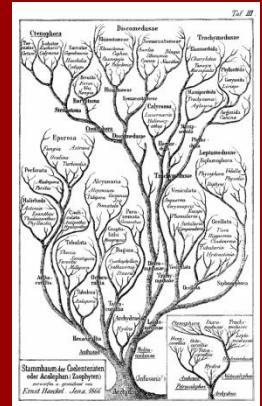
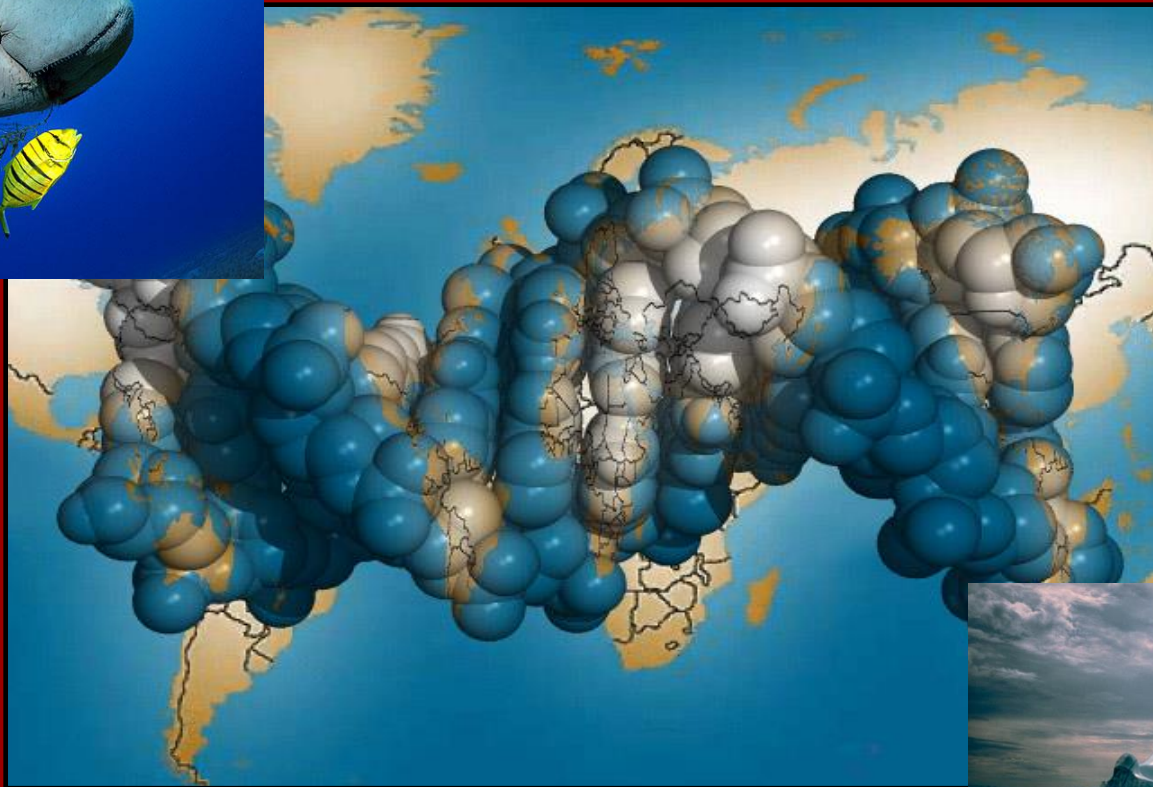


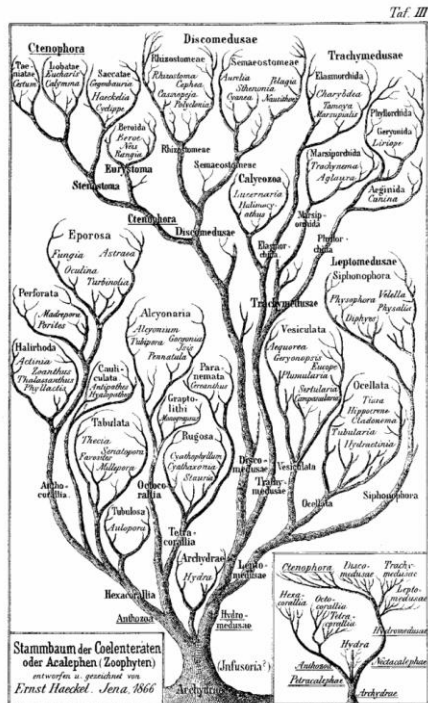
phylogeography

Pavel Hulva

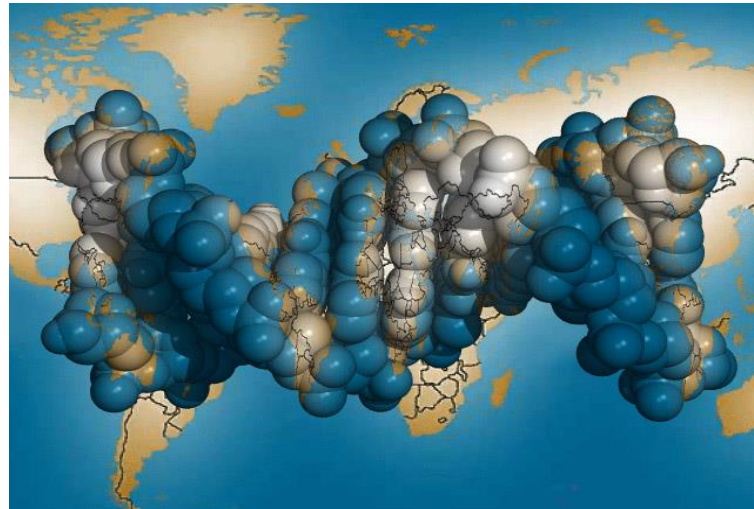


definiton

- Studying mechanisms influencing geographical distribution of genealogies
- Historical, phylogenetic component of spatial distribution of genealogies



