

Rampant gene duplication facilitates Y-linked gene evolution in the *Drosophila simulans* clade

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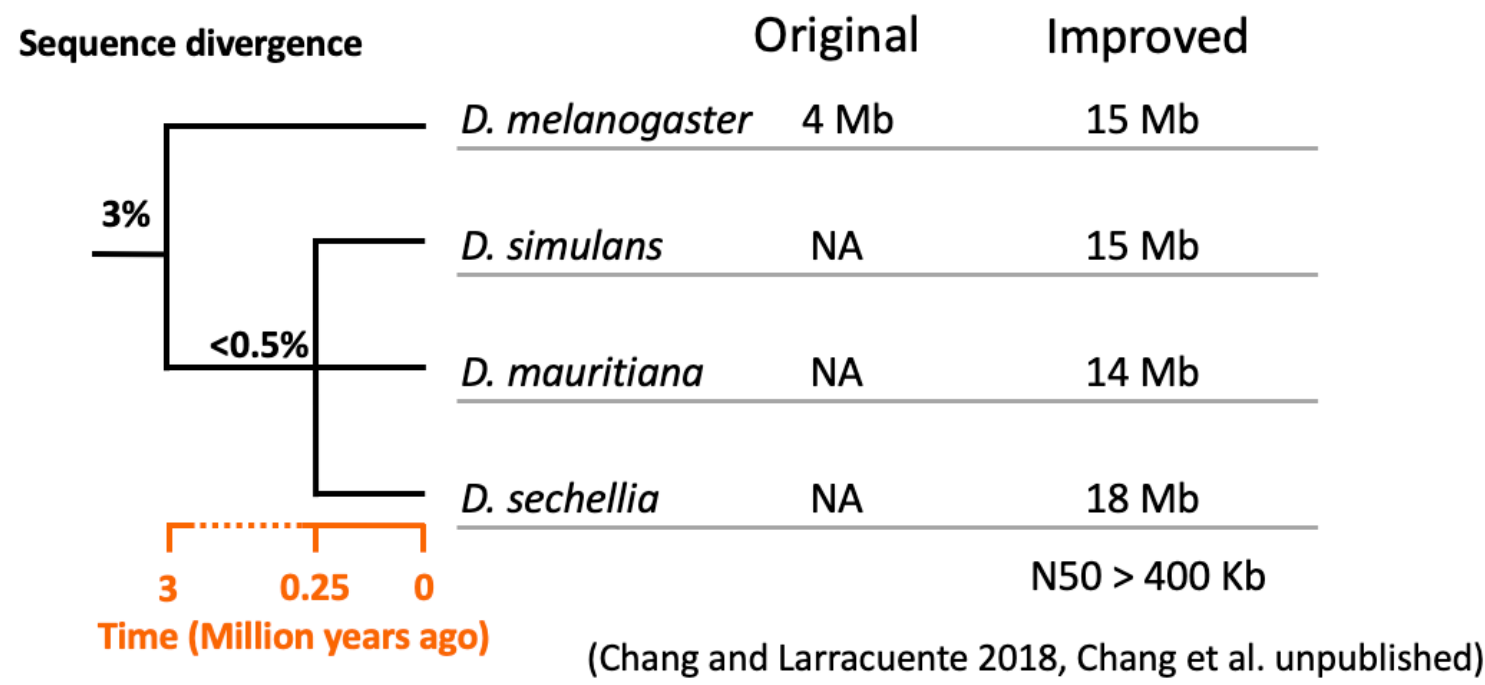
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Summary

