

# Evolución de familias multigénicas 2020

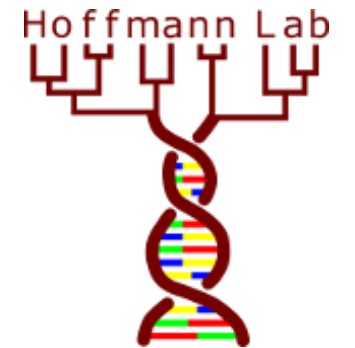
---

Reparto incompleto de linajes,  
introgresión



FACULTAD DE  
**CIENCIAS**

UDELAR | [fcien.edu.uy](http://fcien.edu.uy)



# Genes ortólogos y parálogos (Fitch, 1970)

rect usage. It is not sufficient, for example, when reconstructing a phylogeny from amino acid sequences that the proteins be homologous. It has been pointed out before that a phylogeny of birds and mammals based upon a haphazard mixture of  $\alpha$  and  $\beta$  hemoglobins would be biological nonsense since the initial dichotomy would be on the distinction between the  $\alpha$  and  $\beta$  genes rather than between the birds and the mammals (Fitch and Margoliash, 1967). Therefore, there should be two subclasses of homology. Where the homology is the result of gene duplication so that both copies have descended side by side during the history of an organism, (for example,  $\alpha$  and  $\beta$  hemoglobin) the genes should be called *paralogous* (para = in parallel). Where the homology is the result of speciation so that the history of the gene reflects the history of the species (for example  $\alpha$  hemoglobin in man and mouse) the genes should be called *orthologous* (ortho = exact). Phylogenies require orthologous, not paralogous, genes. Note

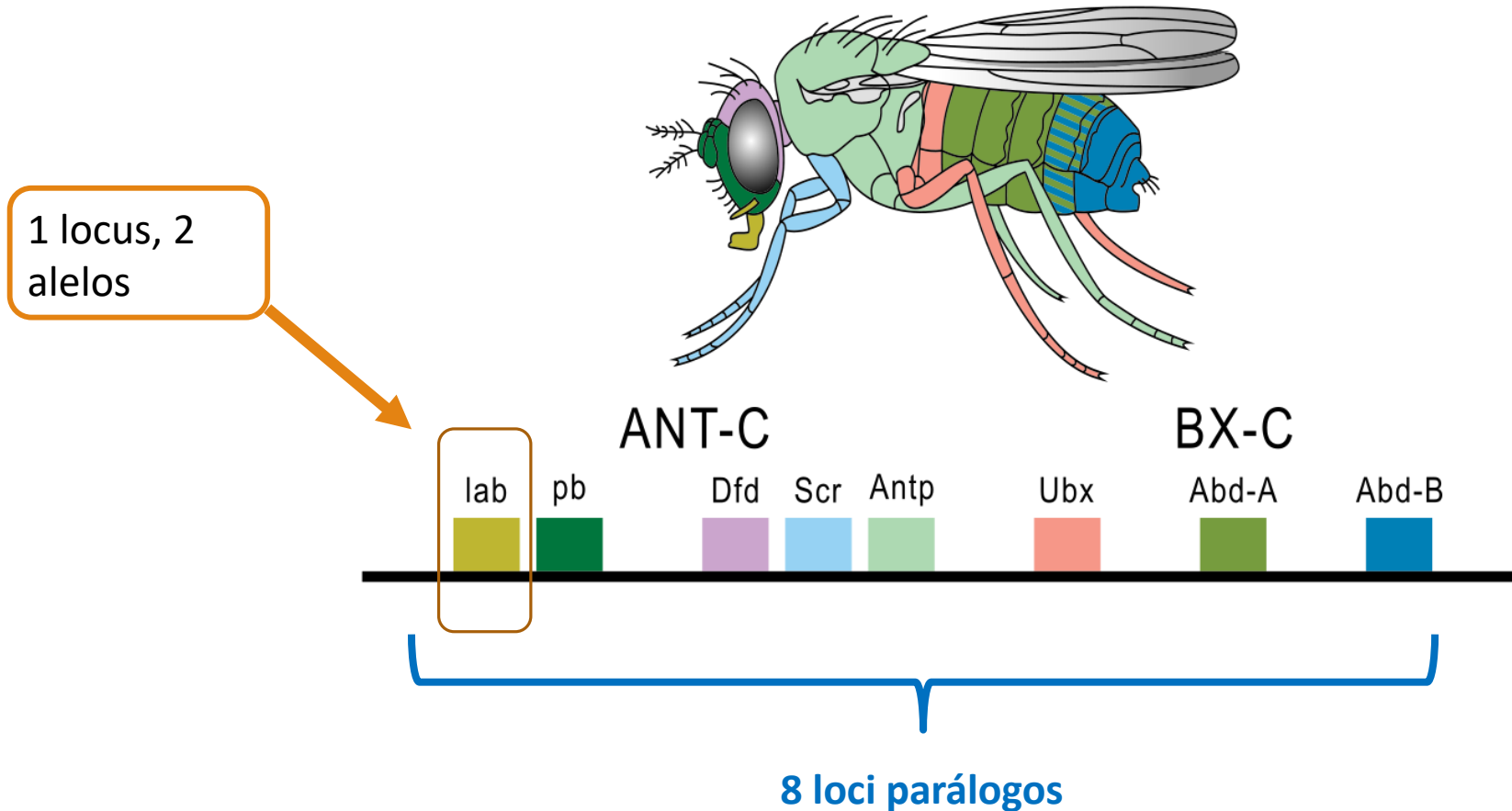
---

Dos subclases de homología:

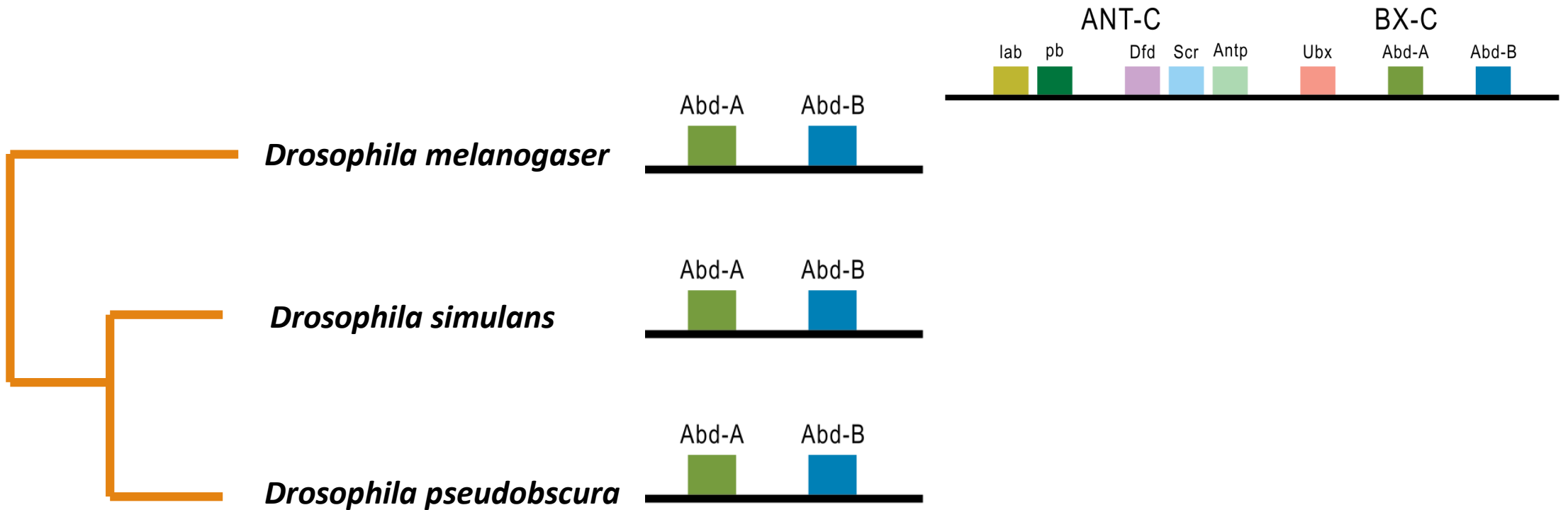
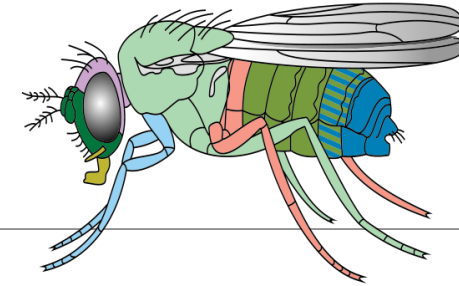
- La resultante de duplicaciones génicas... se transmiten lado a lado (**loci parálogos**).
- La resultante de la especiación... la historia del gen refleja la historia de las especies (**un mismo locus; loci ortólogos**).

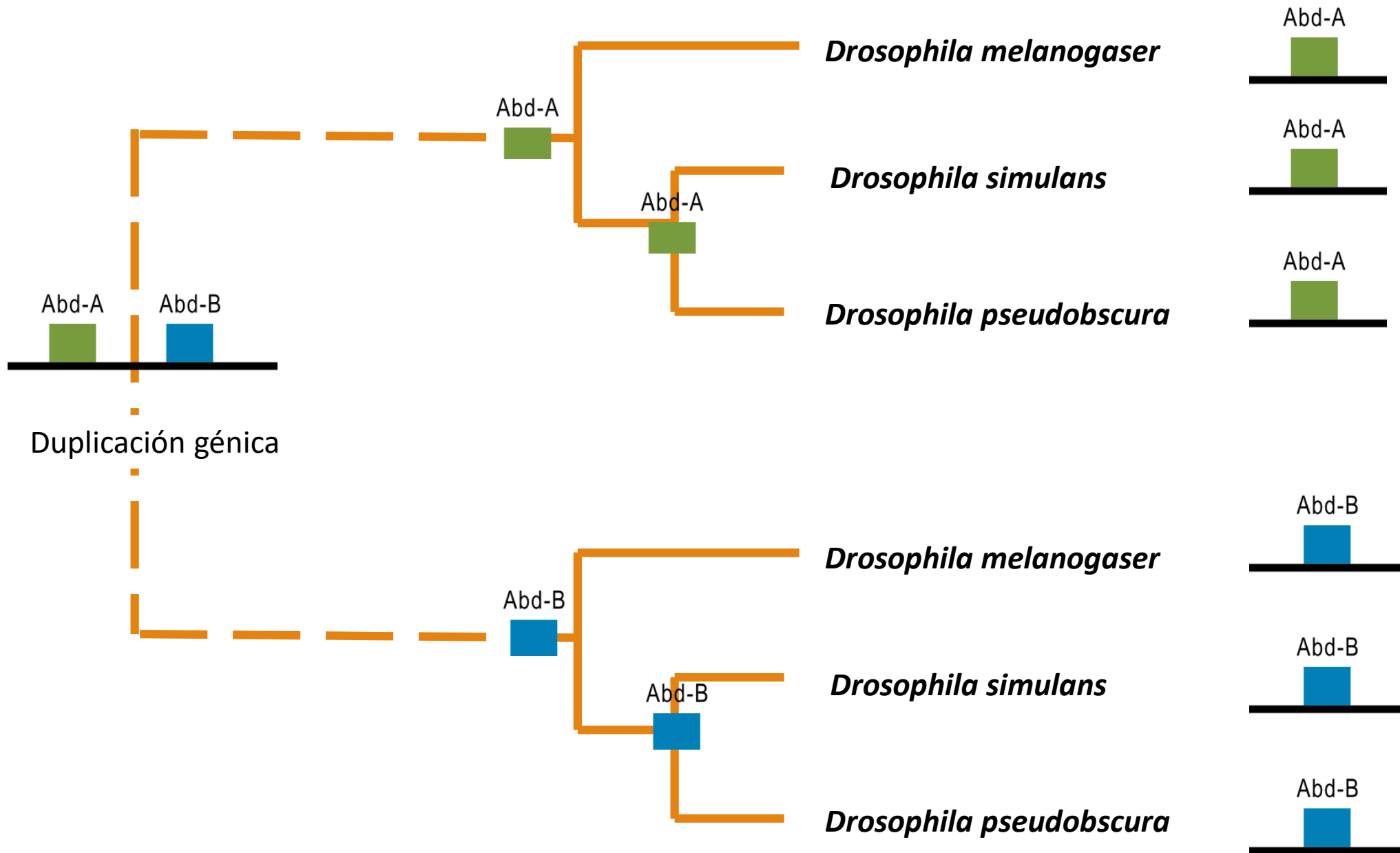
W. M. Fitch. 1970. Distinguishing homologous from analogous proteins. *Systematic Zoology*, 19: 99-113.

# Un solo individuo diploide

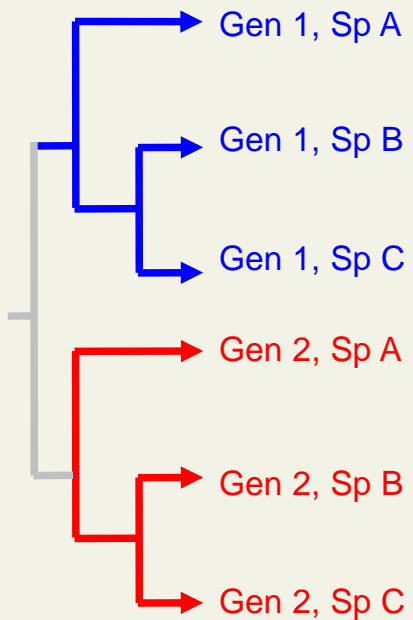


# Escala filogenética

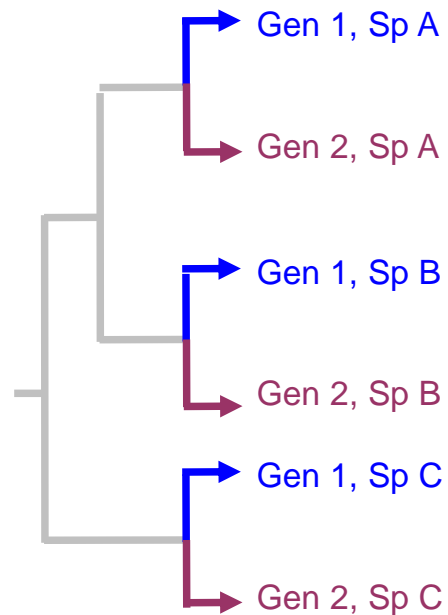
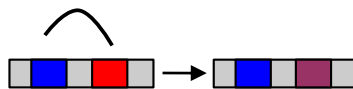




