

Curso de Evolución 2023
Facultad de Ciencias
Montevideo, Uruguay

<http://eva.fcien.universidad.edu.uy/>

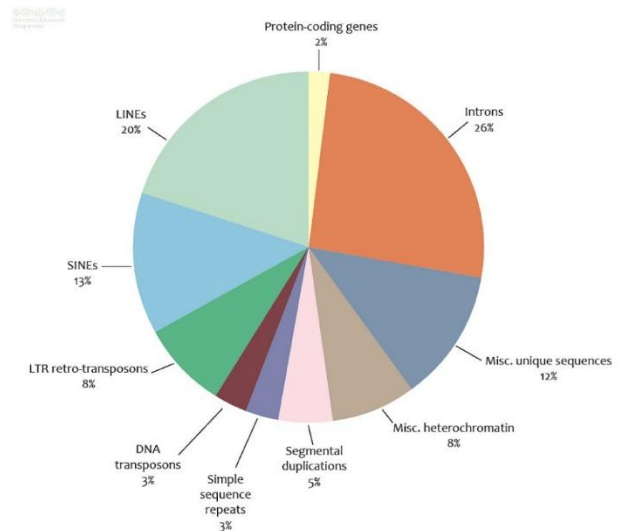
<https://www.youtube.com/@CursoEvolucion/videos>



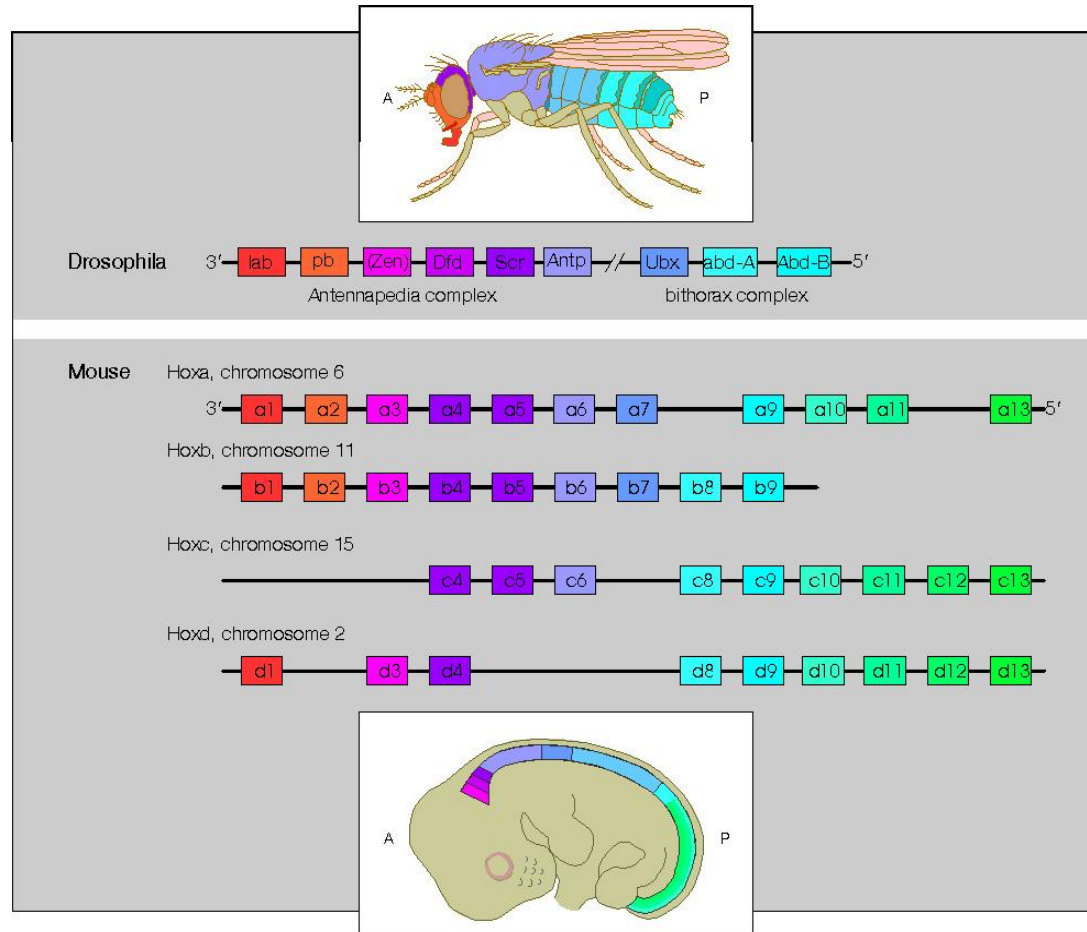
9. La evolución a nivel genómico. Organización y evolución del genoma. Evolución de familias multigénicas.

Evolución a escala genómica: algunos ejemplos

- Vamos a ilustrar, pero no a cubrir, todo el espectro de análisis de la evolución a escala genómica.
- Duplicación genómica
- Evolución de genes que codifican pigmentos visuales en vertebrados.
- El genoma del tuatara.
- Reparto incompleto de linajes e introgresión en humanos y parientes cercanos.

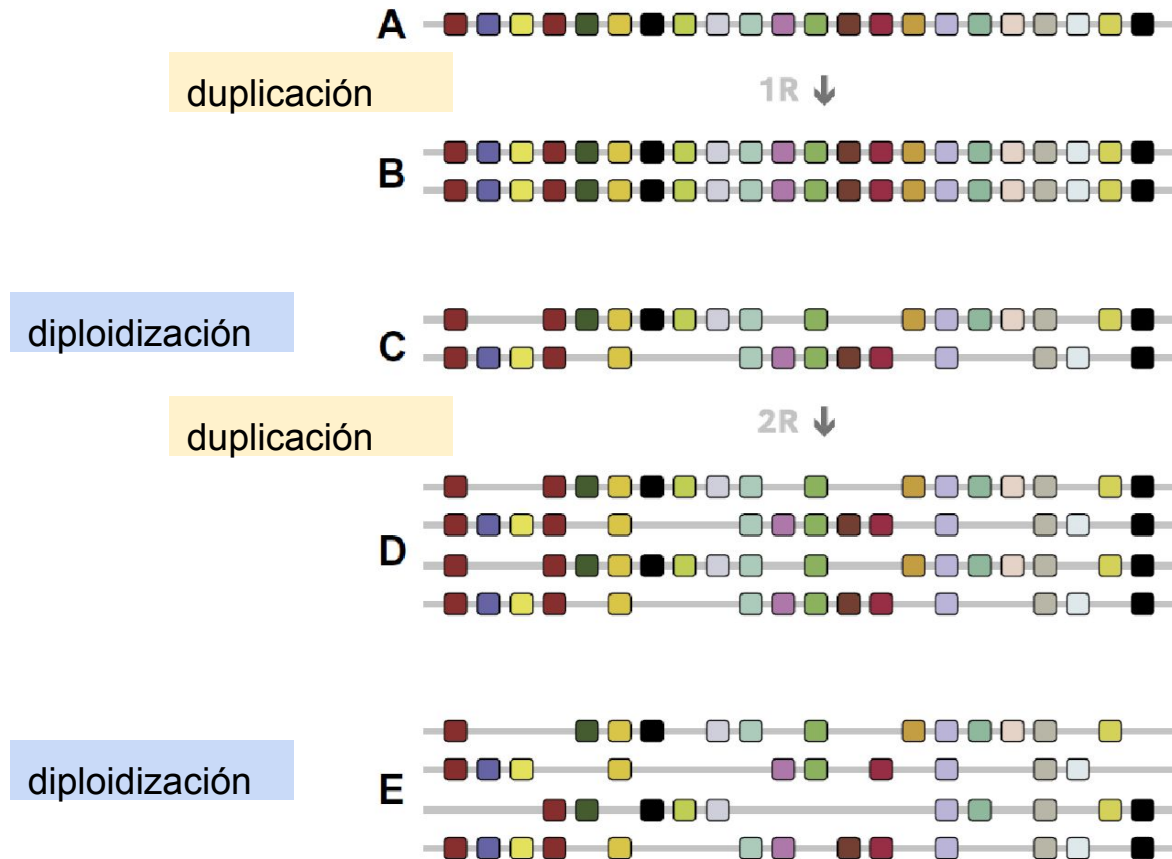


Dos rondas de duplicación genómica en vertebrados: genes Hox



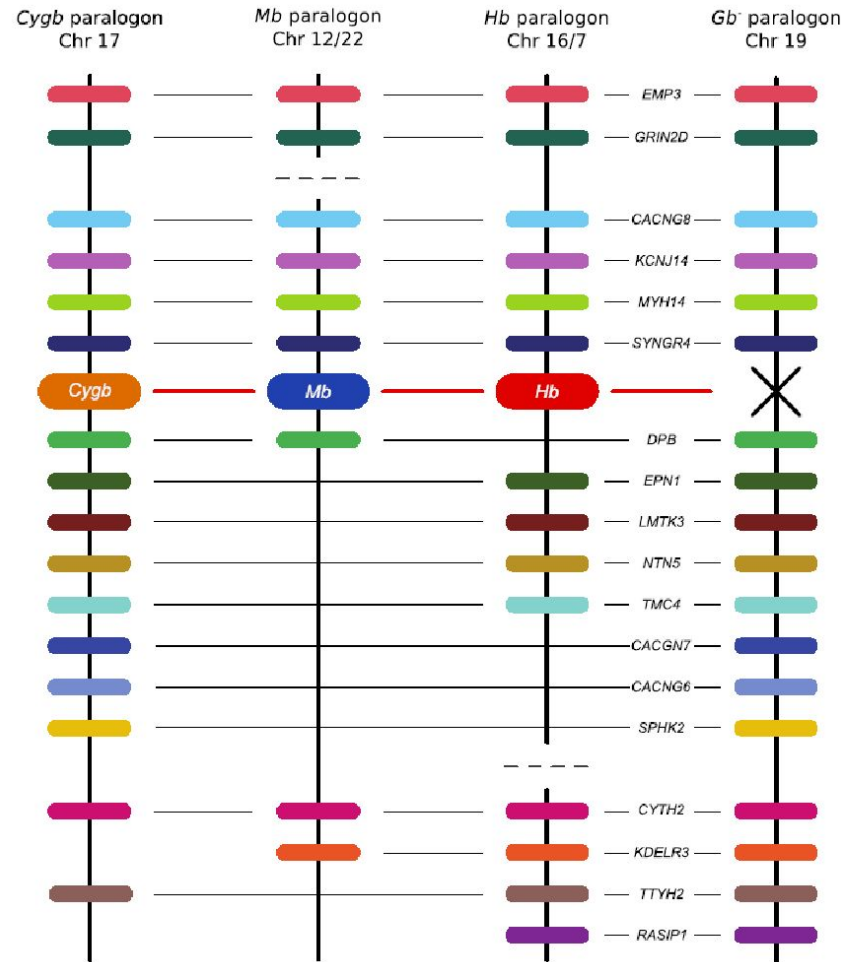
Ejemplo: familia de las globinas

Dos rondas de duplicación genómica en vertebrados
Predicción: “Paralogones”