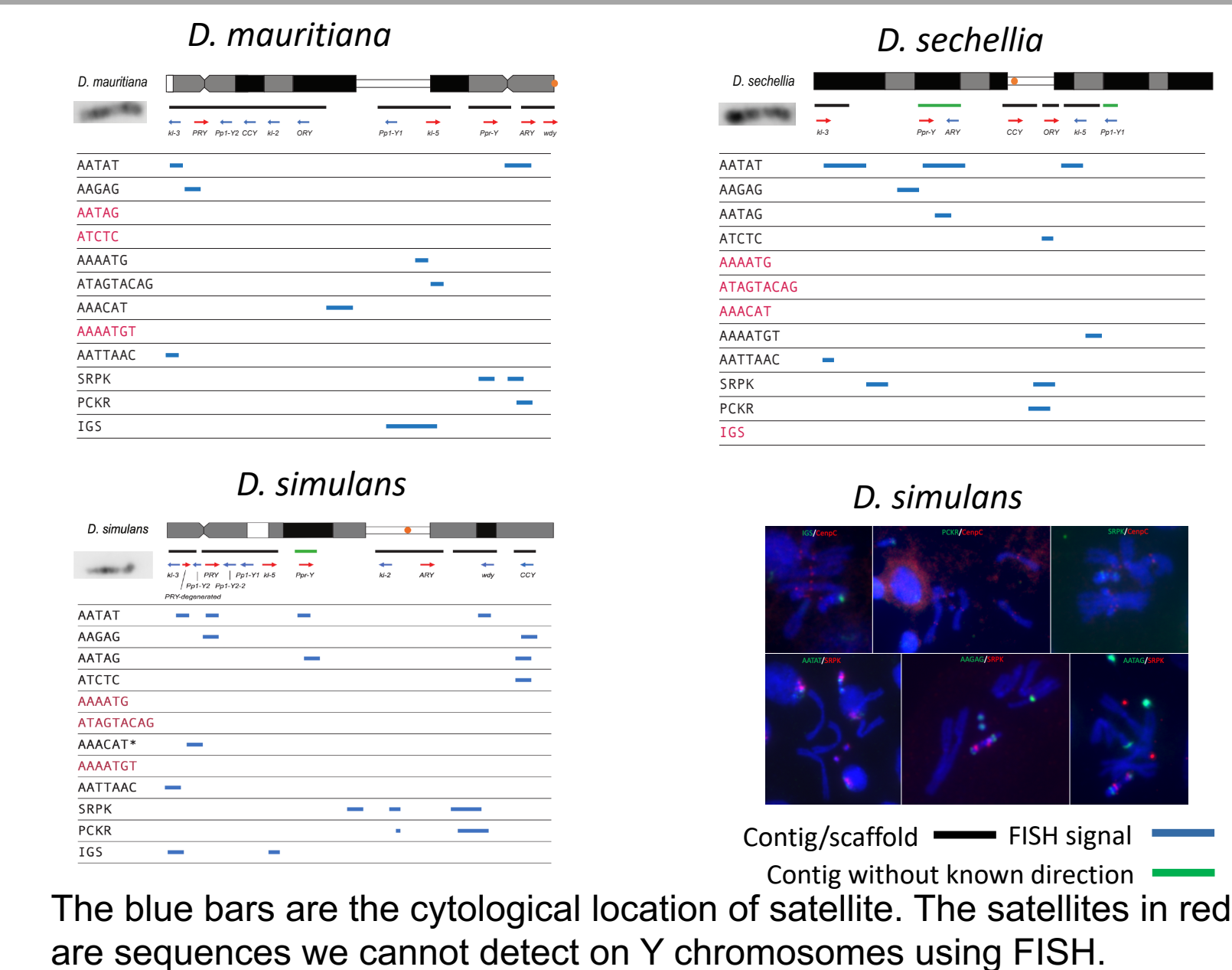
- I. Long sequencing and *de novo* assembly of the Y chromosomes in *D. melanogaster* and *simulans* clade species
- II. Rapid evolution of structure and gene content on Y chromosomes
- III. Intralocus genetic conflicts and gene duplication