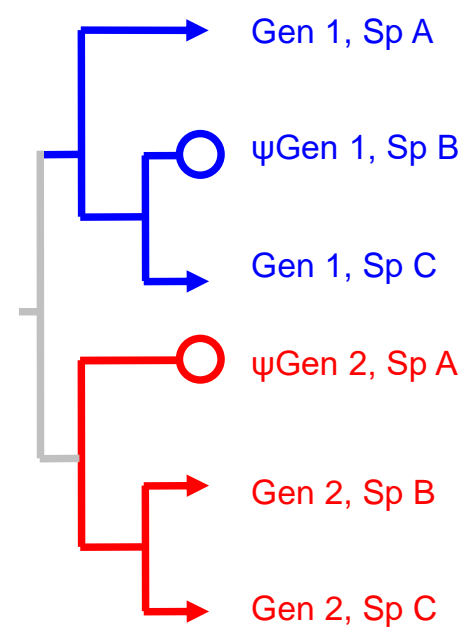
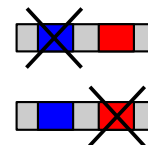
**Nacimiento puro  
(pure birth)  
Evolución divergente**



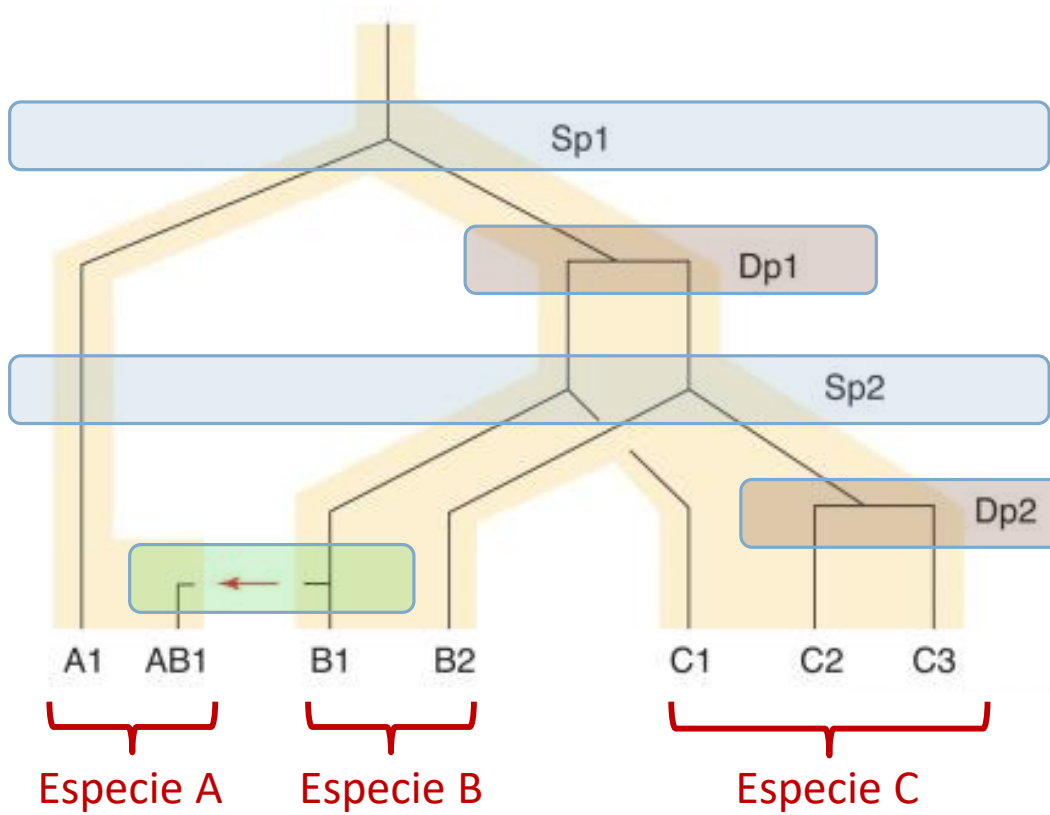
**Transferencia horizontal  
de información  
(evolución concertada)**



**Nacimiento y  
muerte  
(birth-and-death)**



Adaptado de Futuyma (2005)

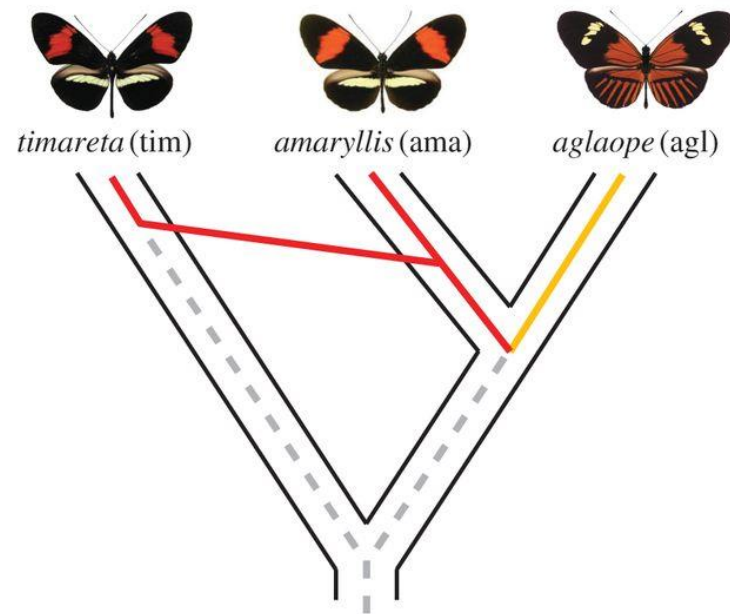


The idealized evolution of a gene (lines) is shown from a common ancestor in an ancestral population (the gray background), descending to three populations labelled A, B and C. There are two speciation events (Sp1 and Sp2), each occurring at the junctions shown as an upside down Y. There are also two gene-duplication events (Dp1 and Dp2), depicted by a horizontal bar. Two genes whose common ancestor resides at a Y junction (speciation) are orthologous. Two genes whose common ancestor resides at a horizontal bar junction (gene duplications) are paralogous. Thus, C2 and C3 are paralogous to each other but are orthologous to B2. Both are paralogous to B1 but orthologous to A1. The red arrow denotes the transfer of the B1 gene from species B to species A. As a result, the AB1 gene is xenologous to all six other genes. All three subtype relationships are reflexive, that is,  $A1 \Rightarrow B1$  implies  $B1 \Rightarrow A1$  where  $\Rightarrow$  should be read, for example, as 'is orthologous to.' However, the relationships are not transitive. Thus,  $C2 \Rightarrow A1 \Rightarrow C3$  might be true, but it is not necessarily therefore true that  $C2 \Rightarrow C3$ , as indeed it is not in the figure if  $\Rightarrow$  is read as 'is orthologous to.' A different non-transitivity occurs for 'is paralogous to' with  $B2 \Rightarrow C1 \Rightarrow C2$ .

