

Curso de Evolución 2020

Facultad de Ciencias

Montevideo, Uruguay

<http://evolucion.fcien.edu.uy/>

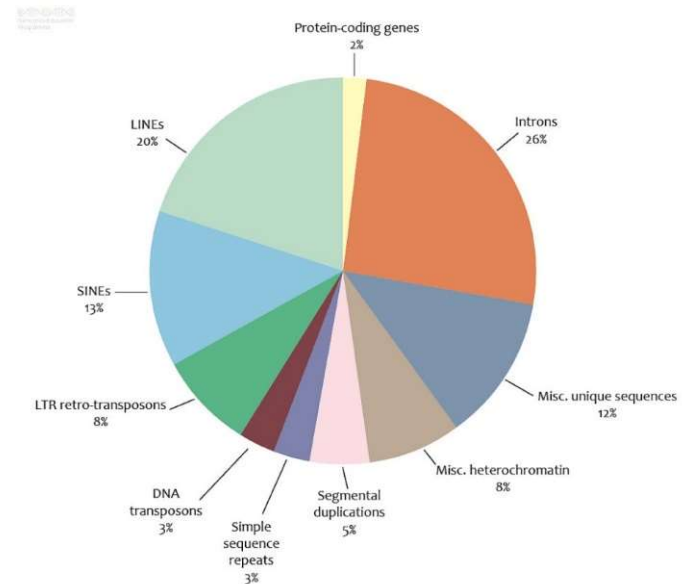
<http://eva.fcien.udelar.edu.uy/>



10. Evolución a nivel genómico. Genómica comparada y divergencia genómica. Aproximaciones genómicas al estudio de la selección natural. Reparto incompleto de linajes e introgresión.

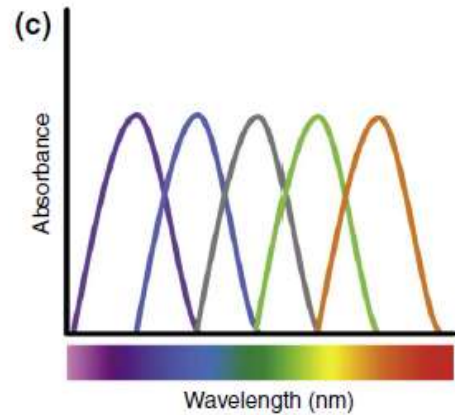
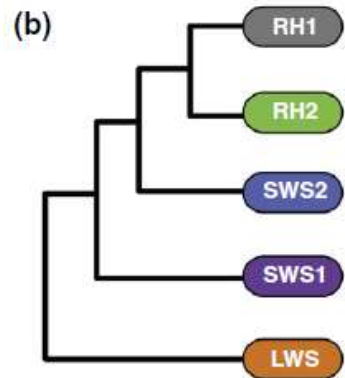
Evolución a escala genómica: algunos ejemplos

- Vamos a ilustrar, pero no a cubrir, todo el espectro de análisis de la evolución a escala genómica.
- Ejemplo 1: evolución de genes que codifican pigmentos visuales en vertebrados.
- Ejemplo 2: el genoma del tuatara.
- Ejemplo 3: reparto incompleto de linajes e introgresión en humanos y parientes cercanos.