Storz et al. Molecular Phylogenetics and Evolution 66 (2013) 469–478

Ejemplo: familia de las globinas



Cromosomas humanos

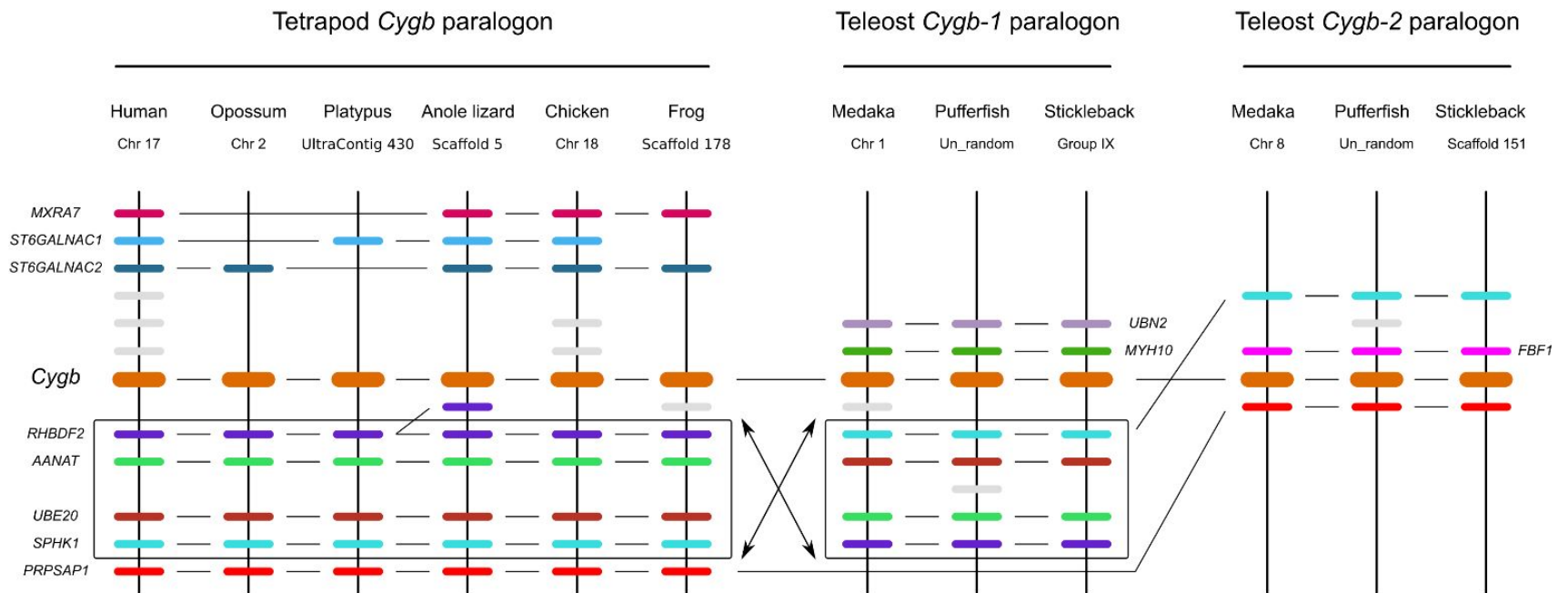


Fig. 7. Patterns of conserved synteny in the chromosomal region that harbors the *Cygb* gene in gnathostome vertebrates. Horizontal lines denote orthologous relationships. From Hoffmann et al. (2011).

Duplicación genómica en teleósteos

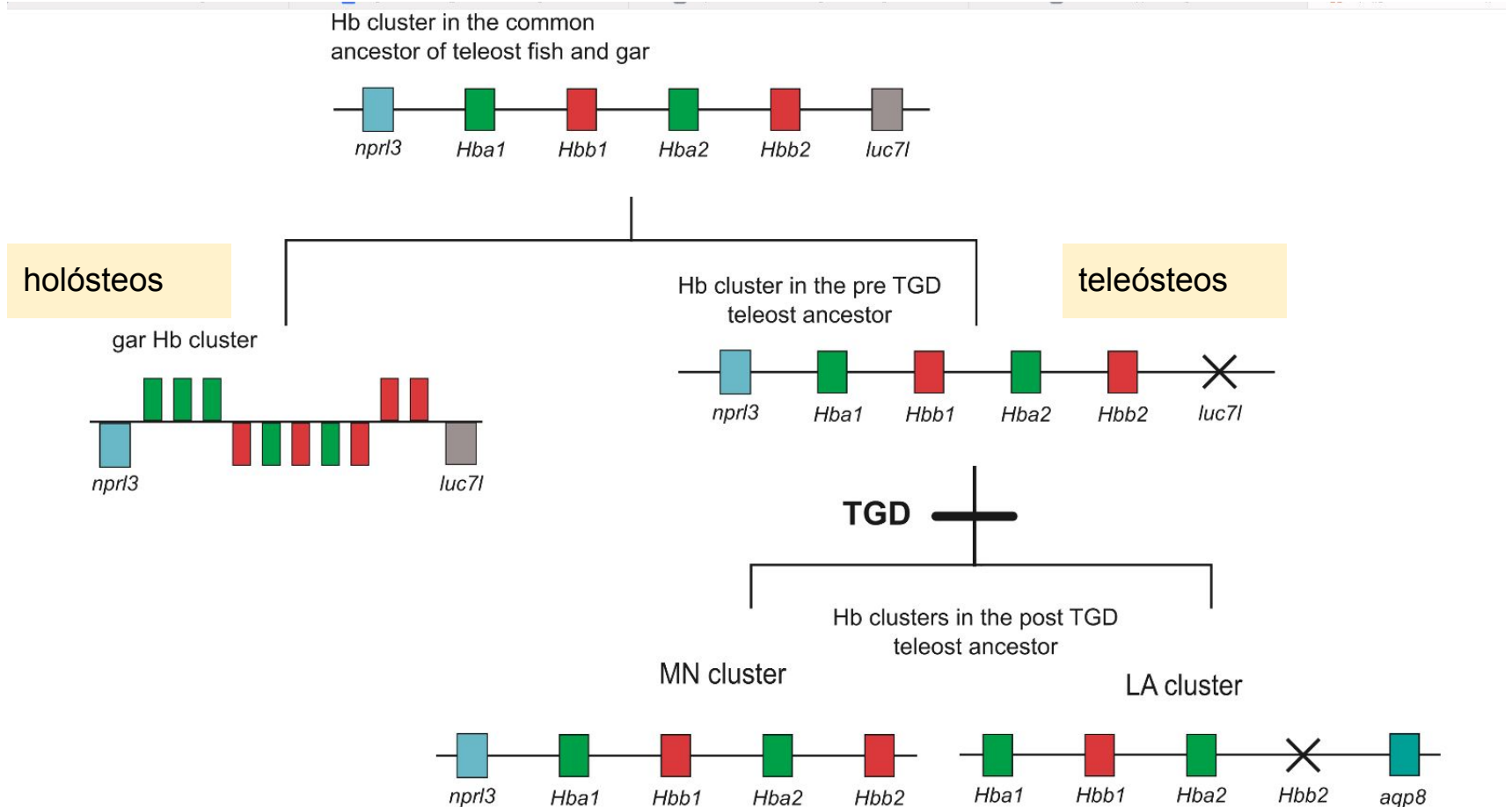
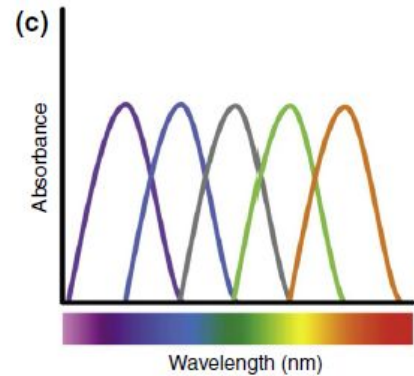
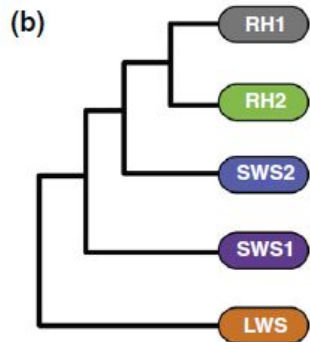


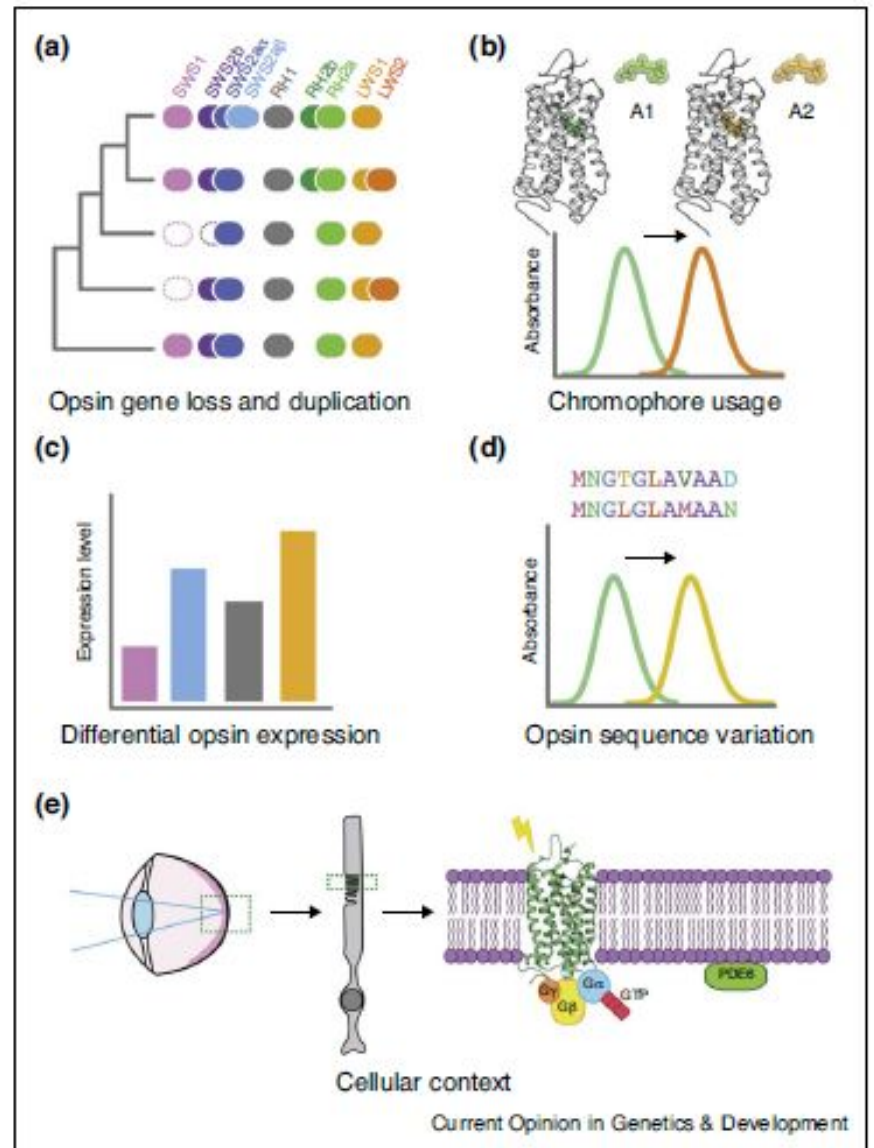
FIG. 4. Evolutionary model describing the duplicative origins of the LA and MN globin gene clusters of teleost fish and the inferred globin gene repertoire in the common ancestor of teleosts and gar, a nonteleost ray finned fish. All clusters depicted are hypothetical with the exception of the gar cluster. The order of the α - and β -globin genes on the hypothetical clusters is arbitrary.