Fitch, W. M. 2000. Homology: a personal view of some of the problems. Trends in Genetics 16: 227-231.

# Transferencia horizontal

(a) introgression (*melpomene* into *timareta*)



introgresión

## Orthology

The relationship of any two homologous characters whose common ancestor lies in the cenancestor of the taxa from which the two sequences were obtained.

## Paralogy

The relationship of any two homologous characters arising from a duplication of the gene for that character.

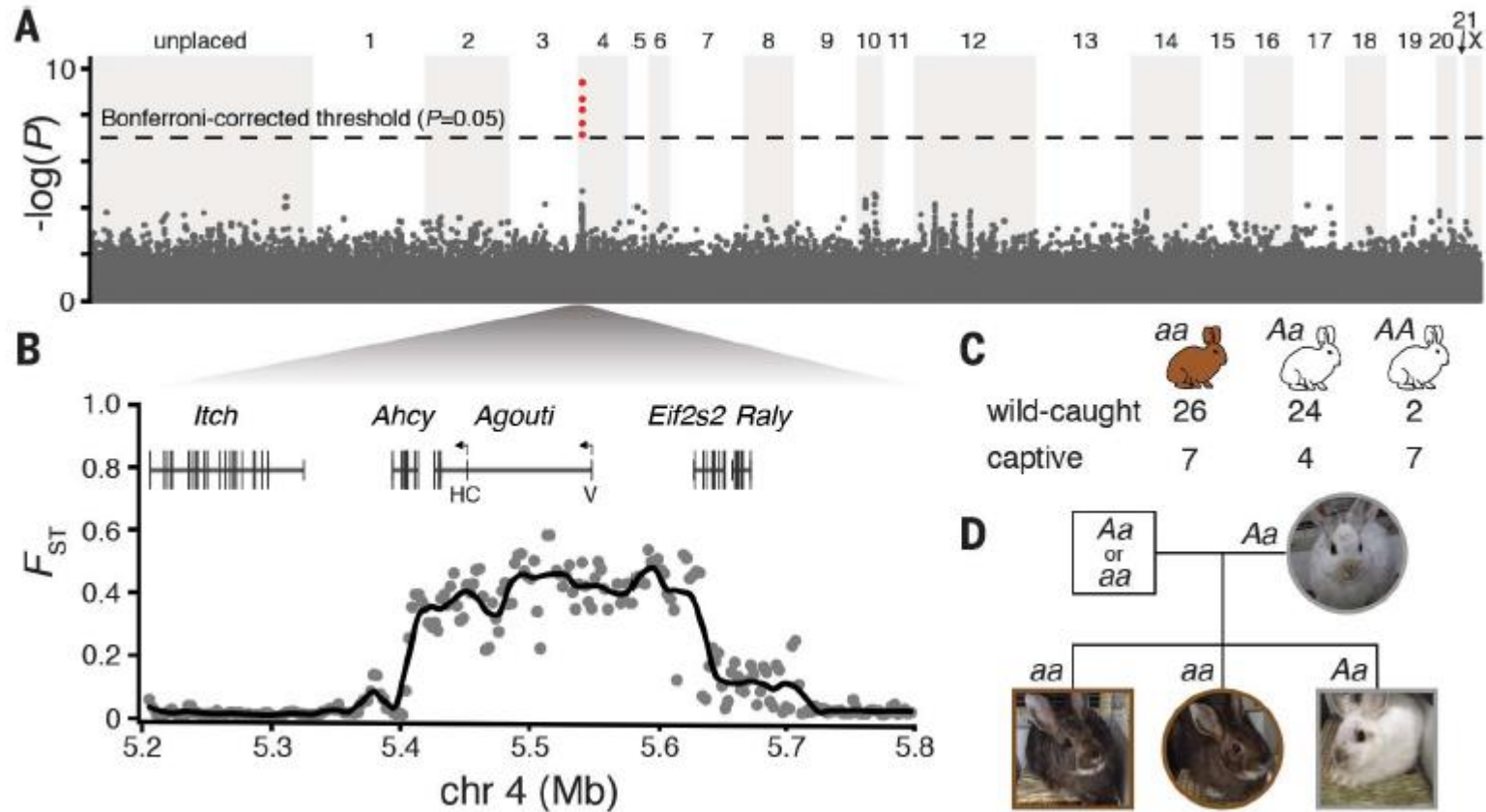
## Xenology

The relationship of any two homologous characters whose history, since their common ancestor, involves an interspecies (horizontal) transfer of the genetic material for at least one of those characters.

Fitch, W. M. 2000. Homology: a personal view of some of the problems. Trends in Genetics 16: 227-231.



# Introgresión adaptativa en liebres



Jones et al. 2018. Science 360: 1355-1358.

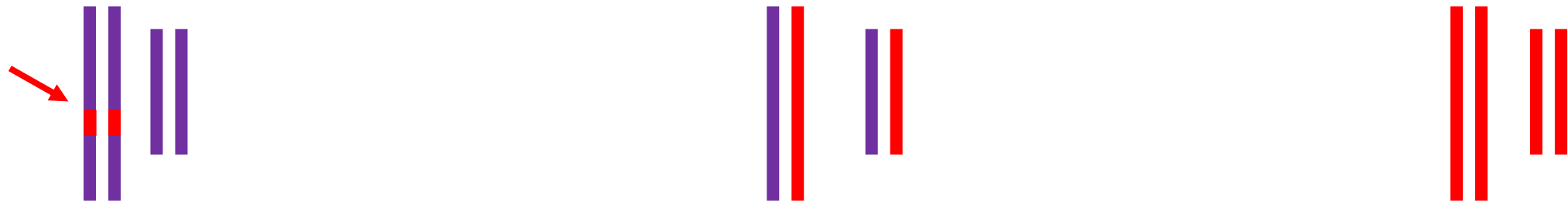
# Introgresión

---

La introgresión resulta en una discordancia entre el árbol de las especies y el árbol de los genes.

La evolución de los genes sigue siendo “vertical”, pero ocurre transferencia horizontal de alelos (no confundir con conversión génica).

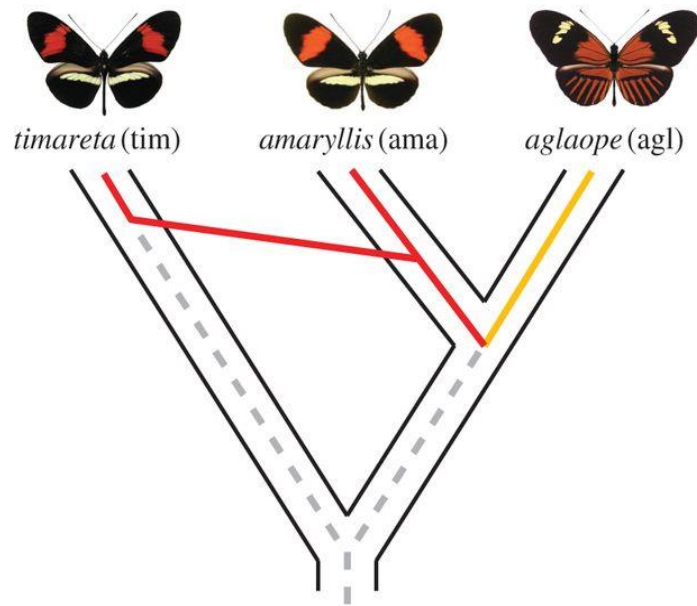
La hibridización (formación de híbridos en sentido amplio, incluyendo retrocruzadas, etc.) puede ser necesaria, pero no suficiente para que ocurra introgresión.