Improving Y assemblies

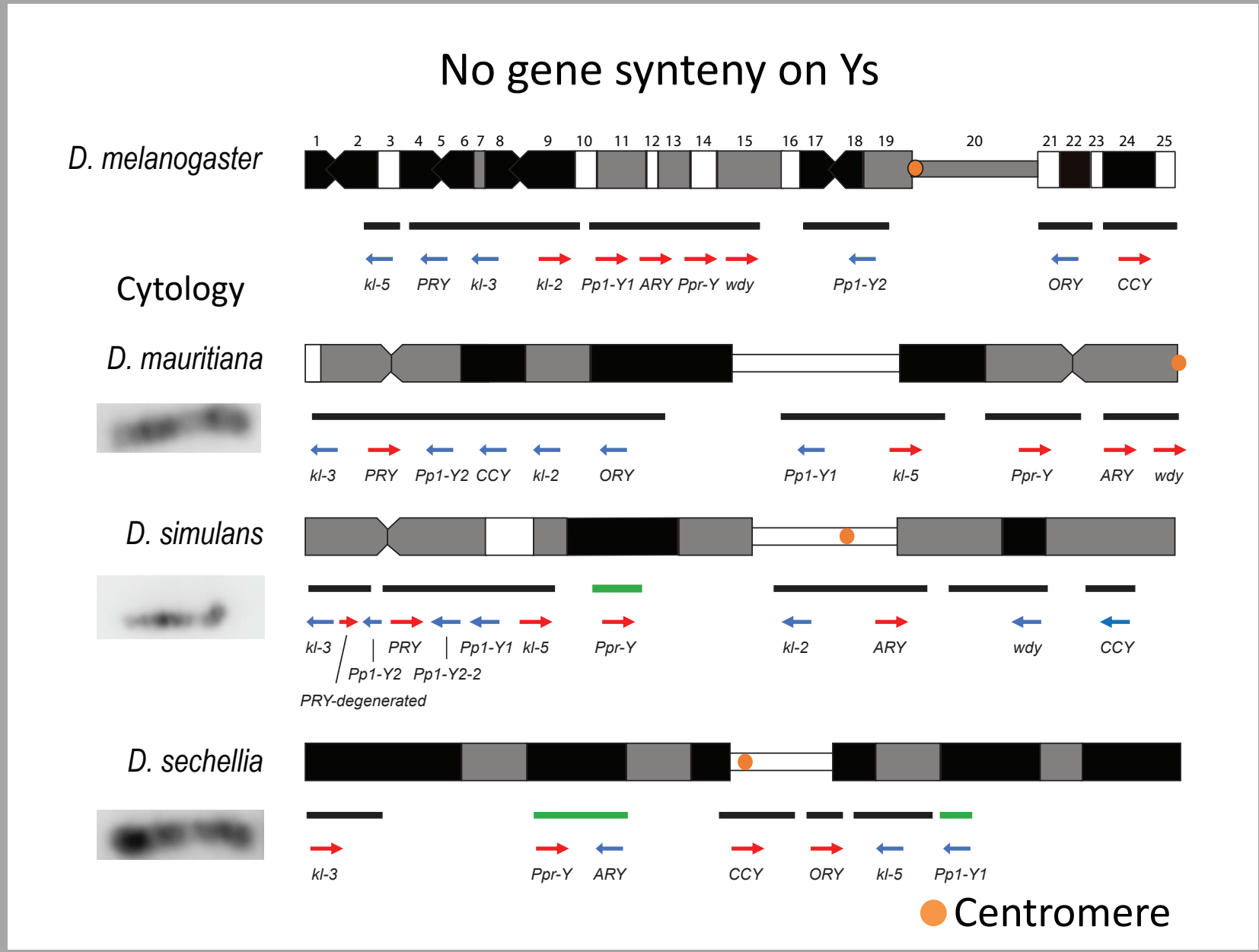


Y Chromosome Assembly	# of Contigs	Total size	Contig N50
D. melanogaster	80	14,578,684	416,887
D. simulans	38	13,717,056	1,031,383
D. sechellia	63	14,899,148	555,130
D. mauritiana	55	17,880,069	1,628,994