Evolución de pigmentos visuales

- Una familia multigénica: beneficios de cobertura del problema al usar datos genómicos.
 - Cobertura completa, incluyendo pseudogenes.
 - Estructura genómica de la familia.
 - Regiones reguladoras.
 - ...

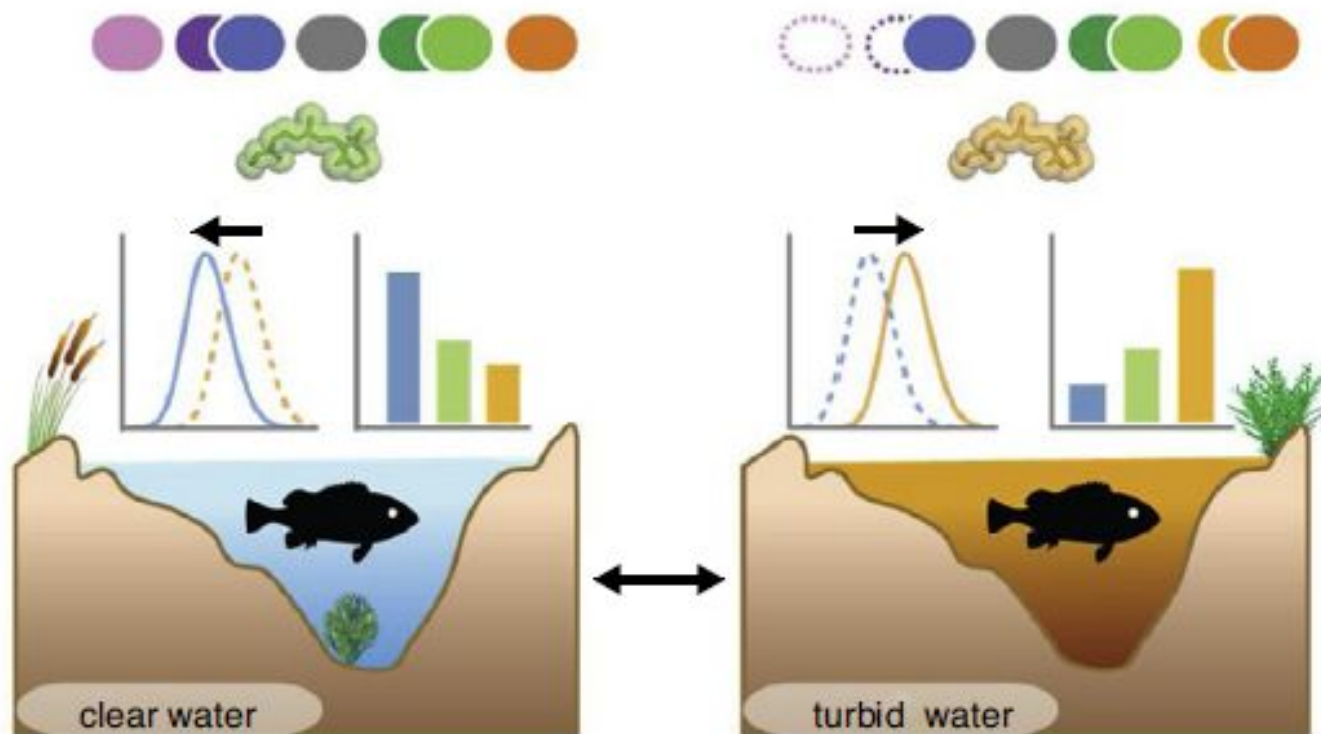


Current Opinion in Genetics & Development



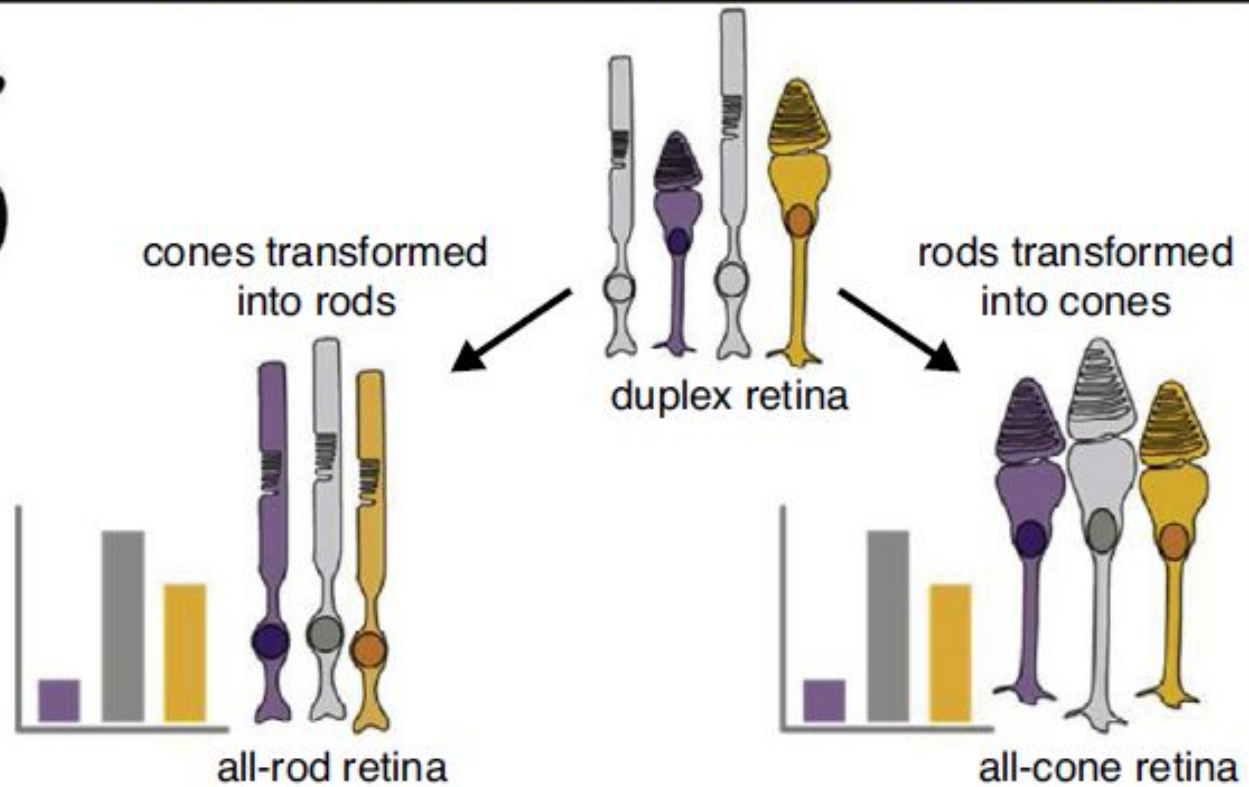
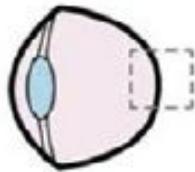
Hauser & Chang 2017.
Current Opinion in Genetics and Development.