Introgresión y adaptación: la introgresión puede implicar fracciones muy pequeñas del genoma, pero éstas pueden ser adaptativas.

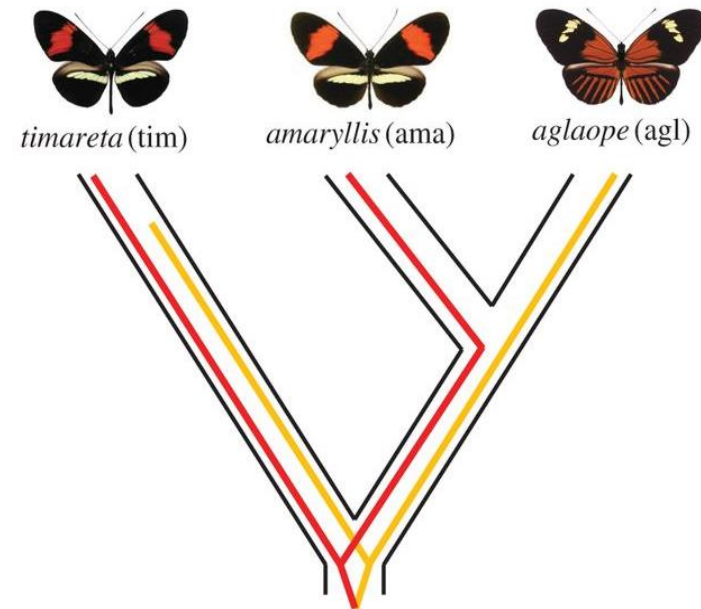
# Transferencia horizontal vs. reparto incompleto de linajes (ILS: incomplete lineage sorting)

(a) introgression (*melpomene* into *timareta*)

