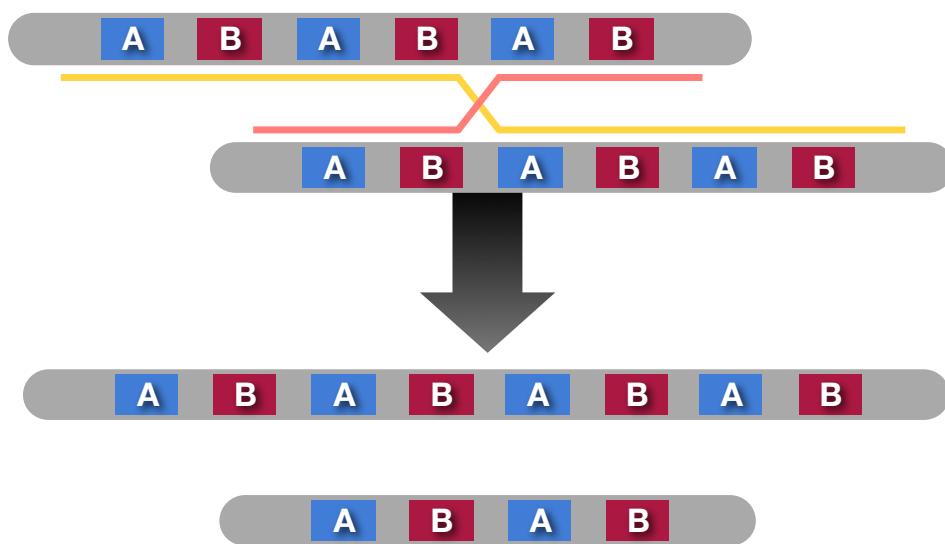
## Gene and Genome Duplications

### Unequal crossing over: misalignment



## Gene and Genome Duplications

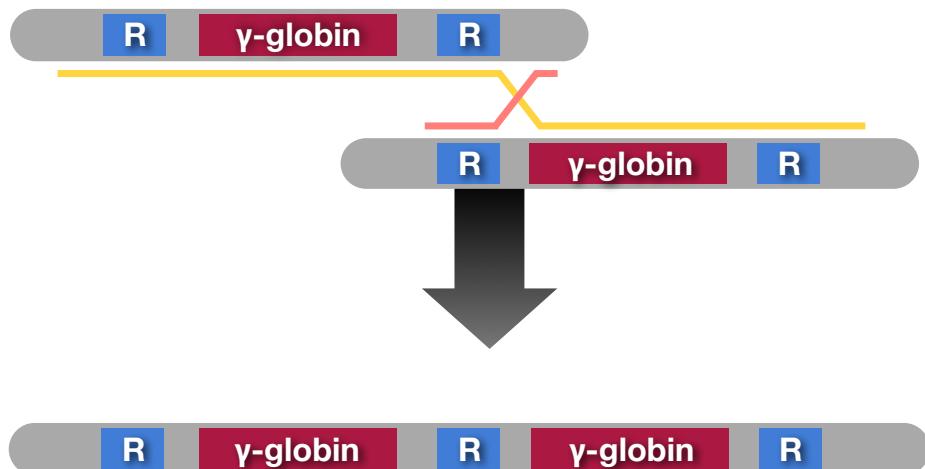
### Unequal crossing over: misalignment



Misalignment is more likely when several copies of similar sequences are already present

# Gene and Genome Duplications

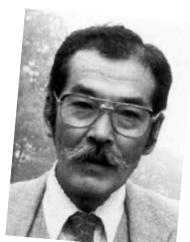
## Unequal crossing over: misalignment



Unequal crossing over is likely at repetitive sequence motives

# Gene and Genome Duplications

## Genome duplication

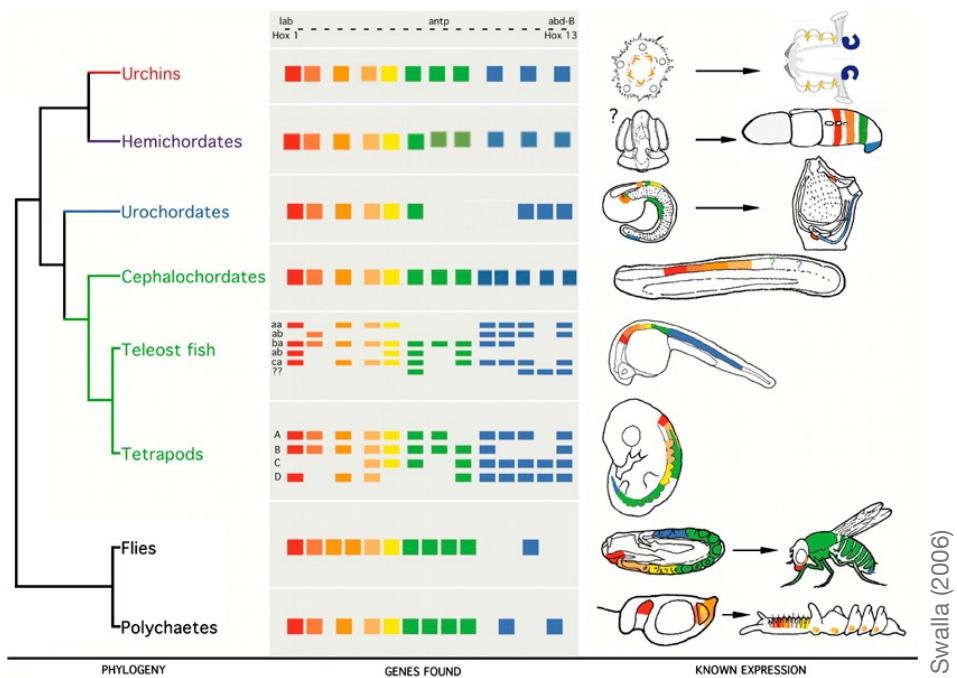


Sosumo Ohno  
(1928-2000)

- ▶ Sosumo Ohno (1970):  
*Evolution by Gene Duplication*
- ▶ Ohno also postulated that whole genome duplications exist and argued that the whole genome had duplicated twice near the origin of the vertebrates (**2R hypothesis**)
- ▶ Many more genome duplication events have been suggested since, e.g., the fish-specific genome duplication (**3R hypothesis**)