Placing contigs on the Y using FISH

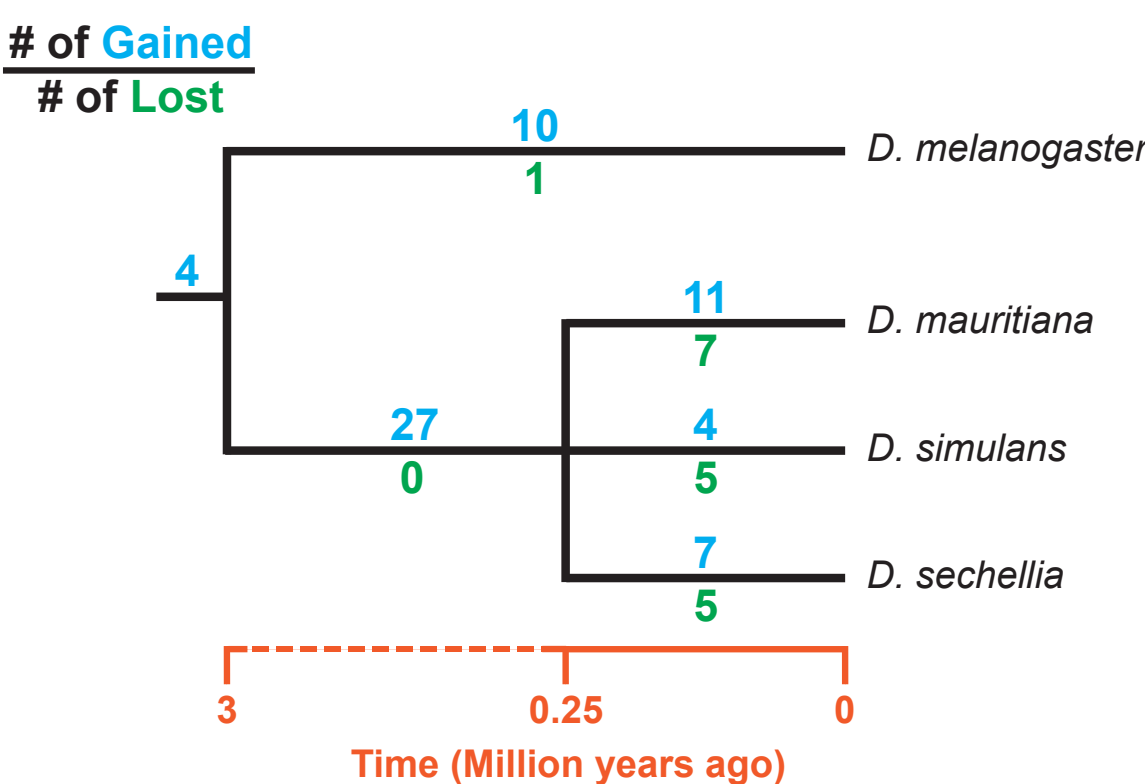


Extensive rearrangement of Y-linked genes



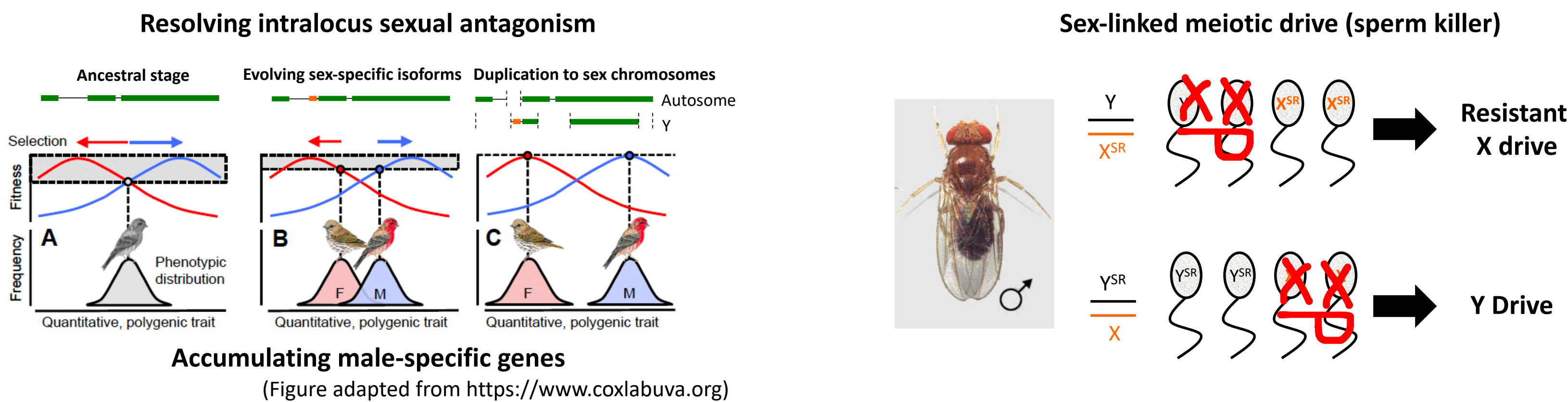
Rapid turnover of duplicated genes on Ys

