

Genome

Walter Salzburger

Zoological Institute
University of Basel, Switzerland



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 doscoideum</i>	34	9000
<i>Arabidopsis thaliana</i>	125	25498
<i>Oryza sativa</i>	466	60256
<i>Lotus japonicus</i>	472	26000

Lynch (2006)

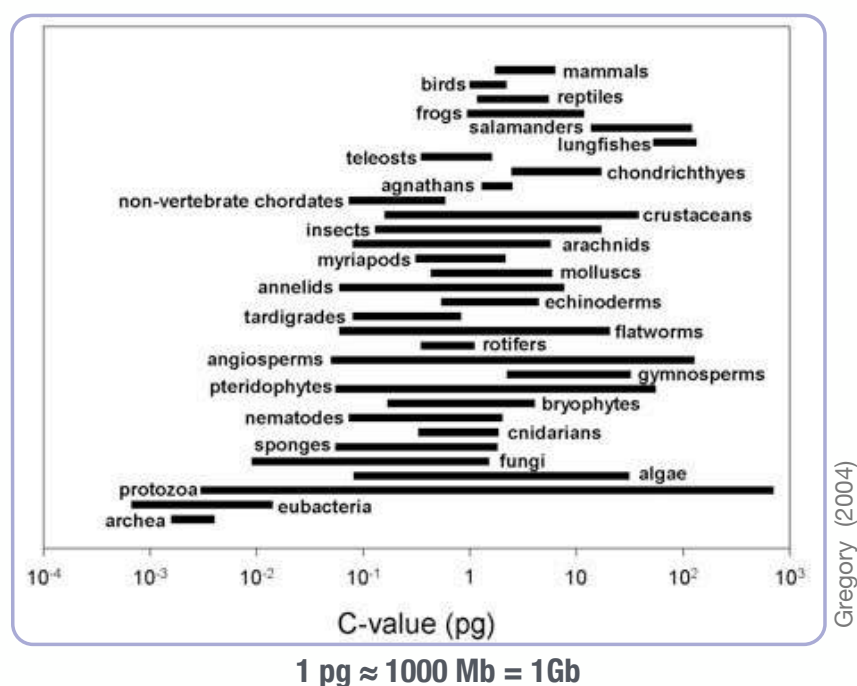
Genome Size Evolution

<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

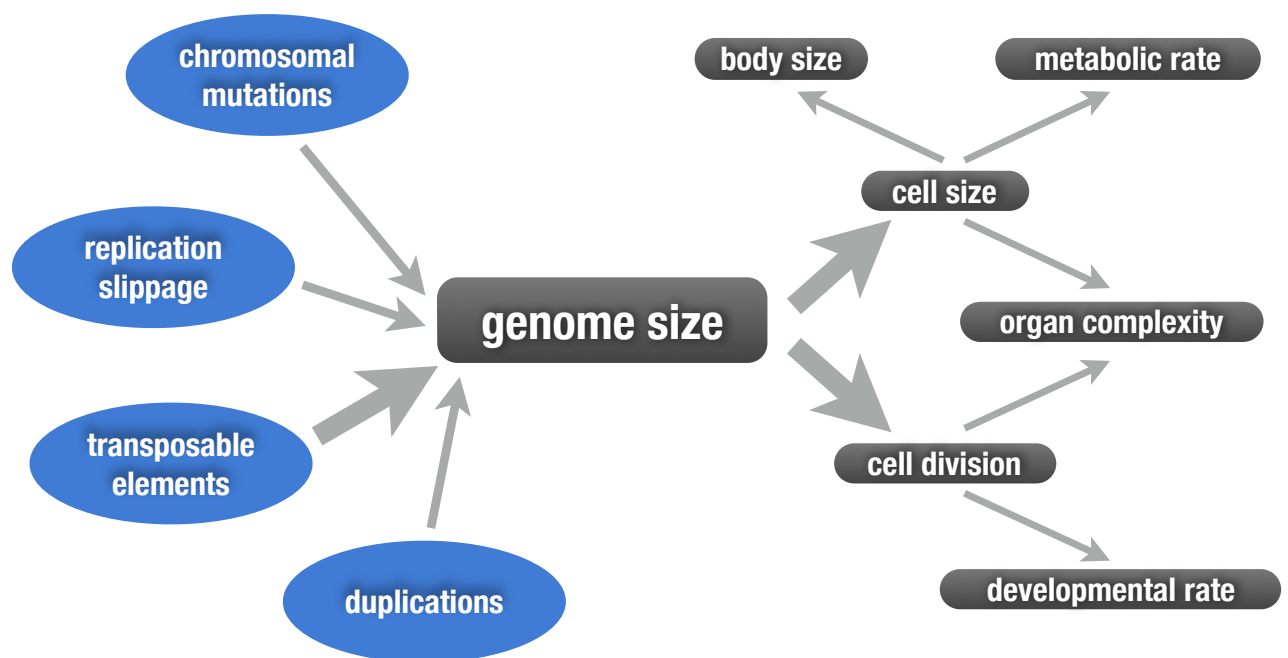
Lynch (2006)

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

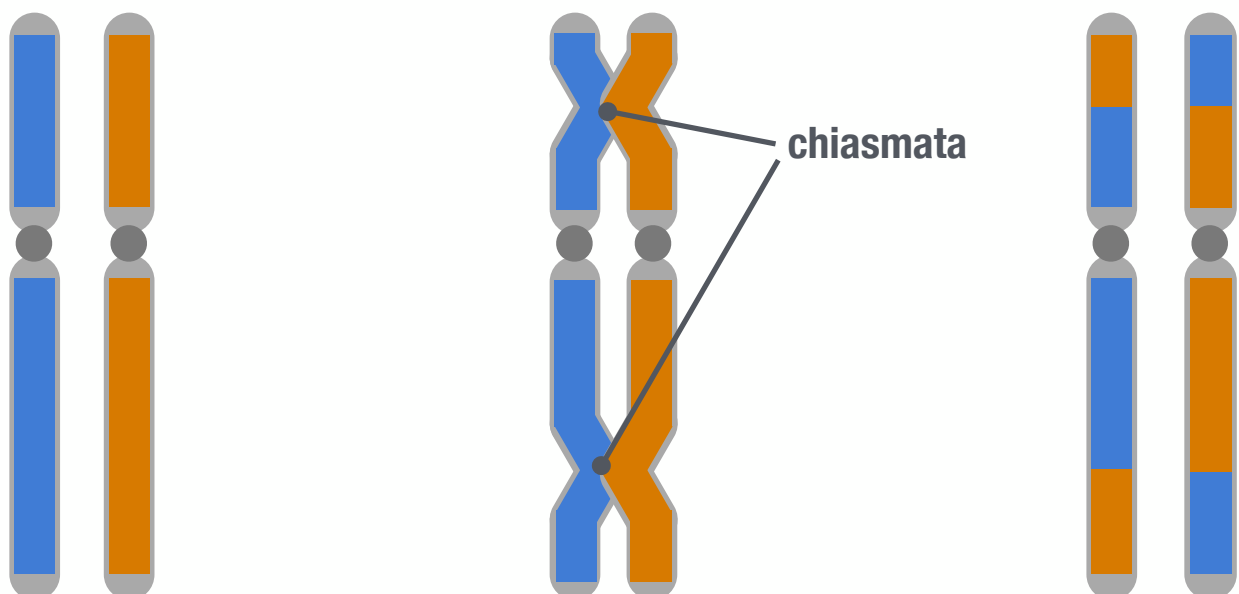
Chromosomal Rearrangements

Recombination

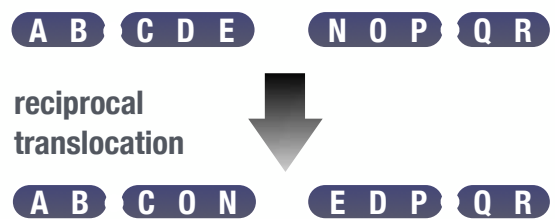
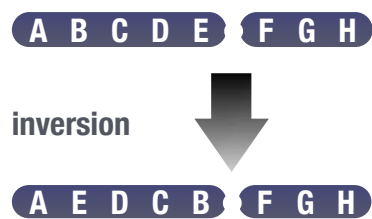
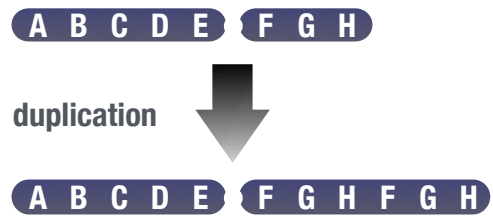
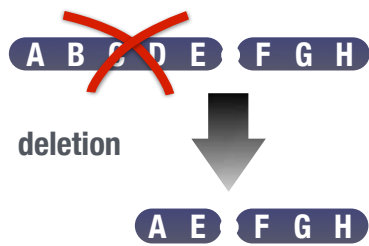
- ▶ ...occurs by the **crossing-over** of homologous chromosomes during **meiosis**
- ▶ ...leads to the **exchange of DNA** between a pair of chromosomes
- ▶ As a consequence, two previously unlinked genes may become linked or *vice versa*
- ▶ ...may lead to rearrangements