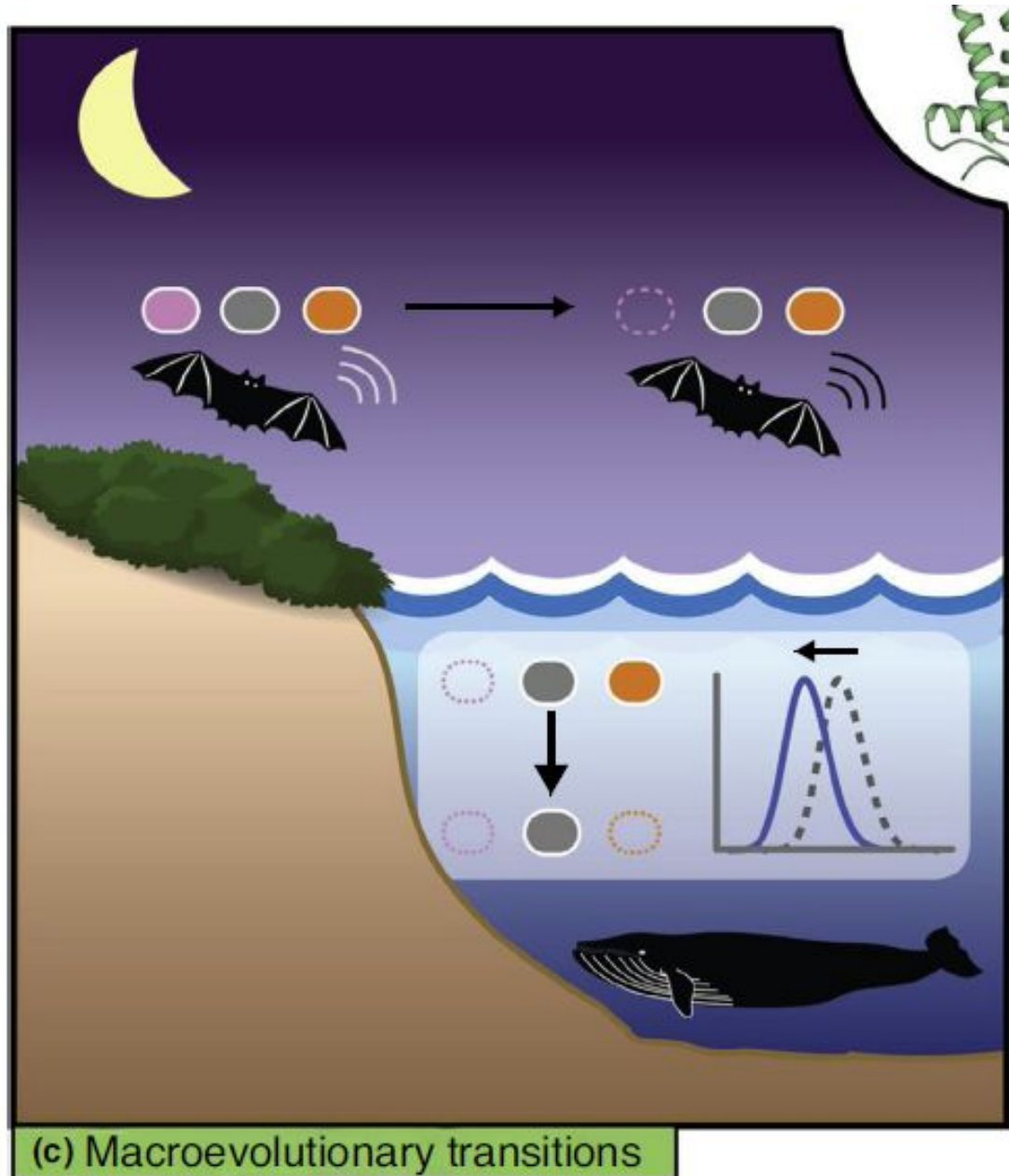
(a) Ambient light environment



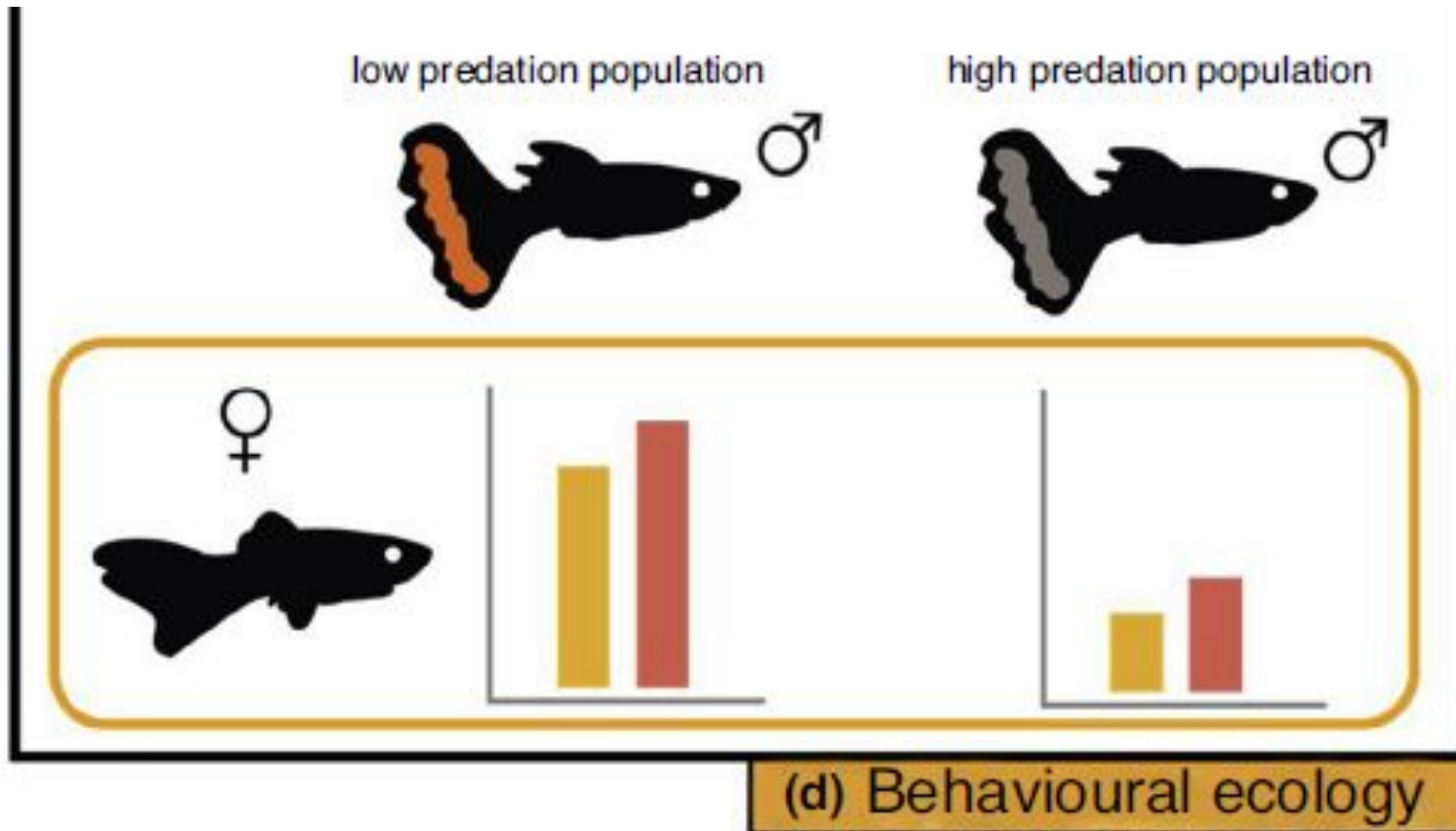
Hauser & Chang 2017.
Current Opinion in Genetics and Development.

(b) Eye anatomy and physiology



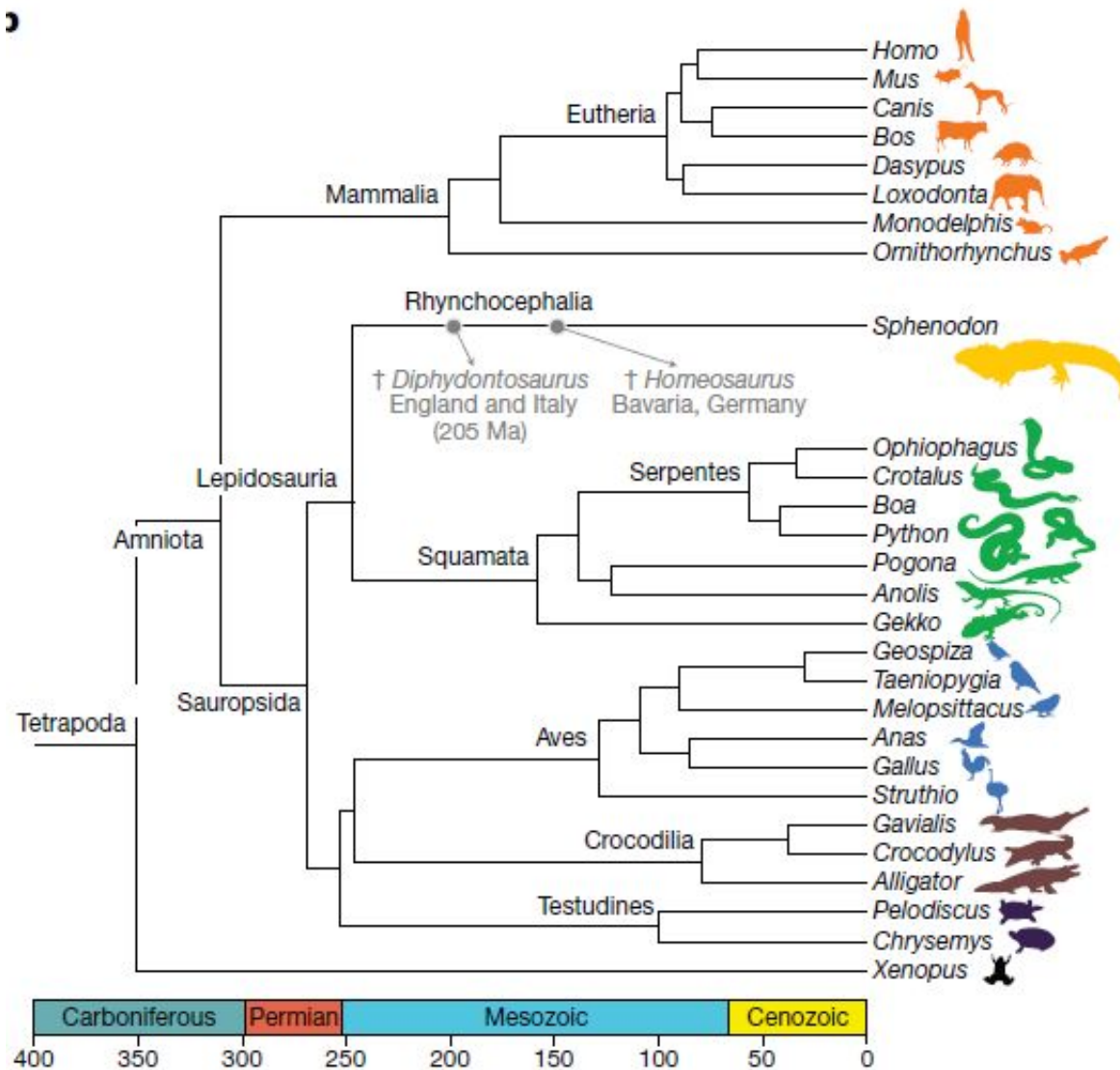


Hauser & Chang
2017.
Current Opinion in
Genetics and
Development.



Hauser & Chang 2017.
Current Opinion in Genetics and Development.

Genoma del tuatara: posición filogenética



Gemmel et al. 2020. The tuatara genome reveals ancient features of amniote evolution. Nature