Many genes duplicated to Y from other chromosomes
>50% of new Y-linked genes are degenerated



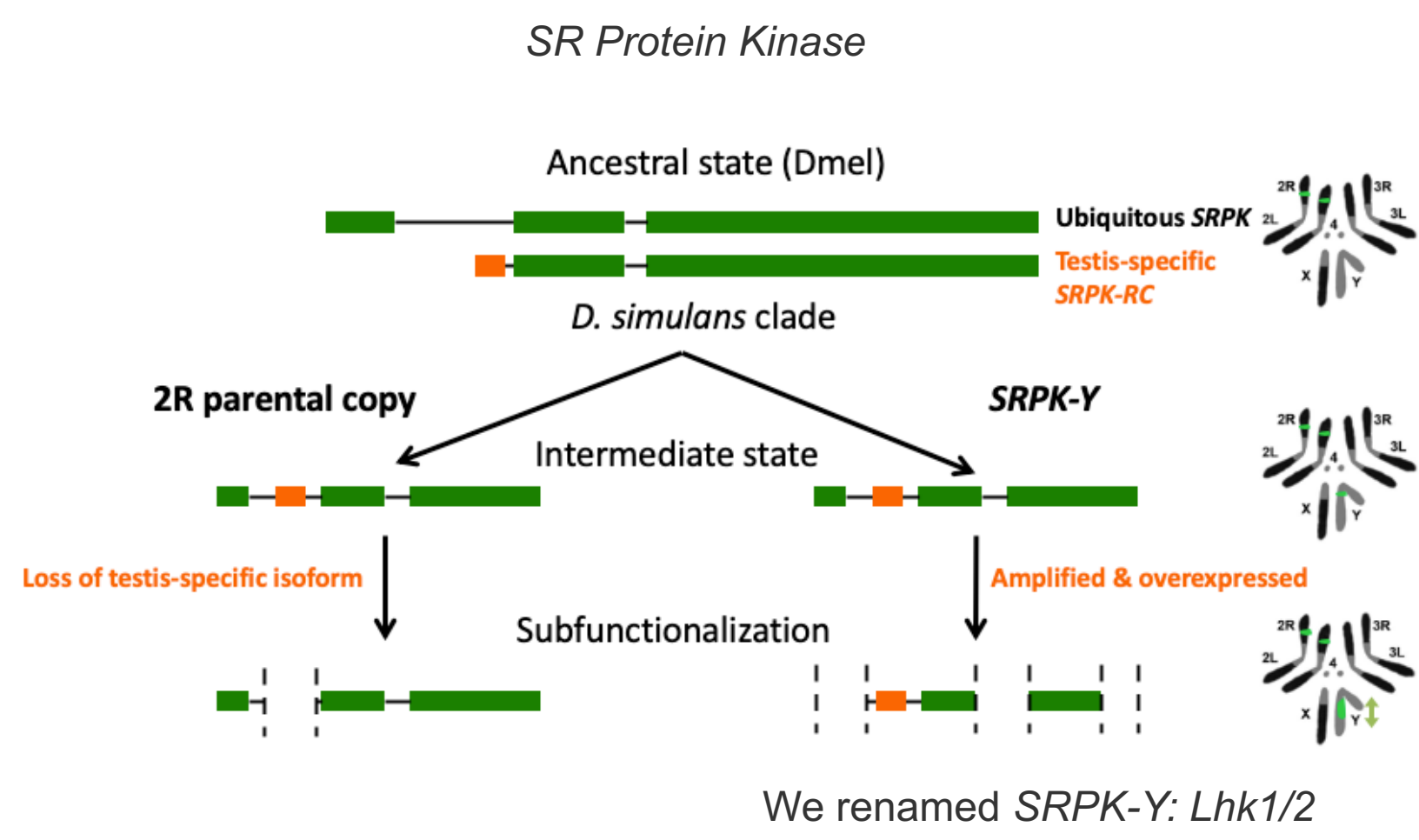
*whole mitochondrial genome inserted in the ancestral *simulans* clade Y