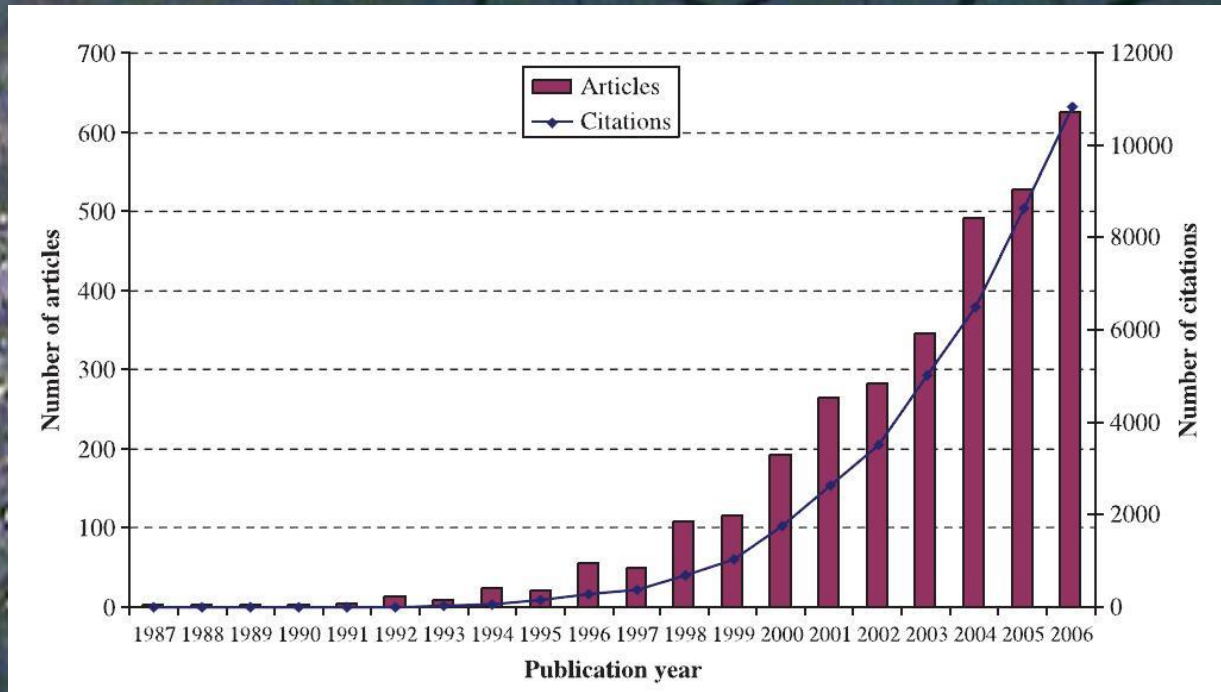
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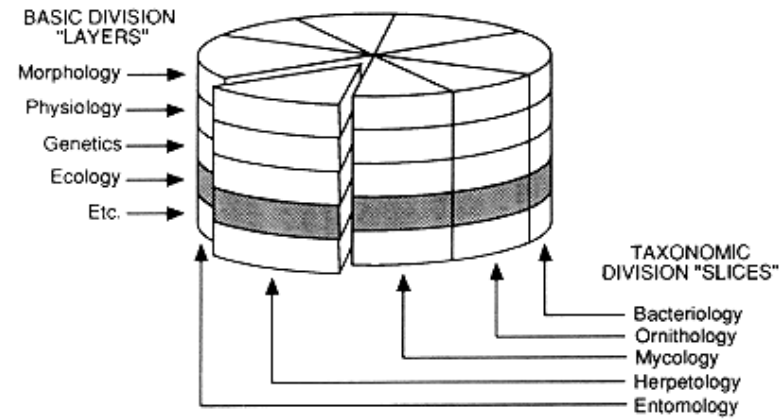
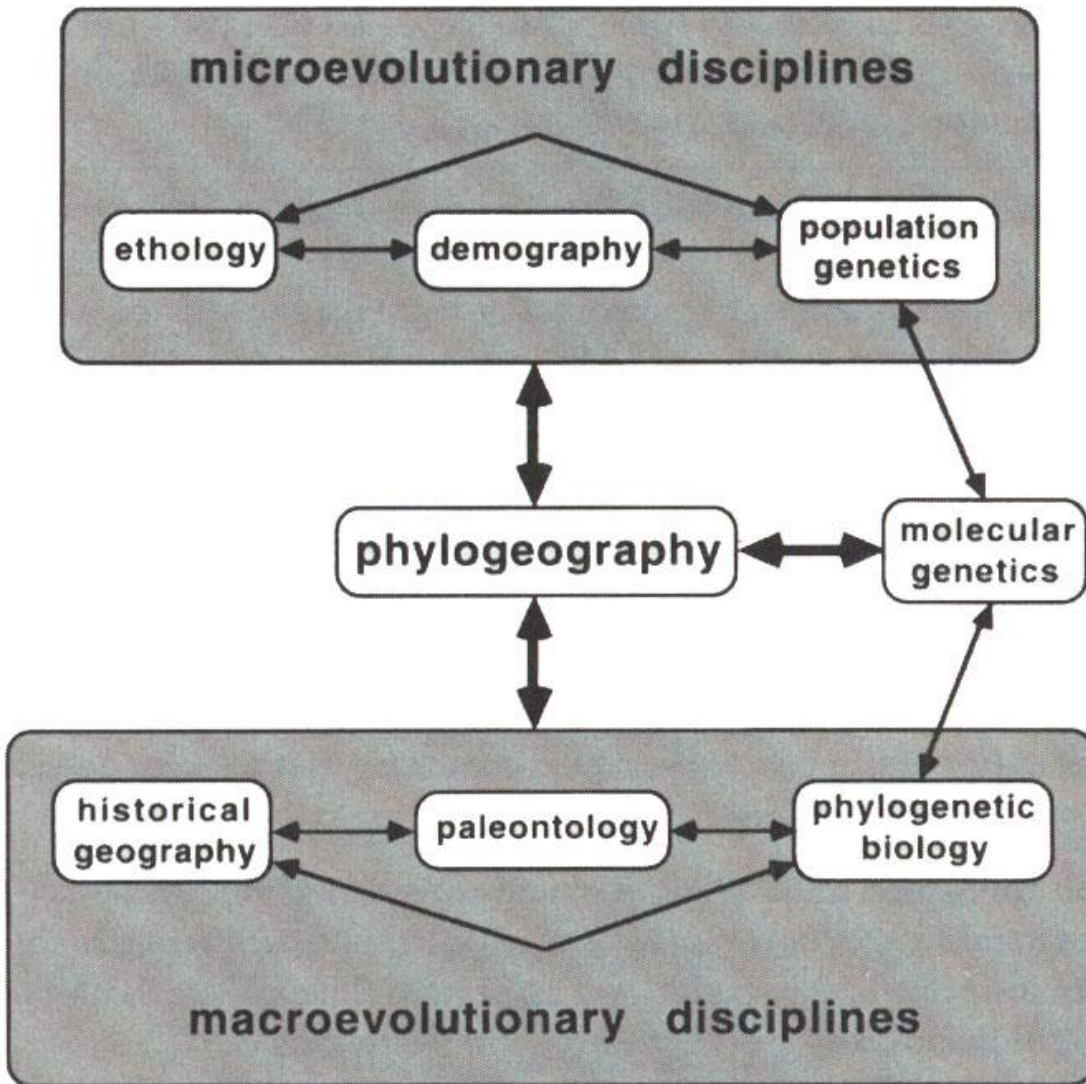