Chromosomal Rearrangements

Recombination

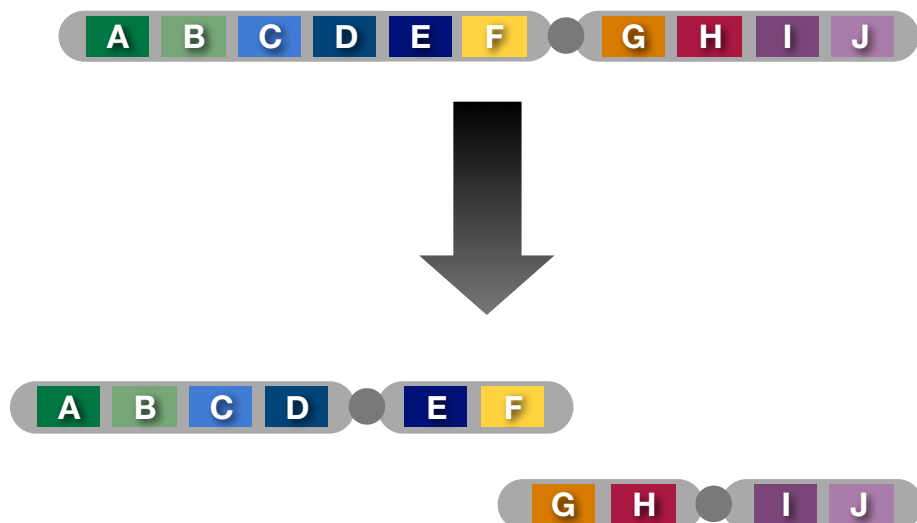


Chromosomal Rearrangements



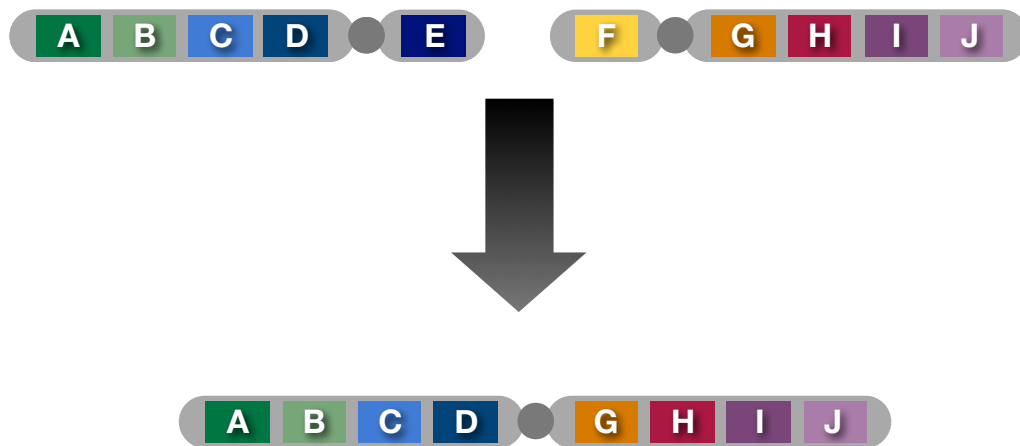
Chromosomal Rearrangements

Chromosomal Fission



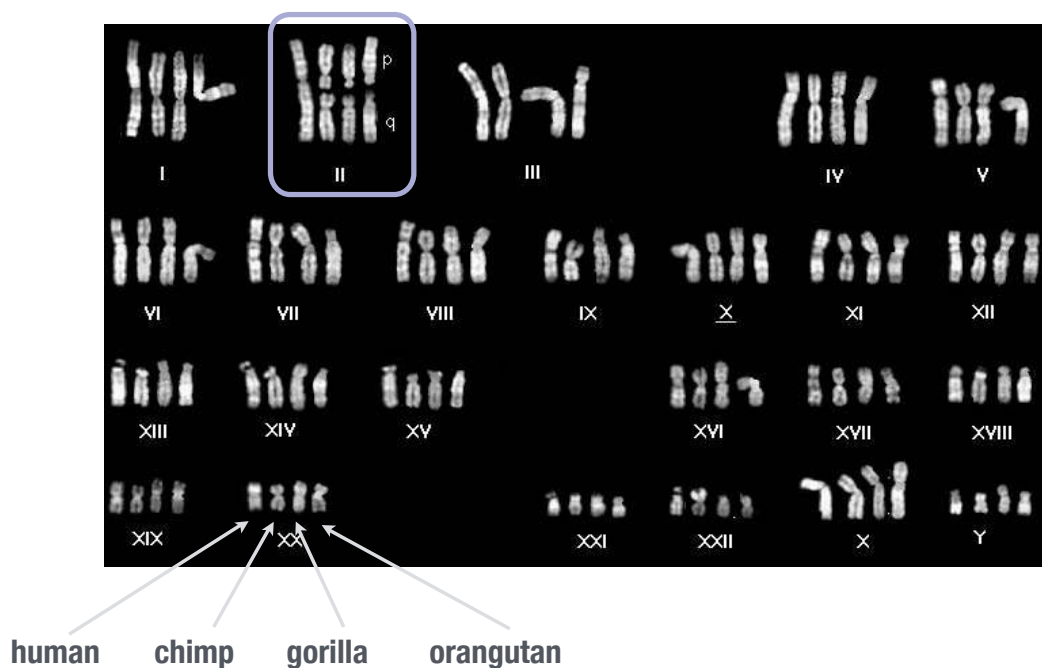
Chromosomal Rearrangements

Chromosomal Fusion



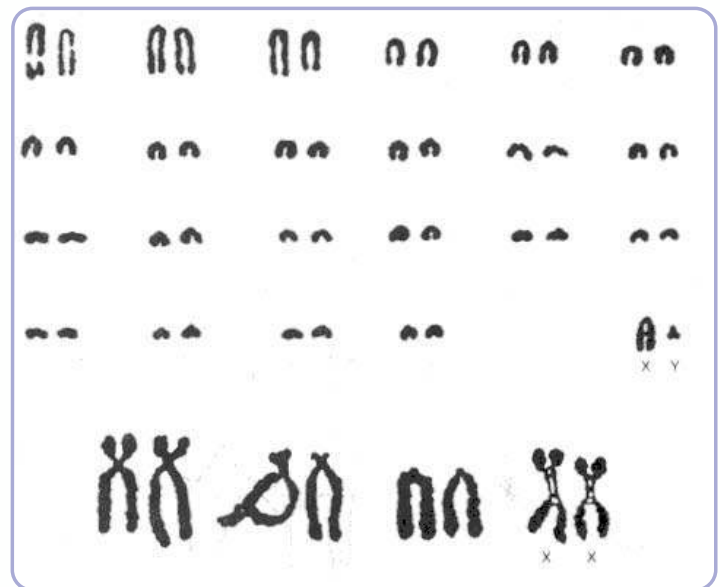
Chromosomal Rearrangements

Chromosomal fusion: human chromosome 2



Chromosomal Rearrangements

An extreme case: muntjak deers



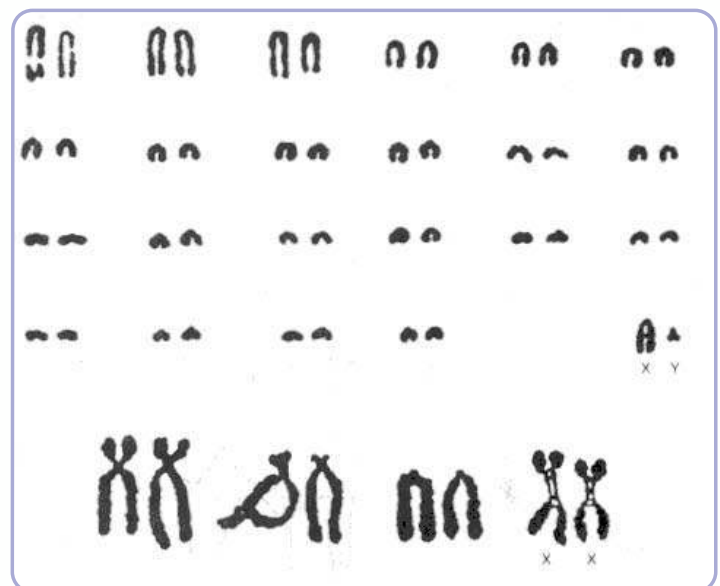
Chromosomal Rearrangements

An extreme case: muntjak deers

chinese muntjac deer

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

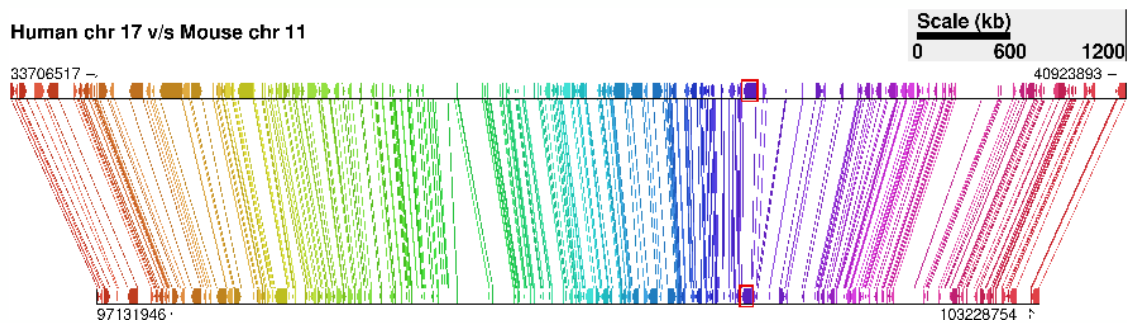
indian muntjac deer



Chromosomal Rearrangements

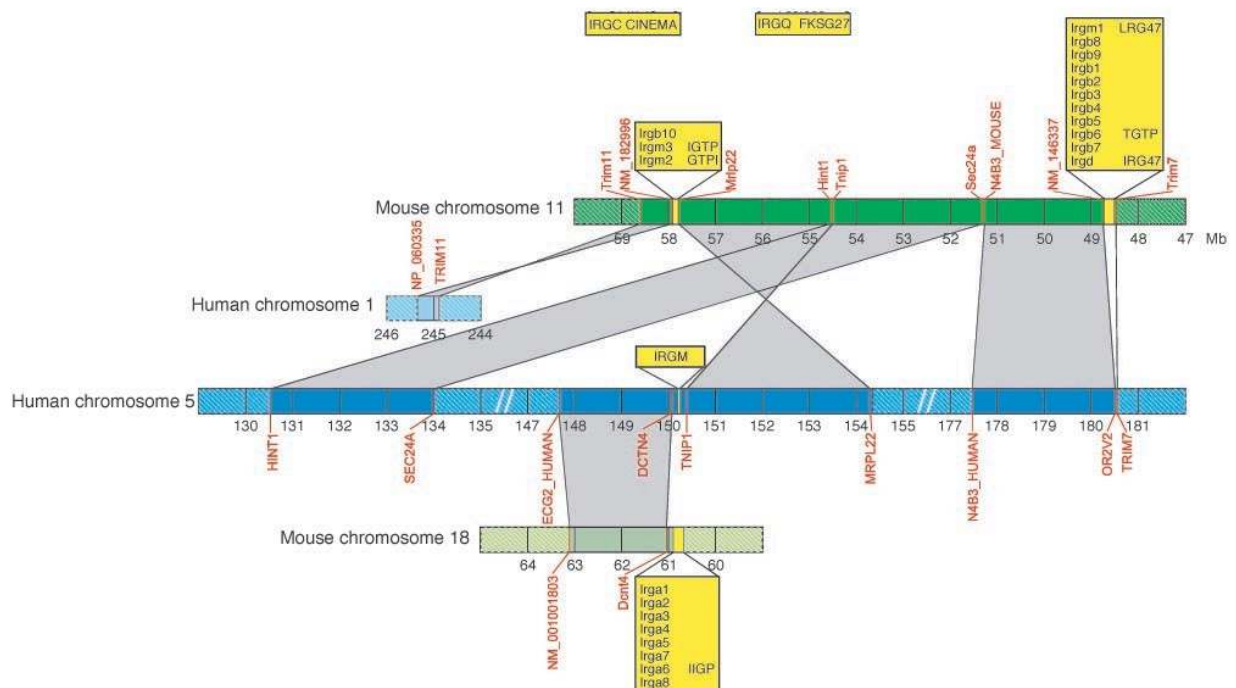
Synteny

- ▶ 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

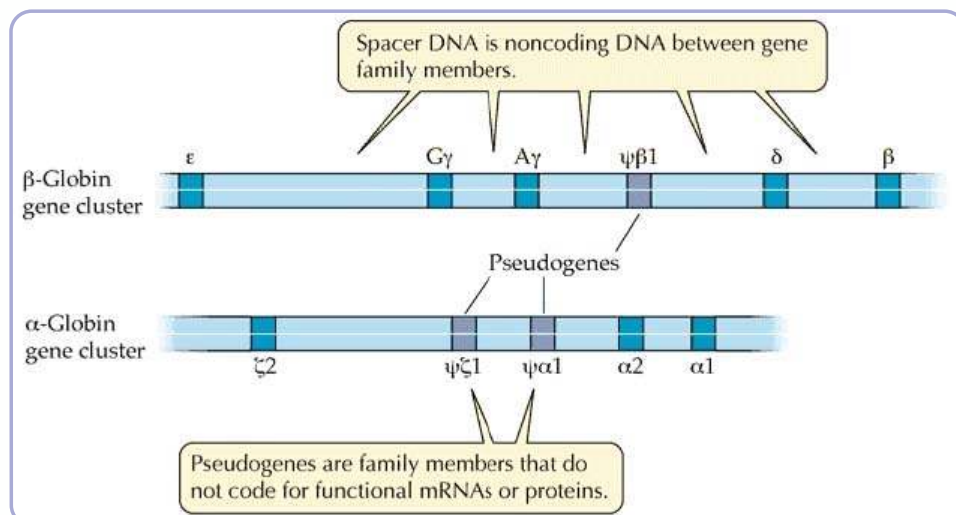