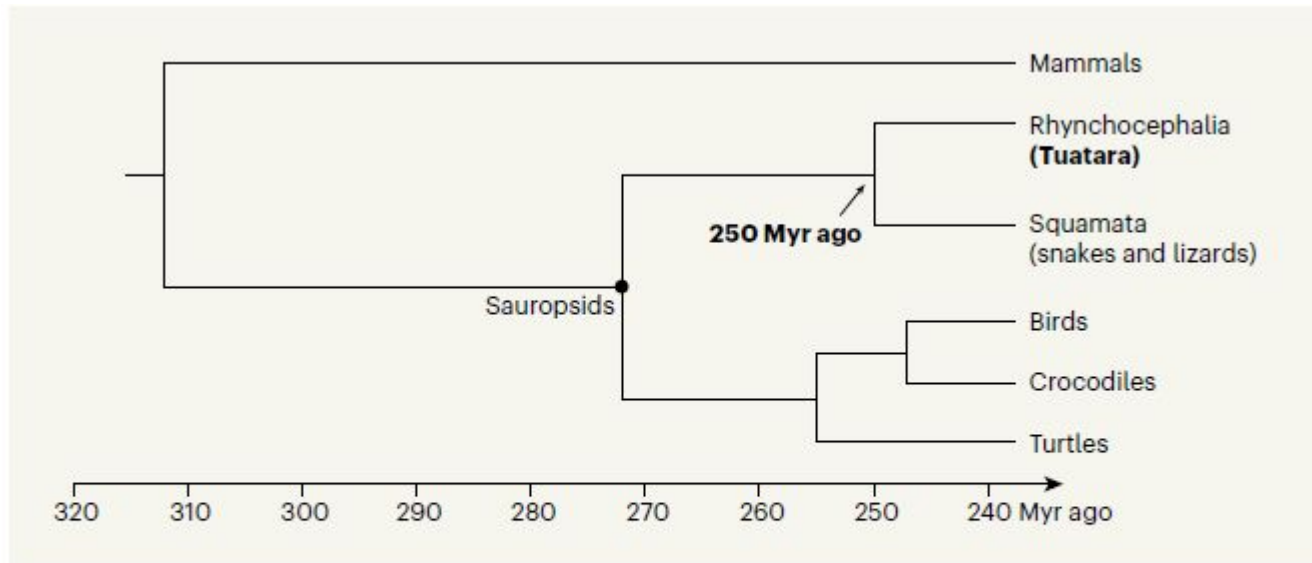
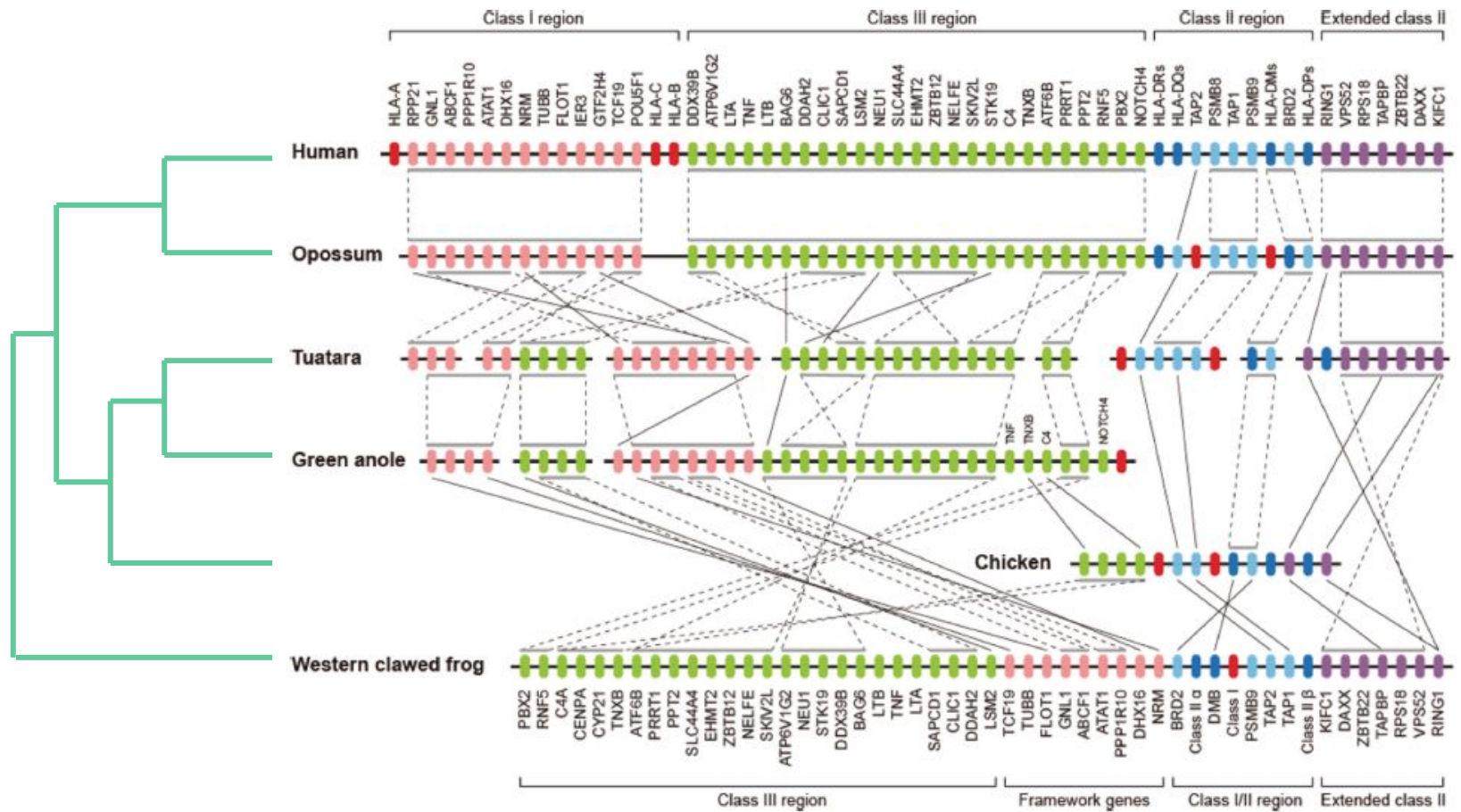


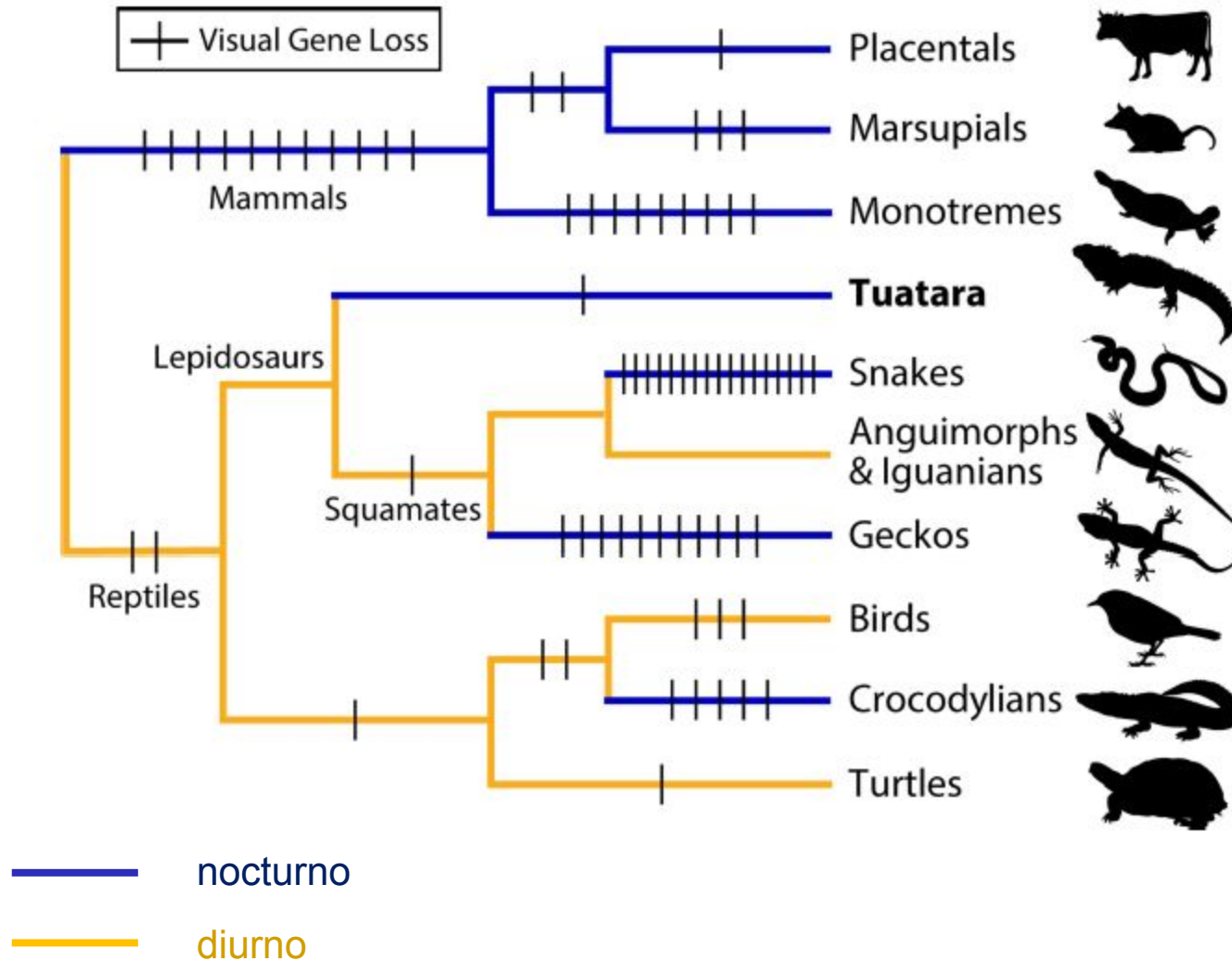
Figure 2 | Refining the evolutionary tree for reptiles, birds and mammals. This phylogenetic tree includes six branches: mammals and five branches within a clade called sauropsids, which comprises reptiles and birds. One of these, the Rhynchocephalia, has only one living member, the tuatara. Gemmell and colleagues date the divergence of the Rhynchocephalia from the Squamata to about 250 million years (Myr) ago.

- Genoma 50% mayor que el humano.
- Gran cantidad de secuencias repetidas (muchas “activas”).
- Posición filogenética y antigüedad de ancestros comunes.
- Baja tasa de evolución (tiempo generacional, baja temperatura corporal).