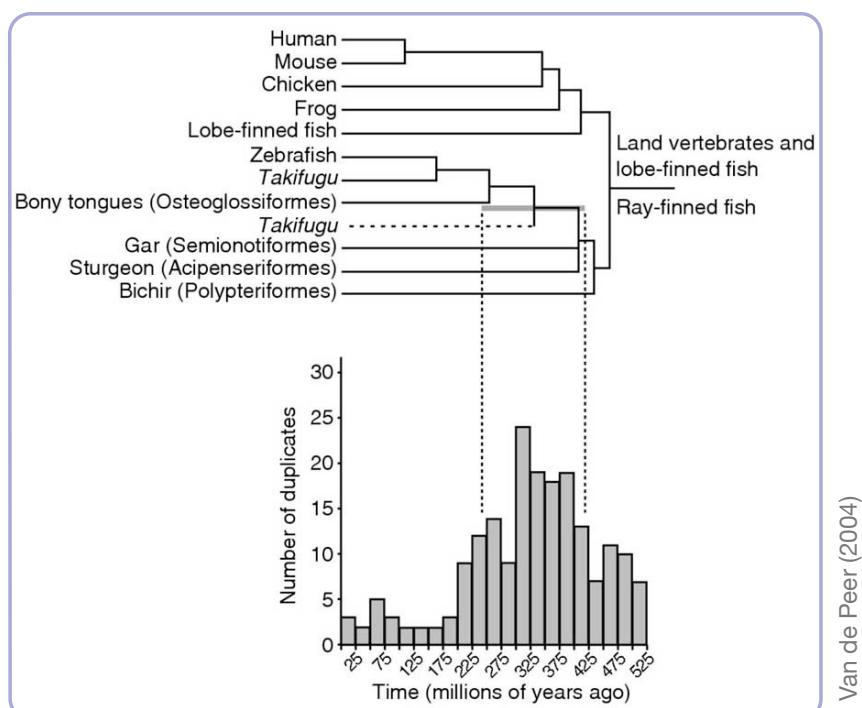
# Gene and Genome Duplications

## Hox gene clusters



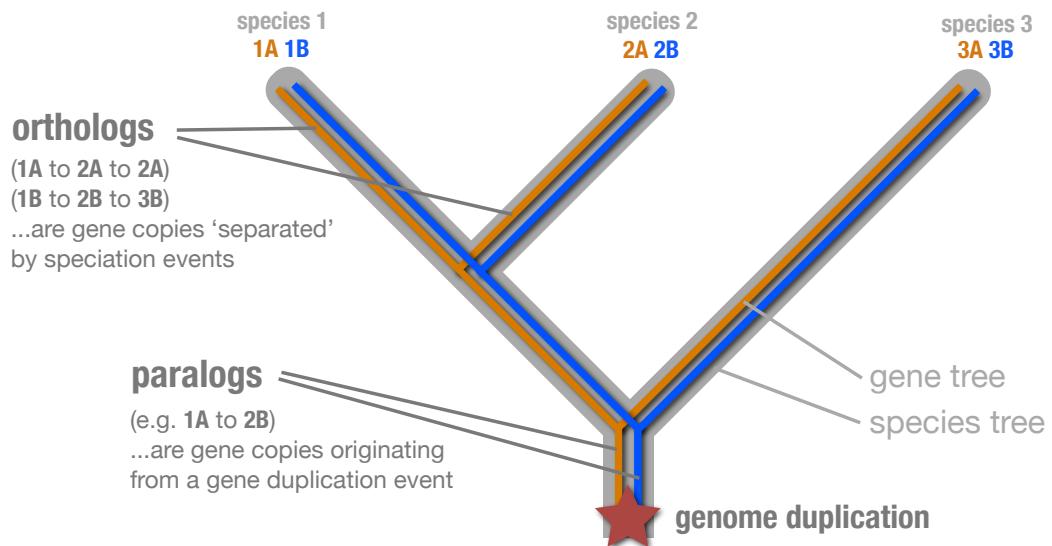
# Gene and Genome Duplications

## fish specific genome duplication



# Gene and Genome Duplications

## paralogs, orthologs and ohnologs

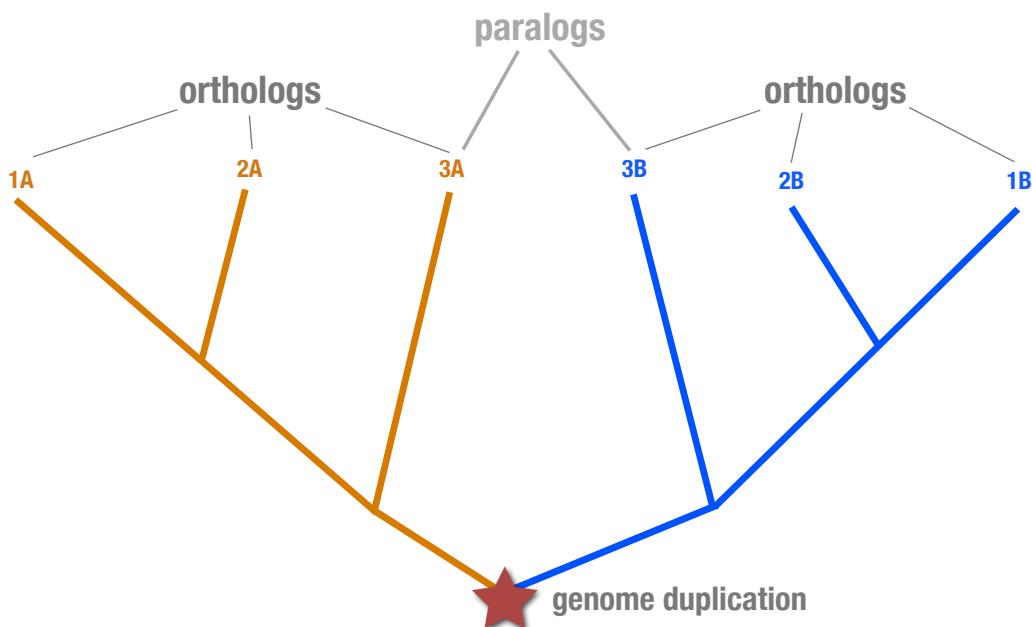


## ohnologs

...are paralogs that go back to the same whole genome duplication event

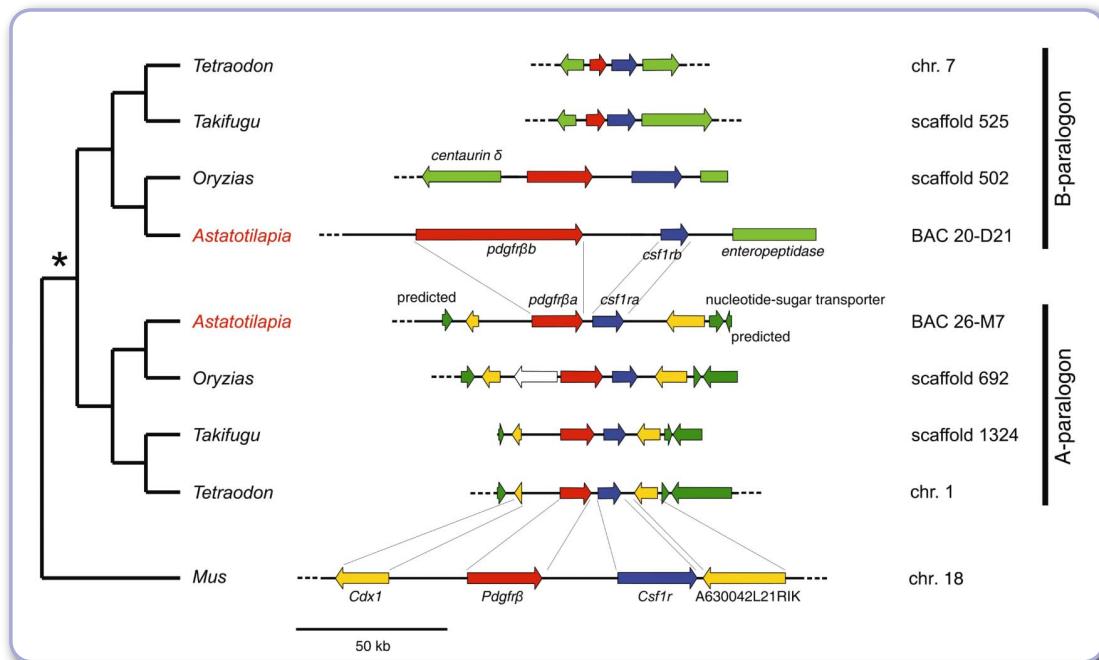
# Gene and Genome Duplications

## paralogs, orthologs and ohnologs



# Gene and Genome Duplications

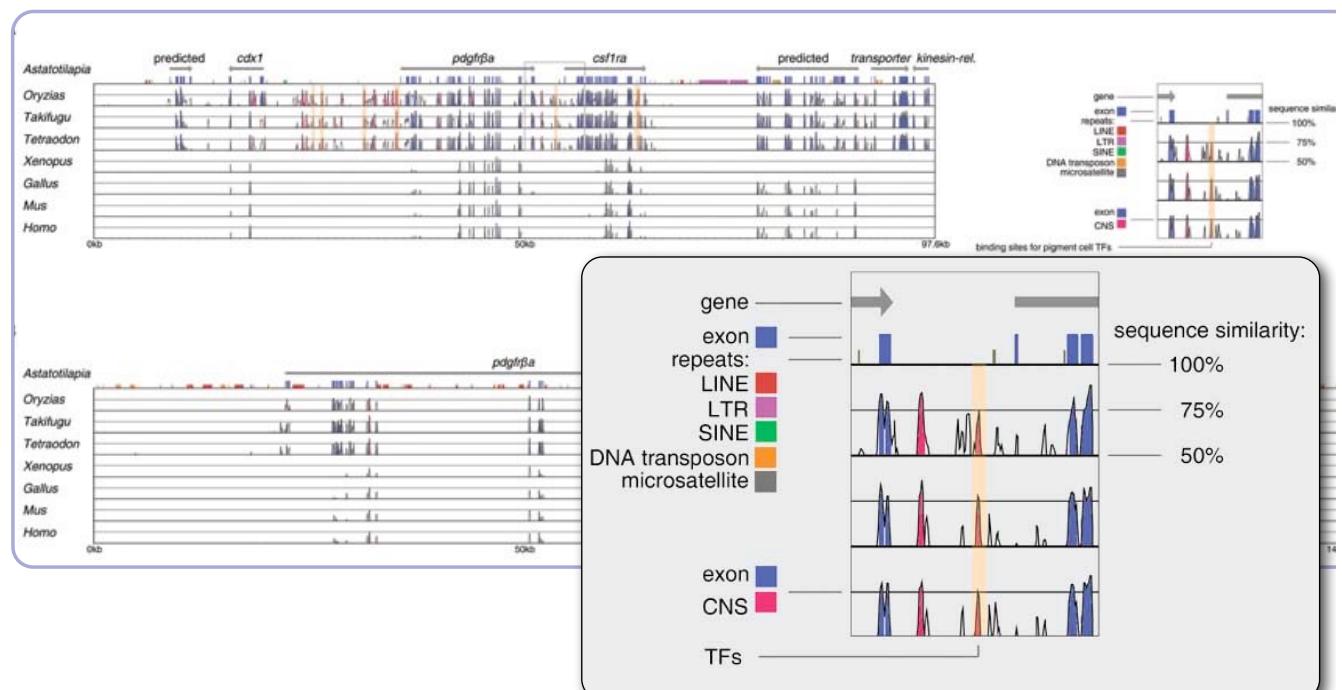
## 'paralogons'