Gene duplication

- ▶ 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

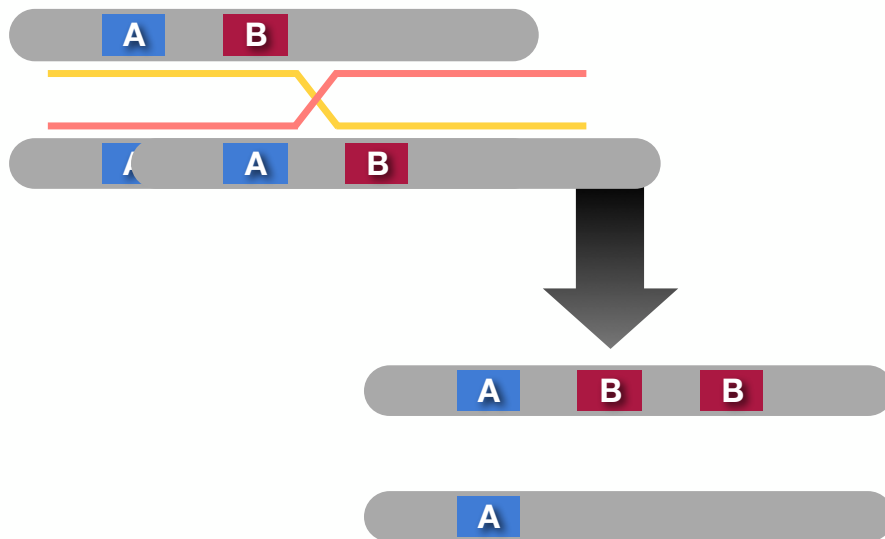
Gene clusters

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



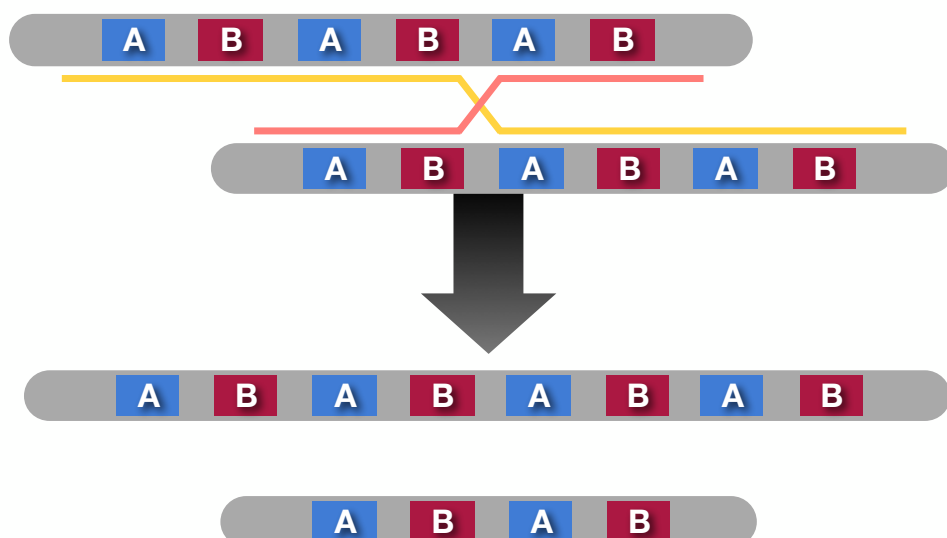
Gene and Genome Duplications

Unequal crossing over: misalignment



Gene and Genome Duplications

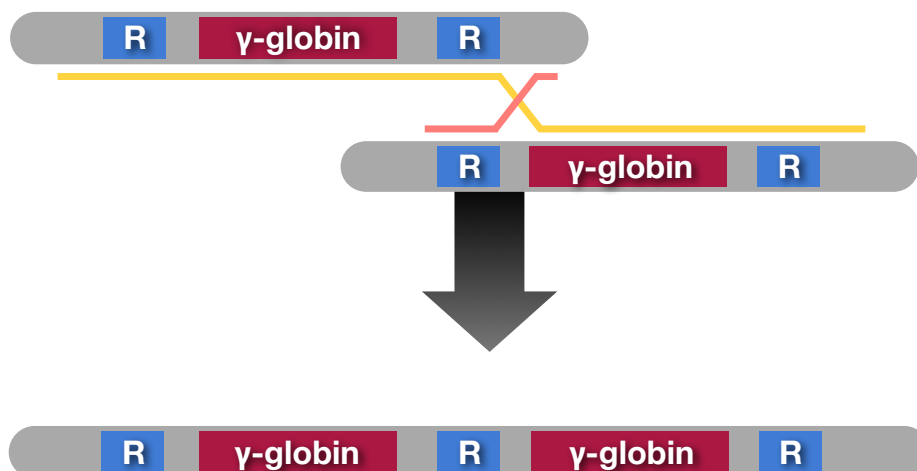
Unequal crossing over: misalignment



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

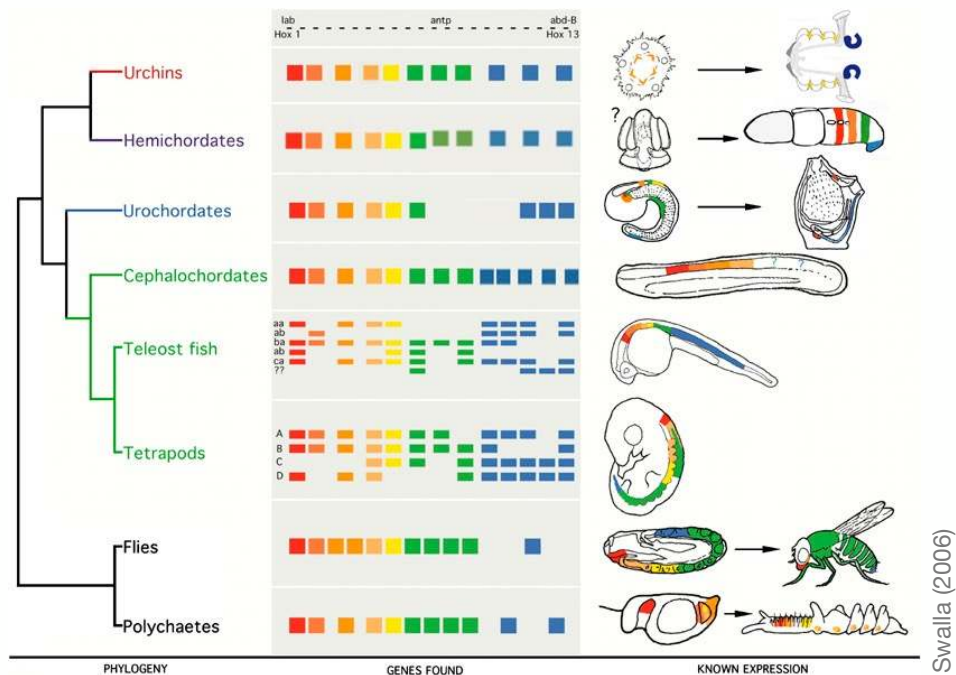
- ▶ Sosumo Ohno (1970):
Evolution by Gene Duplication
- ▶ Ohno also postulated that whole genome duplications exist and argued that the whole genome had duplicated twice near the origin of the vertebrates (**2R hypothesis**)
- ▶ Many more genome duplication events have been suggested since, e.g., the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