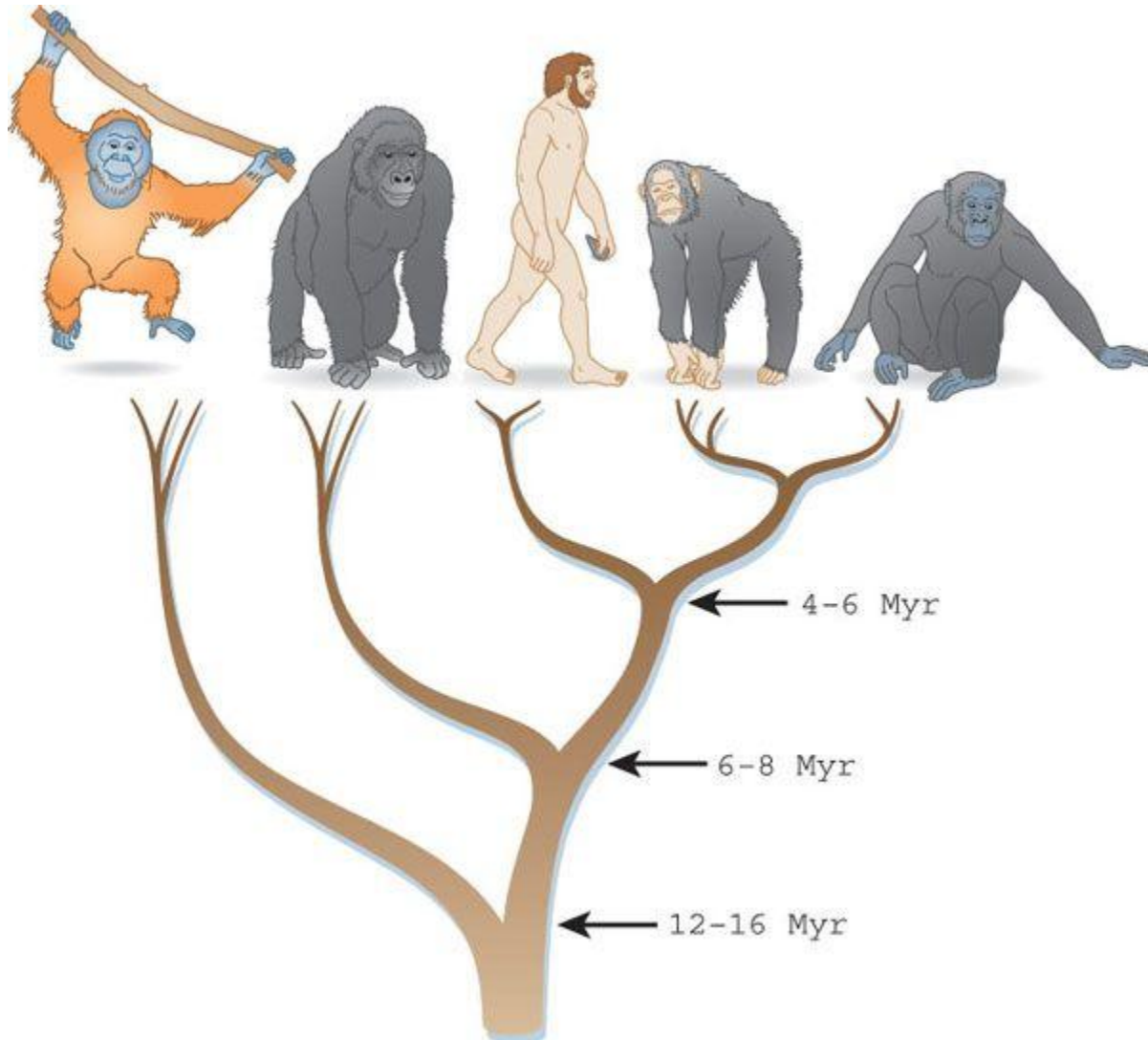
Genes MHC



Pérdida de genes del sistema visual

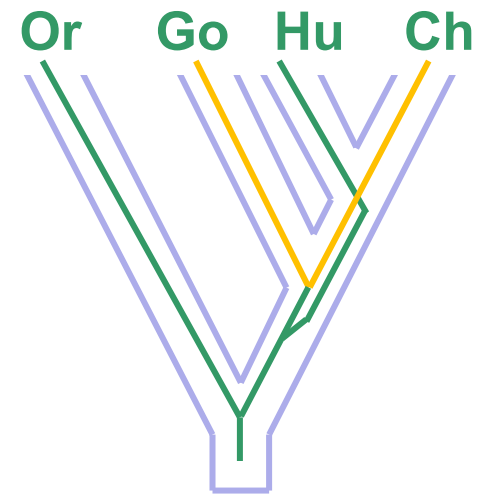
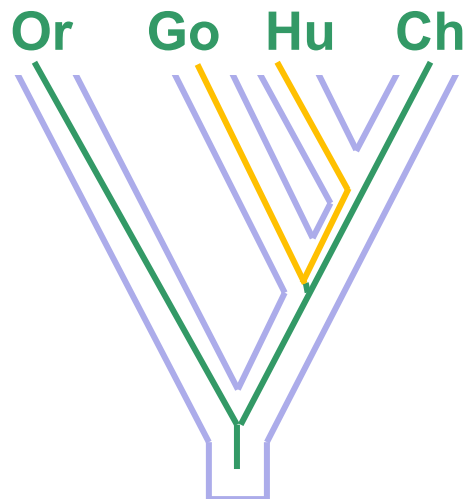
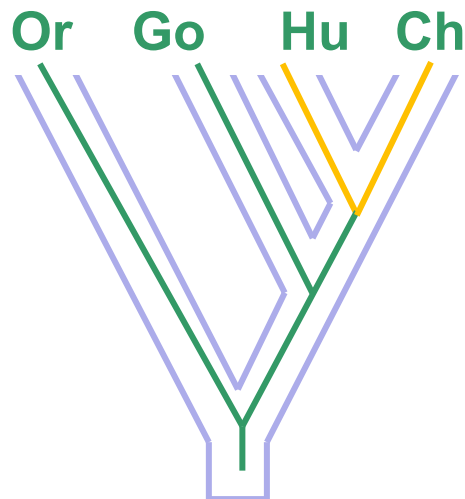
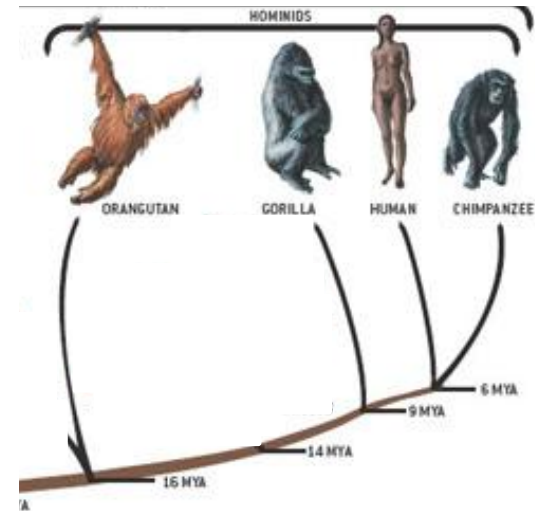


Reparto incompleto de linajes e introgresión

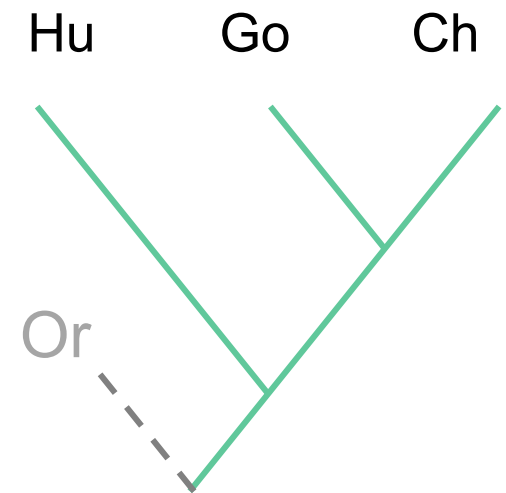
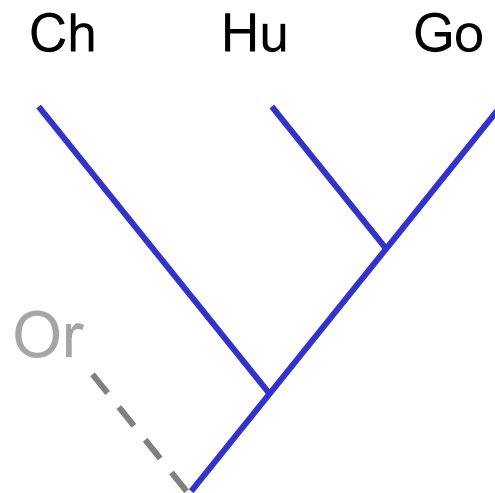
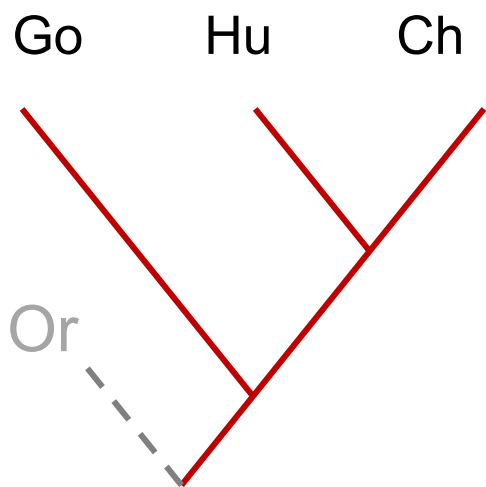
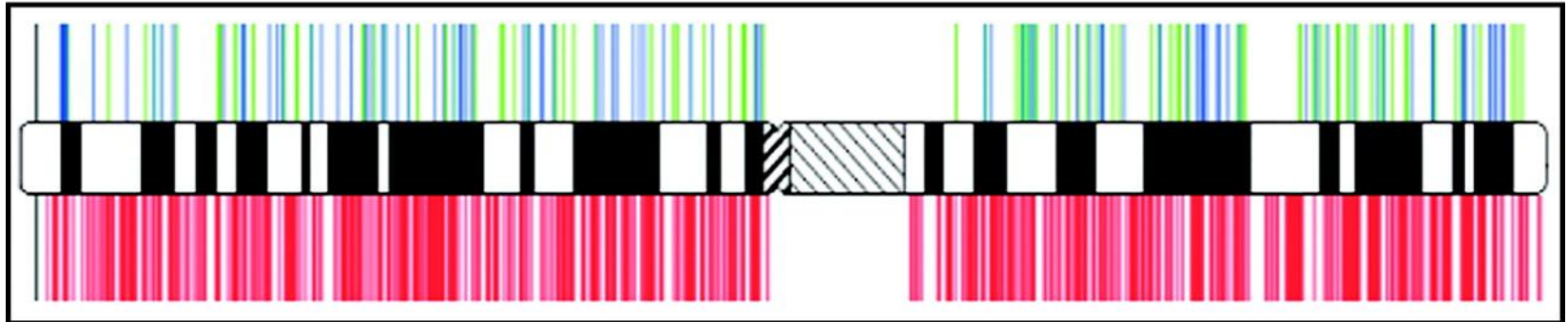


Árboles de genes en un árbol de especies

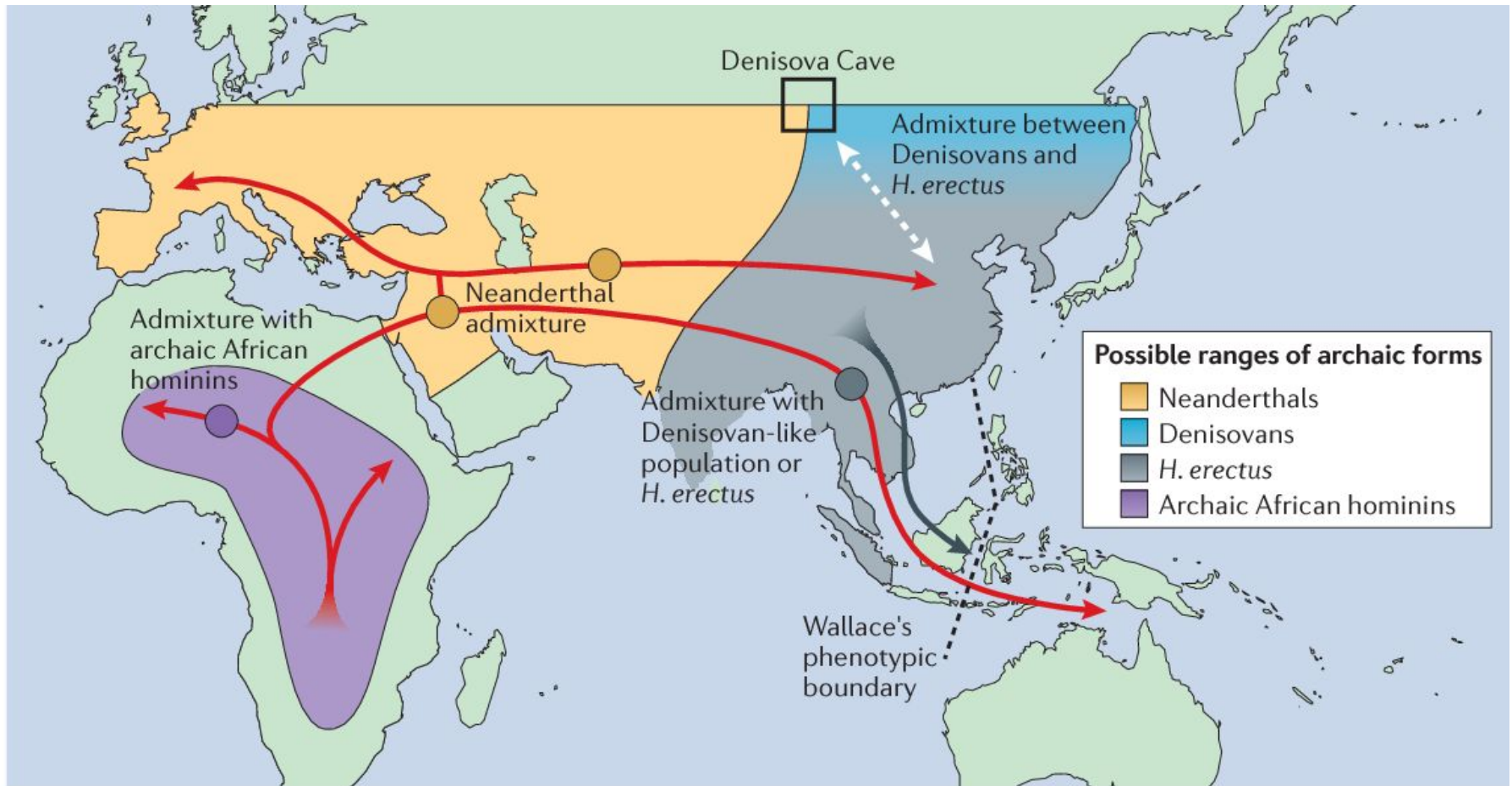
Reparto incompleto de linajes:
ILS: incomplete lineage sorting