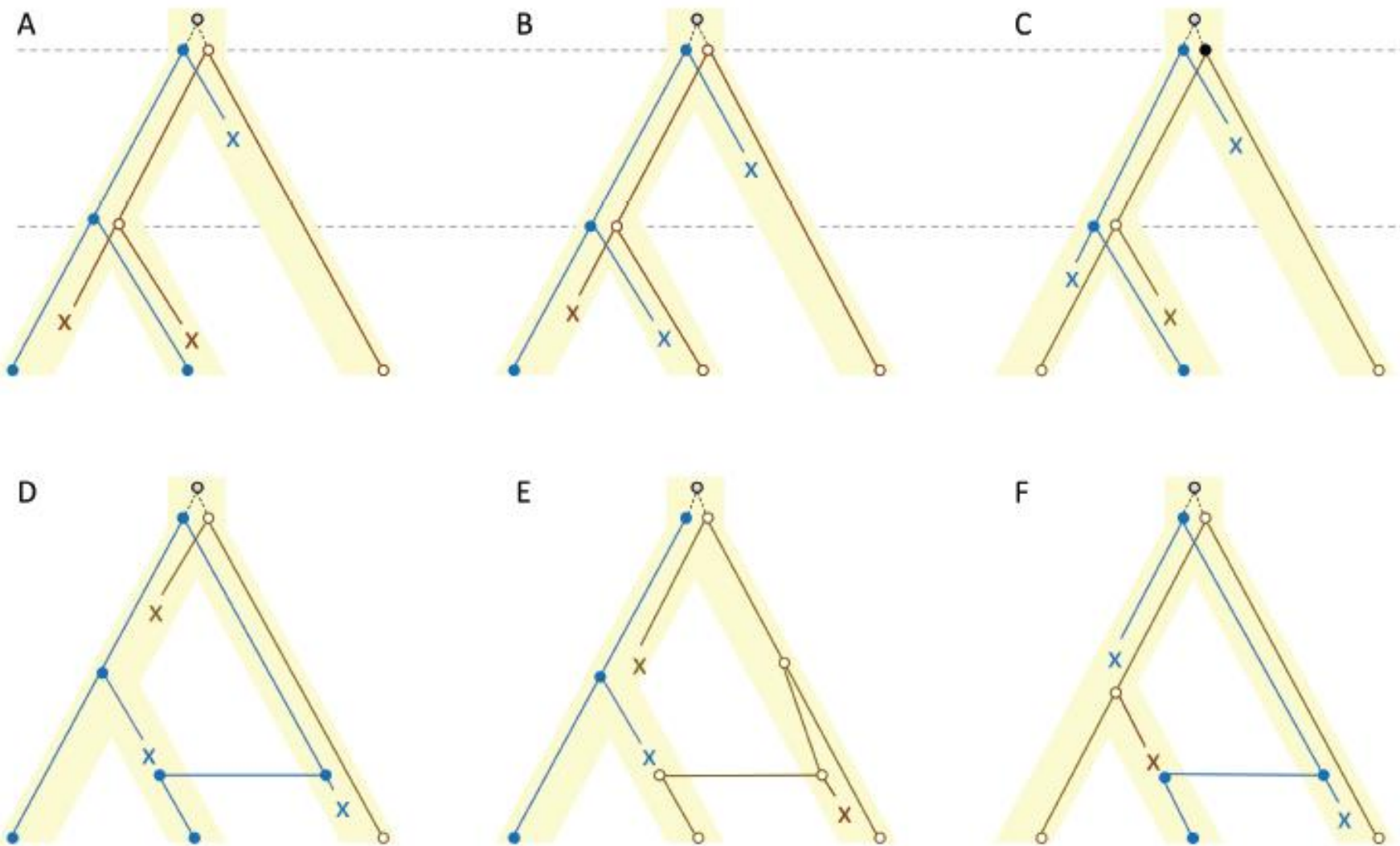


introgresión

(c) ancestral polymorphism



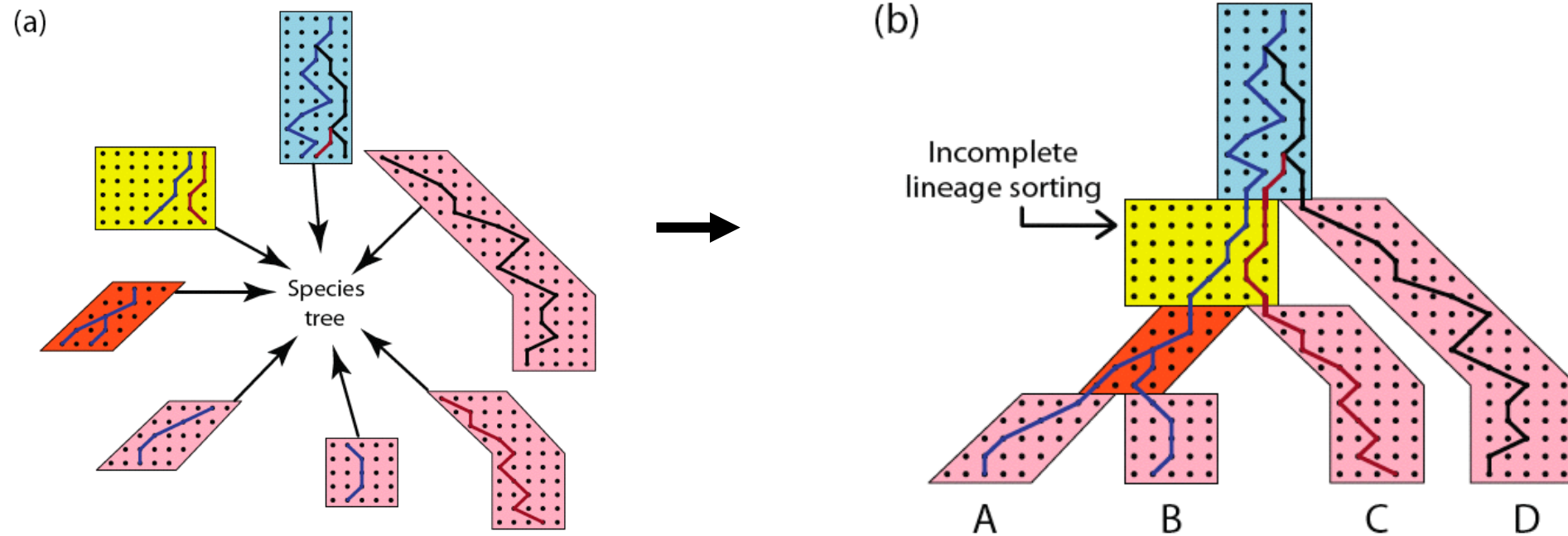
Reparto incompleto de linajes  
("incomplete lineage sorting")



**Fig. 1.**—Relationships between gene (allele) trees (lines) and species trees (shaded area). The ancestry of alleles (circles) of three species, resulting from two successive speciation events, is tracked to two ancestral alleles. The symbol X signals allele extinction. A) a gene tree congruent with the species tree. B, C) gene trees incongruent as a result of incomplete lineage sorting. D–F) the same gene tree topologies of A–C, but resulting from introgression.

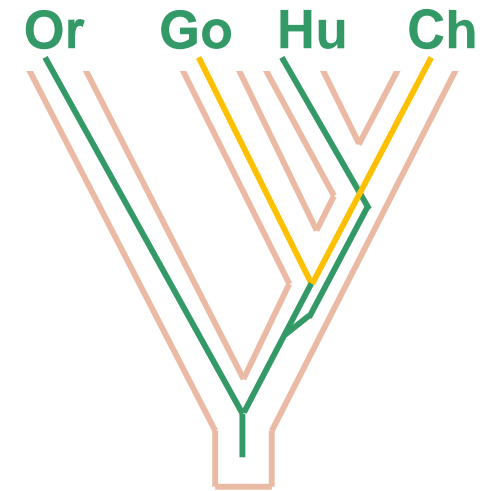
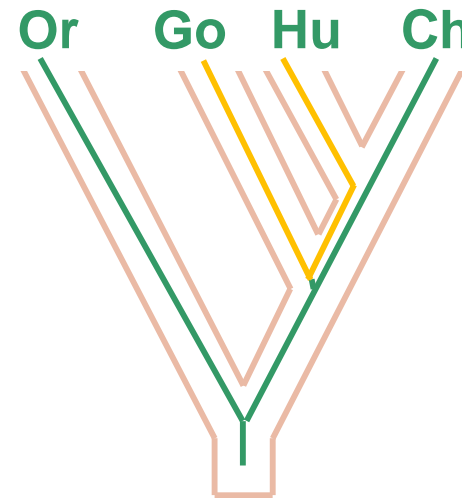
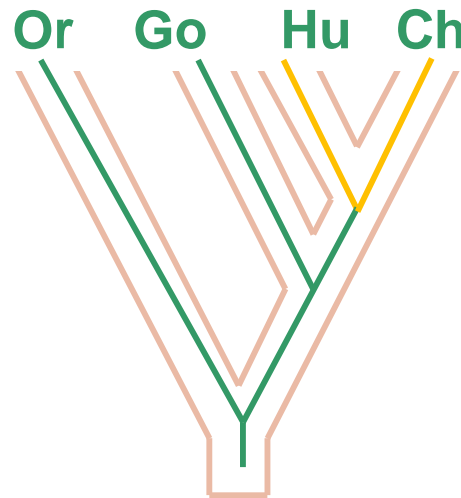
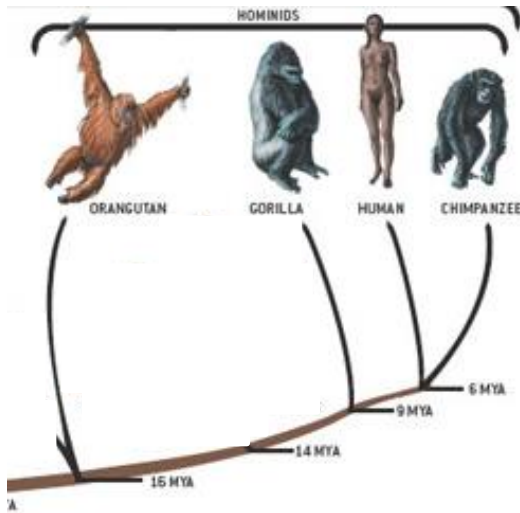
D’Elía et al. 2019. Rodent systematics in an age of Discovery: recent advances and prospects. *Journal of Mammalogy* 100: 852-871.