Sosumo Ohno
(1928-2000)

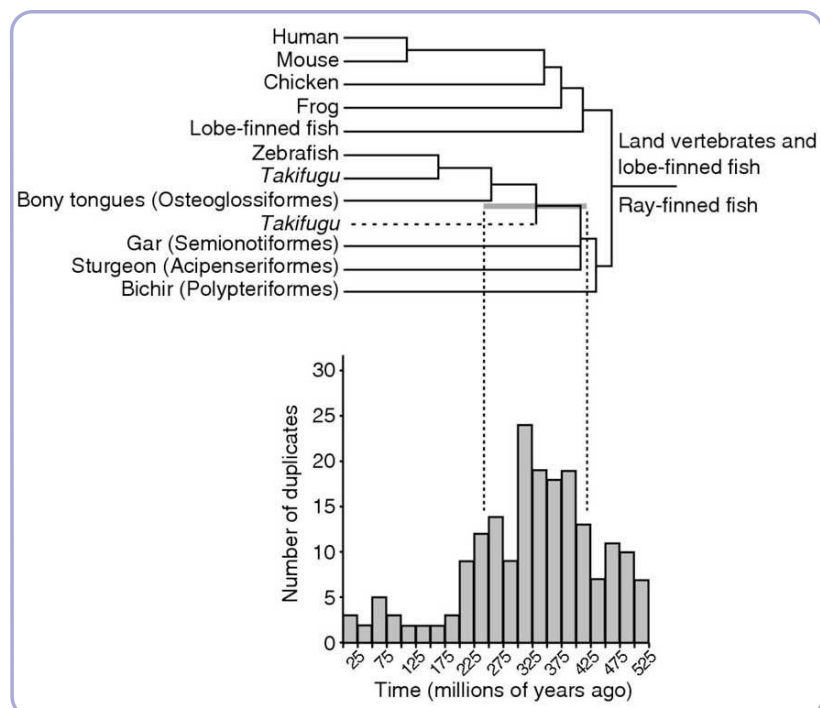
Gene and Genome Duplications

Hox gene clusters



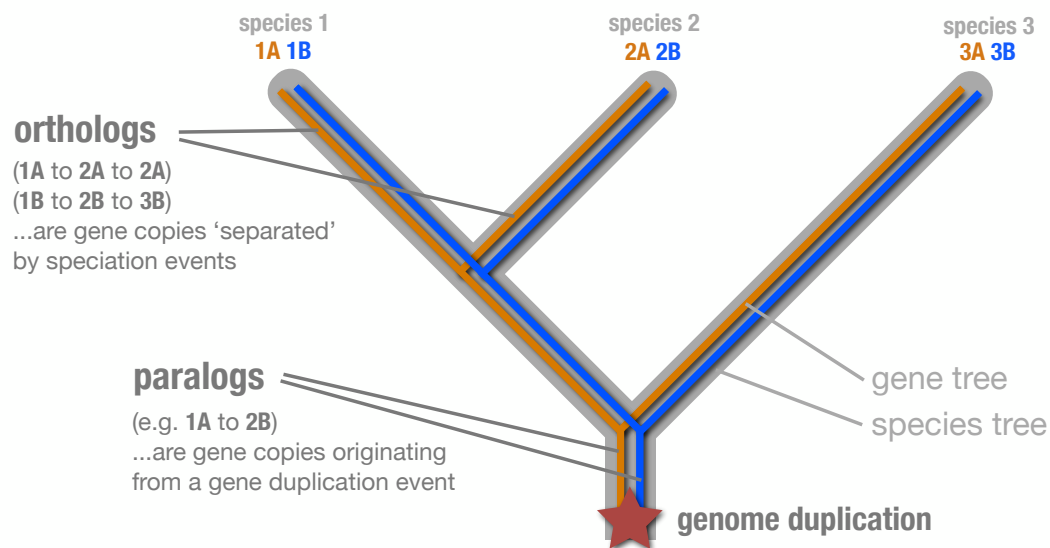
Gene and Genome Duplications

fish specific genome duplication



Gene and Genome Duplications

paralogs, orthologs and ohnologs

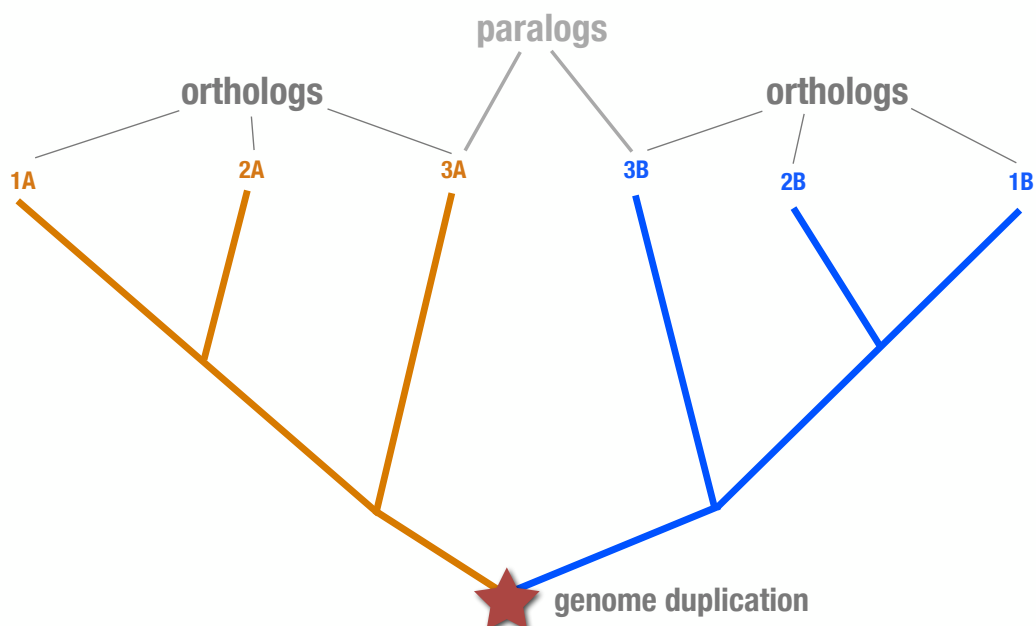


ohnologs

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