Conflicts and Y chromosomes

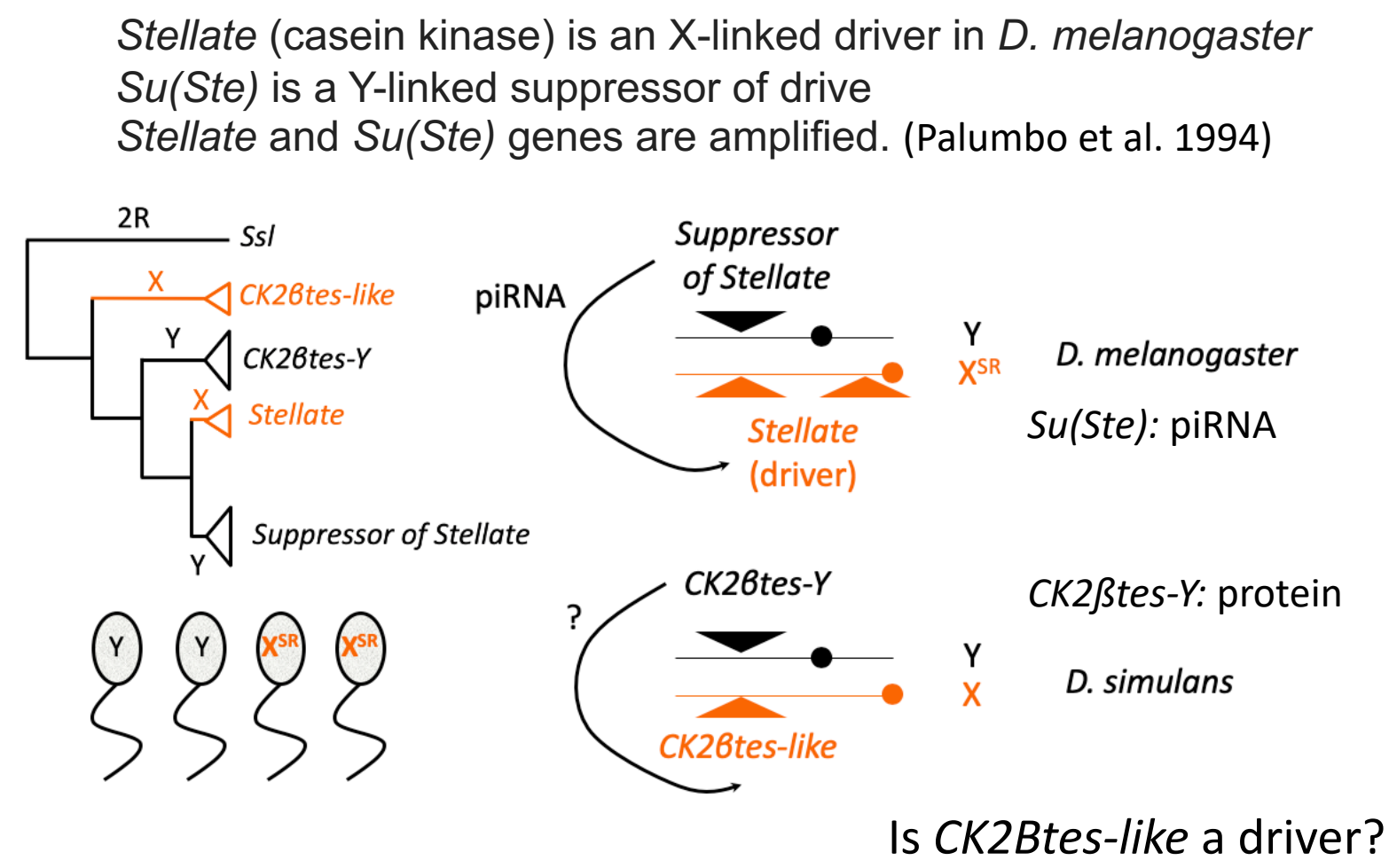


Y-linked ampliconic genes specific to the *simulans* clade

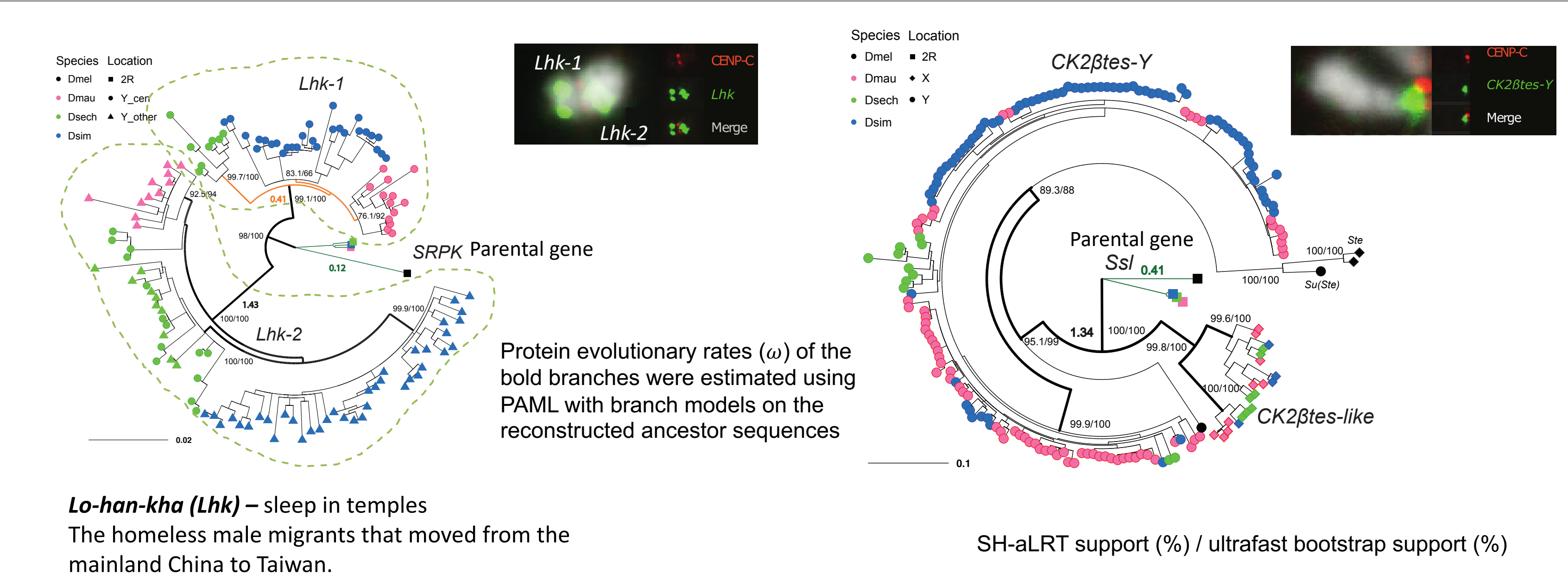
Translocation of a testis-specific isoform



Co-amplification of a potential driver



Strong positive selection on Y ampliconic genes ($\omega = 1.3-1.5$)



SRPK has different roles in each sex

- I. Testis-specific isoform, *SRPK-RC*, is recently acquired in melanogaster group
- II. *SRPK* knockout is sterile in both sexes
- III. Two *SRPK* mutants are only sterile in females
- IV. Ubiquitous *SRPK* isoforms are expressed premeiotically and are localized to cytoplasm and nucleolus in testes
- V. *SRPK-RC* is only expressed in primary spermatocytes

Ongoing

- I. Exploring functional divergence between *SRPK-RC*, *Lhk-1*, and *Lhk-2*
- II. What drives positive selection and amplification of multicopy Y-linked genes?
- III. Do the sequence and copy number divergences between species contribute to hybrid sterility in *D. simulans* clade?

References

Chakraborty et al. 2016. Nucleic Acids Res 44(19):e147.
Chin et al. 2013. Nat Methods 10:563–569.
Hoskins et al. 2015. Genome Res 25(3):445–458.
Koren et al. 2017. Genome Res 27(5):722–736.
Loh et al. 2012. J Cell Sci 125(Pt 19):4457–62.
Palumbo et al. 1994. Genetics 138:1181–1197.