PHYLOGEOGRAPHY

THE HISTORY AND FORMATION OF SPECIES

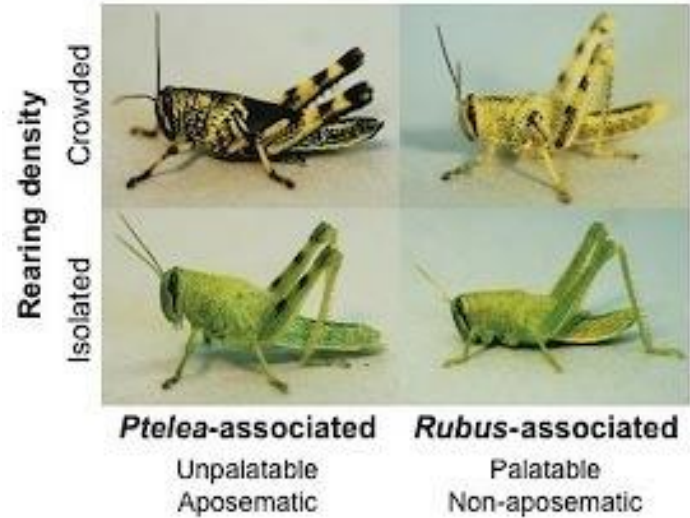


Avise, 2000



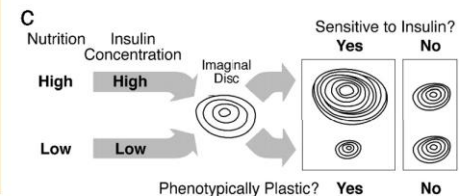
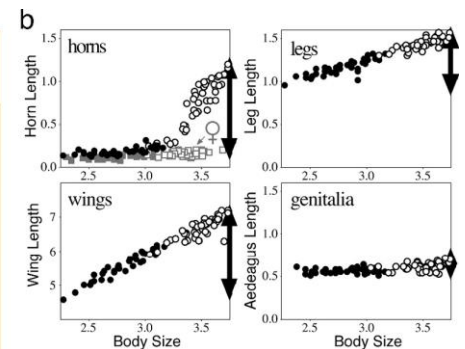
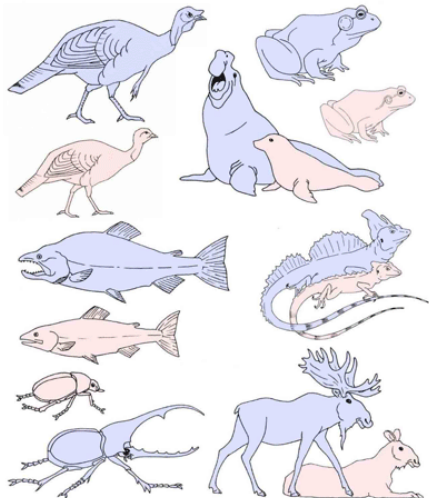
Avise, 2000

Variation related to phenotypic plasticity, age, sexual dimorphism, ecotypes, etc. – role of genetics in studying variation and range history



Males are the larger sex in many species...

yet females are the larger sex in many others



molecular biology

phenotype

often studied directly
proximate mechanisms
focused to extreme phenotypes
(loss of function)

genetic variability and genotype

simple approach to variation
(wild type / mutant etc.)

molecular evolution

phenotype

seldom studied directly

genetic variability and genotype

complex approach to variability
mechanisms of its origin and
maintenance
complicated mathematical apparatus

Divergences among species

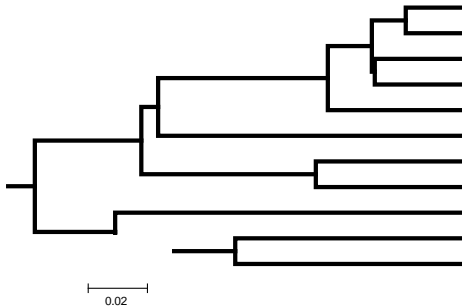
Large genetic distances among samples

Easy to find well supported tree

trees – phylogenies

Tree topology important for hypothesis formulations

Nodes represent speciation events



divergences within species (phylogeography)