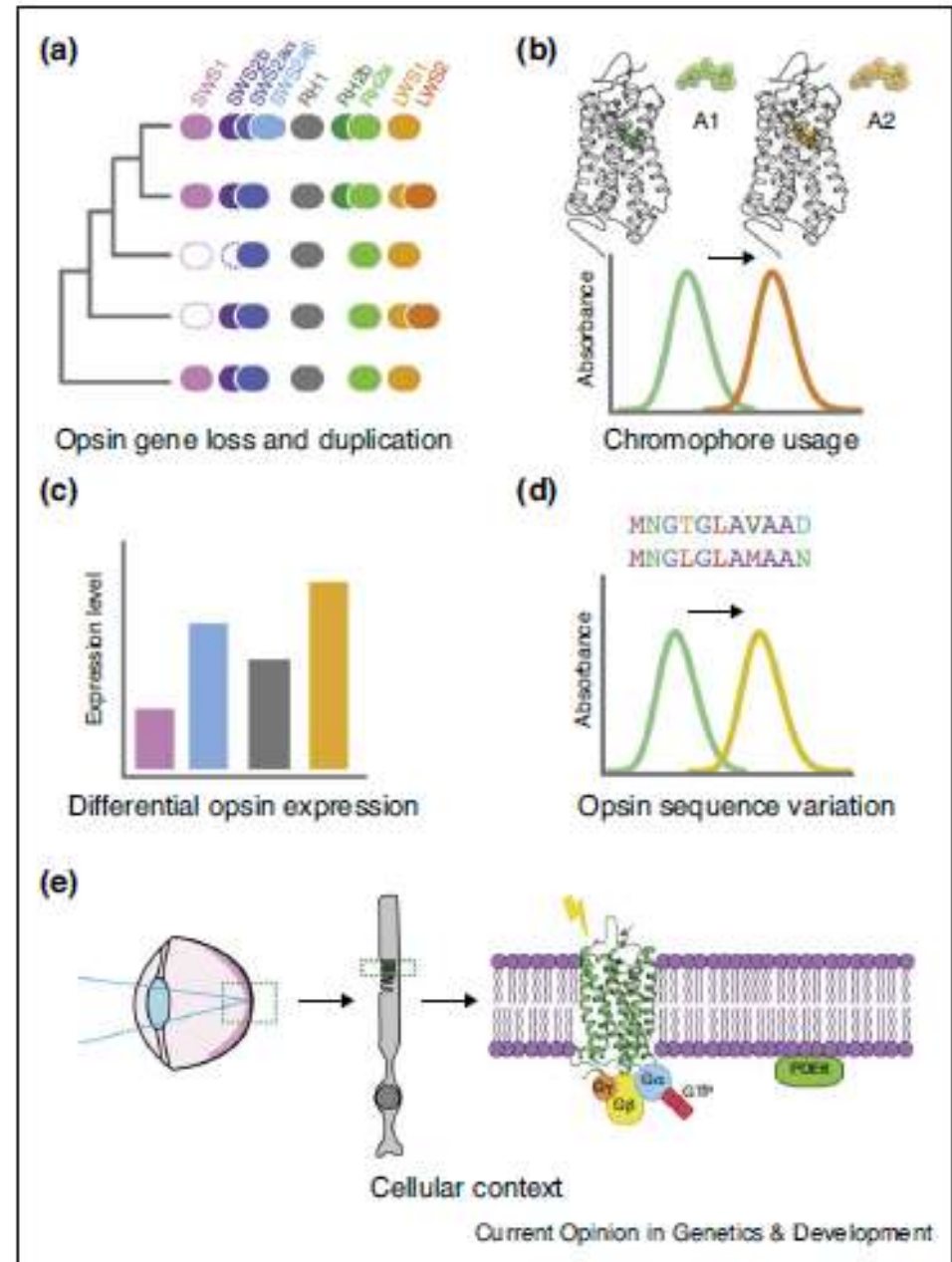
Evolución de pigmentos visuales

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

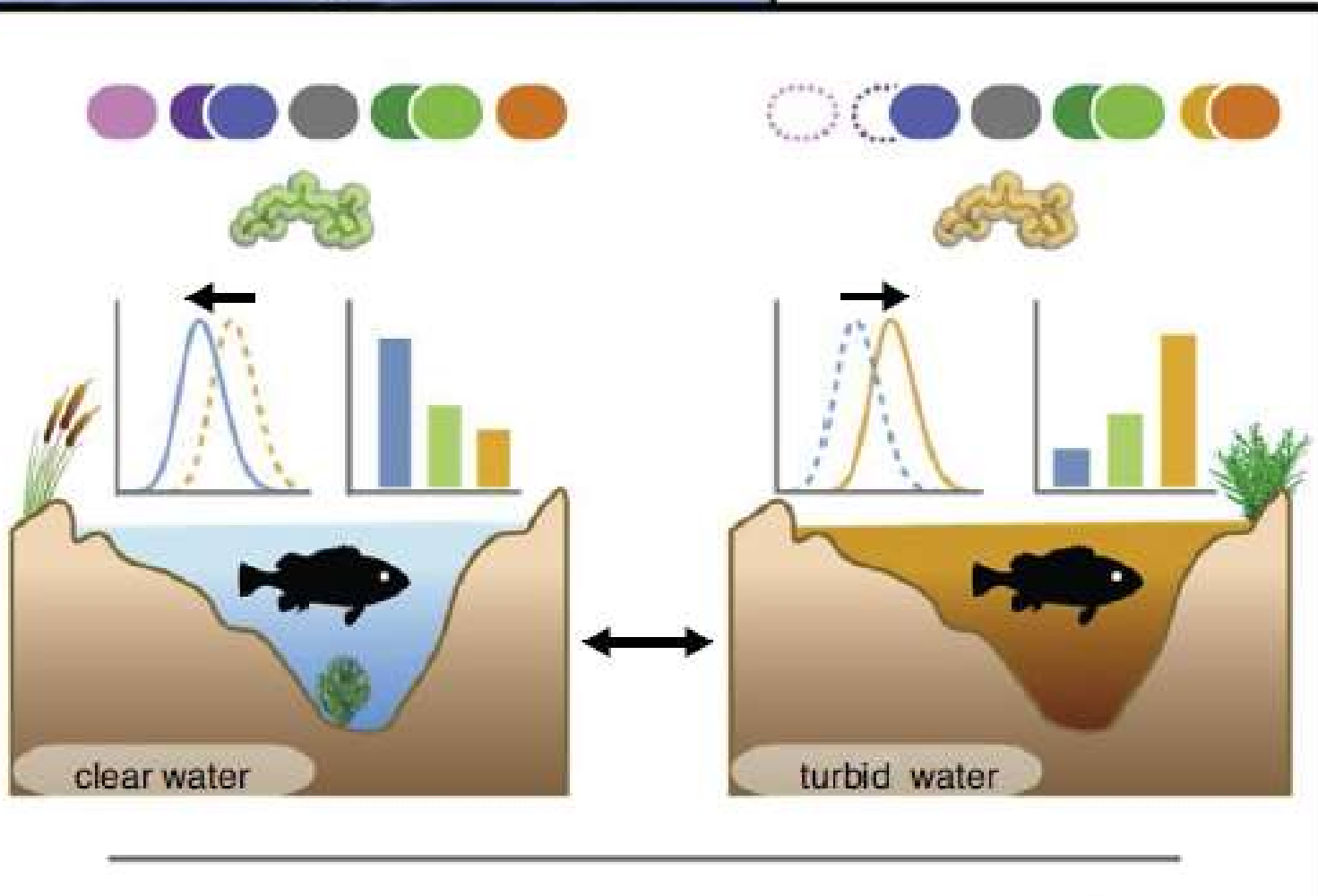


Current Opinion in Genetics & Development

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

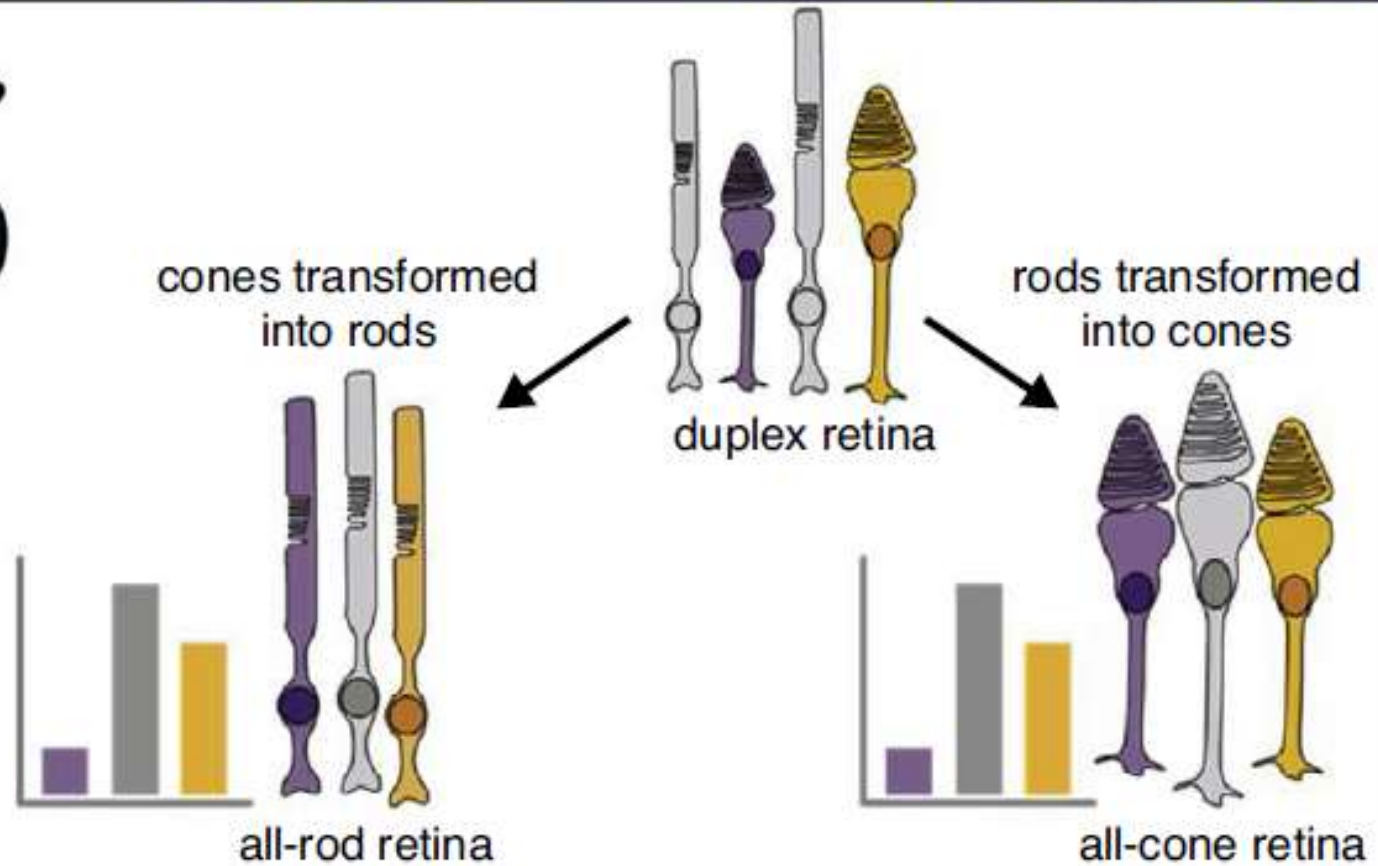
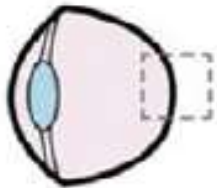