I Braasch, W Salzburger & A Meyer (2006) Molecular Biology and Evolution

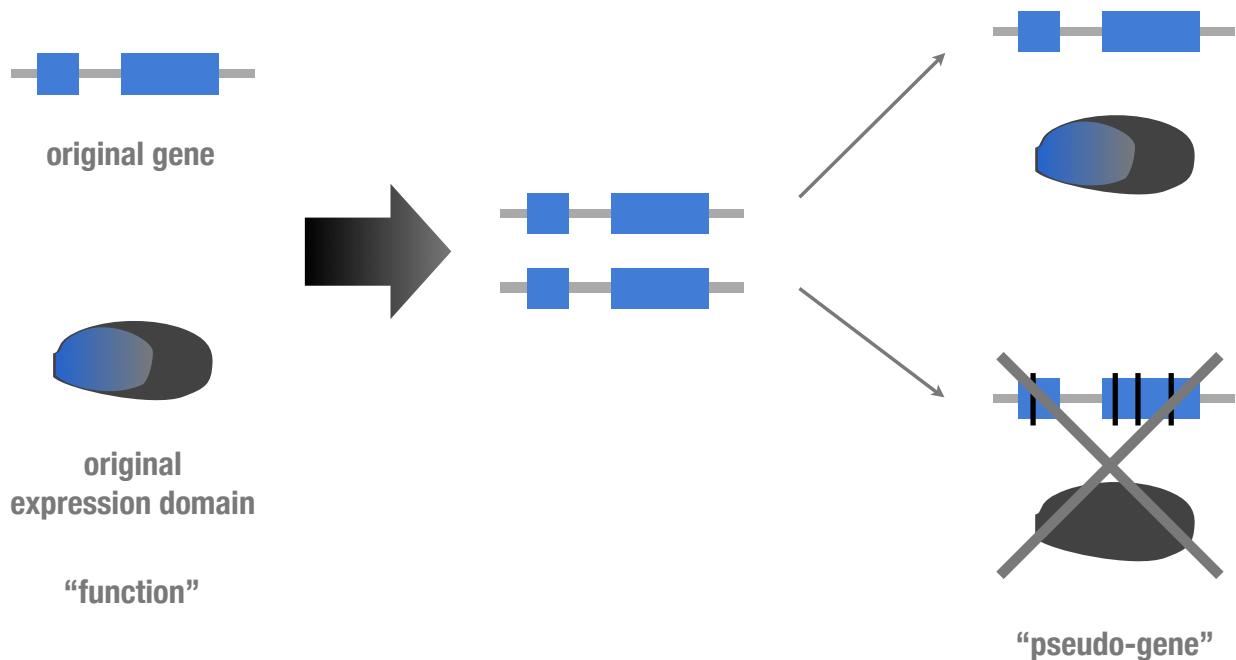
# Gene and Genome Duplications

## 'vista' plots



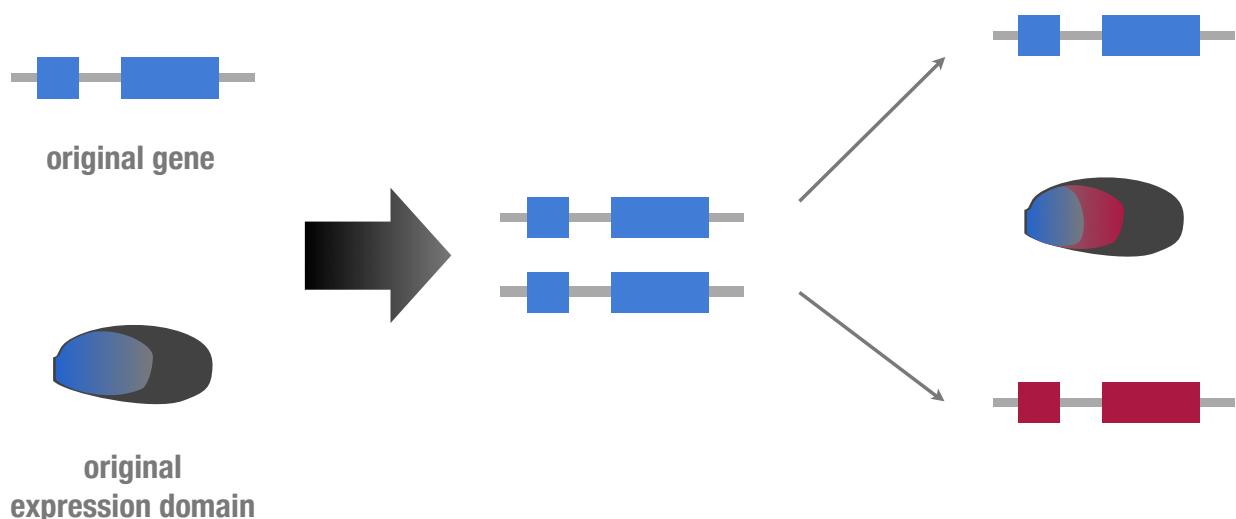
## Gene and Genome Duplications

### non-functionalization

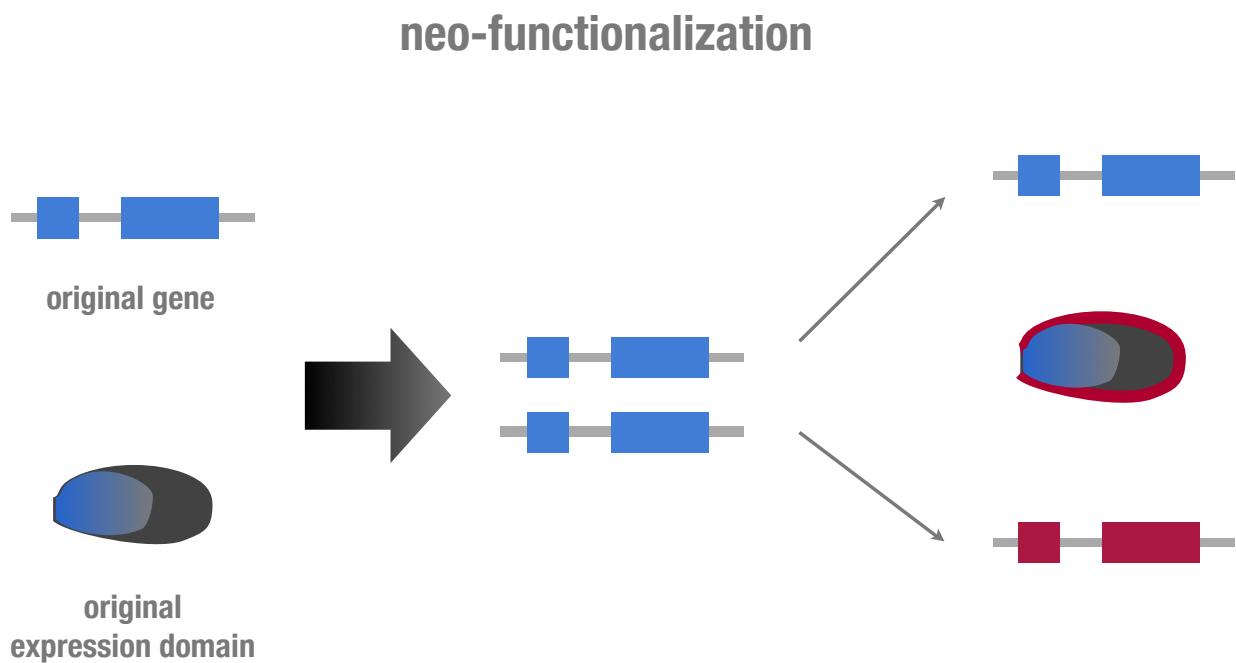


## Gene and Genome Duplications

### sub-functionalization

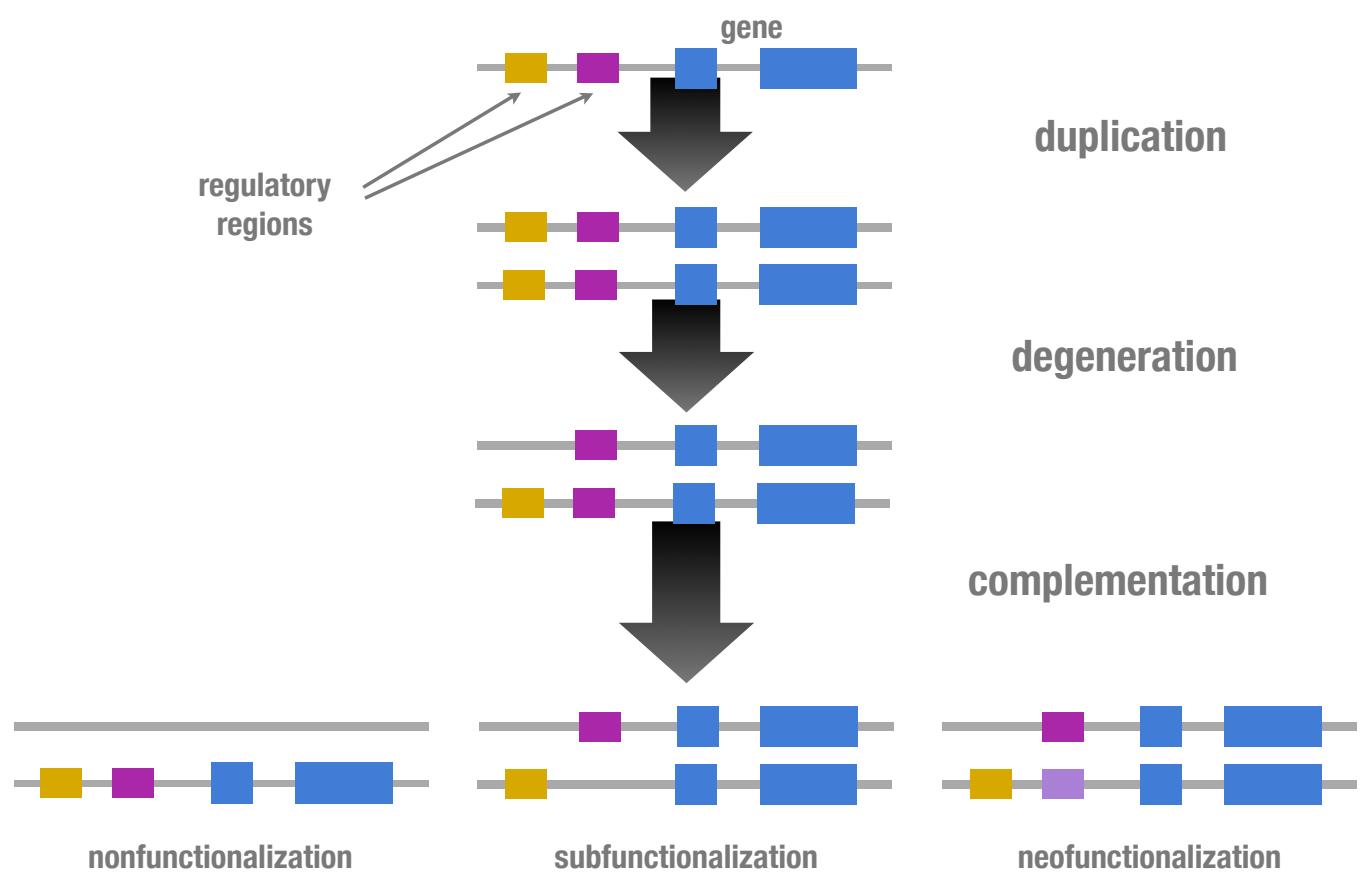


## Gene and Genome Duplications



## Gene and Genome Duplications

### DDC model

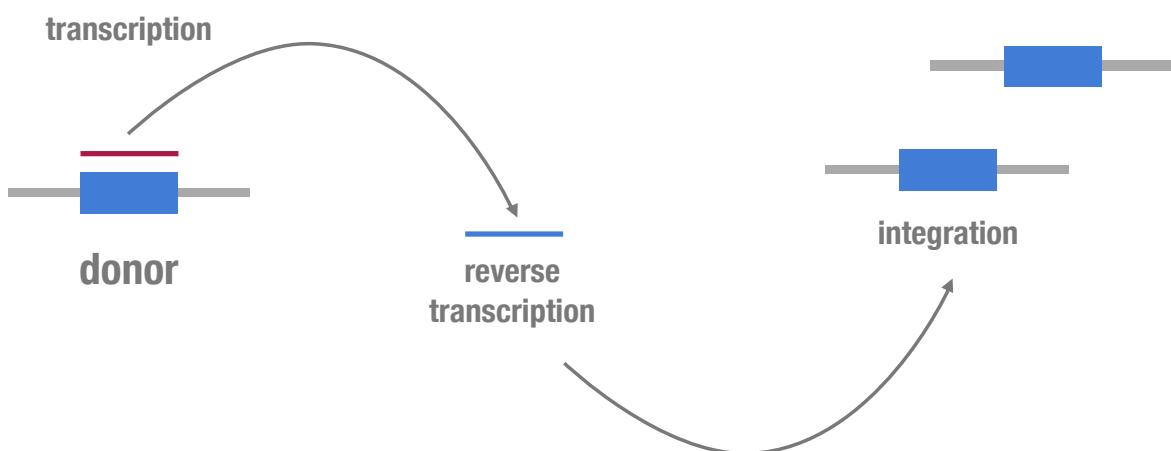


## Transposable Elements

### TEs

- ▶ Transposable elements (TEs) are discrete DNA sequences that move from one location to another within the genome
- ▶ TEs were discovered by Barbara McClintock (1902-1992) in the 1940s and 1950s. She received the Nobel Prize in 1983.