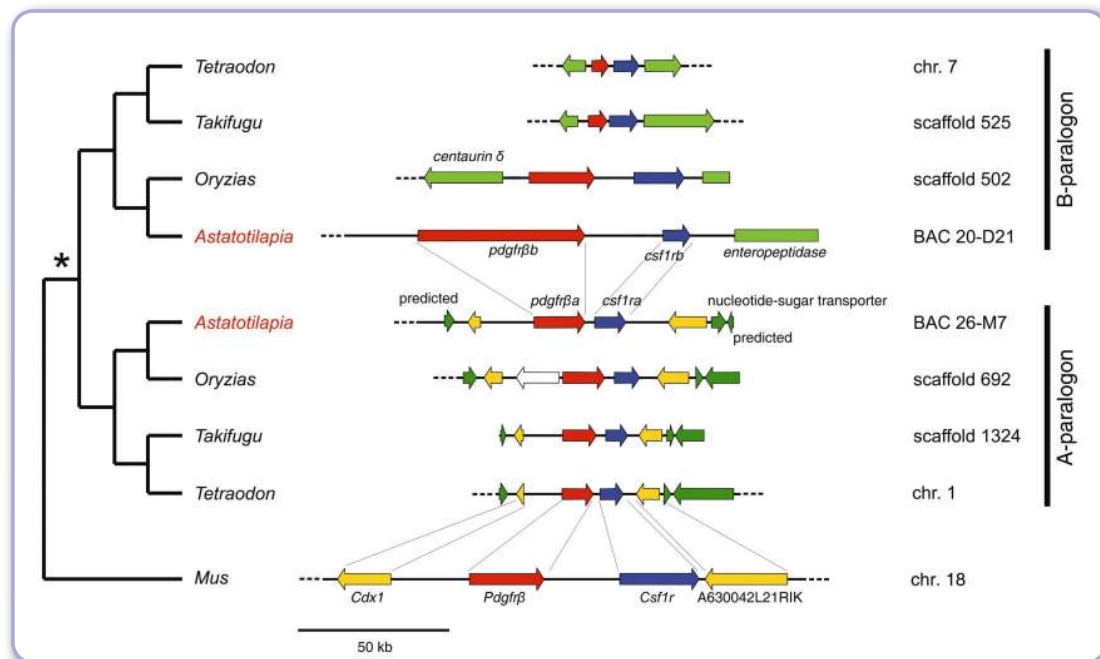
Gene and Genome Duplications

paralogs, orthologs and ohnologs



Gene and Genome Duplications

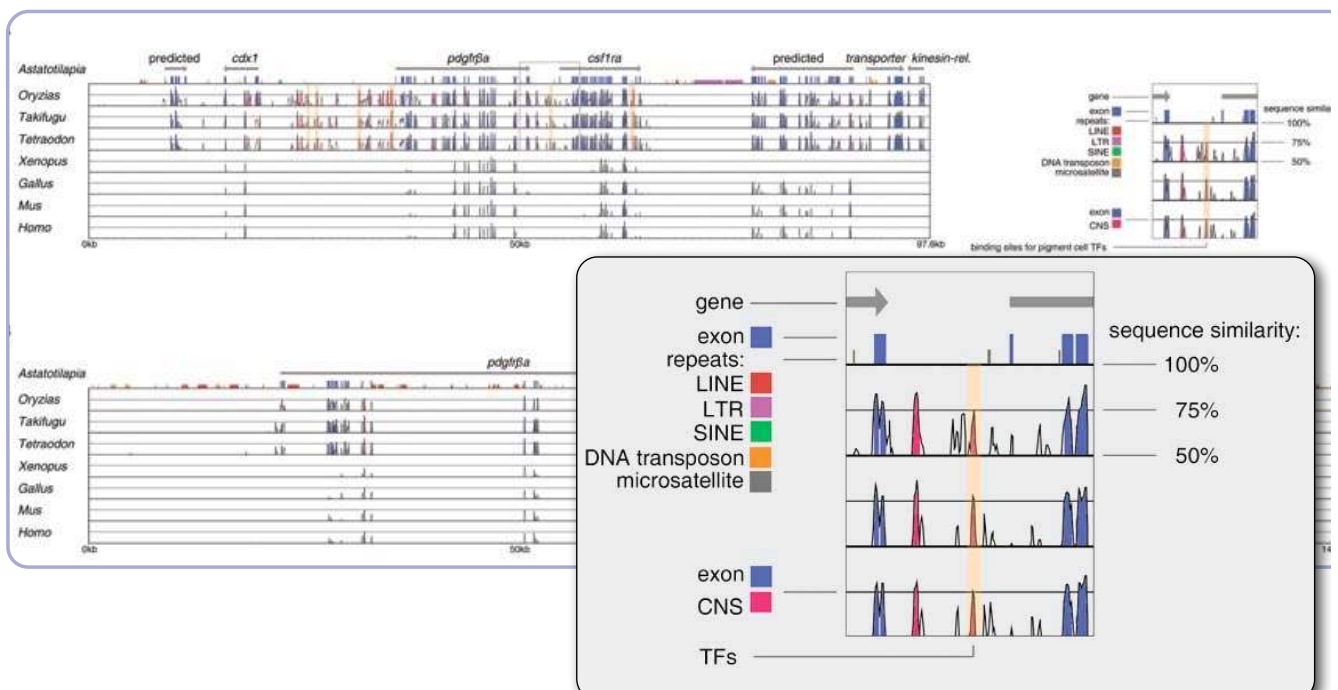
'paralogons'



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

Gene and Genome Duplications

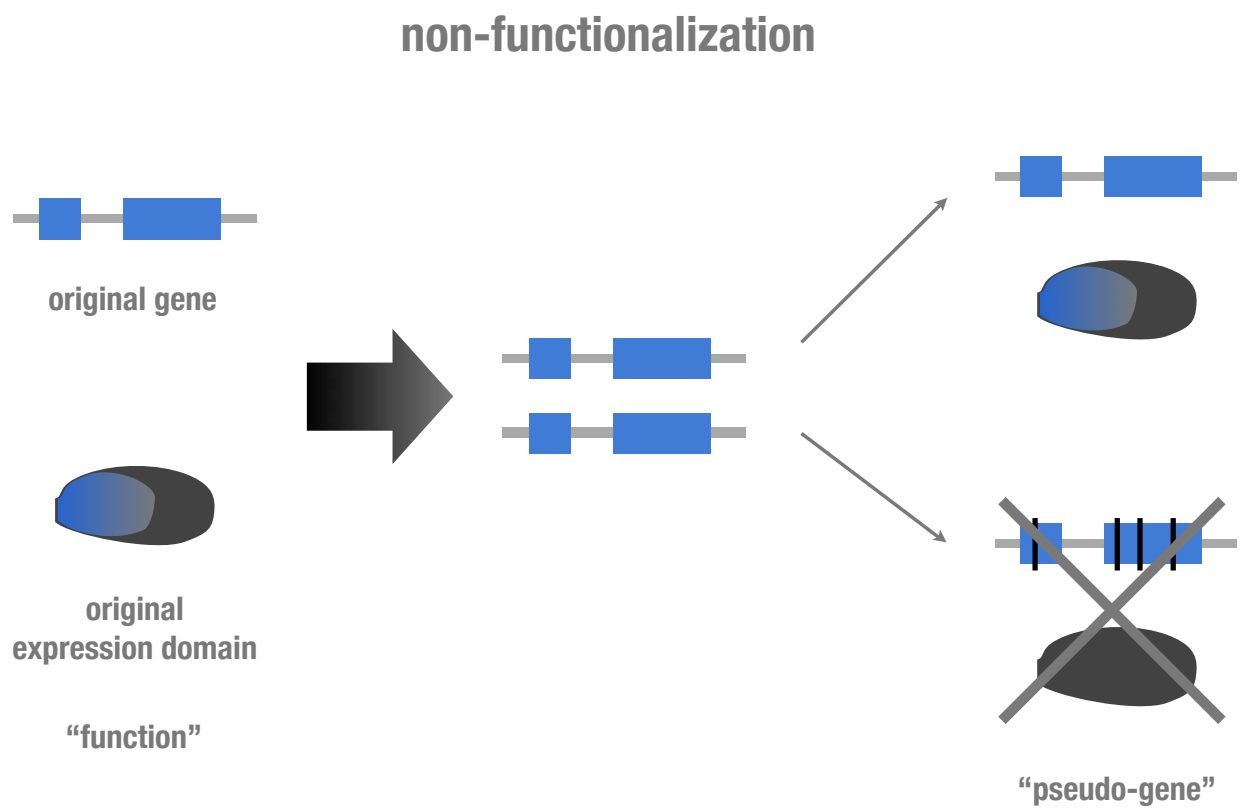
'vista' plots



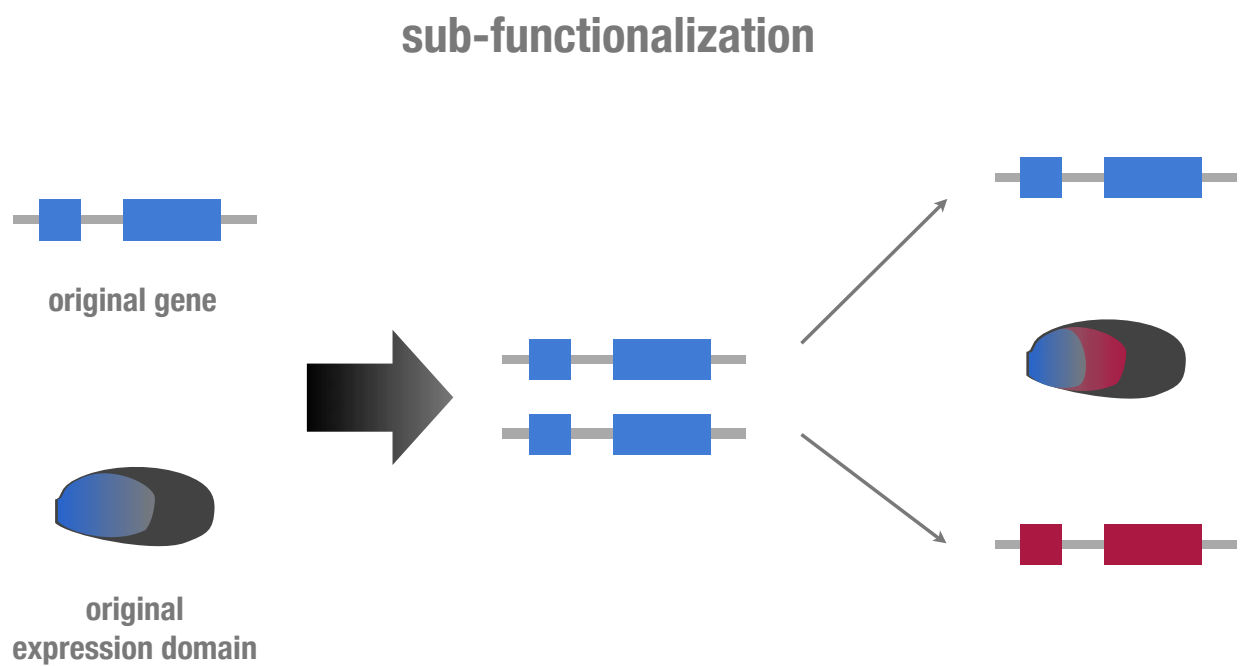
<http://genome.lbl.gov/vista/index.shtml>