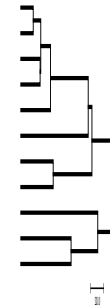
Small genetic distances among samples

Difficult to find well supported tree

trees – genealogies

Tree topology less important for hypothesis formulations

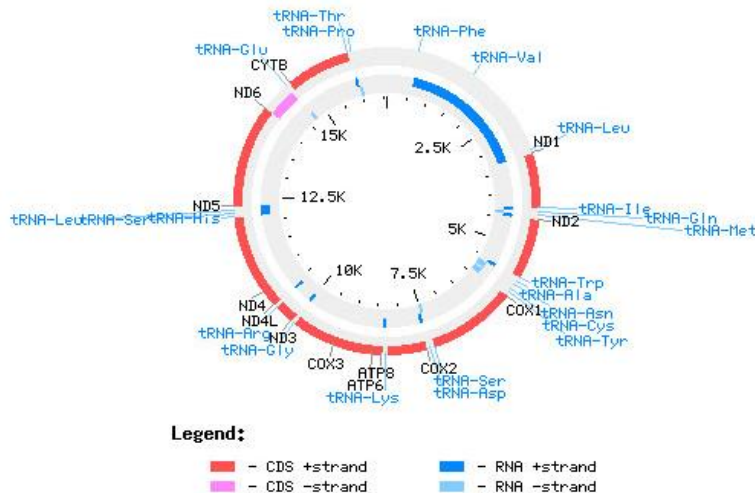
Nodes represent coalescent events



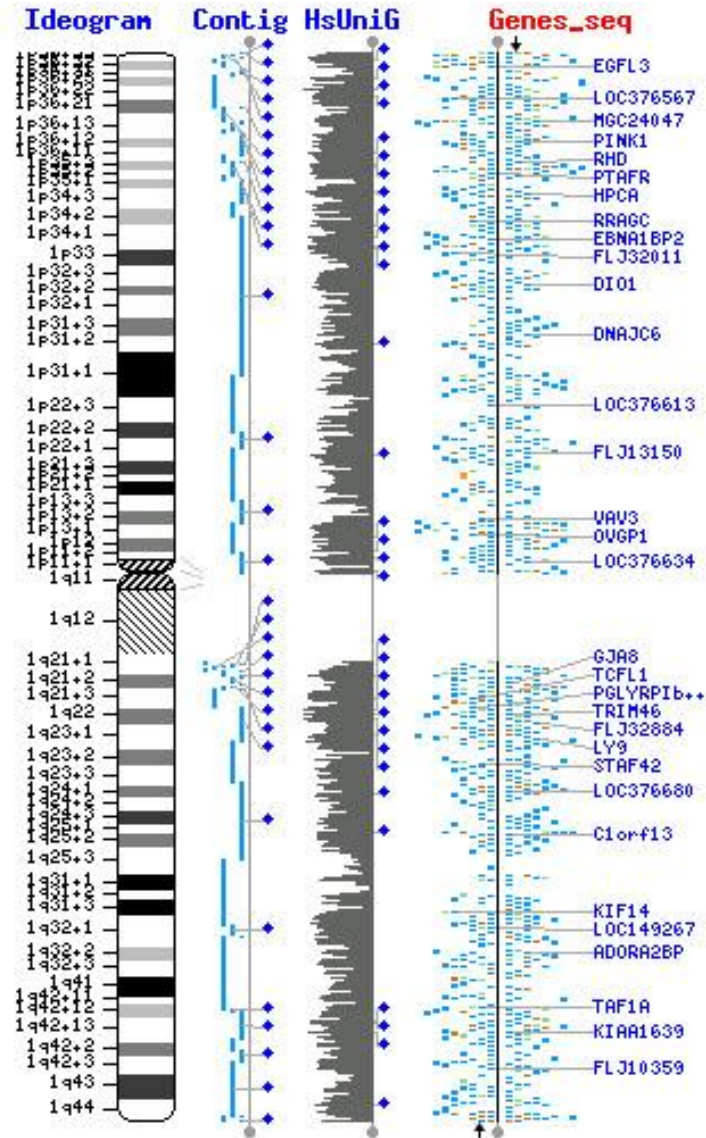
mtDNA x nDNA

properties of mtDNA

small, compact, clonal, stable (two mitochondrial membranes, more copies), high mutation rate – high intraspecific variation - haplotypes, particular regions exhibit different mutation rate, lack of recombination, matrilinear