Cromosoma 1 humano



Oportunidades de introgresión



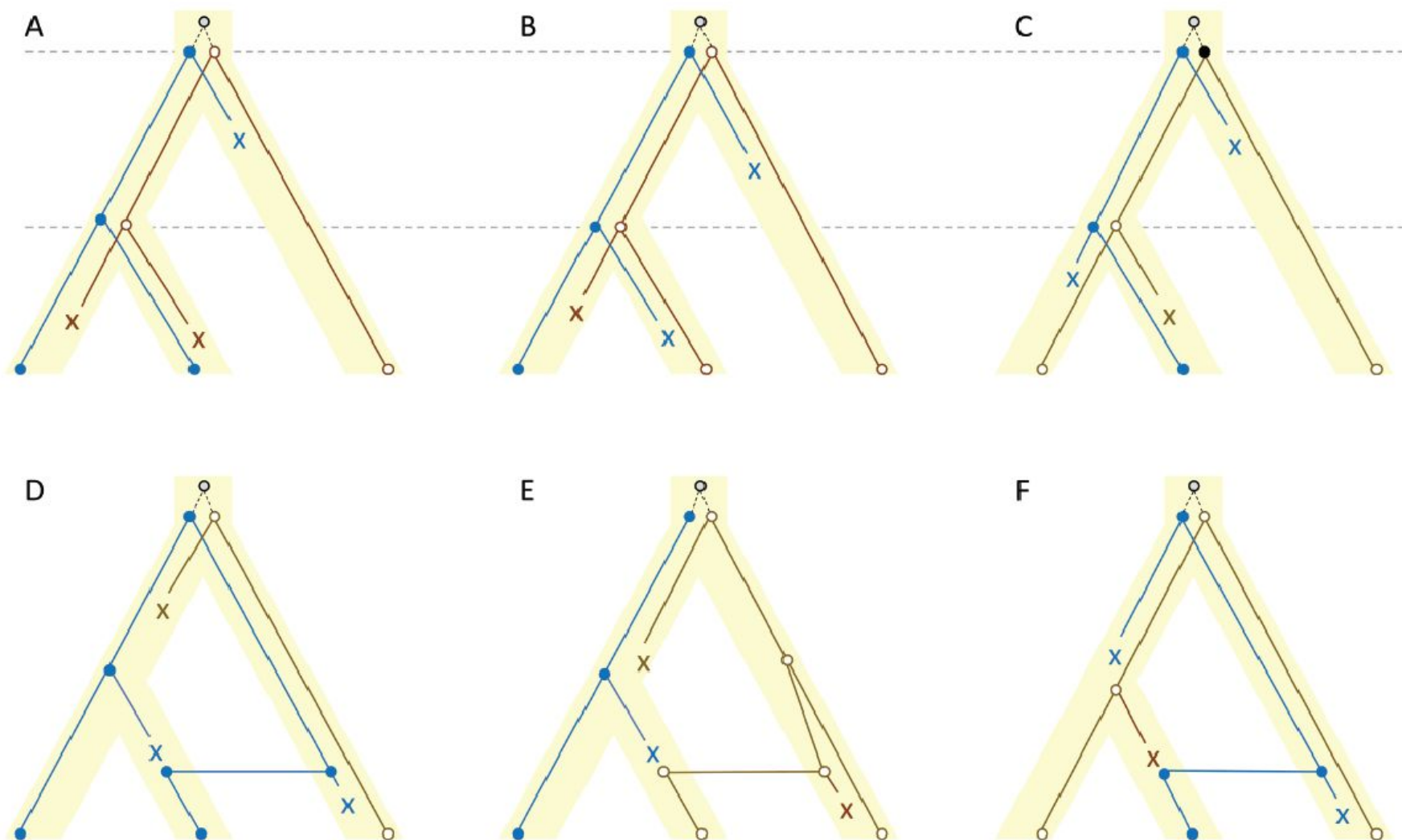


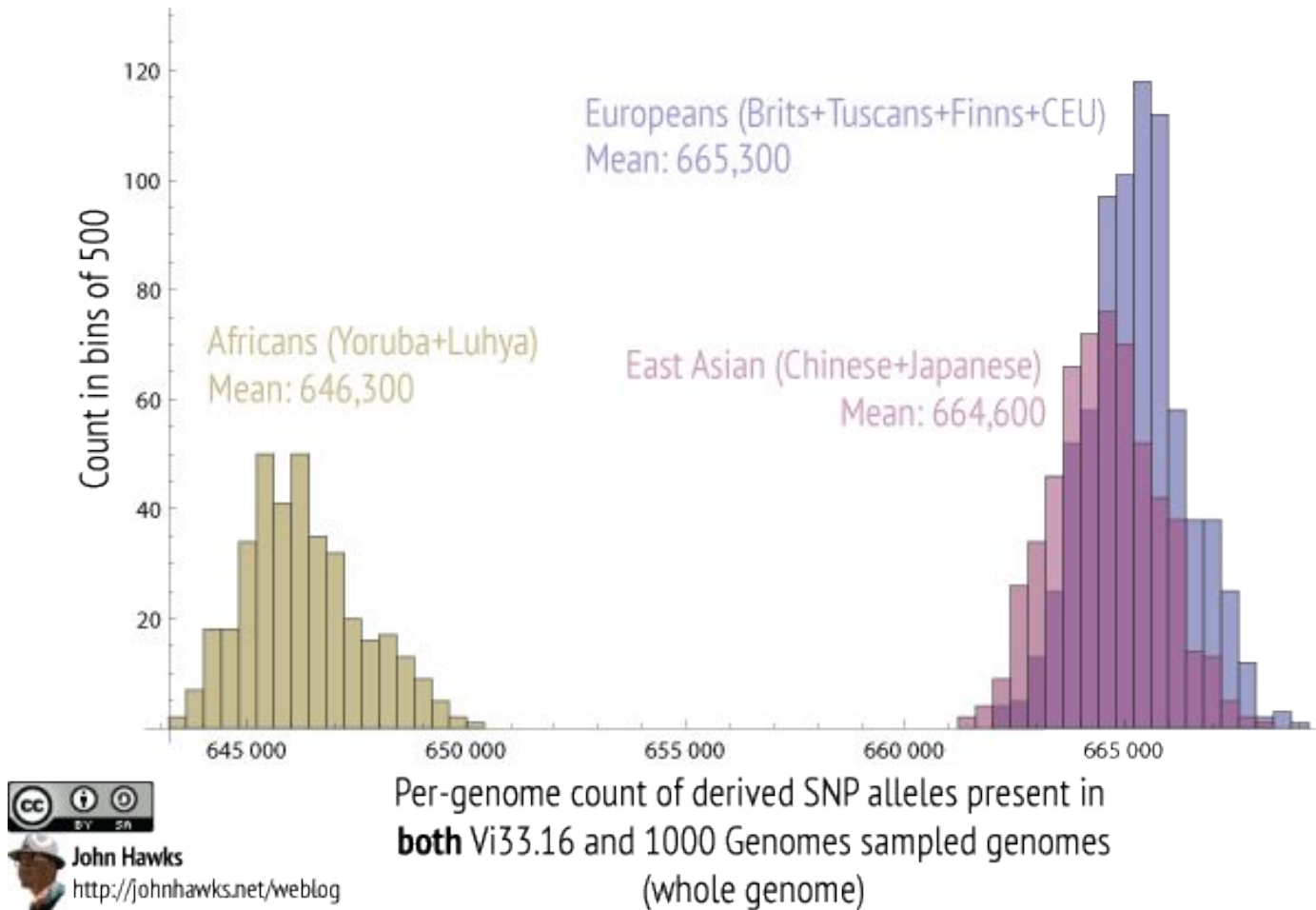
Fig. 1.—Relationships between gene (allele) trees (lines) and species trees (shaded area). The ancestry of alleles (circles) of three species, resulting from two successive speciation events, is tracked to two ancestral alleles. The symbol X signals allele extinction. A) a gene tree congruent with the 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.