- ▶ TEs are found in nearly all species and constitute a large fraction of some genomes, including the human genome
- ▶ TEs can generate variation in the host genome

## Transposable Elements



## Transposable Elements

### “retrotransposons”

#### LTRs

These transposable genetic elements are characterized by flanking **long terminal repeats**. LTRs are similar to retroviruses and contain a group specific antigen (*gag*).

#### LINEs

**Long interspersed nuclear elements** are autonomous retrotransposons. LINEs have two open reading frames (ORFs); ORF2 encodes for a reverse transcriptase (rvt)

#### SINEs

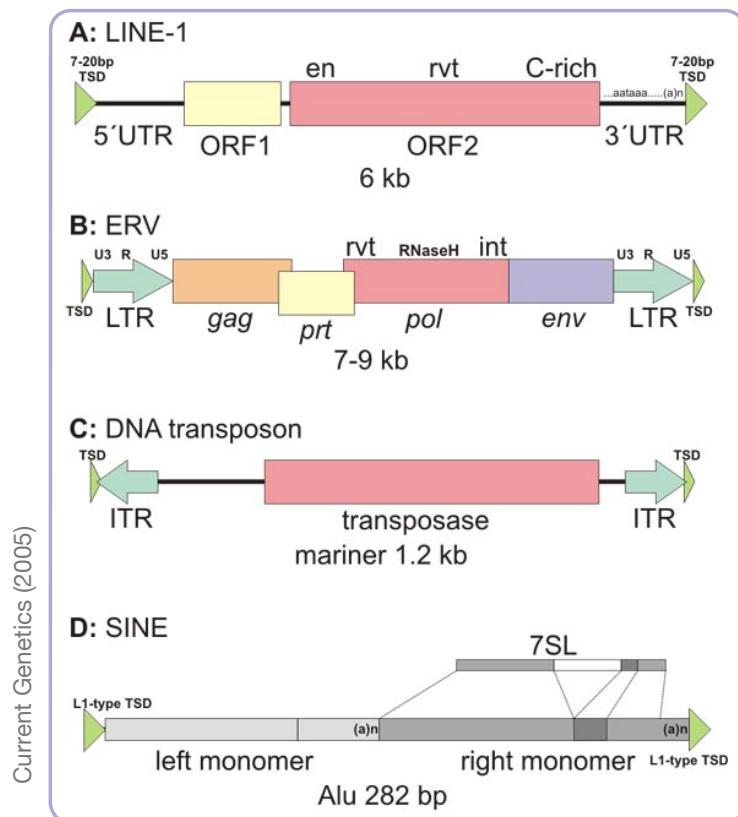
**Short interspersed nuclear elements** are non-autonomous retrotransposons that exploit the enzymatic retrotransposition machinery of LINEs