exceptions: recombination, heteroplazmy, paternal leakage



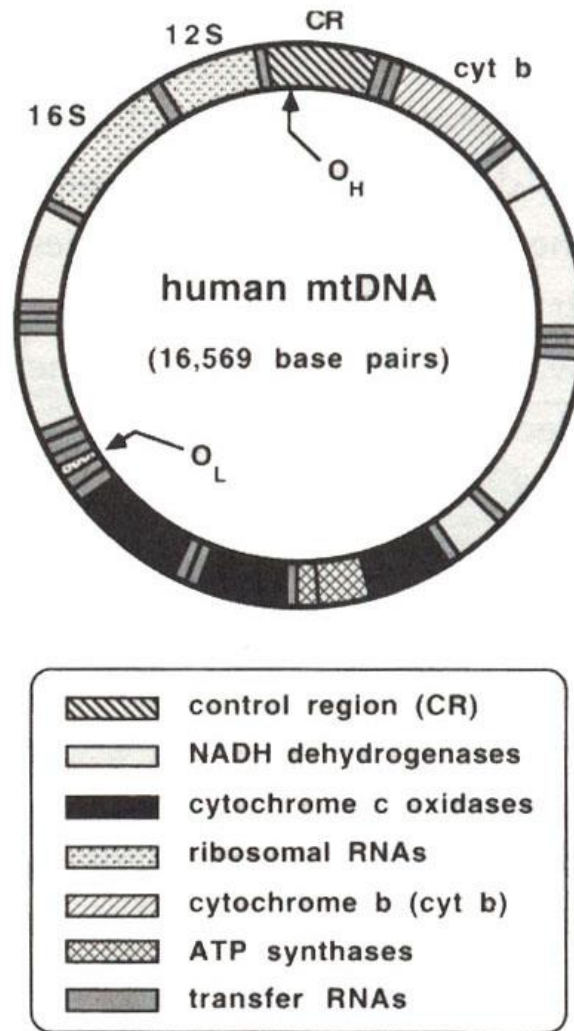
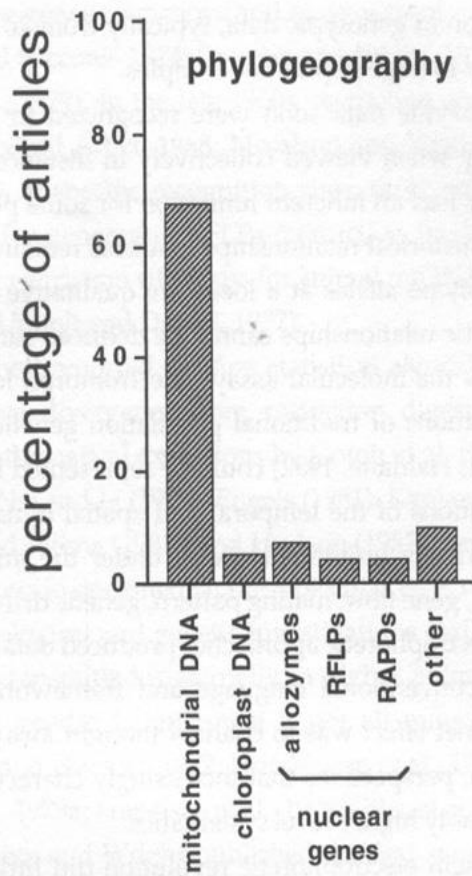
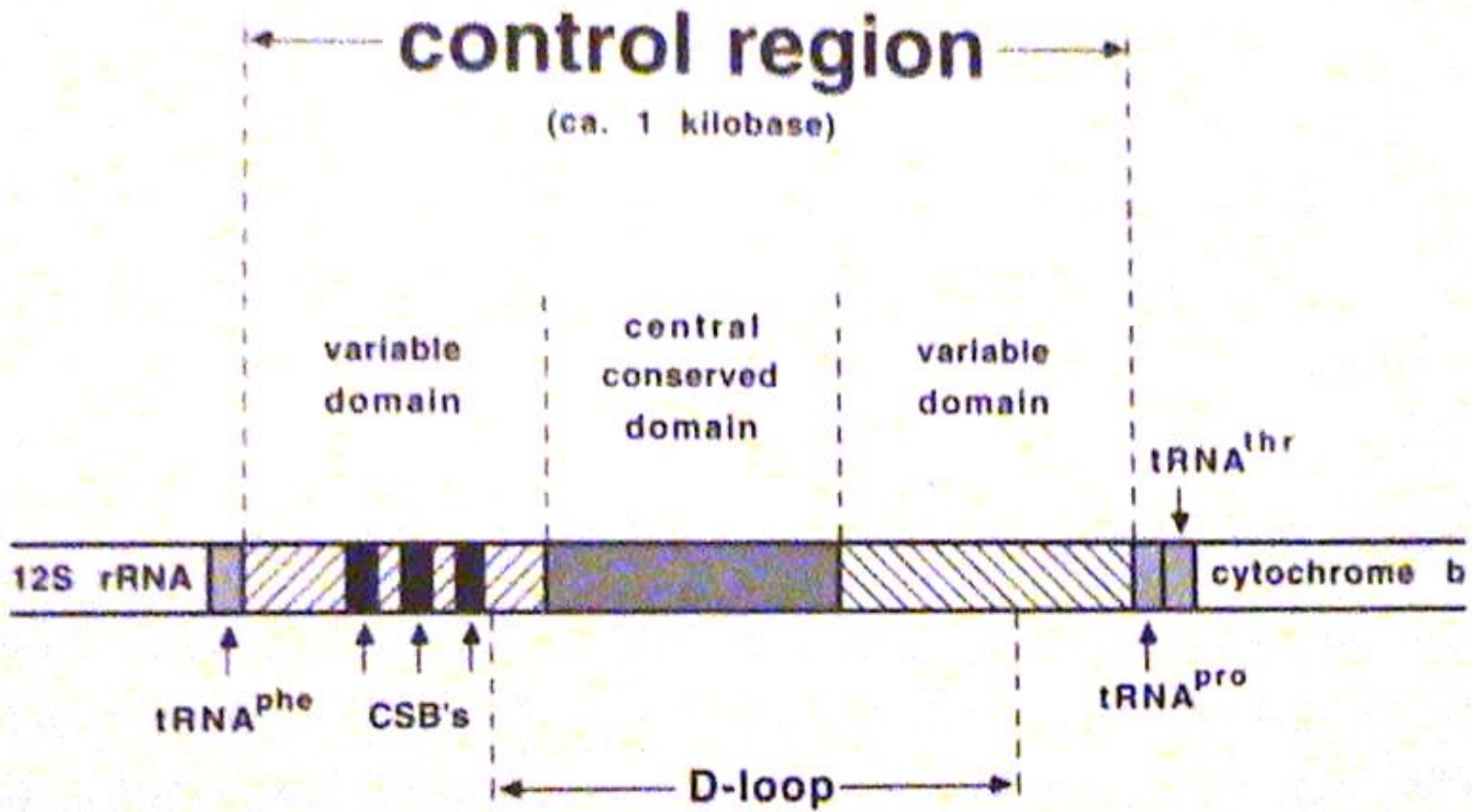


FIGURE 1.10 Structure and organization of genes in mammalian mtDNA. O_H and O_L are origins of replication of the two strands of the molecule. MtDNA molecules normally display identical gene orders within and among related animal species, but occasional rearrangements have occurred in evolution and can be exploited as markers to identify deep branches in macroevolutionary trees (Smith et al., 1993; Boore et al., 1995).