# Árboles de genes en un árbol de especies

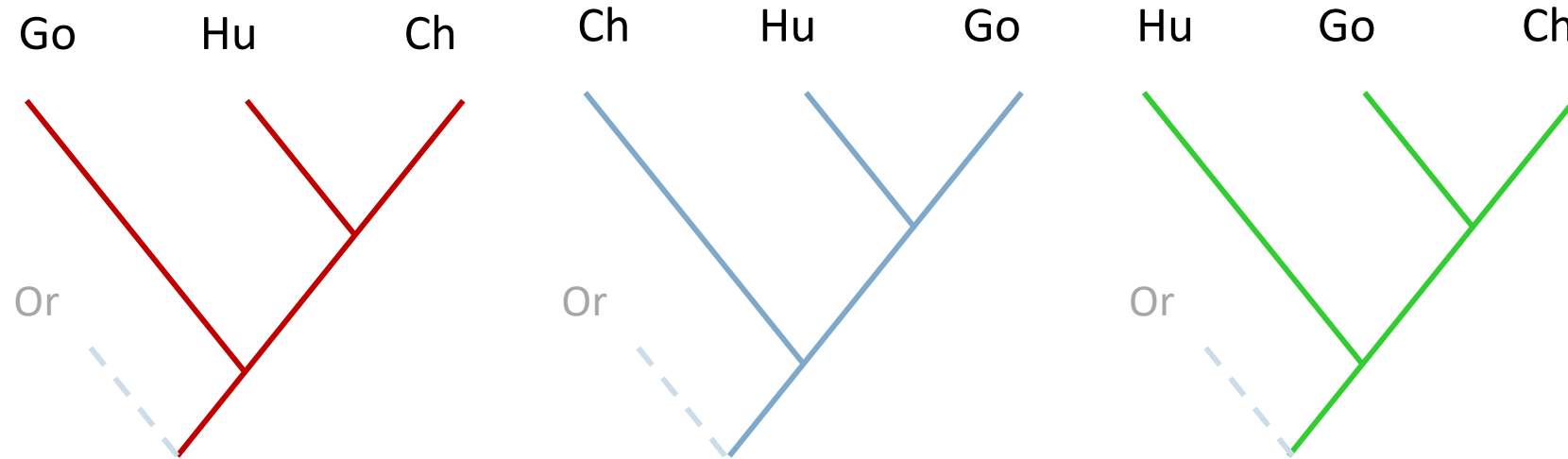
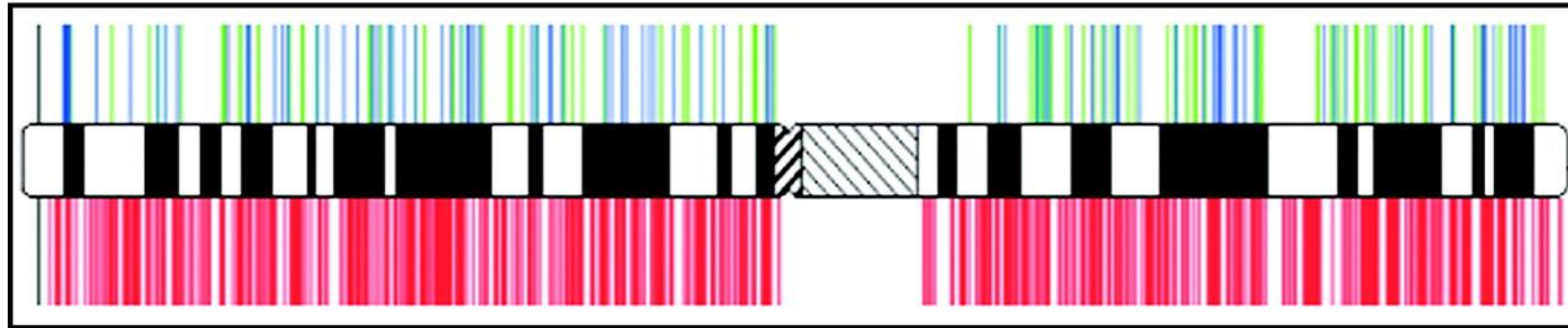