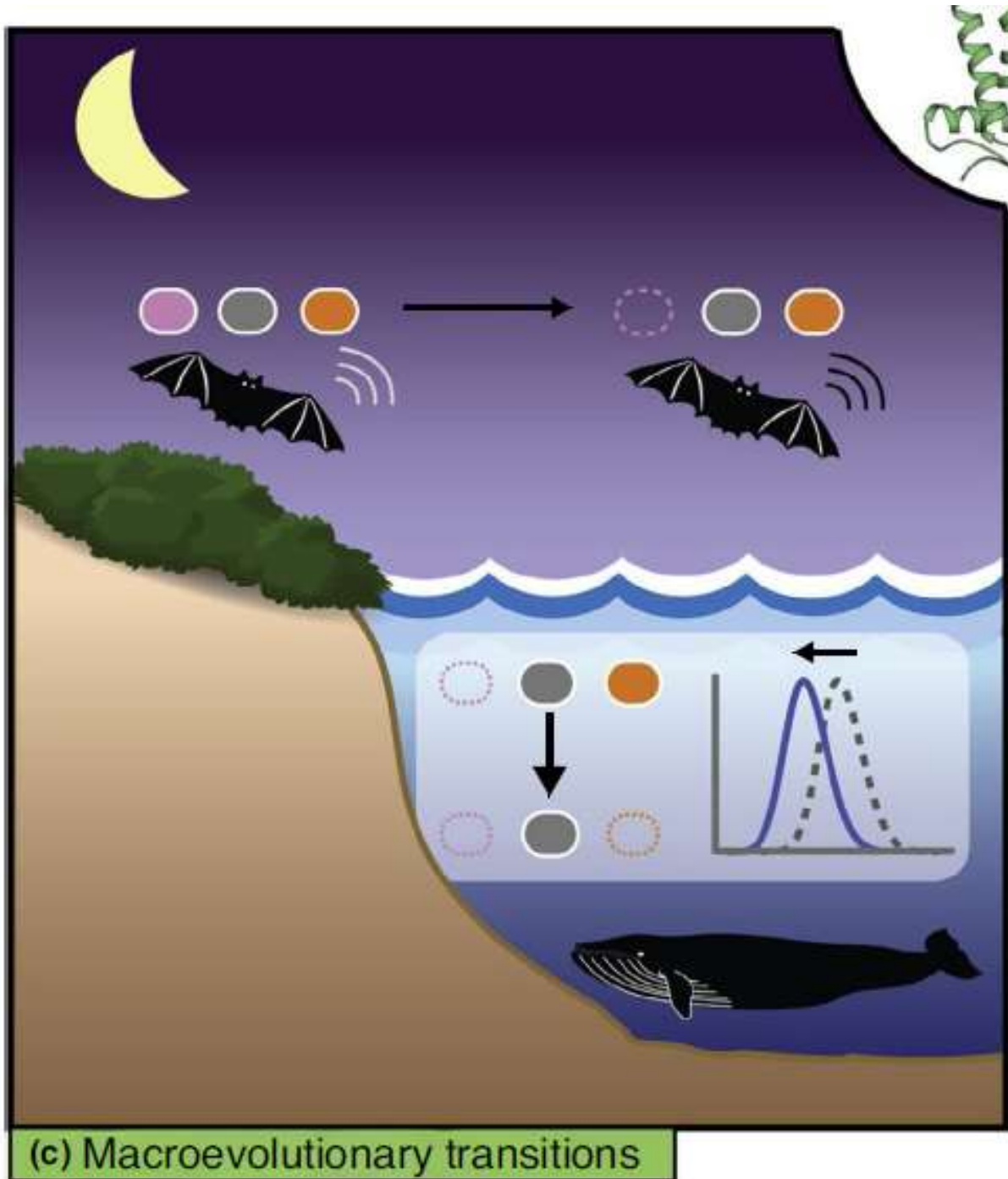
(a) Ambient light environment



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

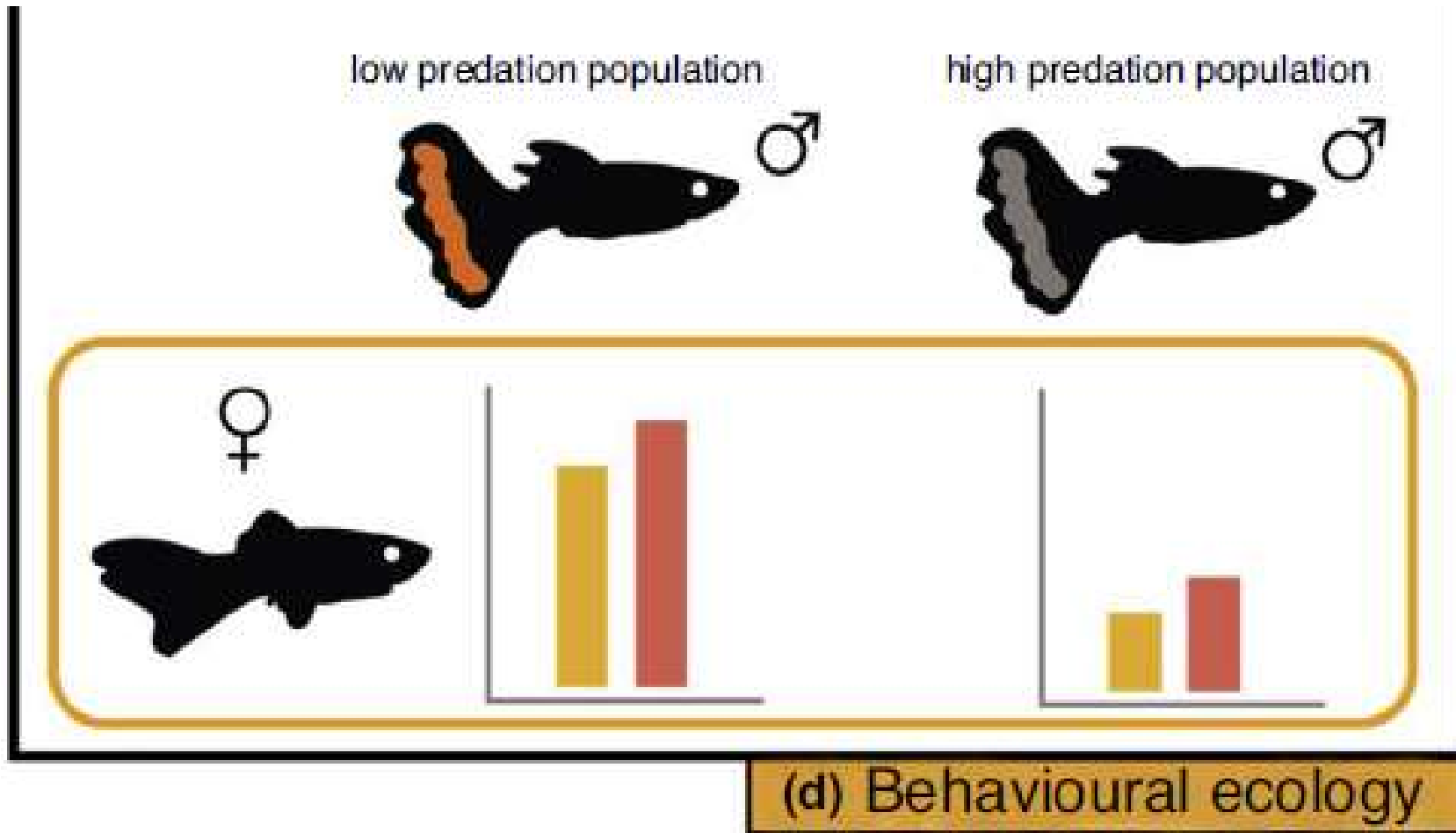
(b) Eye anatomy and physiology





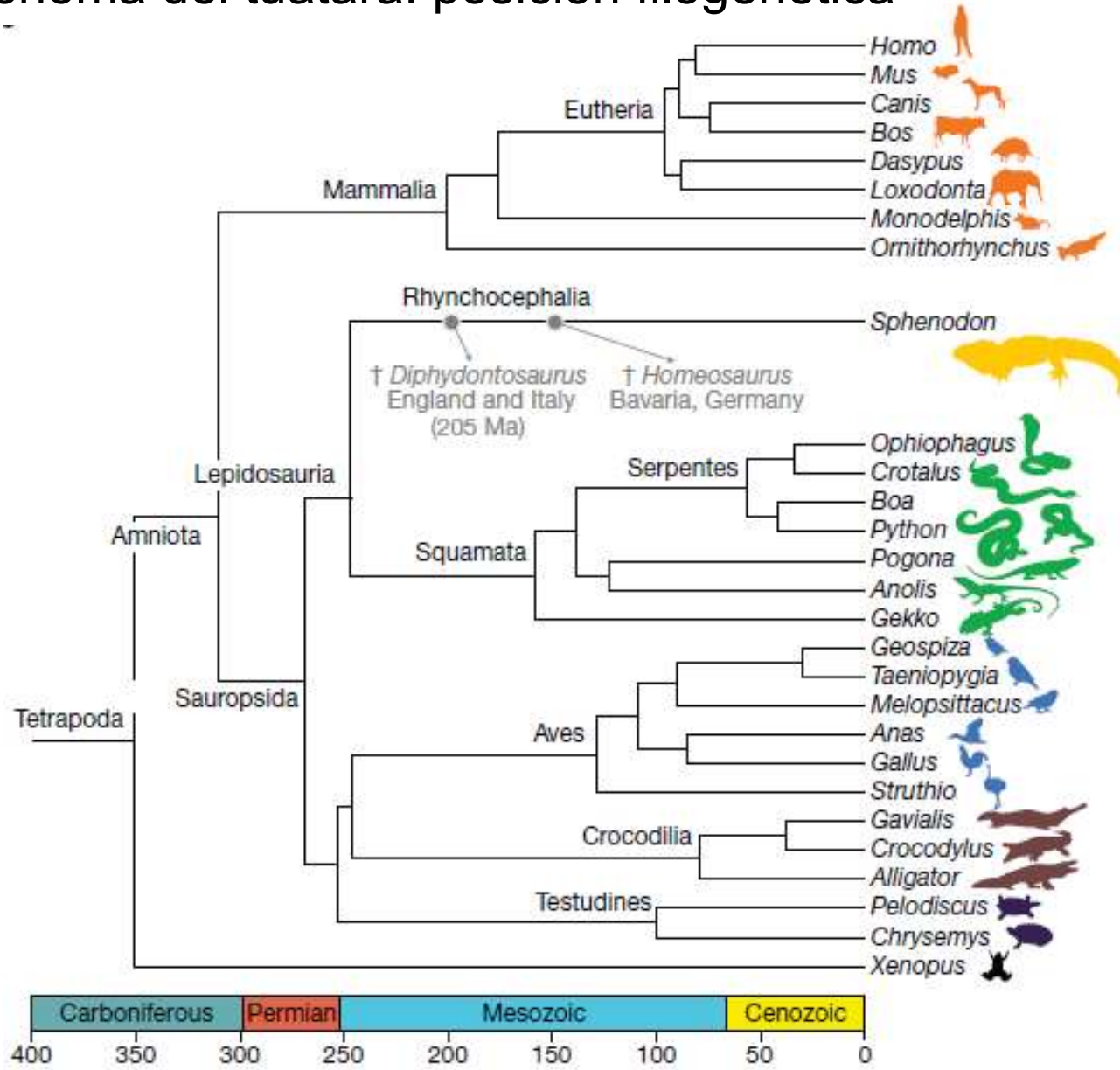
(c) Macroevolutionary transitions

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

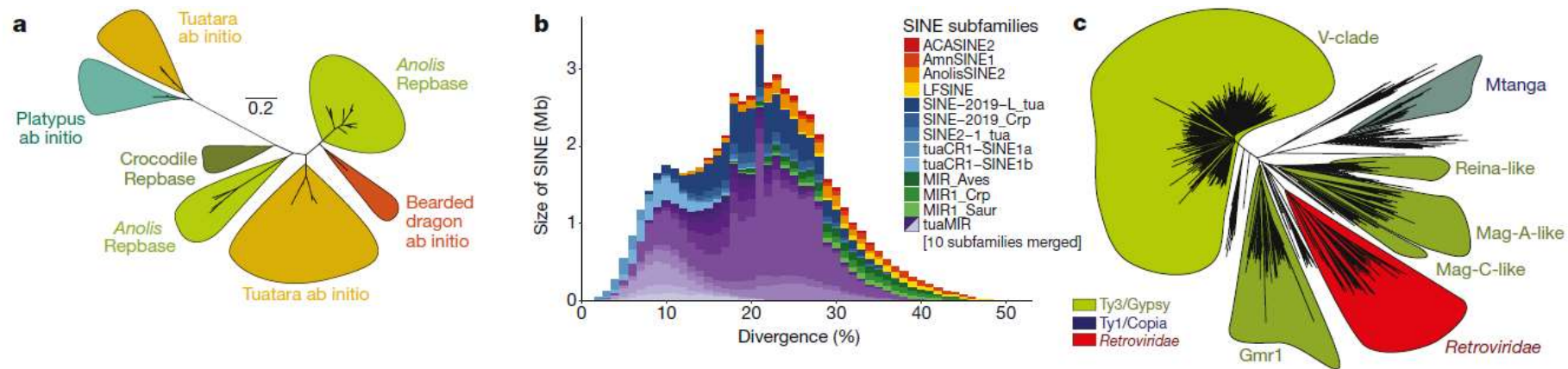


Hauser & Chang 2017.
 Current Opinion in Genetics and Development.

Genoma del tuatara: posición filogenética

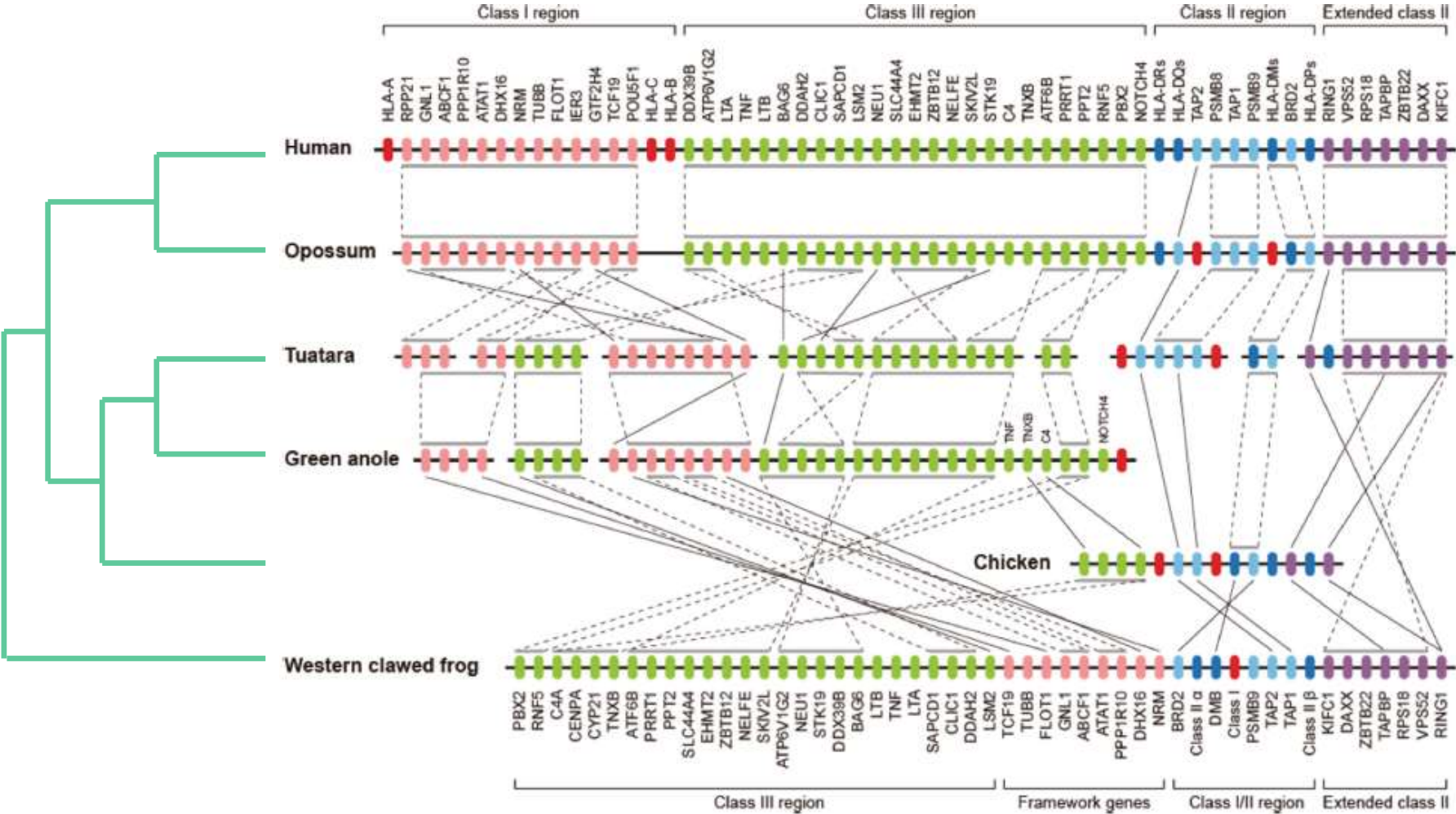


Gemmel et al. 2020. The tuatara genome reveals ancient features of amniote evolution. Nature