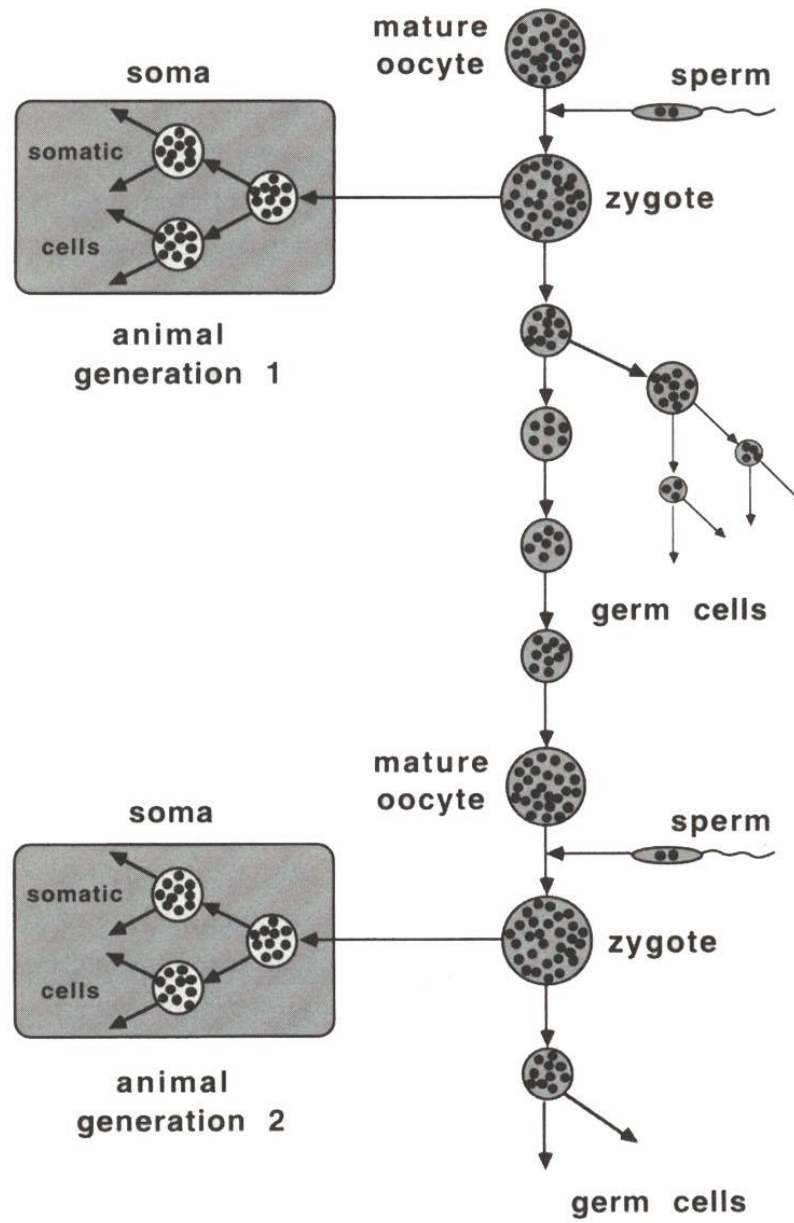
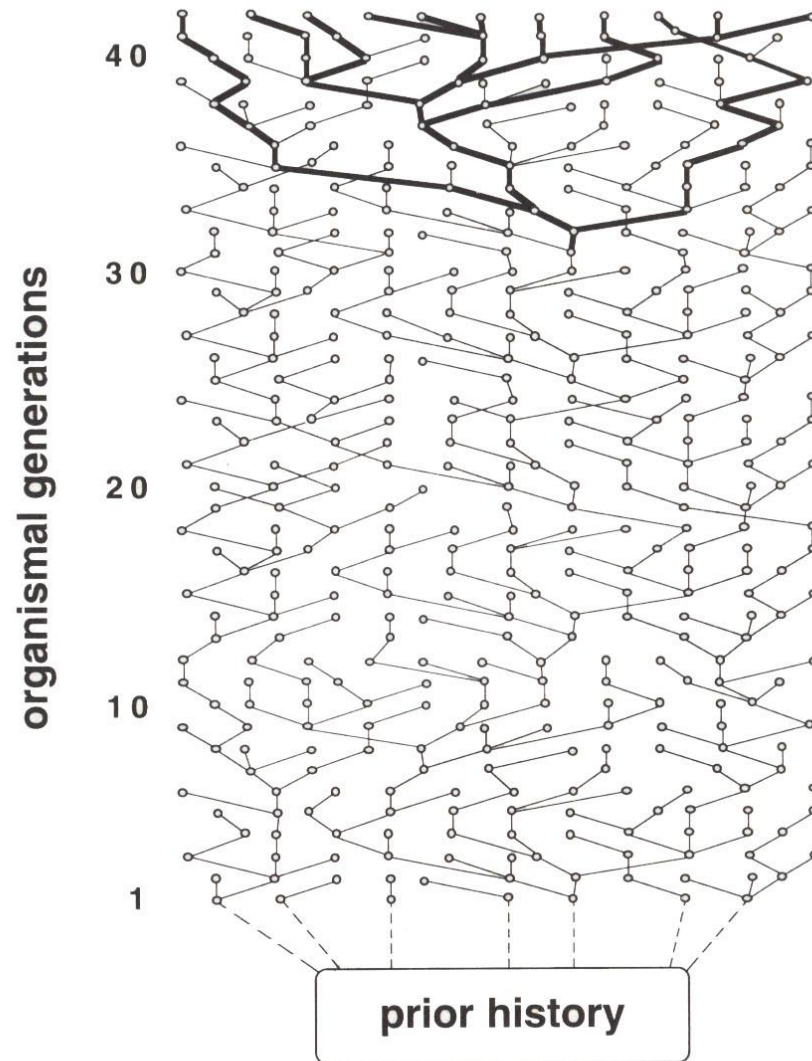


FIGURE 1.11 Mitochondria (black dots) exist as large populations of organelles within the cytoplasmic lineages of somatic cells and germ cells. In most animals, about 20–50 germ-cell generations intervene between organismal generations.