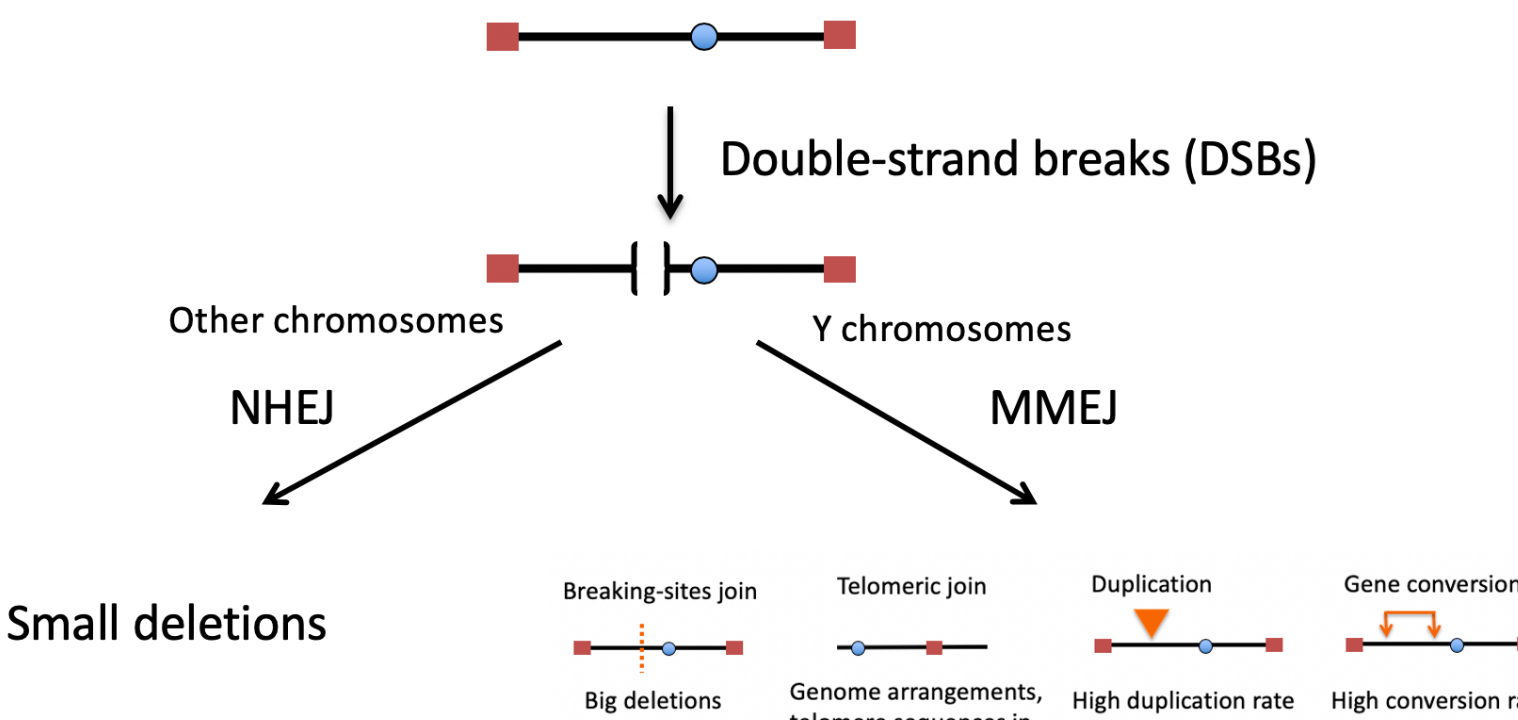
Common features of ampliconic Y genes

- I. Positive selection after duplicating to the Y
- II. Massive copies (>40) on Ys
- III. High gene conversion between Y copies (10^{-4} to 10^{-6} event per bp/generation)
- IV. Nuclear localization and function in serine phosphorylation

Y-specific mutation patterns?

Model: Microhomology-mediated end joining

- I. NHEJ is rare in heterochromatin regions
- II. No homologous recombination



Acknowledgements

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