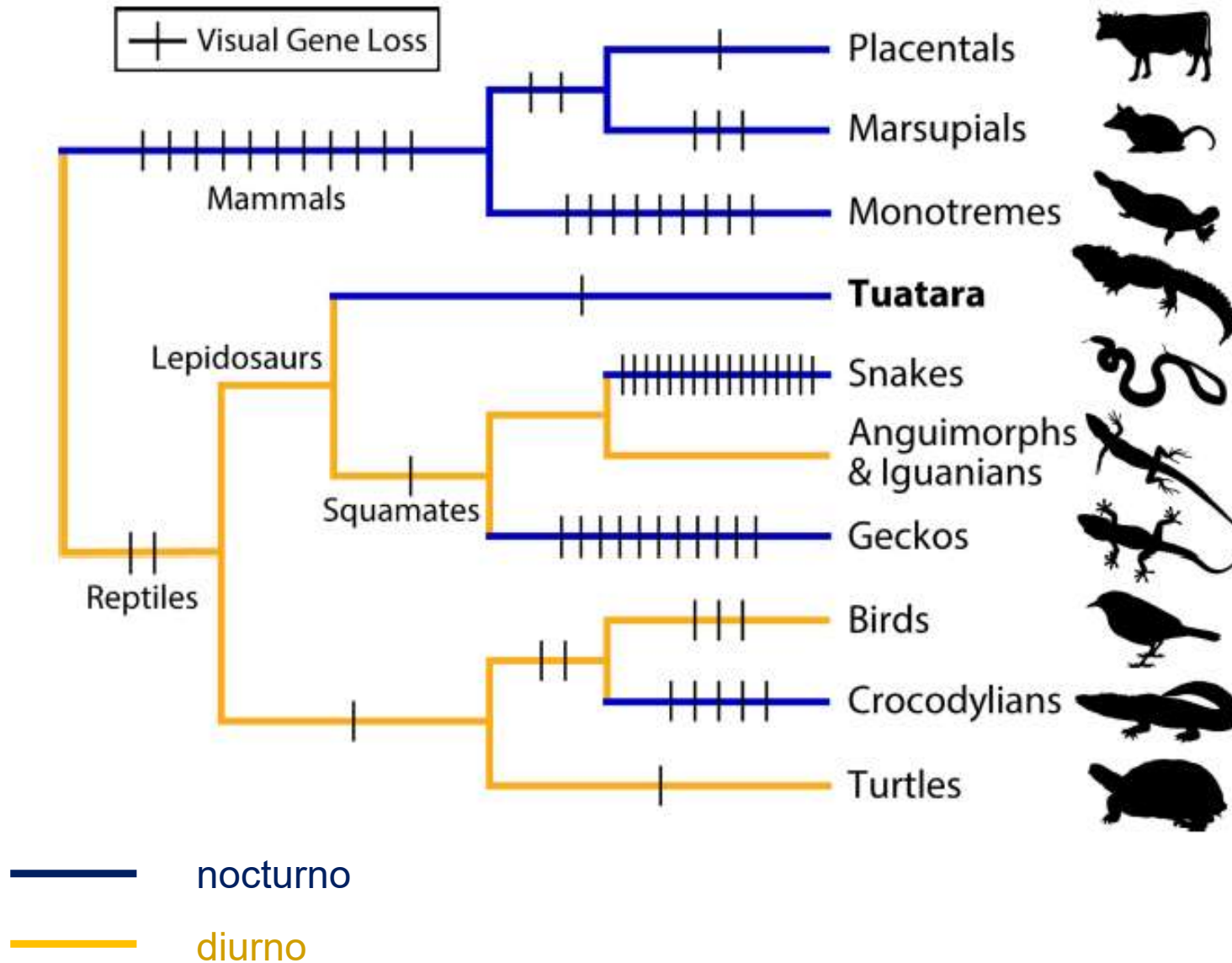


- Genoma 50% mayor que el humano (aumento por elementos transponibles, recientemente?)
- Gran cantidad de secuencias repetidas (muchas “activas”), 64%!! (50% vert), algunas características de mamíferos (MIR1) y otras de reptiles
- Posición filogenética y antigüedad de ancestros comunes.
- Baja tasa de evolución (tiempo generacional, baja temperatura, corporal).
- Genes de importancia en historia de vida (ej. Visión diurna a pesar de ser nocturnos, MHC)
- Diferenciación poblacional - conservación

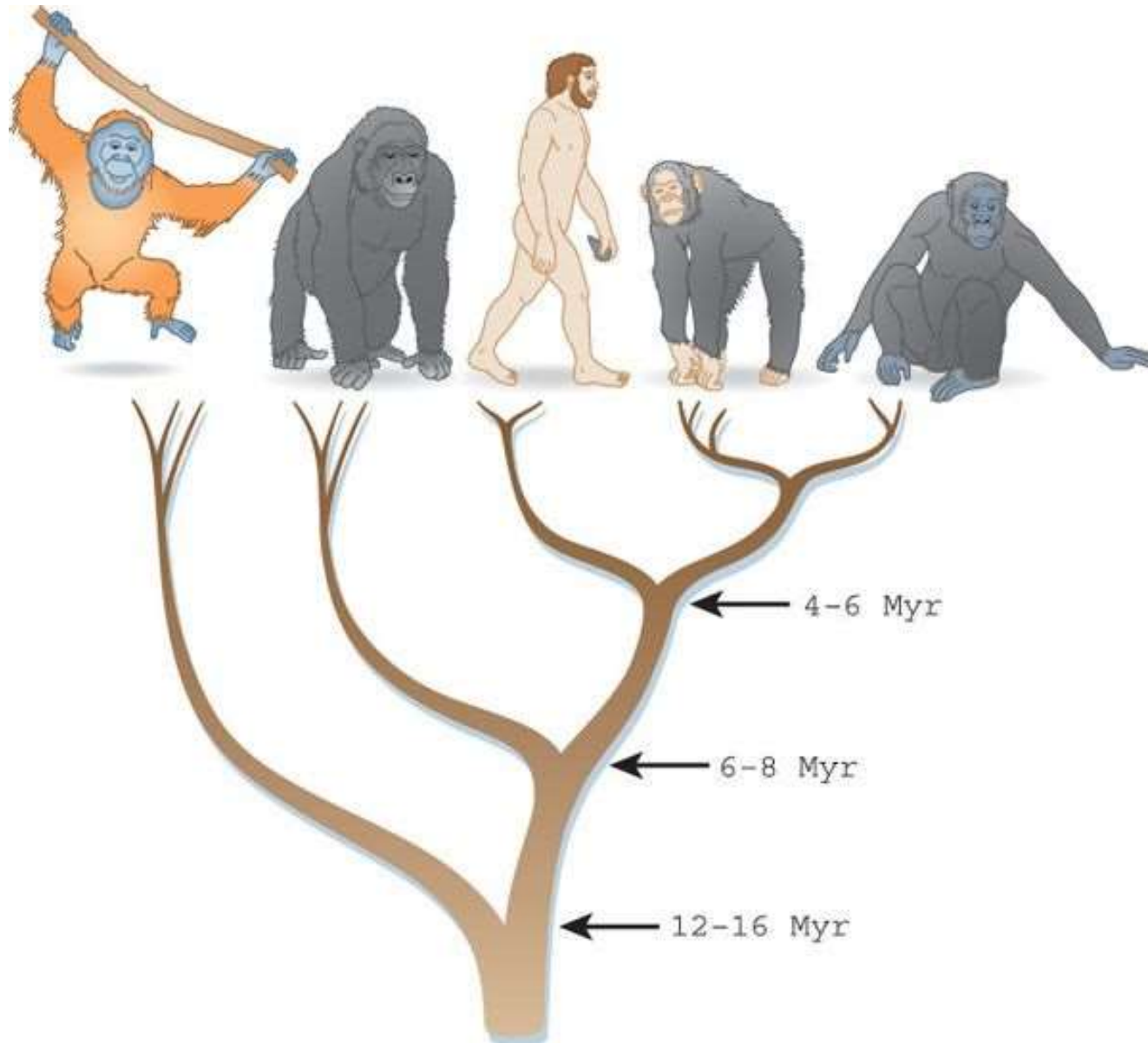
Genes MHC



Pérdida de genes del sistema visual

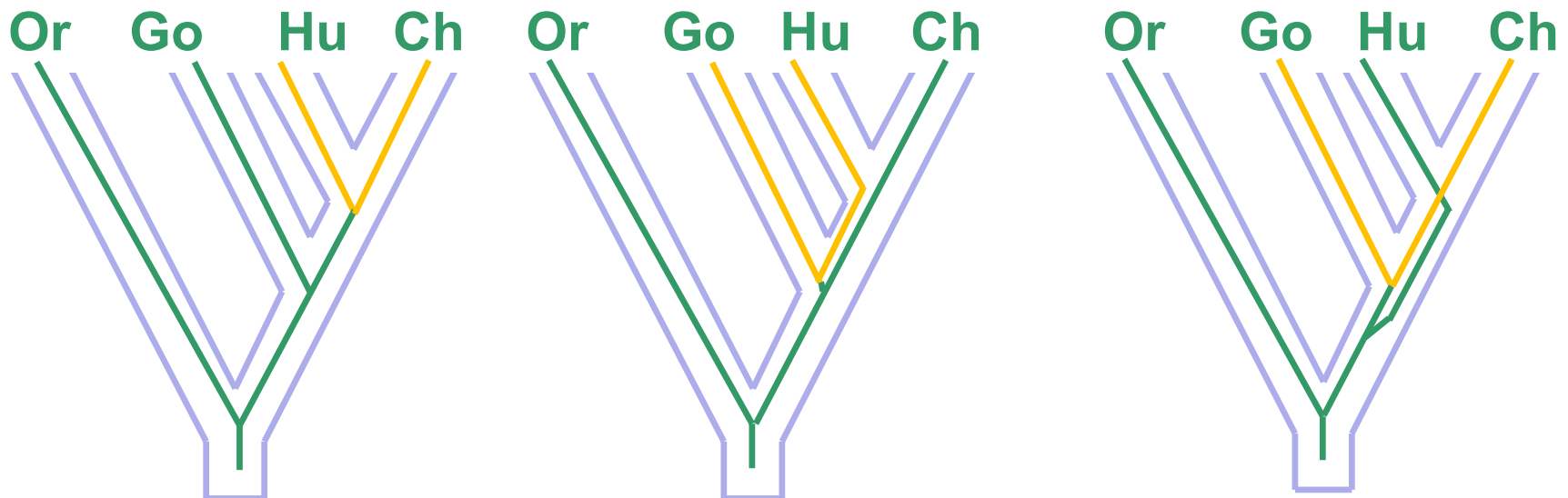
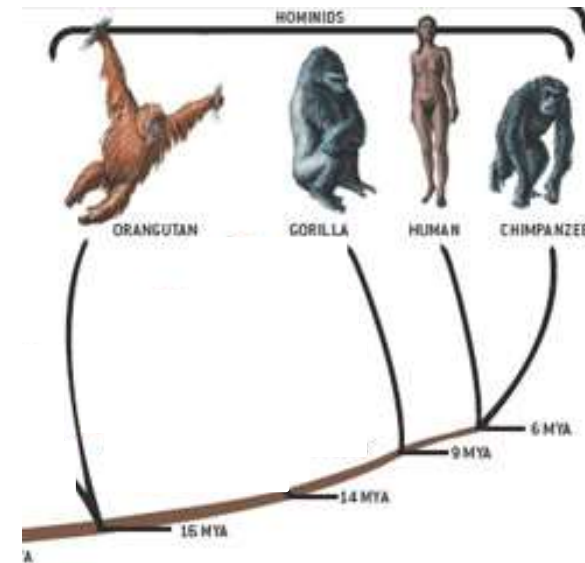