## Transposable Elements

### DNA transposons

- ▶ DNA transposons follow a different way of transposition and do not use an RNA stage and reverse transcription
- ▶ There are two classes of DNA transposons:
  - ▶ The majority of DNA transposons use a **cut-and-paste** mechanism
  - ▶ The other group uses a **rolling circle** (RC) mechanism

# Transposable Elements



# Transposable Elements

	yeast	slime mold	<i>C. elegans</i>	<i>Arabidopsis</i>	human
LTRs	3.1	4.4	0.1	6.4	7.9
SINEs, LINEs	0	3.7	0.4	0.7	31.2
DNA transposons	0	1.5	5.3	6.8	2.8
<b>total</b>	<b>3.1</b>	<b>9.6</b>	<b>6.5</b>	<b>14</b>	<b>44.8</b>

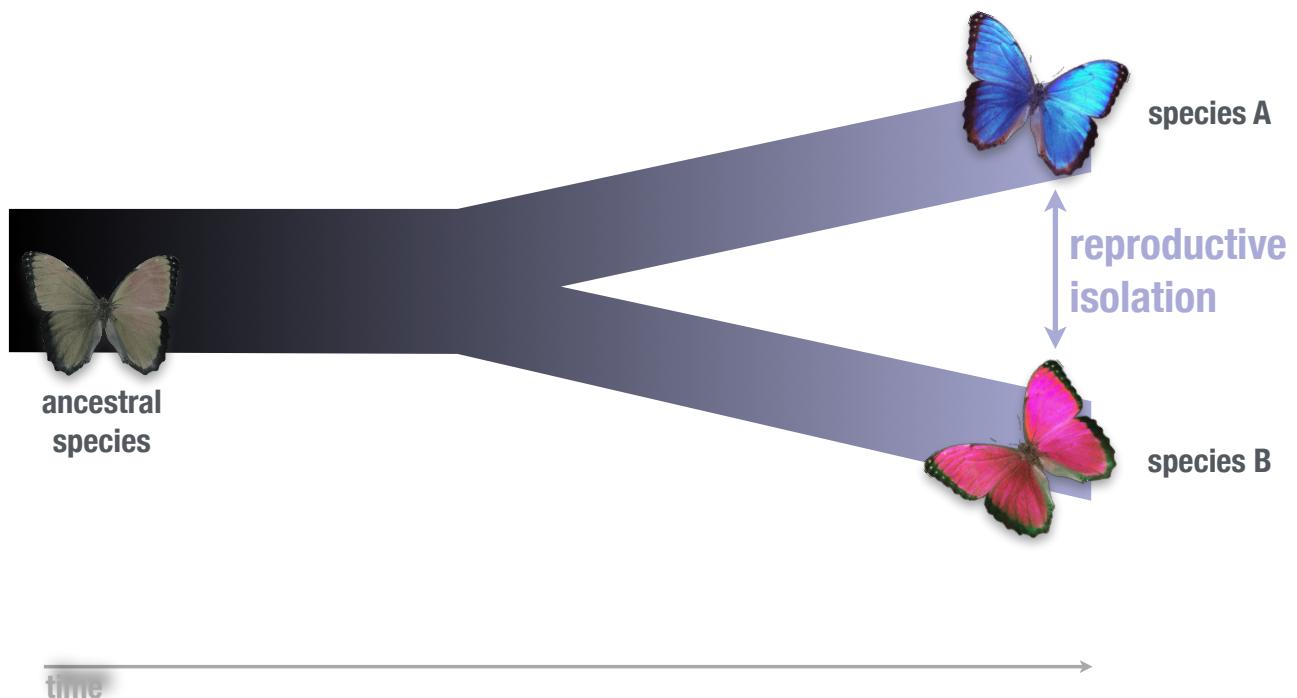
in % of genomes

Kidwell (2005)