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

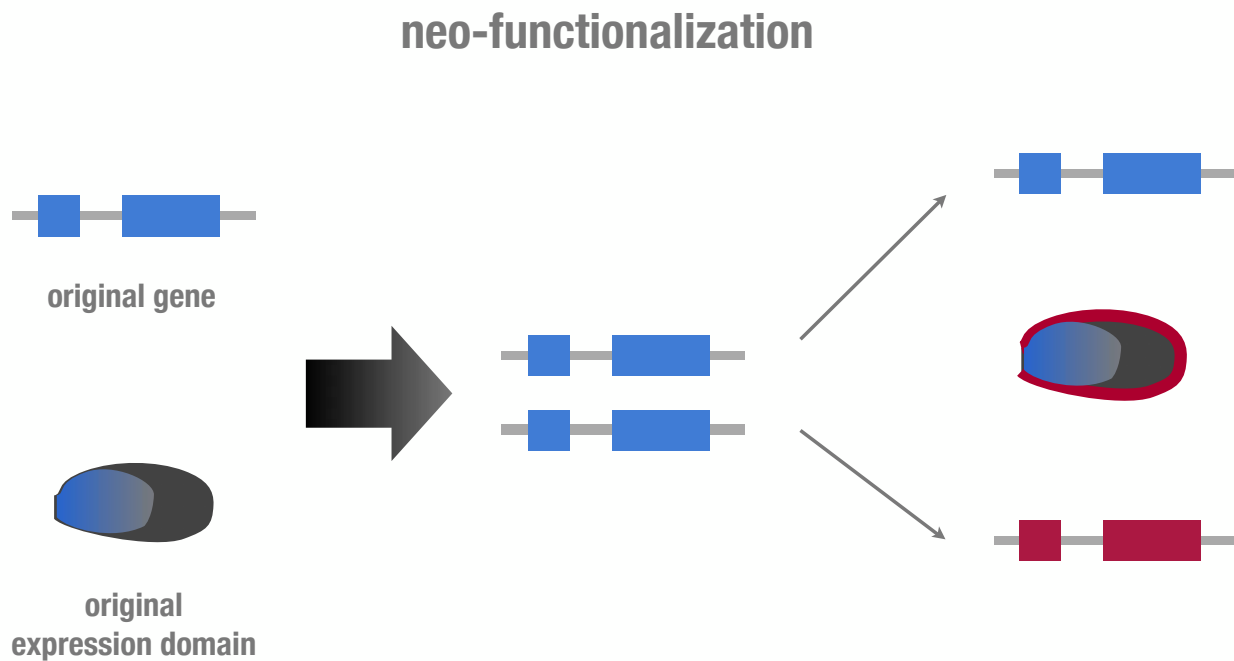
Gene and Genome Duplications



Gene and Genome Duplications

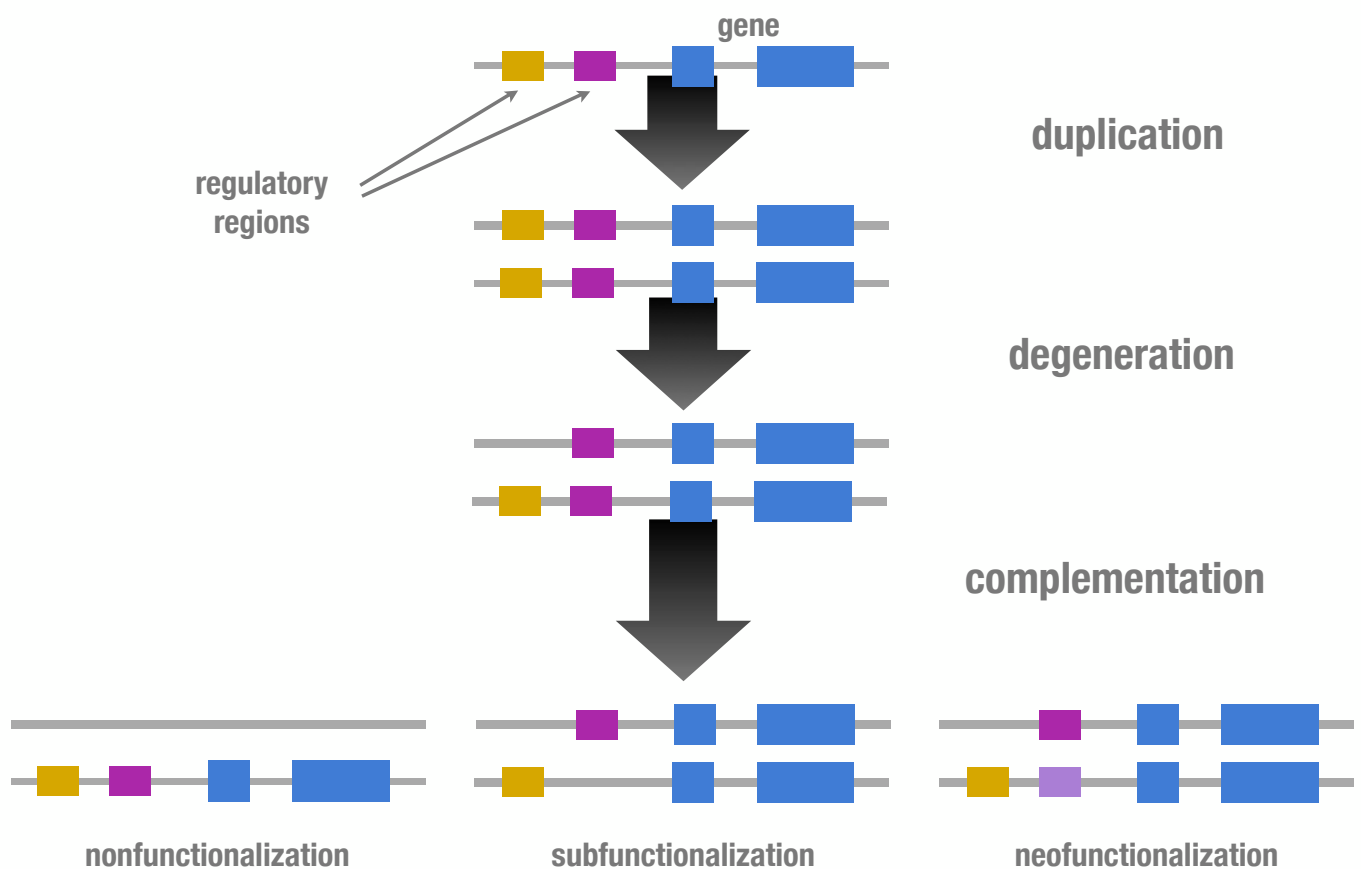


Gene and Genome Duplications



Gene and Genome Duplications

DDC model

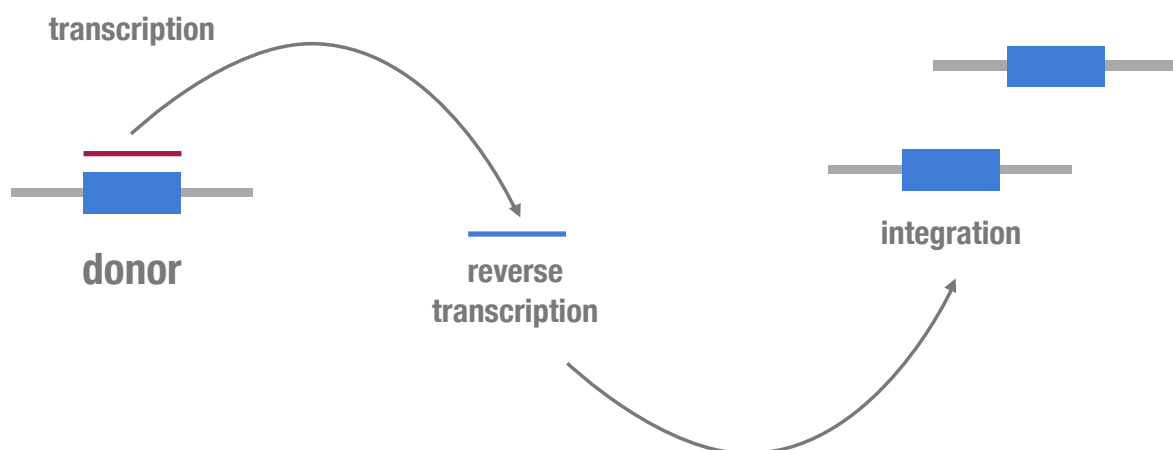


Transposable Elements

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

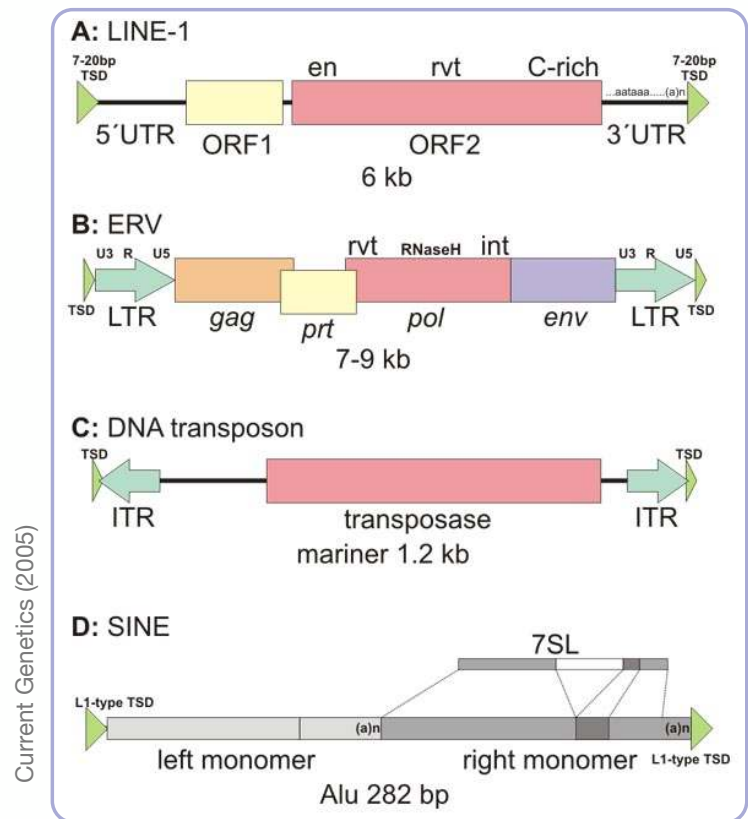
Short interspersed nuclear elements are non-autonomous retrotransposons that exploit the enzymatic retrotransposition machinery of LINEs

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
total	3.1	9.6	6.5	14	44.8

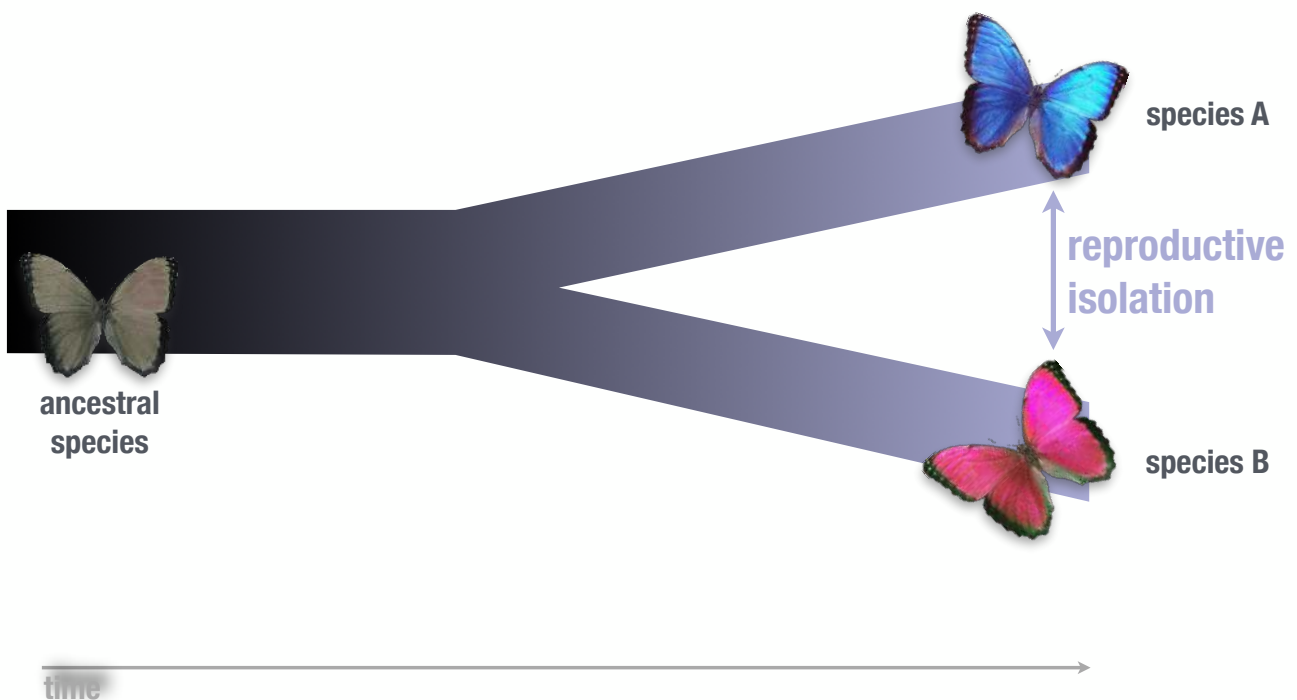
in % of genomes

Kidwell (2005)