Reparto incompleto de linajes e introgresión

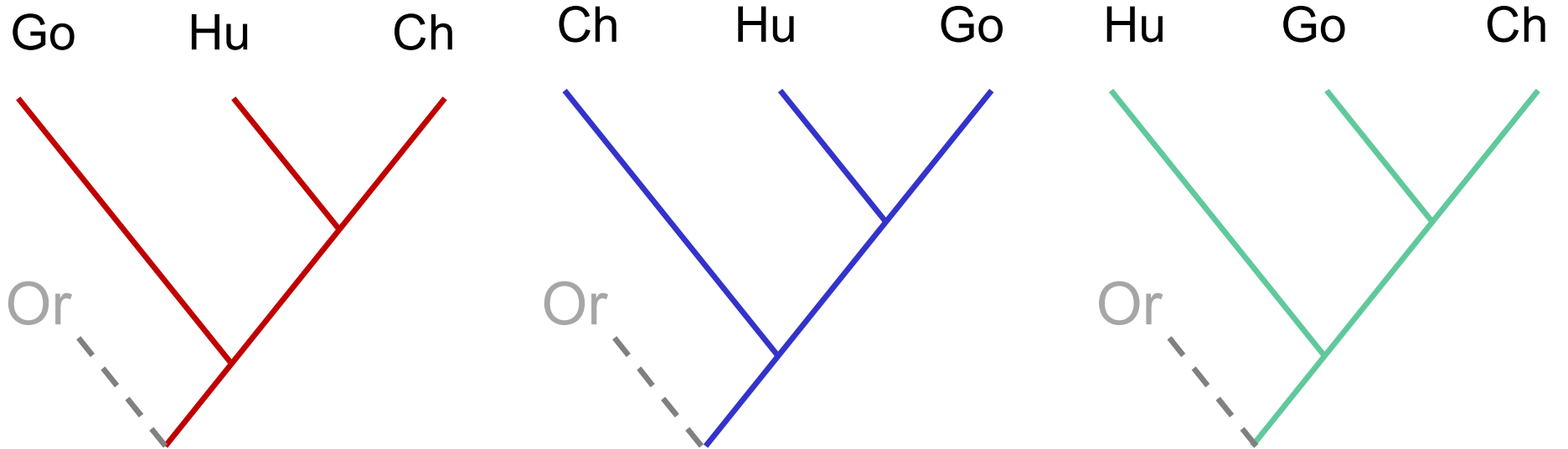
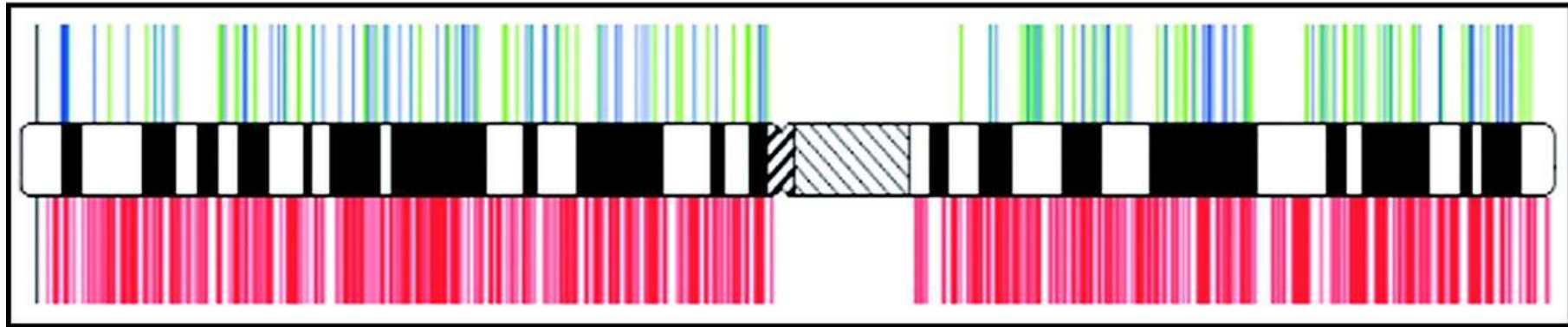


Árboles de genes en un árbol de especies

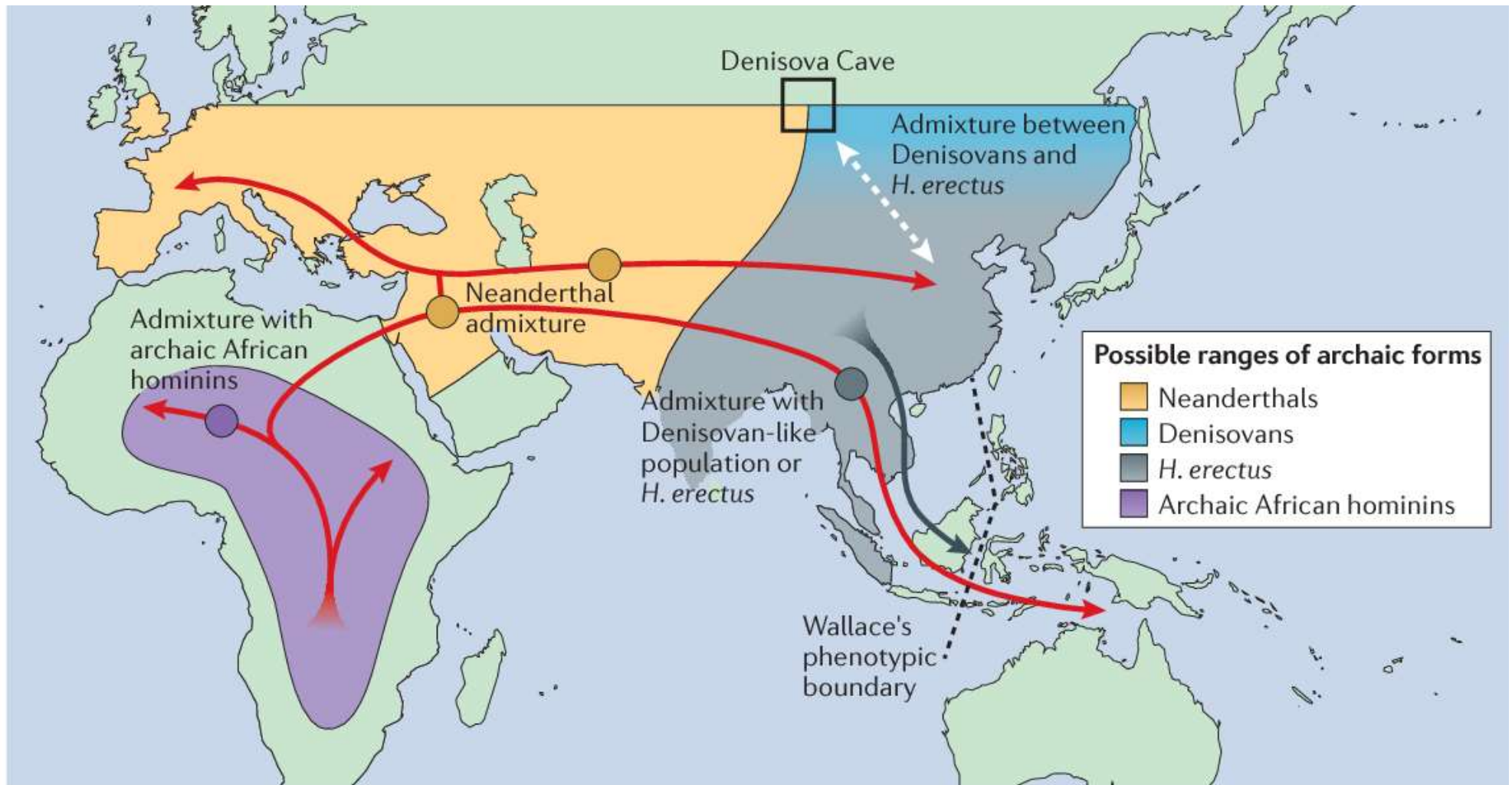
Reparto incompleto de linajes:
ILS: incomplete lineage sorting