Pruebas (“tests”) de introgresión: ABBA-BABA test

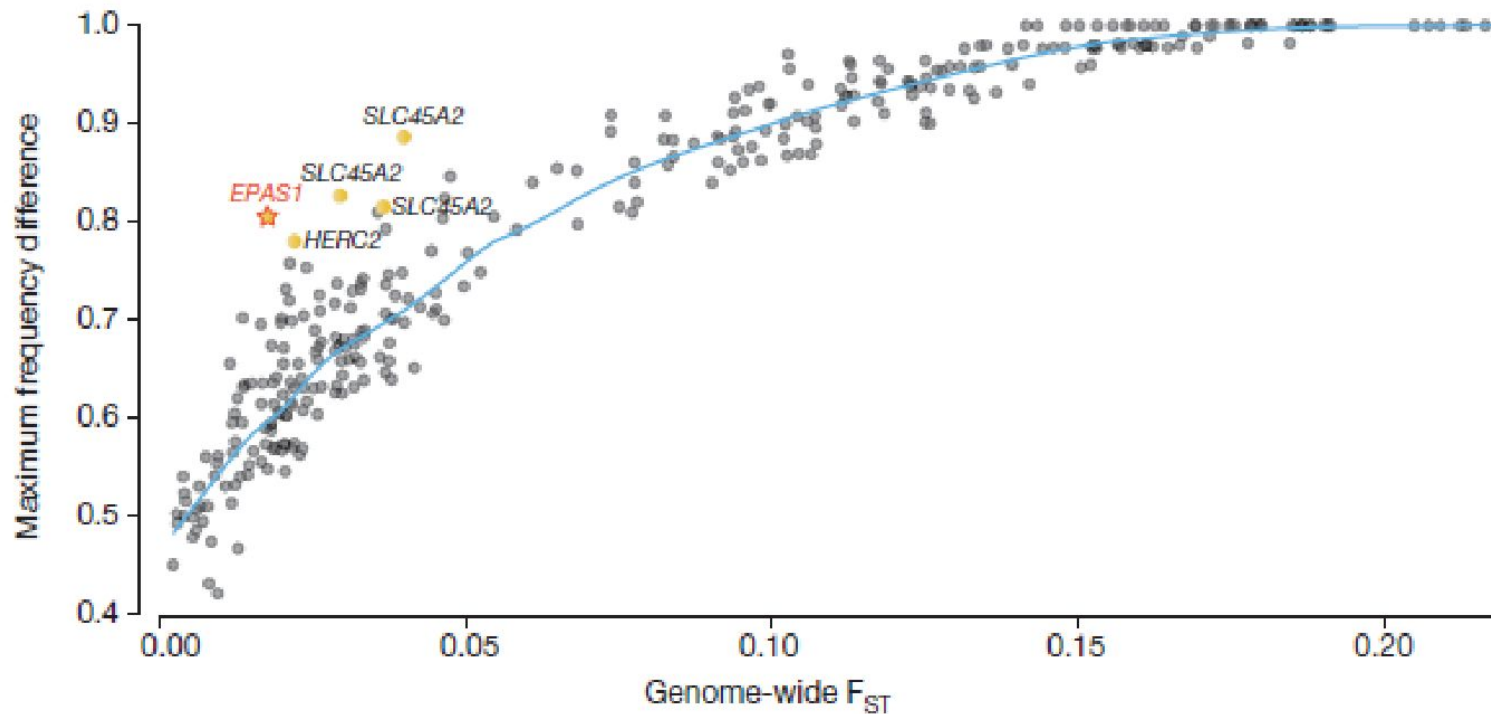
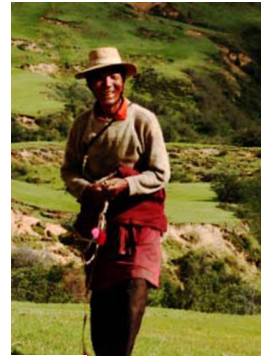


Idea básica: ILS produce frecuencias idénticas de ABBA y BABA; la introgresión de neandertal a europeos produce un exceso de ABBA.

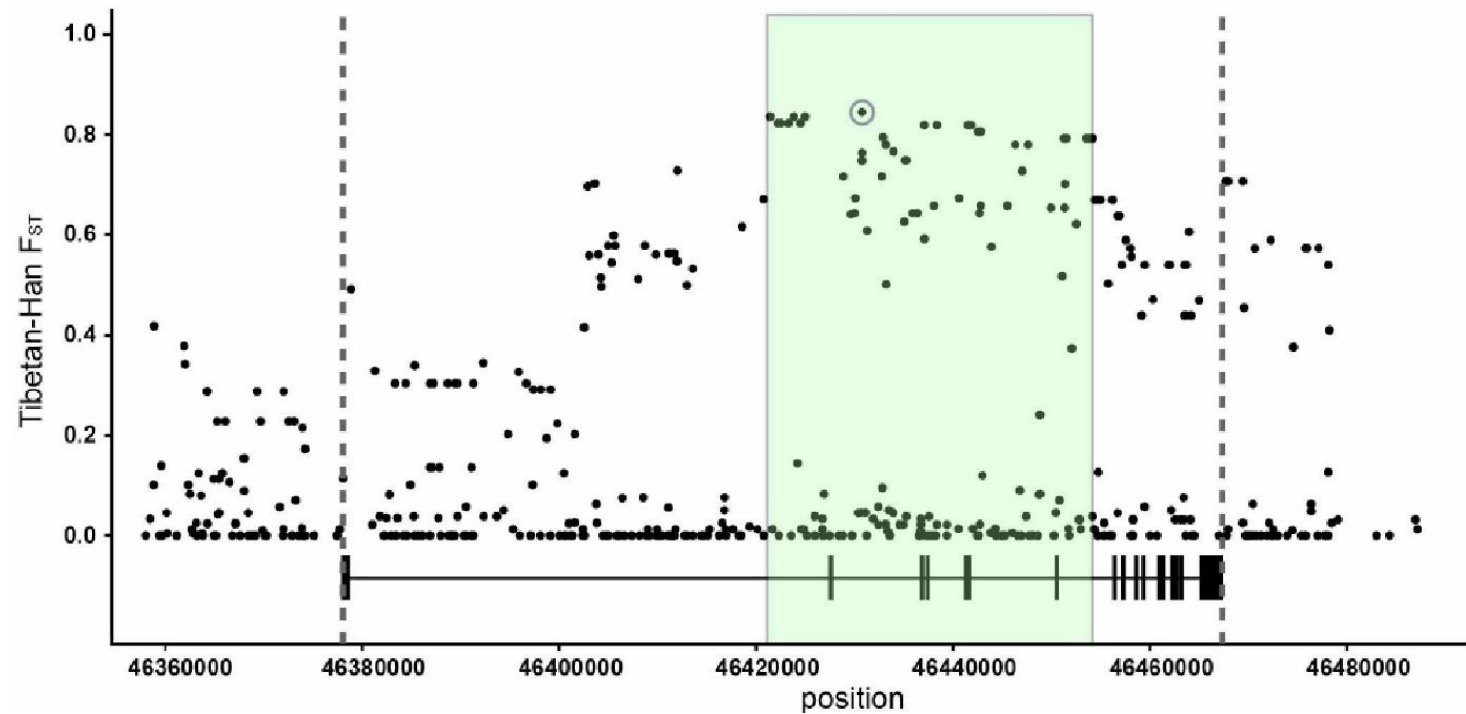
Exceso de “ABBA” en poblaciones no africanas



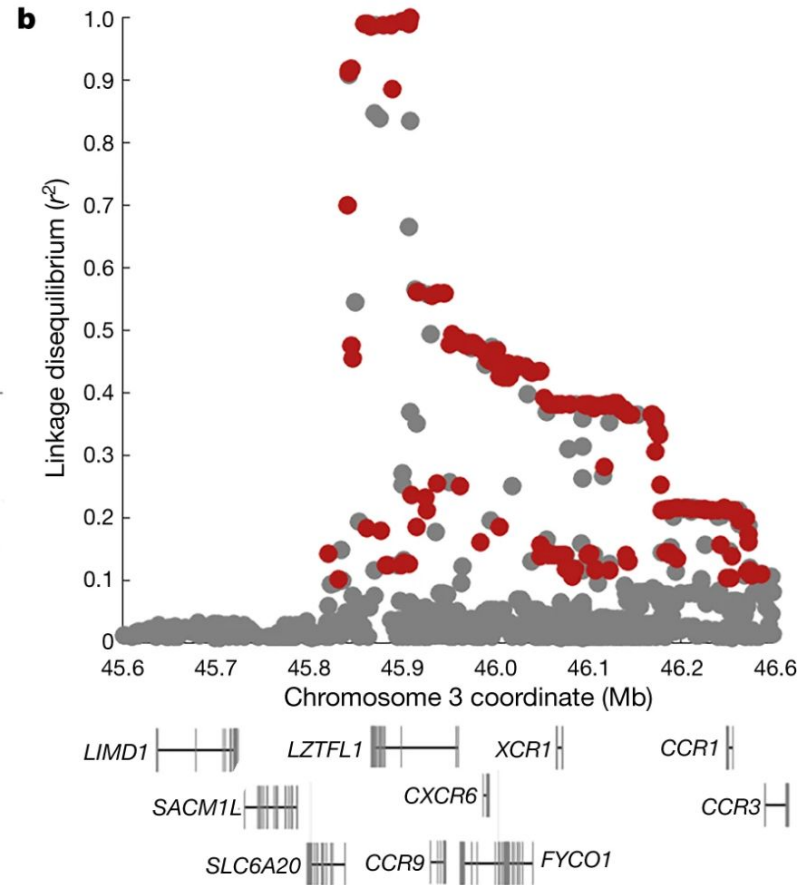
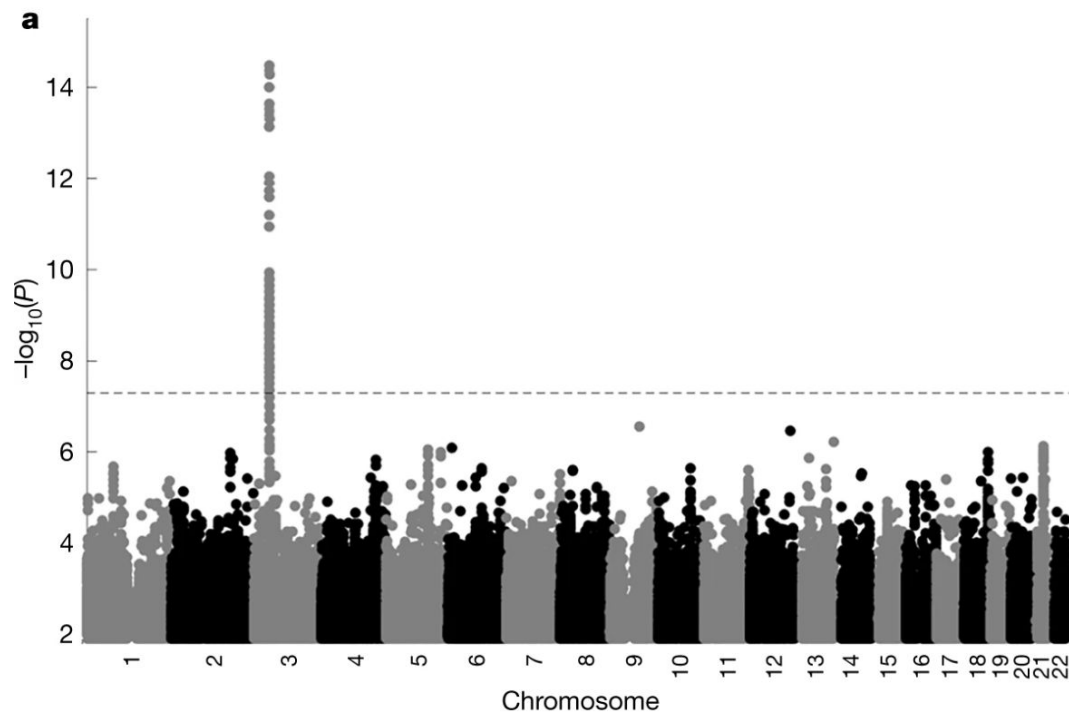
La introgresión, filtrada por la selección natural, puede ser adaptativa



Huerta-Sánchez et al 2014. Altitude adaptation in Tibetans caused by introgression of Denisovan-like DNA. Nature 512: 194-197



Huerta-Sánchez et al 2014. Altitude adaptation in Tibetans caused by introgression of Denisovan-like DNA. *Nature* 512: 194-197



Zeberg & Paabo 2020. The major genetic risk factor for severe COVID-19 is inherited from Neanderthals. Nature