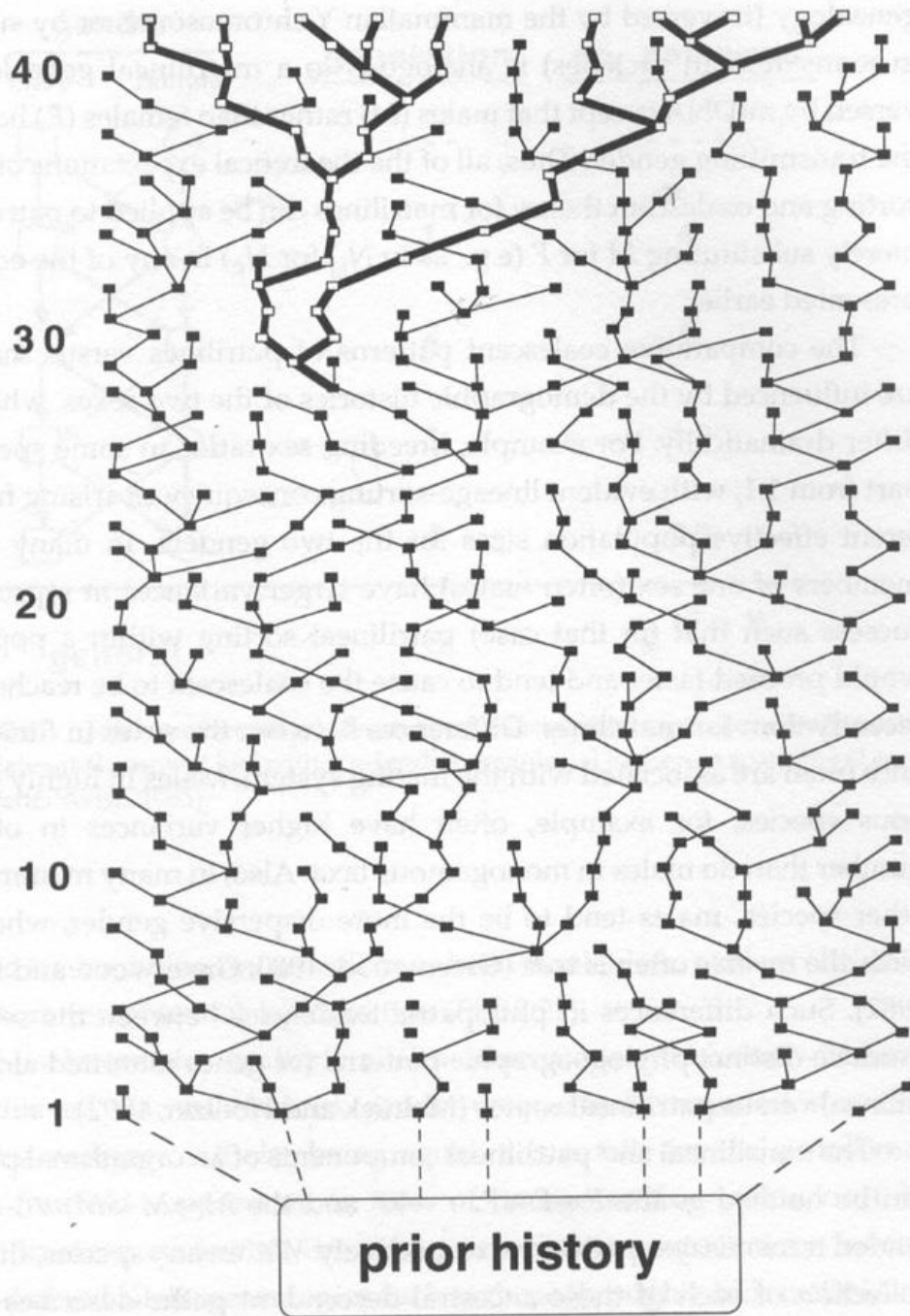
mtDNA
nonreticulate genealogy

x

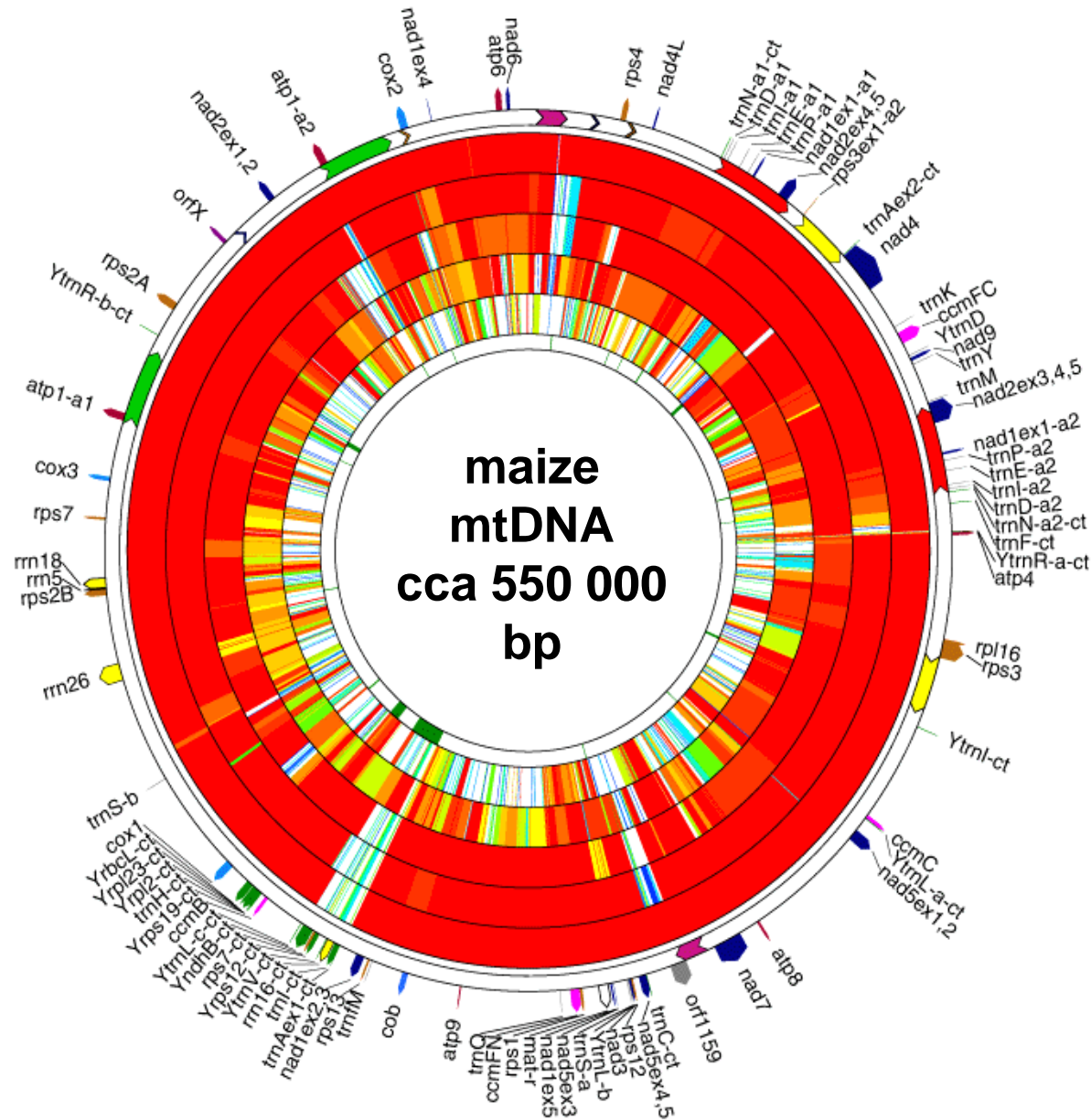
nucDNA
anastomose genealogy

FIGURE 2.1 Schematic representation of the lineage sorting process within a population. Shown are matrilineal pathways connecting mothers and daughters across more than 40 generations. Lines that cross do not imply any genetic event such as recombination. Note that all seven founding lineages except one have gone extinct during this interval, and that all extant lineages (*top*) coalesce to a common female parent 10 generations earlier. Note also that these matrilineal pathways collectively describe a hierarchical and branched gene tree reflecting the hereditary transmission routes traversed by mtDNA molecules.