Cromosoma 1 humano



Oportunidades de introgresión



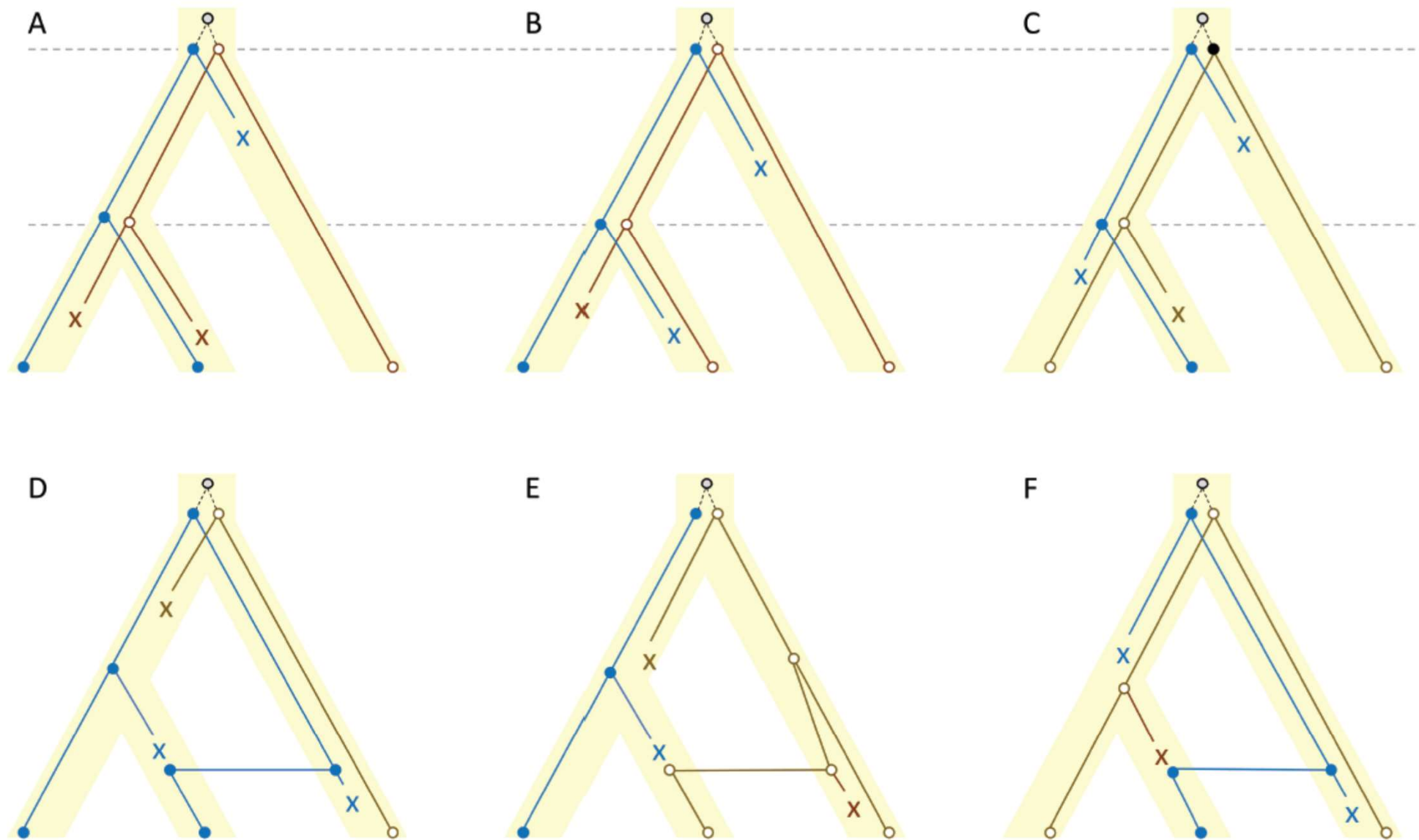


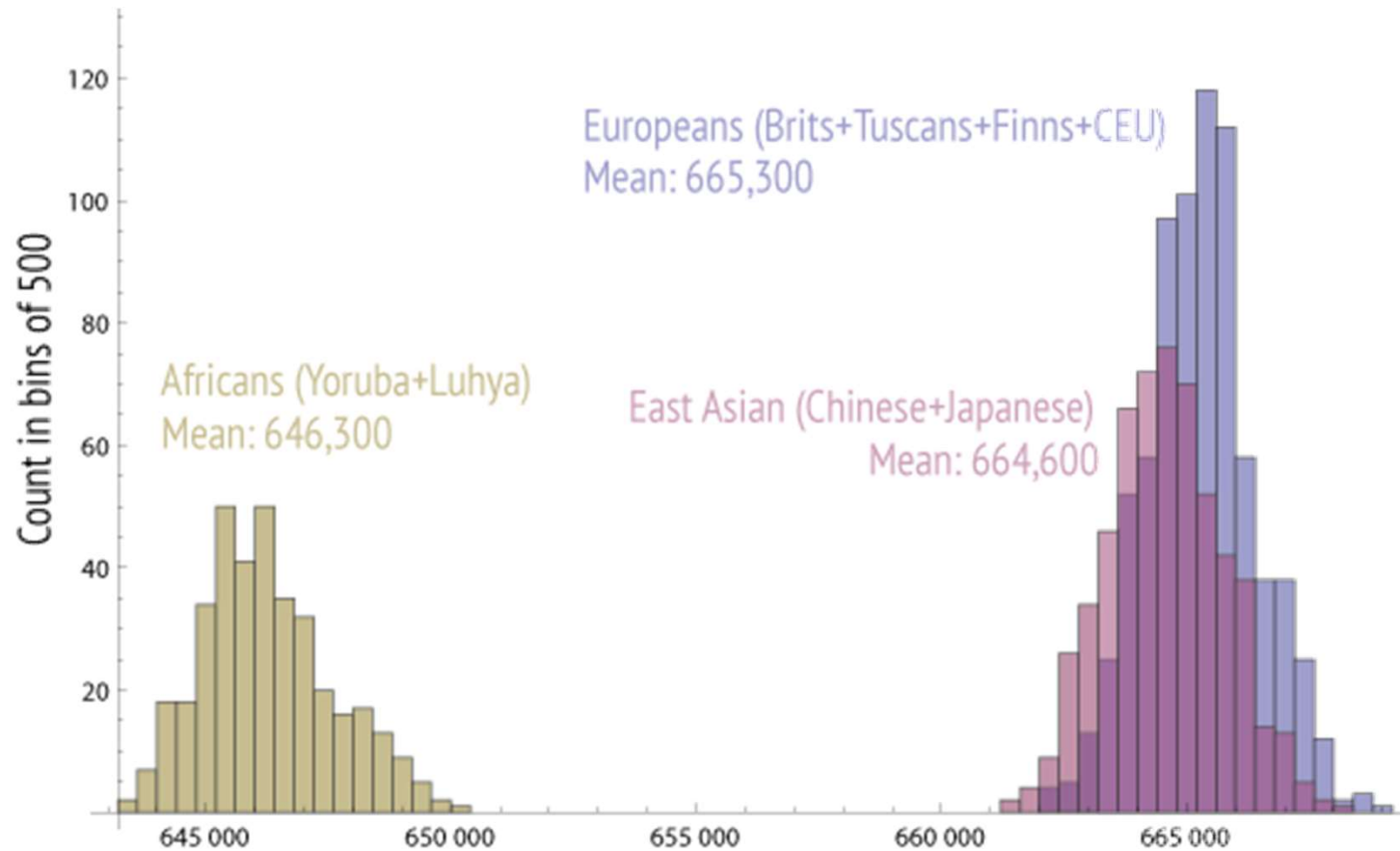
Fig. 1.—Relationships between gene (allele) trees (lines) and species trees (shaded area). The ancestry of alleles (circles) of three species, resulting from two successive speciation events, is tracked to two ancestral alleles. The symbol X signals allele extinction. A) a gene tree congruent with the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

			ILS		introgresión
			<hr/>		
	chimpancé	A	A	B	A
	neandertal	A	B	A	B
	 europeo	B	B	B	B
	africano	B	A	A	A

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

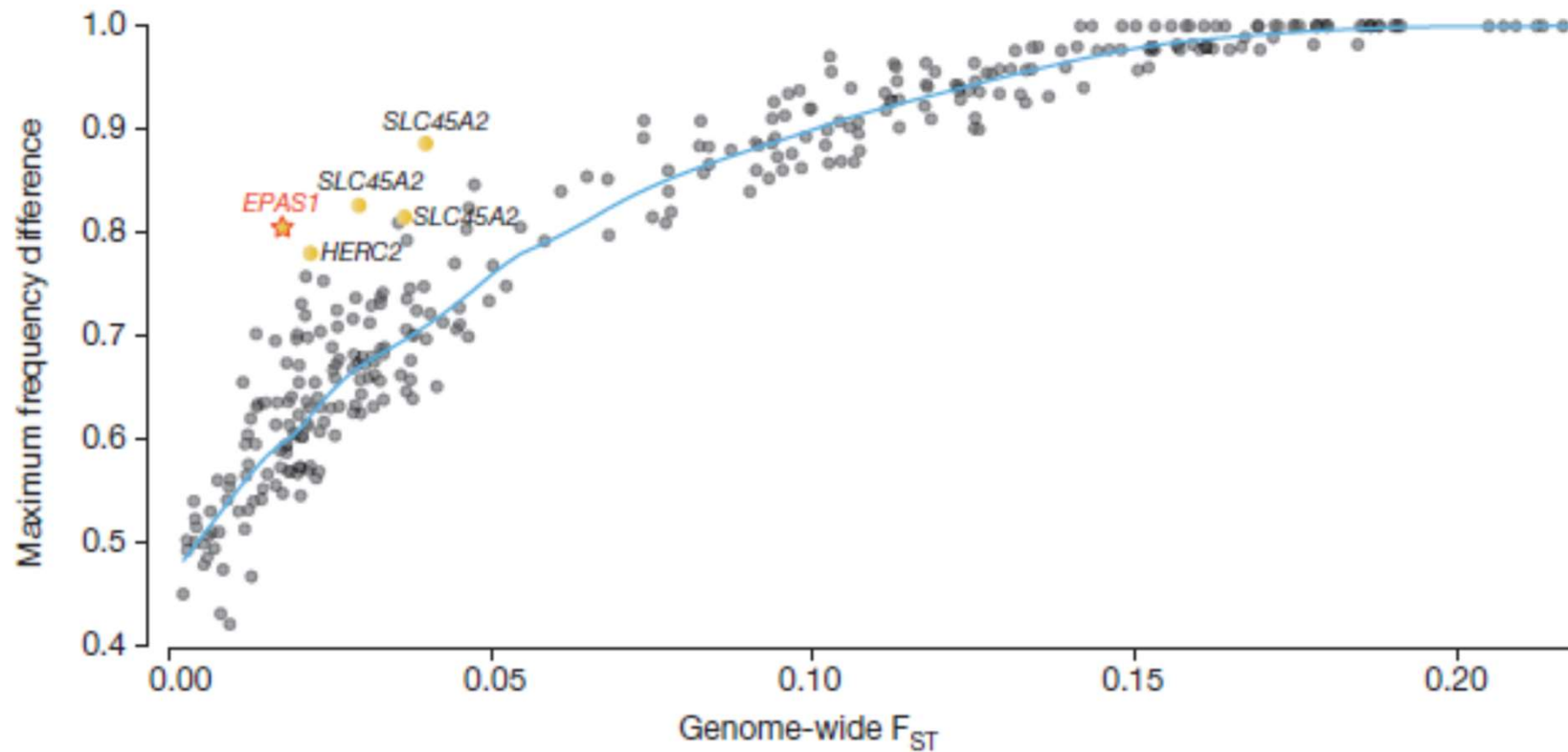
Exceso de “ABBA” en poblaciones no africanas