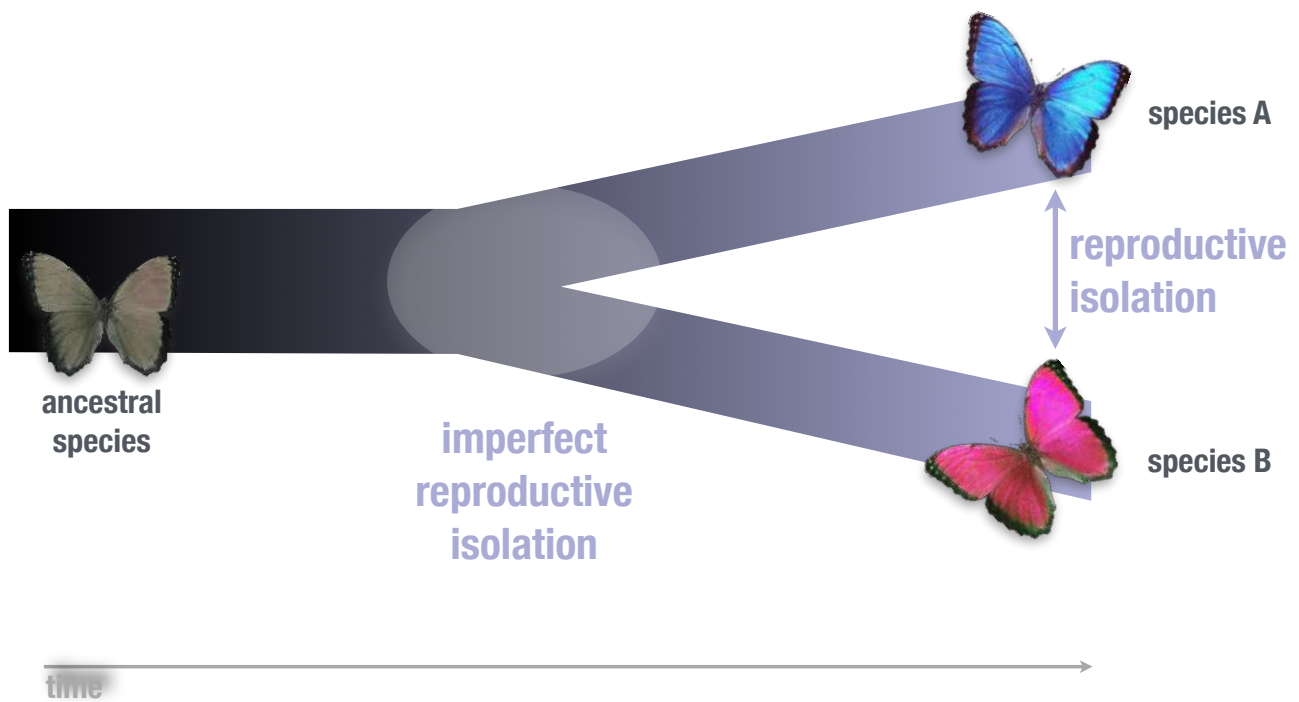
Speciation



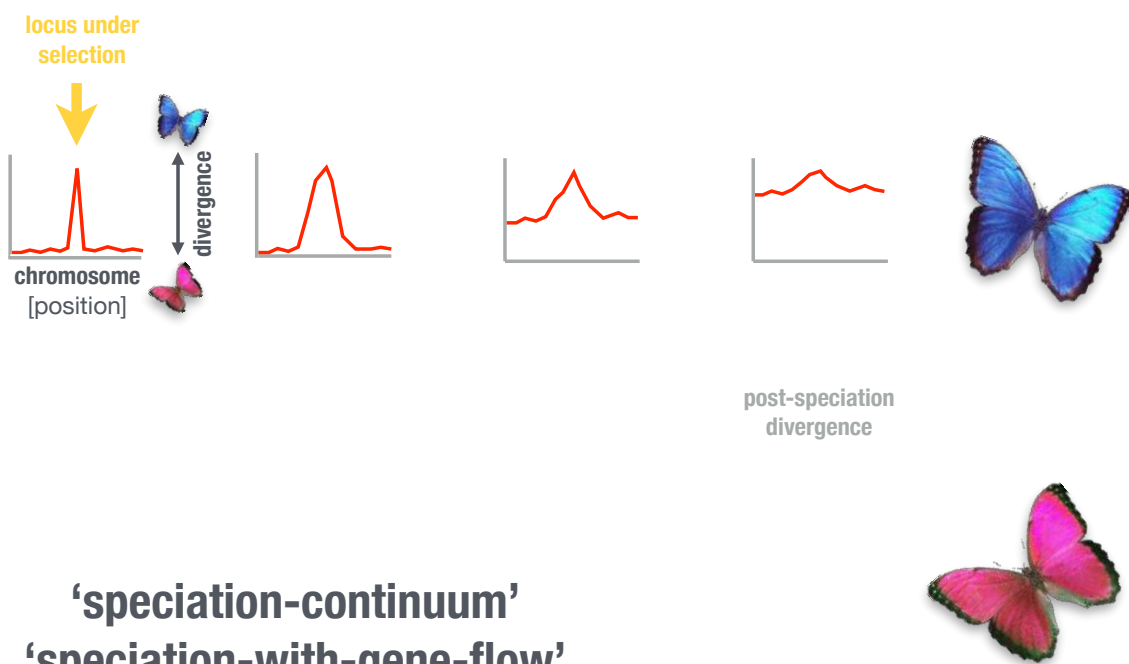
Speciation Genomics



Speciation Genomics



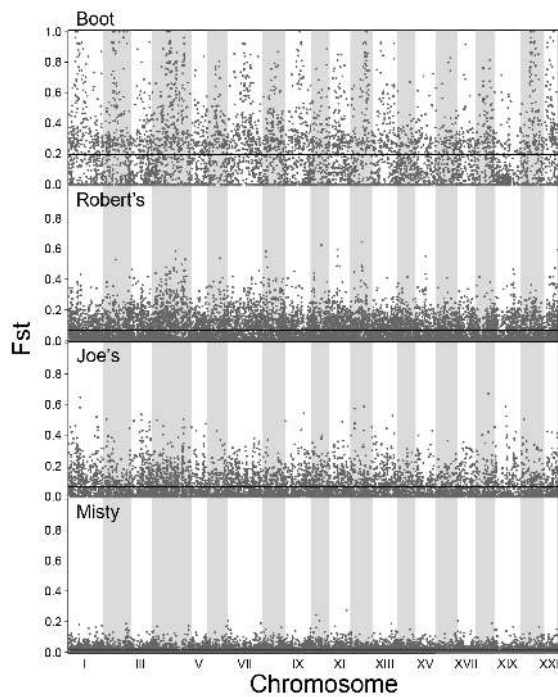
Speciation Genomics



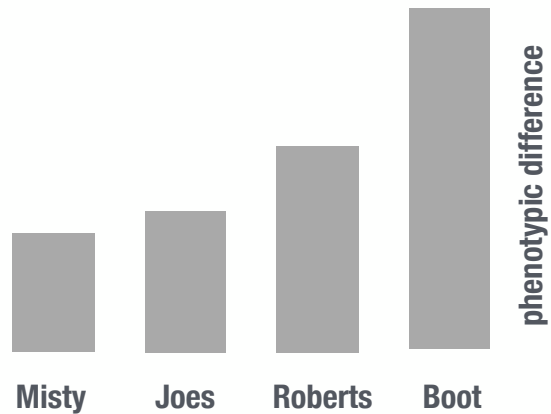
'speciation-continuum'
'speciation-with-gene-flow'

JL Feder, SP Egan & P Nosil (2012) *TREE*

Speciation Genomics:



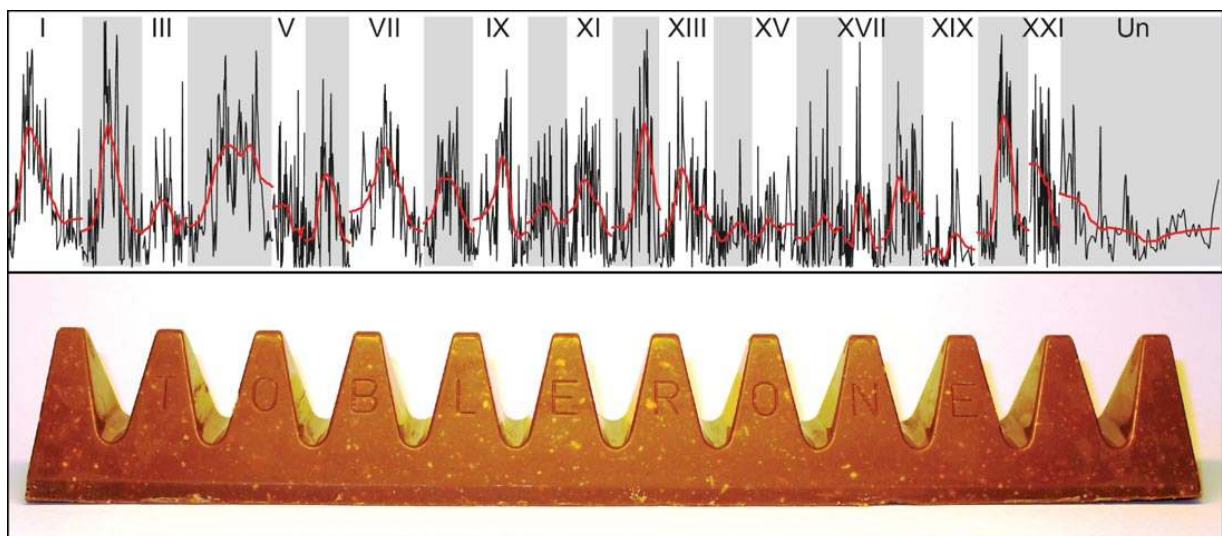
RAD genome scans



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

Speciation Genomics:

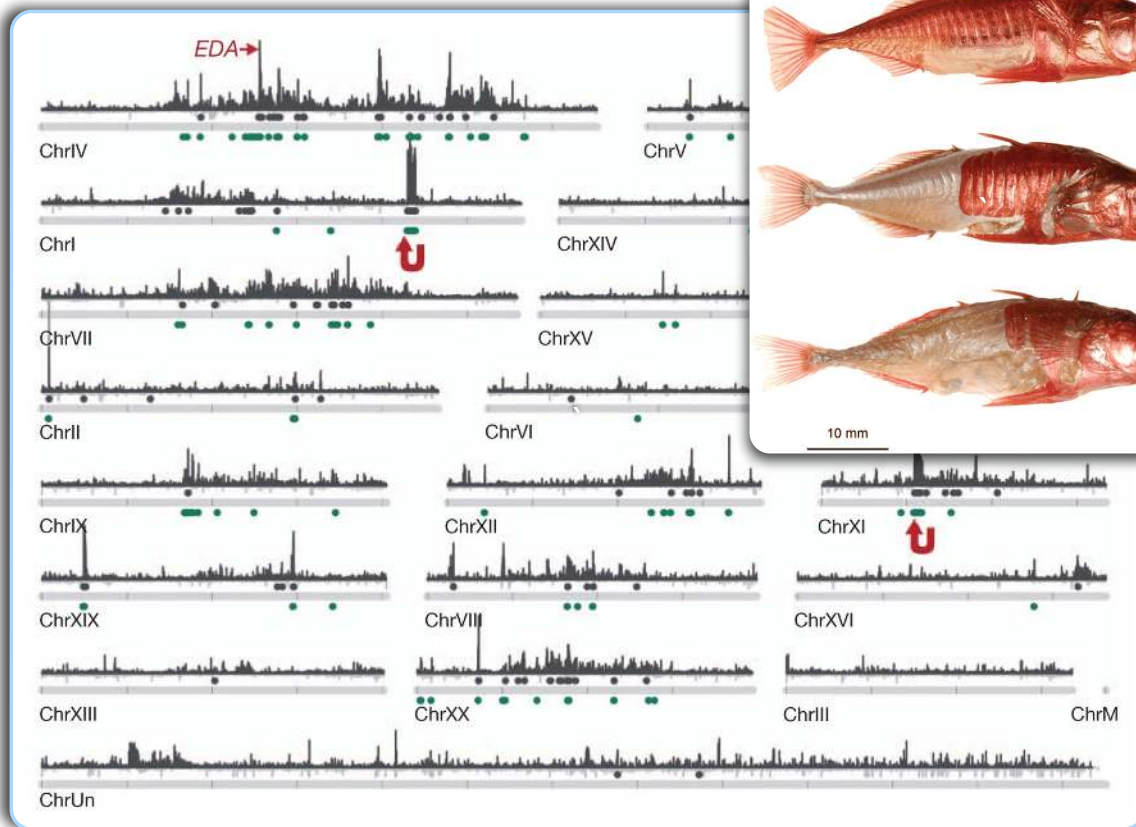
RAD genome scans



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

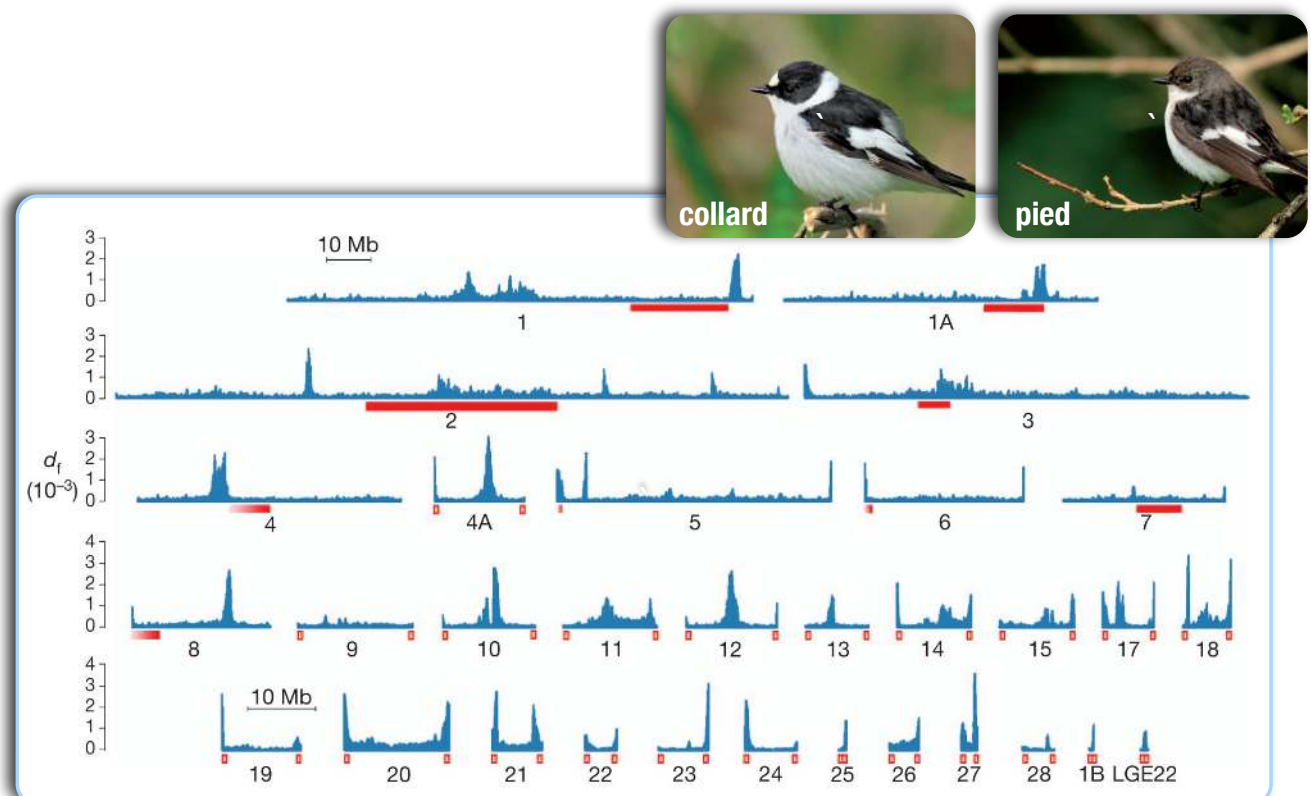
Speciation Genomics:

Barrett et al. (2008) *Science*



Jones et al. (2012) *Nature*

Speciation Genomics:

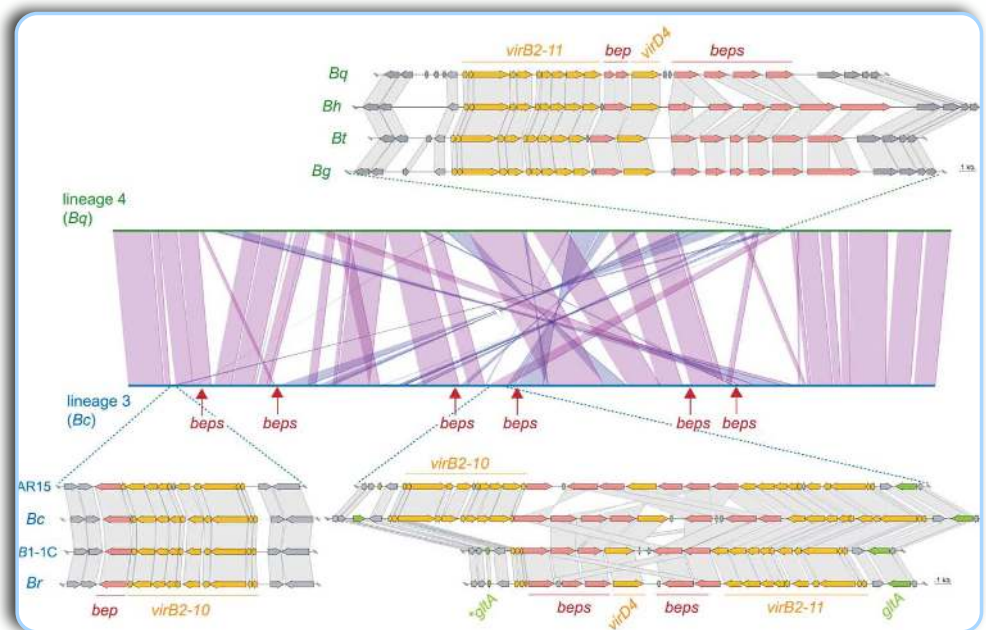


Ellegren et al. (2012) *Nature*

Speciation Genomics



Speciation Genomics



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

Speciation Gen