# Speciation Genomics

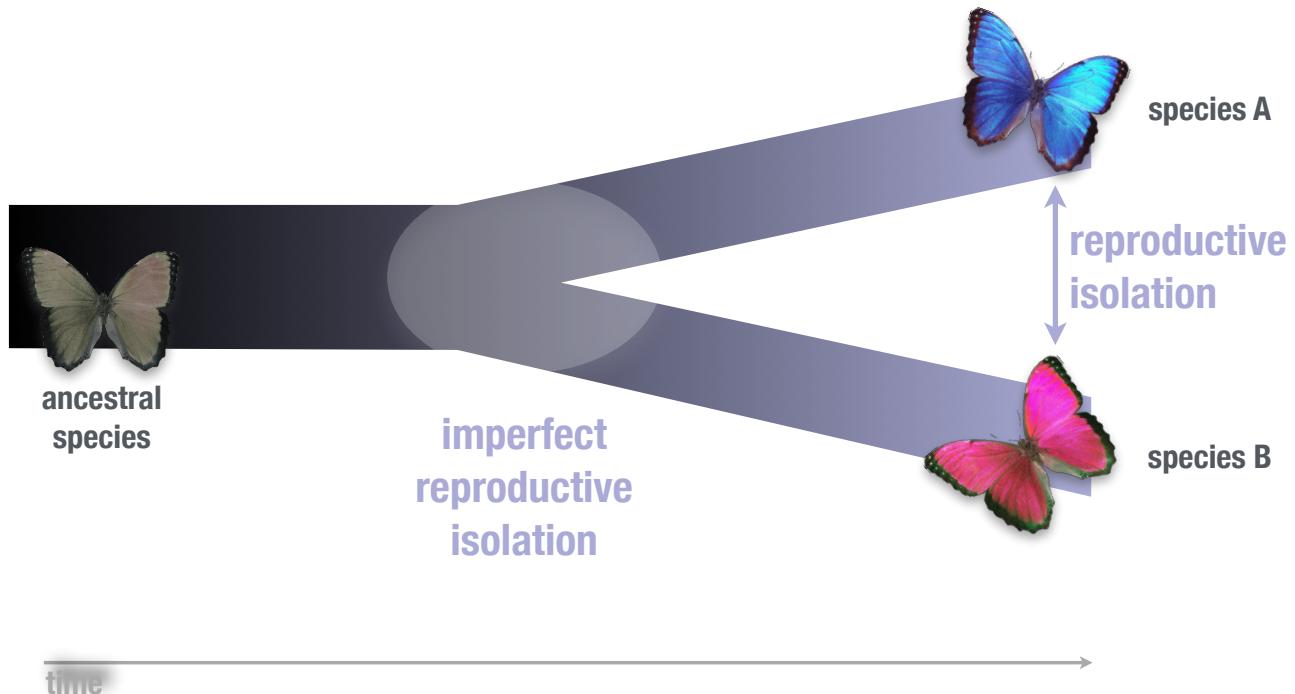


## Speciation Genomics

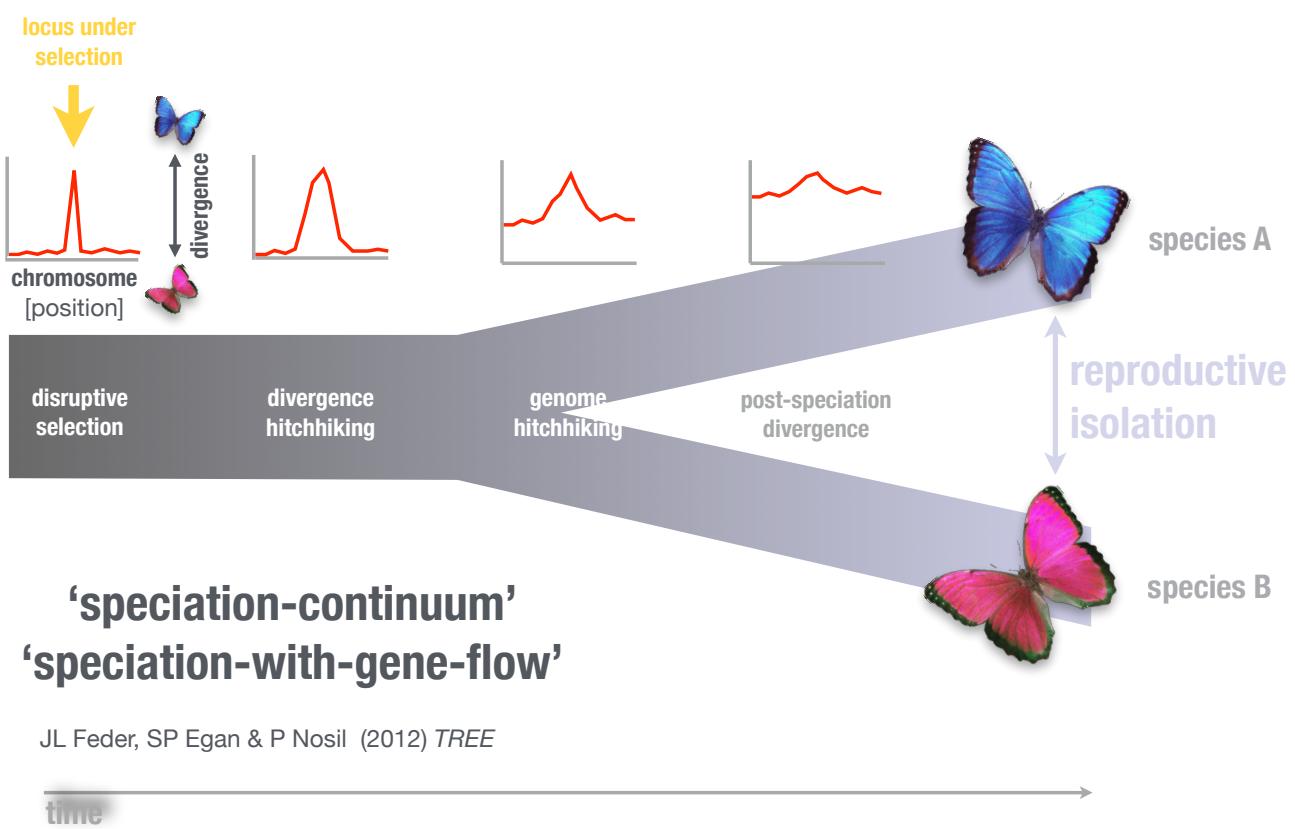


time →

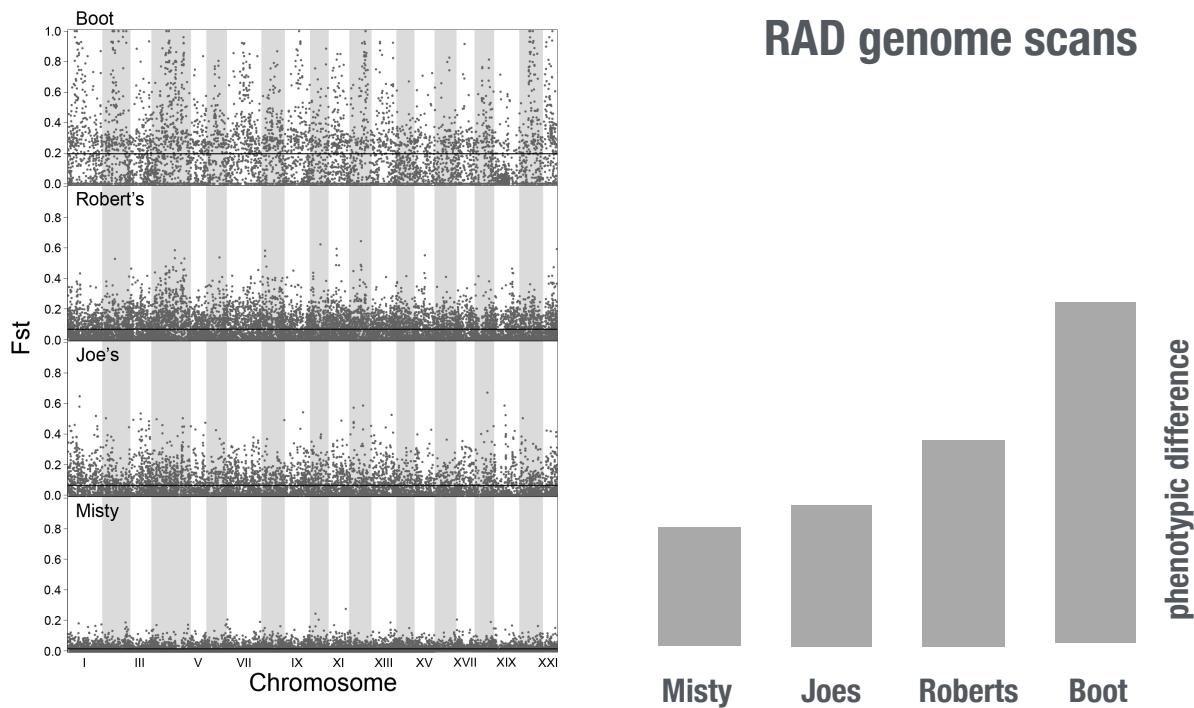
## Speciation Genomics



## Speciation Genomics

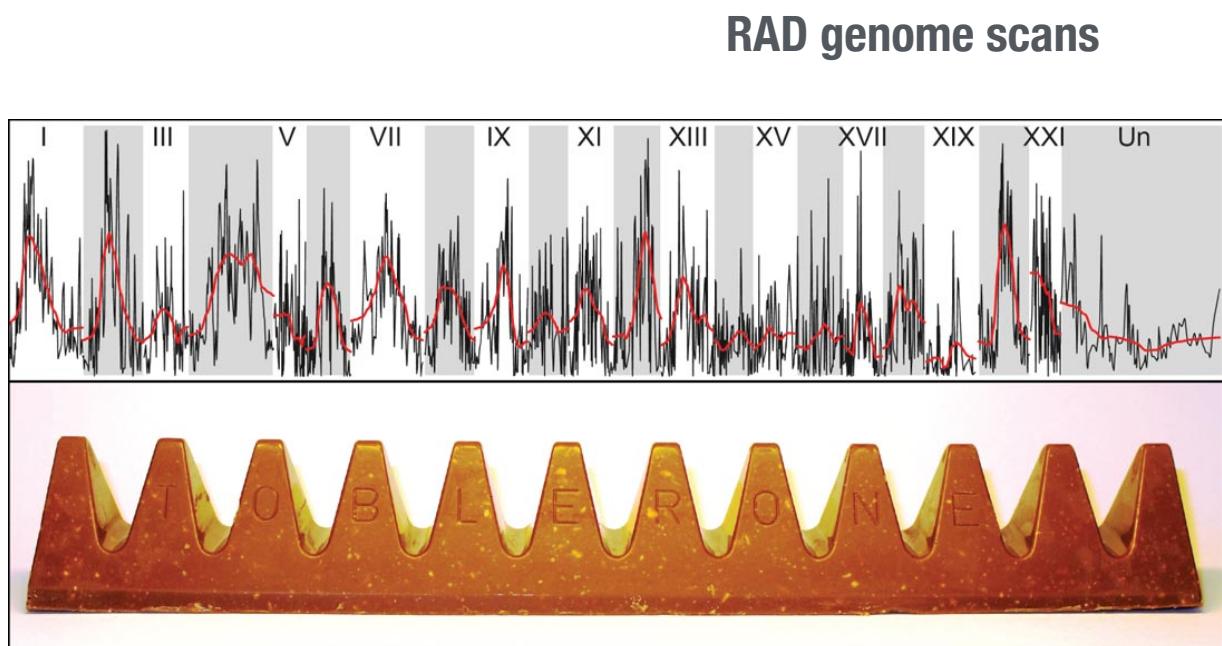