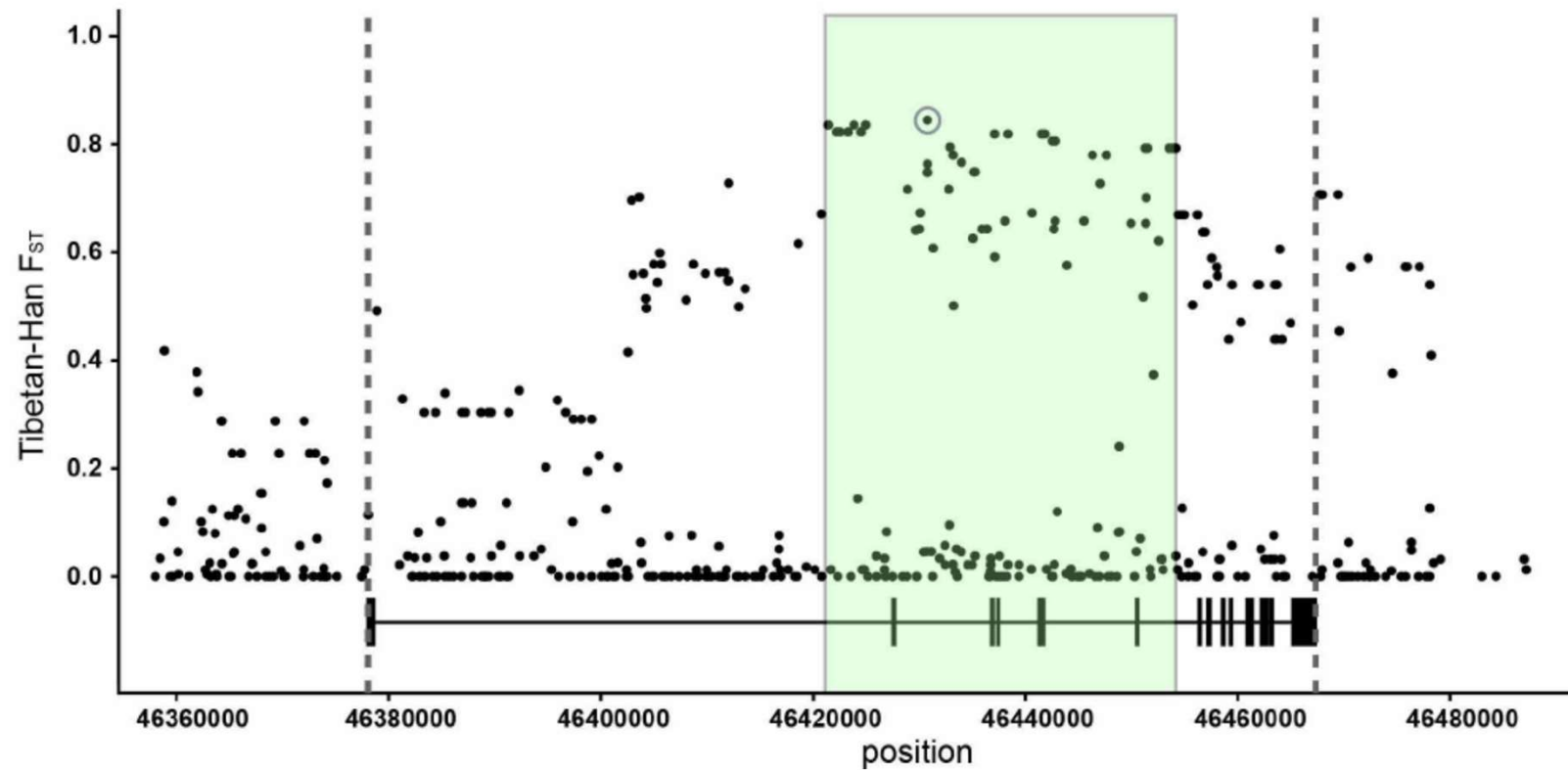
John Hawks
<http://johnhawks.net/weblog>

Per-genome count of derived SNP alleles present in
both Vi33.16 and 1000 Genomes sampled genomes
(whole genome)

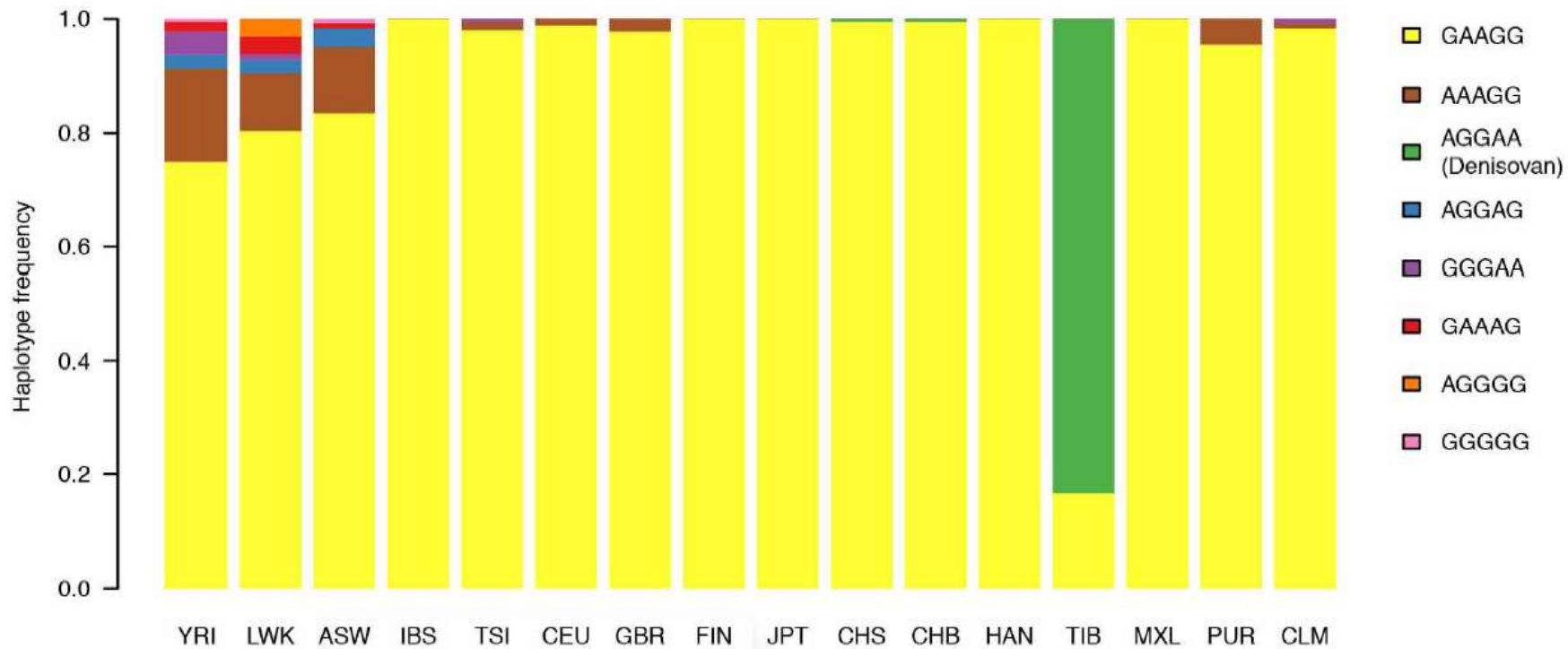
La introgresión, filtrada por la selección natural, puede ser adaptativa



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

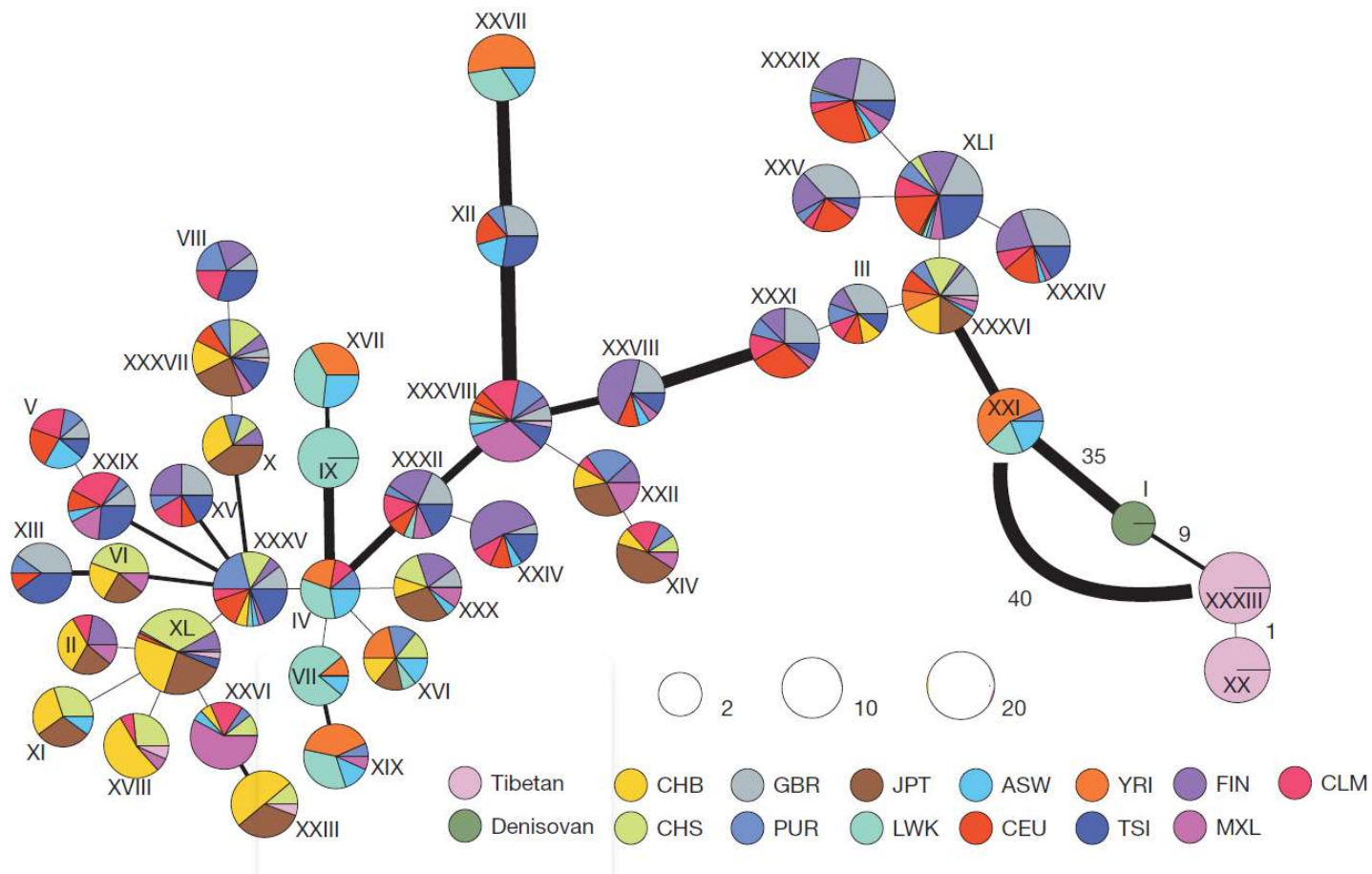


Huerta-Sánchez et al 2014. Altitude adaptation in Tibetans caused by introgression of Denisovan-like DNA. *Nature* 512: 194-197