Degnan & Rosenberg. 2009. *Trends in Ecology and Evolution* 24: 332-340

# Árboles de genes en un árbol de especies



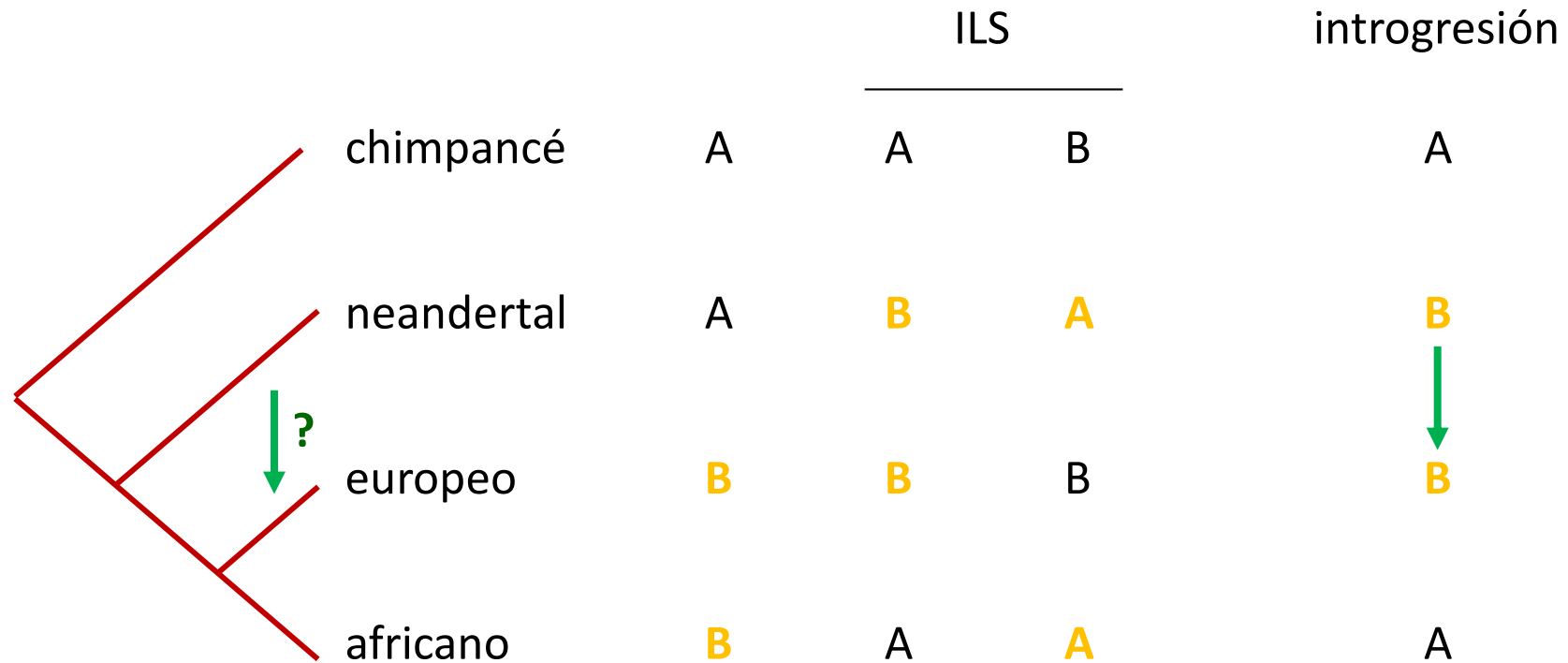
# ILS en cromosoma 1 humano



Ebersberger et al. 2007. *Mol Biol Evol.* 24:2266-2276



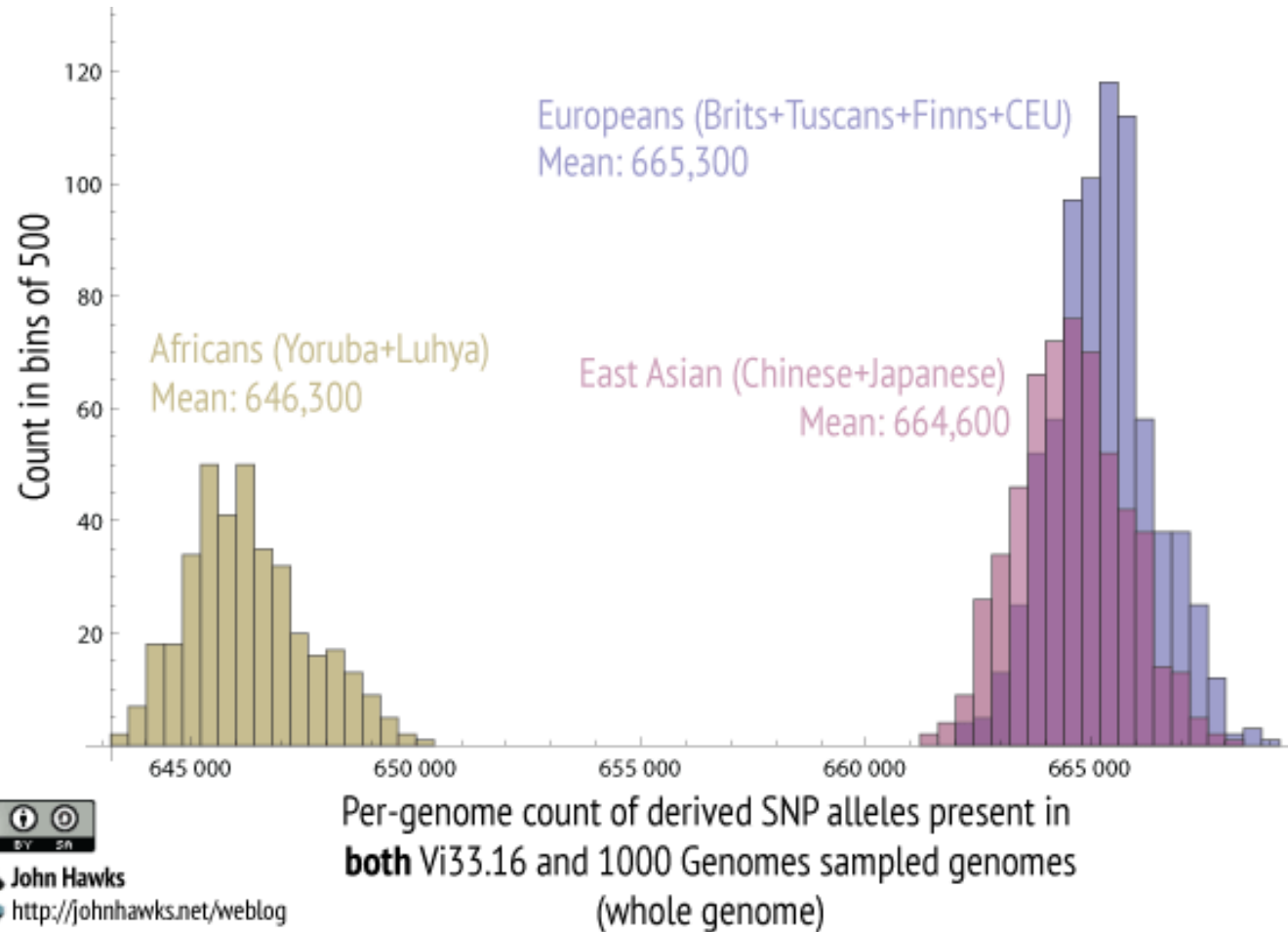
# Pruebas (tests) de introgresión: ABBA-BABA test



Idea básica: ILS produce frecuencias idénticas de ABBA y BABA; la introgresión de neandertal a europeos produce un exceso de ABBA.



# Exceso de “ABBA” en poblaciones humanas no africanas



John Hawks  
<http://johnhawks.net/weblog>

# Reparto incompleto de linajes (ILS)

---

Ocurre naturalmente como consecuencia de que, en todo momento, una población incluye múltiples alelos para un mismo locus.

El árbol de las especies (o poblaciones) condiciona, pero no determina el árbol de los genes.

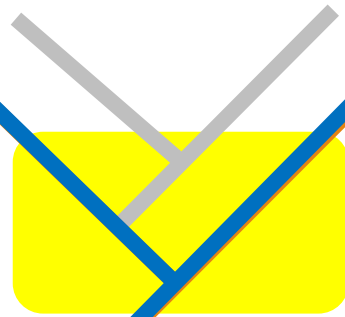
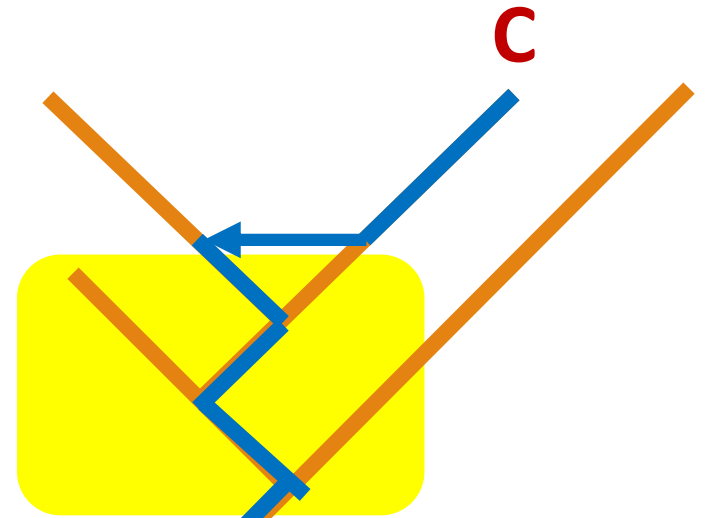
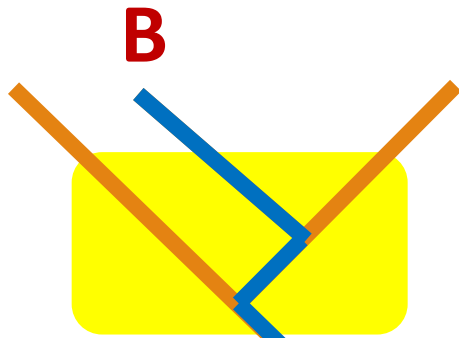
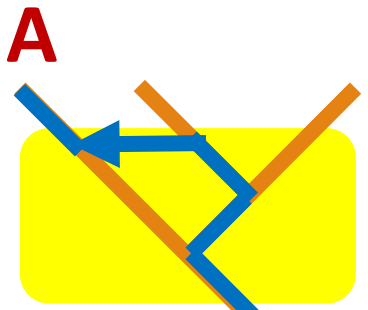
Para que ocurra incongruencia entre ambos tipos de árbol, un polimorfismo ancestral debe persistir entre dos o más eventos de especiación.

Para la variación neutral, la probabilidad de dicha persistencia:

- Aumenta si los tiempos entre eventos de especiación son pequeños.
- Aumenta si los tamaños poblacionales son grandes.

El efecto de la selección natural depende del tipo:

- Selección direccional positiva: reduce la probabilidad de ILS.
- Selección equilibradora: aumenta la probabilidad de ILS.



# Efecto de la escala filogenética

---

La introgresión y el reparto incompleto de linajes ocurren de manera permanente.

A escalas filogenéticas “grandes” (pensar que significan en la práctica), la extinción y el espaciamiento del muestreo pueden eliminar virtualmente sus efectos.

Otros mecanismos de transferencia horizontal pueden operar a mayores escalas que la introgresión mediada por hibridización.

Factores adicionales: clase de organismos, tipos de selección.