## Speciation Genomics:



••• M Roesti, A Hendry, W Salzburger & D Berner (2012) Molecular Ecology

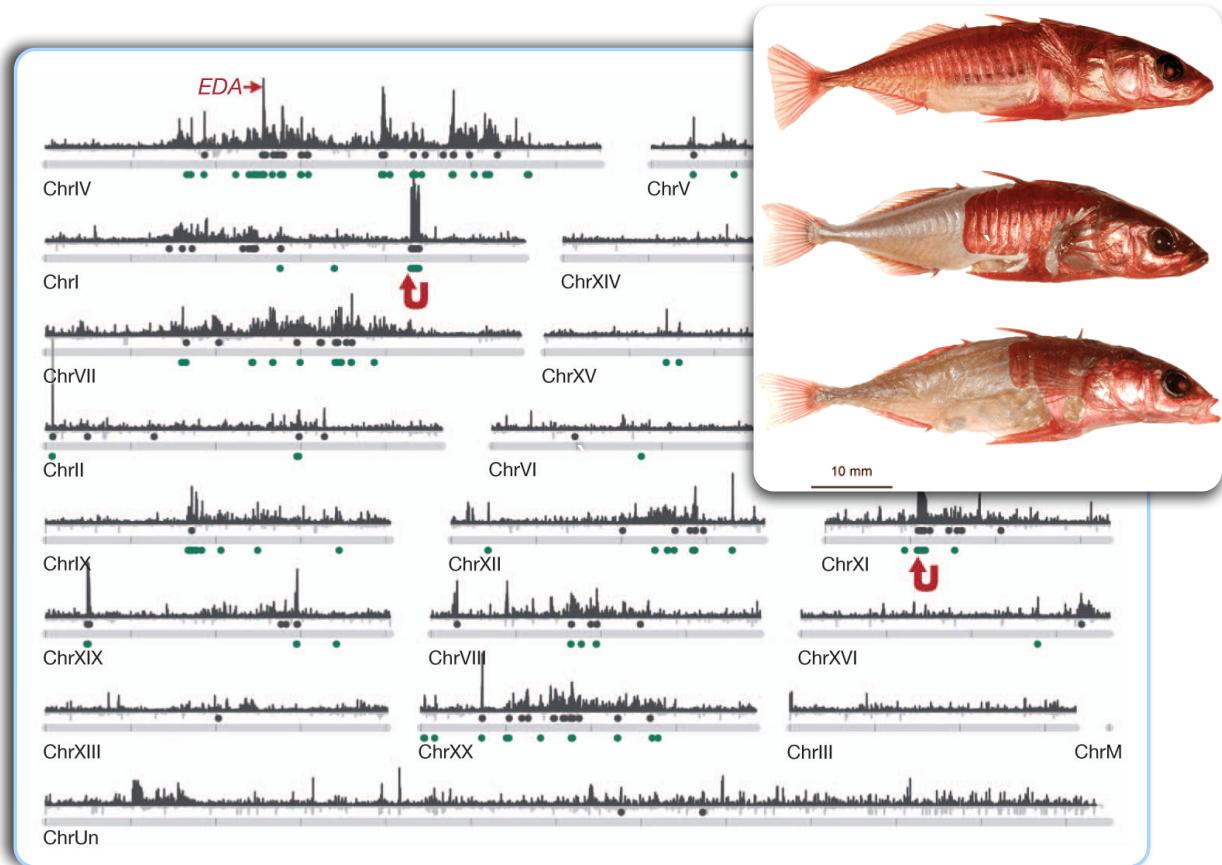
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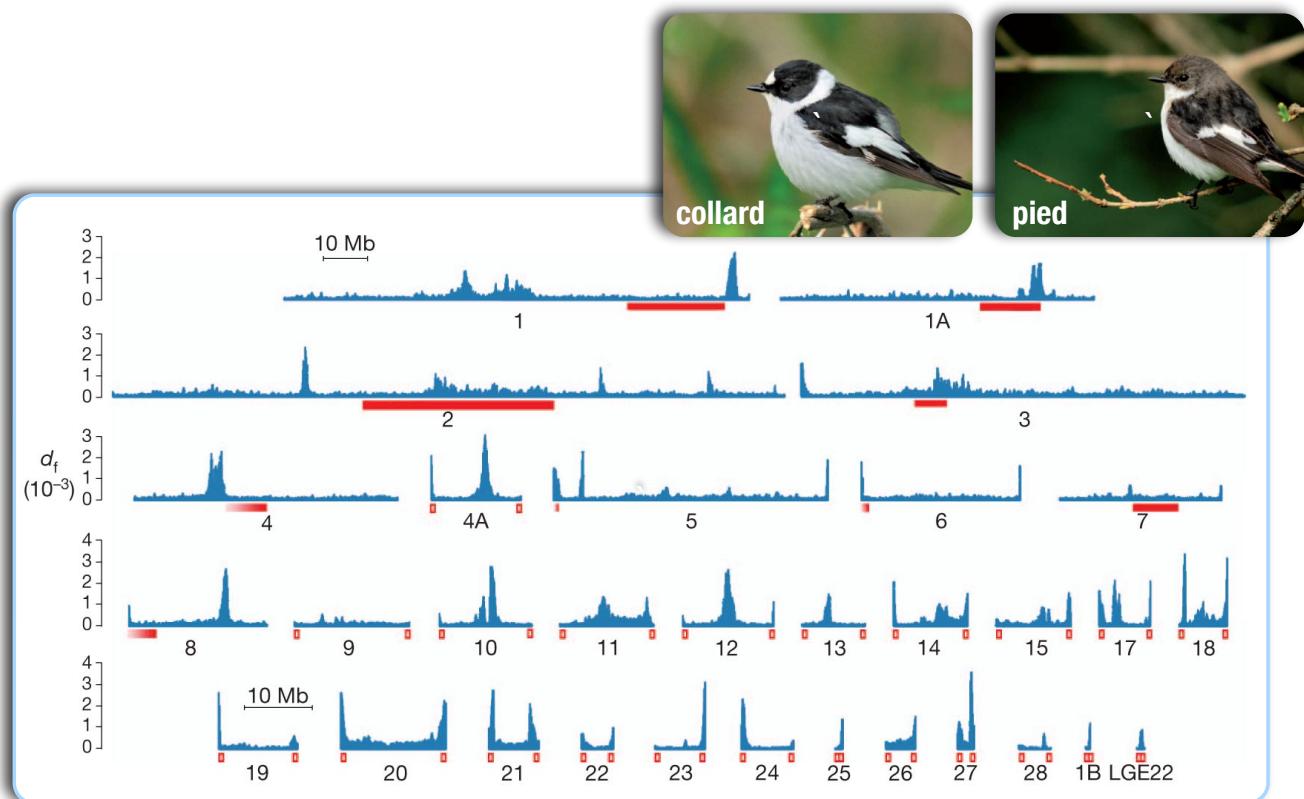
## Speciation Genomics:

Barrett et al. (2008) *Science*



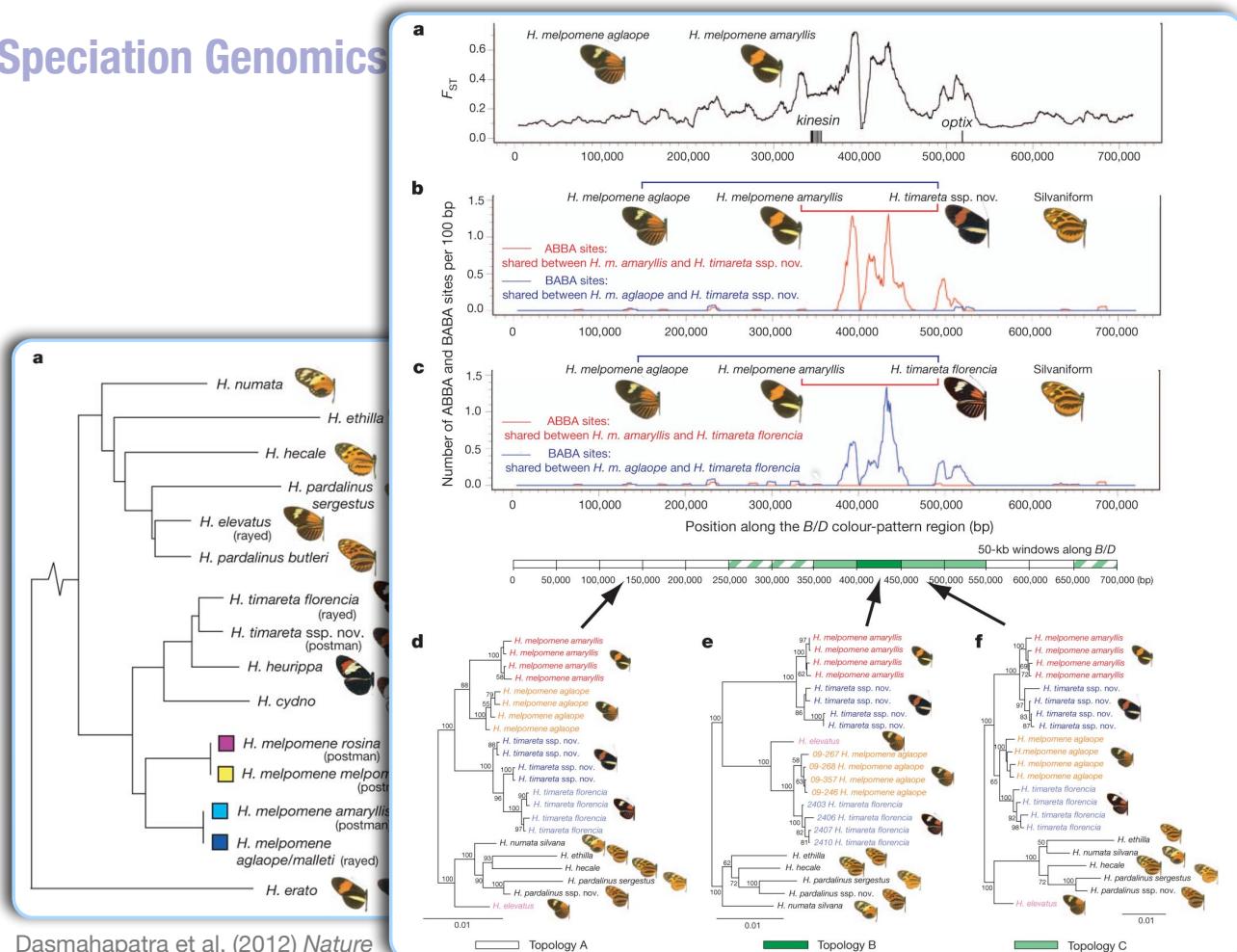
Jones et al. (2012) *Nature*

## Speciation Genomics:



Ellegren et al. (2012) *Nature*

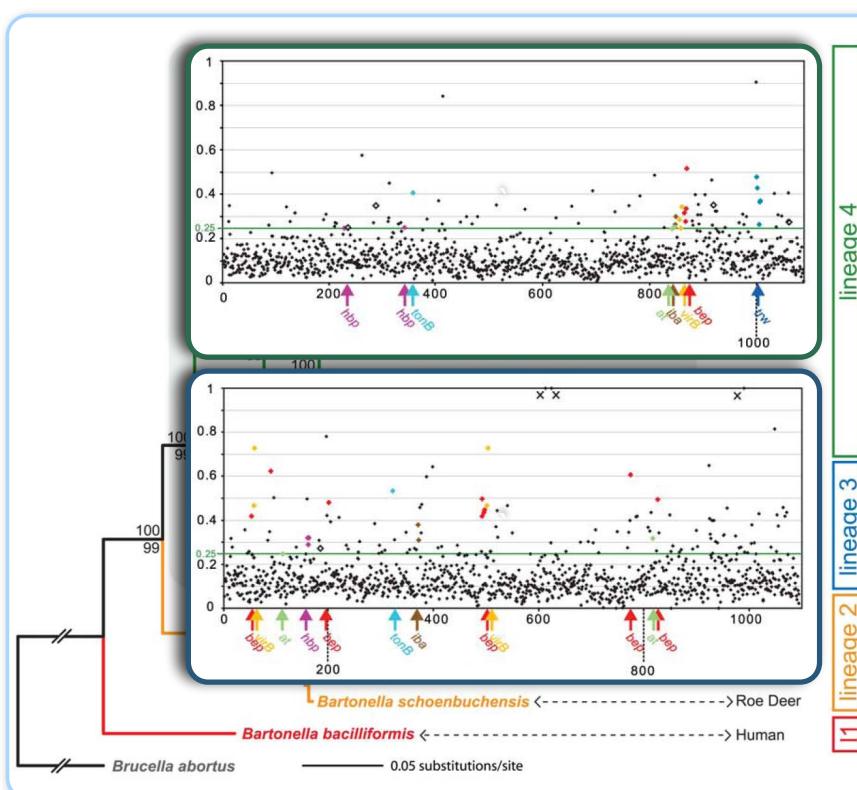
# Speciation Genomics



Dasmahapatra et al. (2012) *Nature*

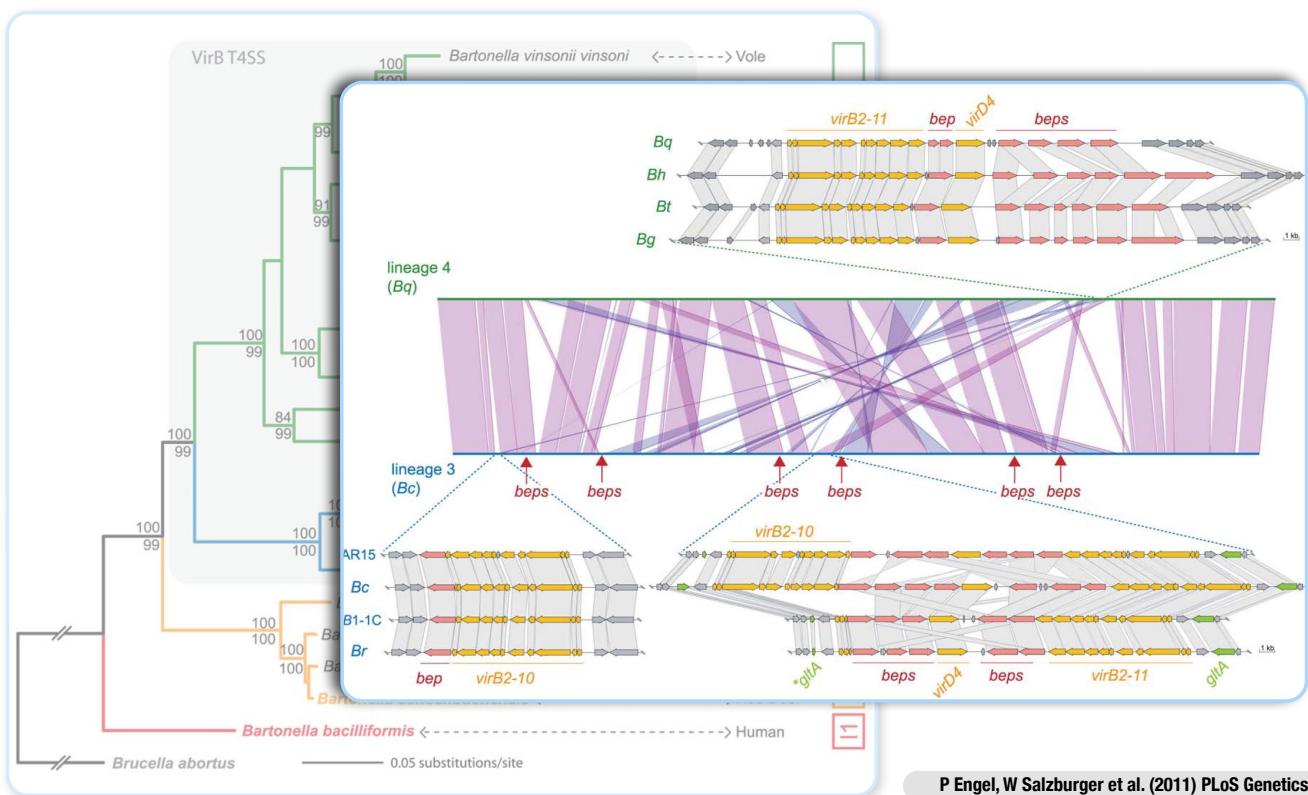
# Speciation Genomics

## pathogen evolution



P Engel, W Salzburger et al. (2011) PLoS Genetics

# Speciation Genomics



# Speciation Genomics