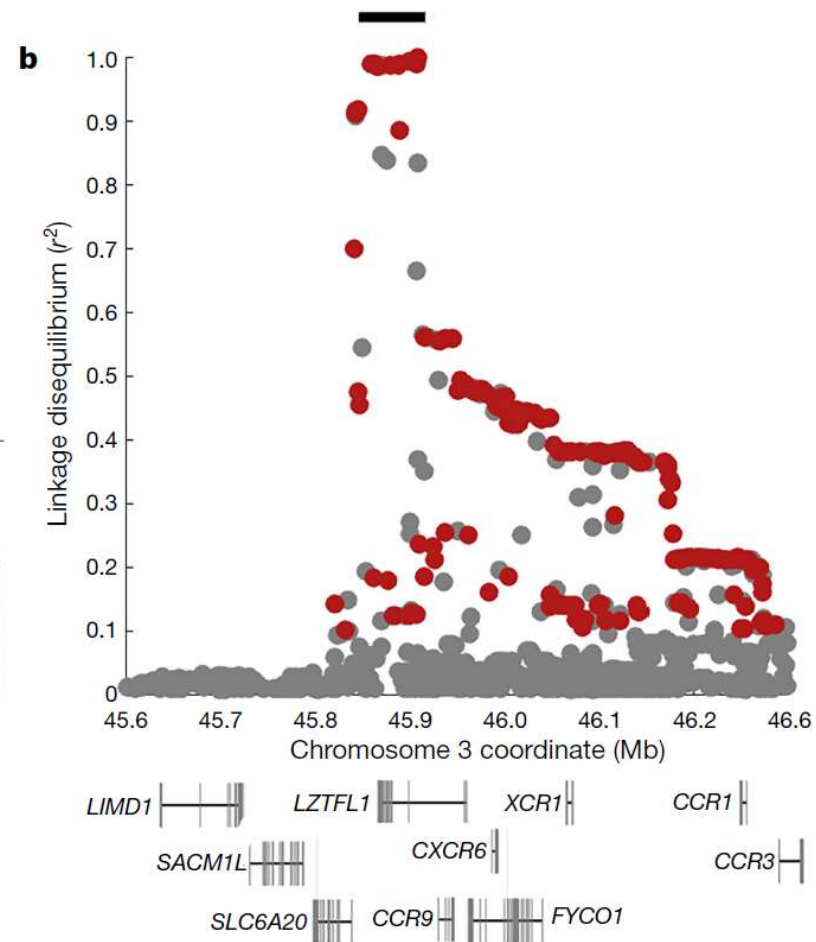
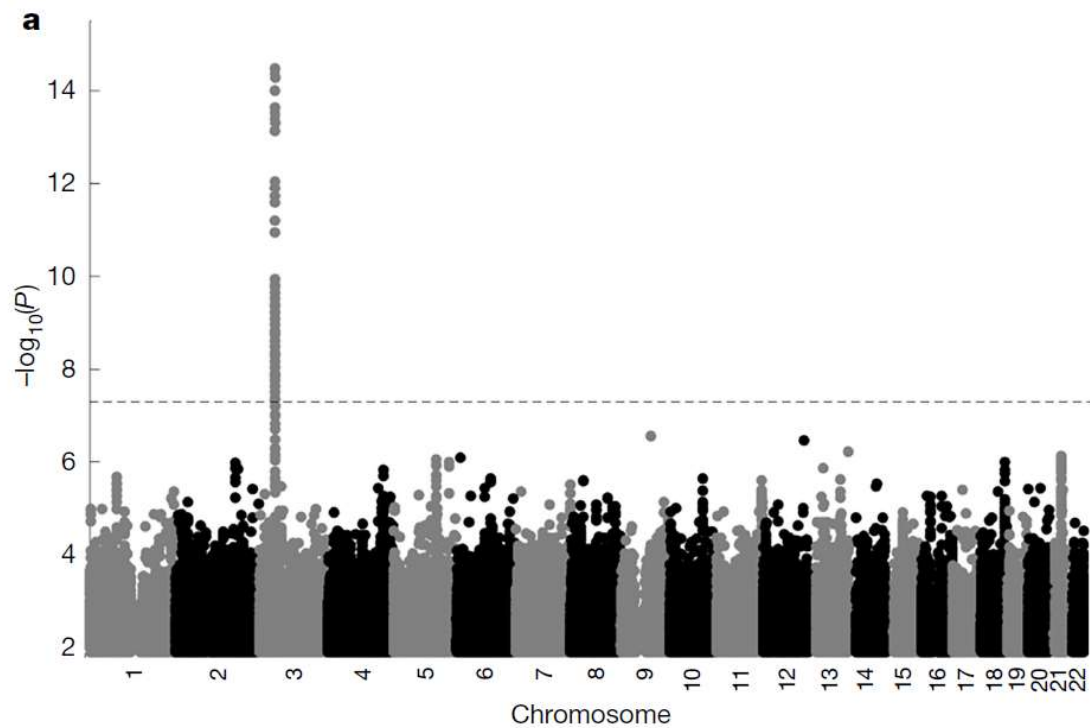


Extended Data Figure 3 | Haplotype frequencies for Tibetans, our Han samples and the populations from the 1000 genomes project for the five-SNP motif in the *EPAS1* region. The y axis is the haplotype frequency. The legend shows all the possible haplotypes for the region considered among these populations: ASW, African American from the south western United

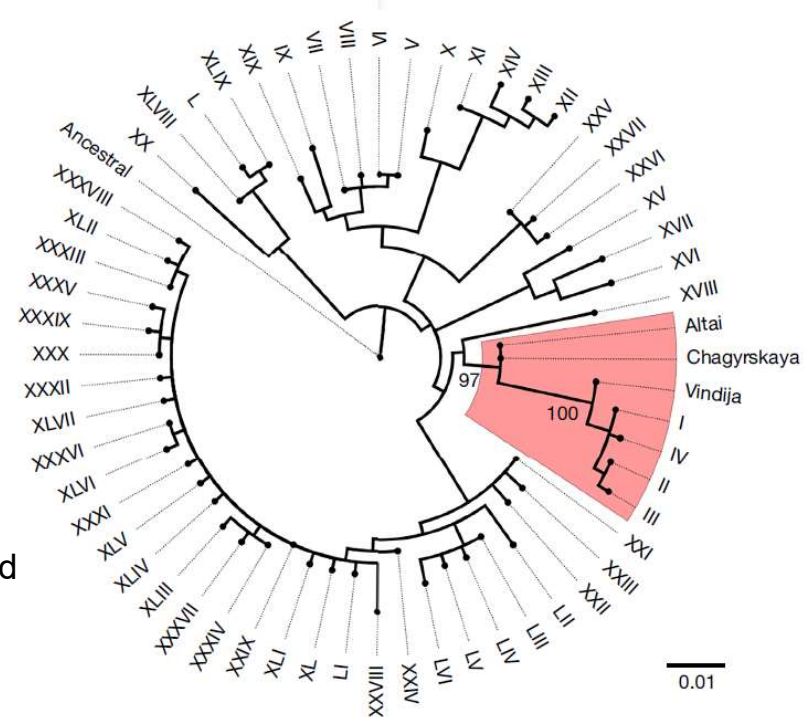
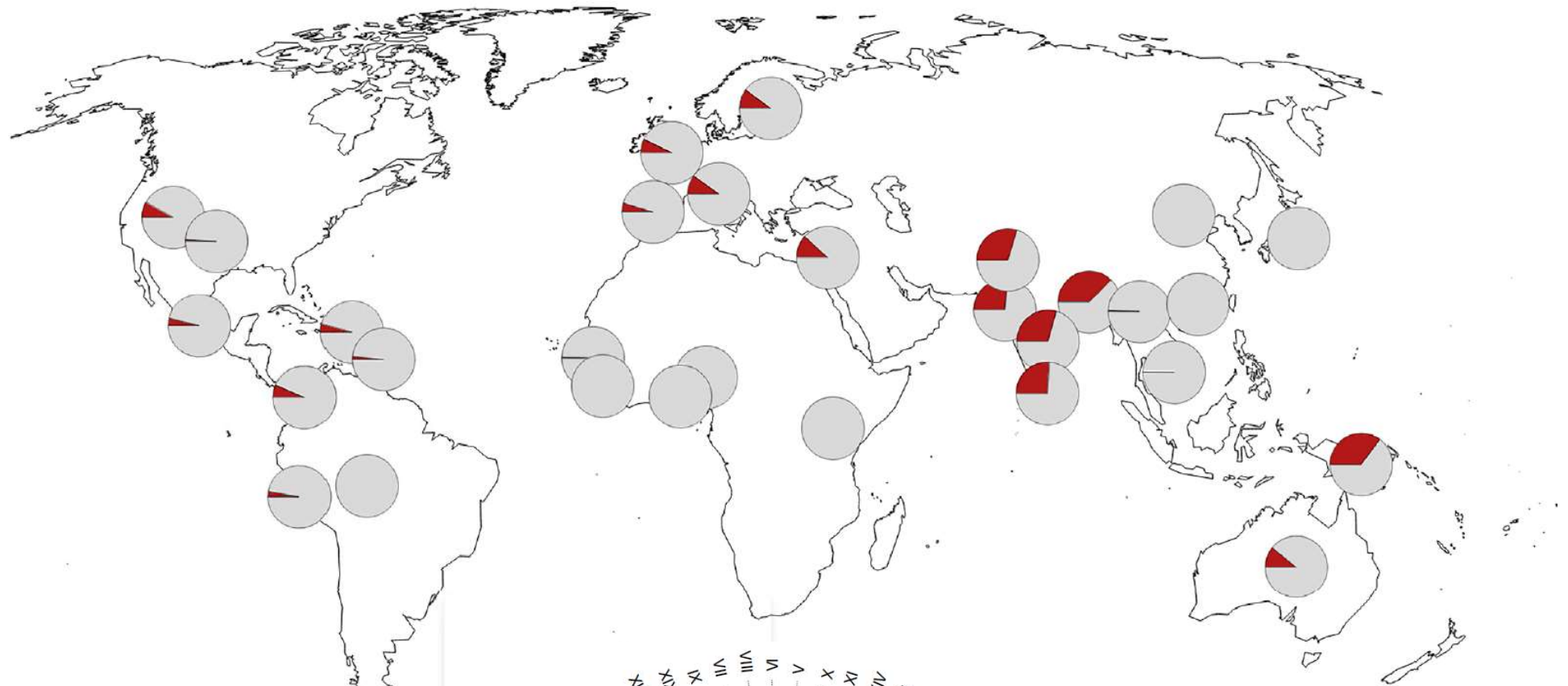
States; CEU, Utah Residents with Northern and Western European ancestry; CHB, Han Chinese from Beijing; CHS, Southern Han Chinese; CLM, Colombian; FIN, Finnish; GBR, British; HAN, Han Chinese from Beijing; IBS, Iberian; JPT, Japanese; MXL, Mexican; PUR, Puerto Rican; LWK, Luhya; TSI, Toscani; TIB, Tibetan; YRI, Yoruban (see Methods).



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



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

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