organismal generations



plant mtDNA – high rate of rearrangements, slow sequence evolution
 (ca 100x lower mutation rate compared to animal mtDNA)

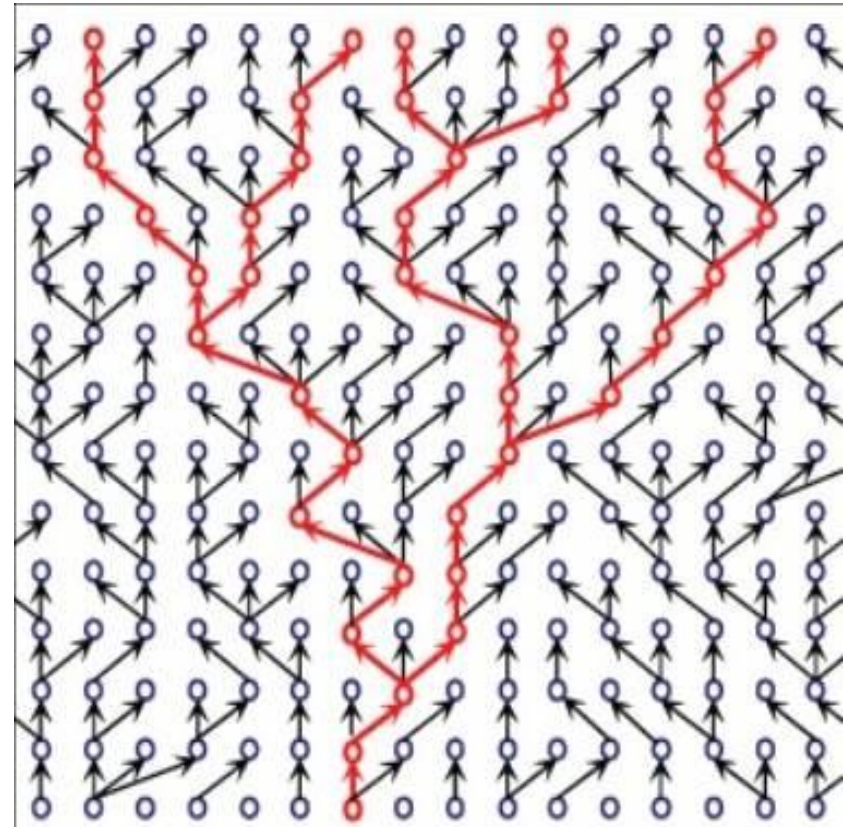
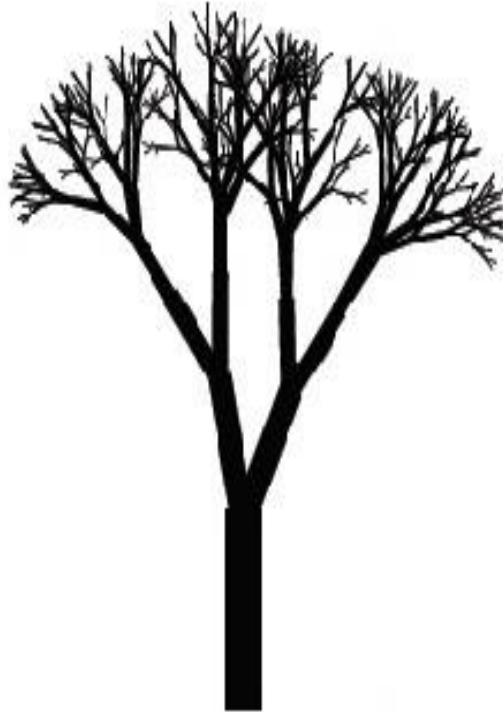
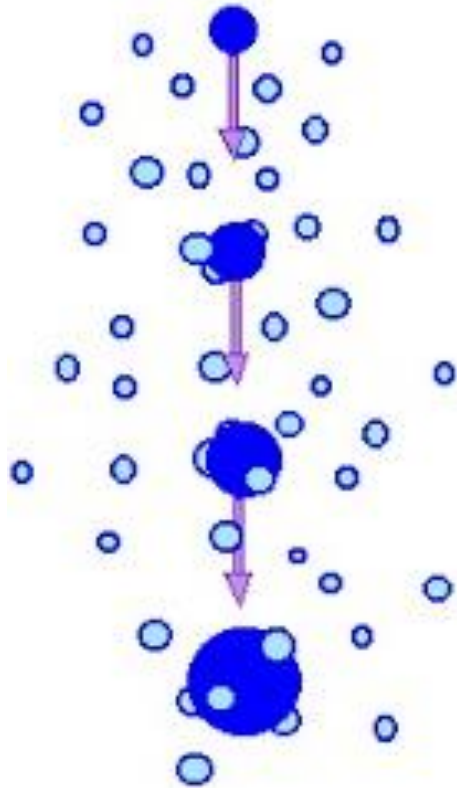


coalescent theory

aerosols physics

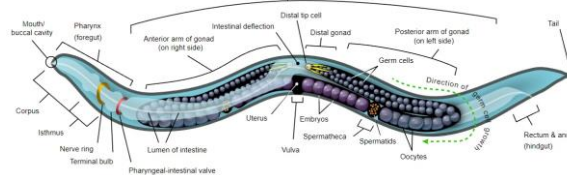
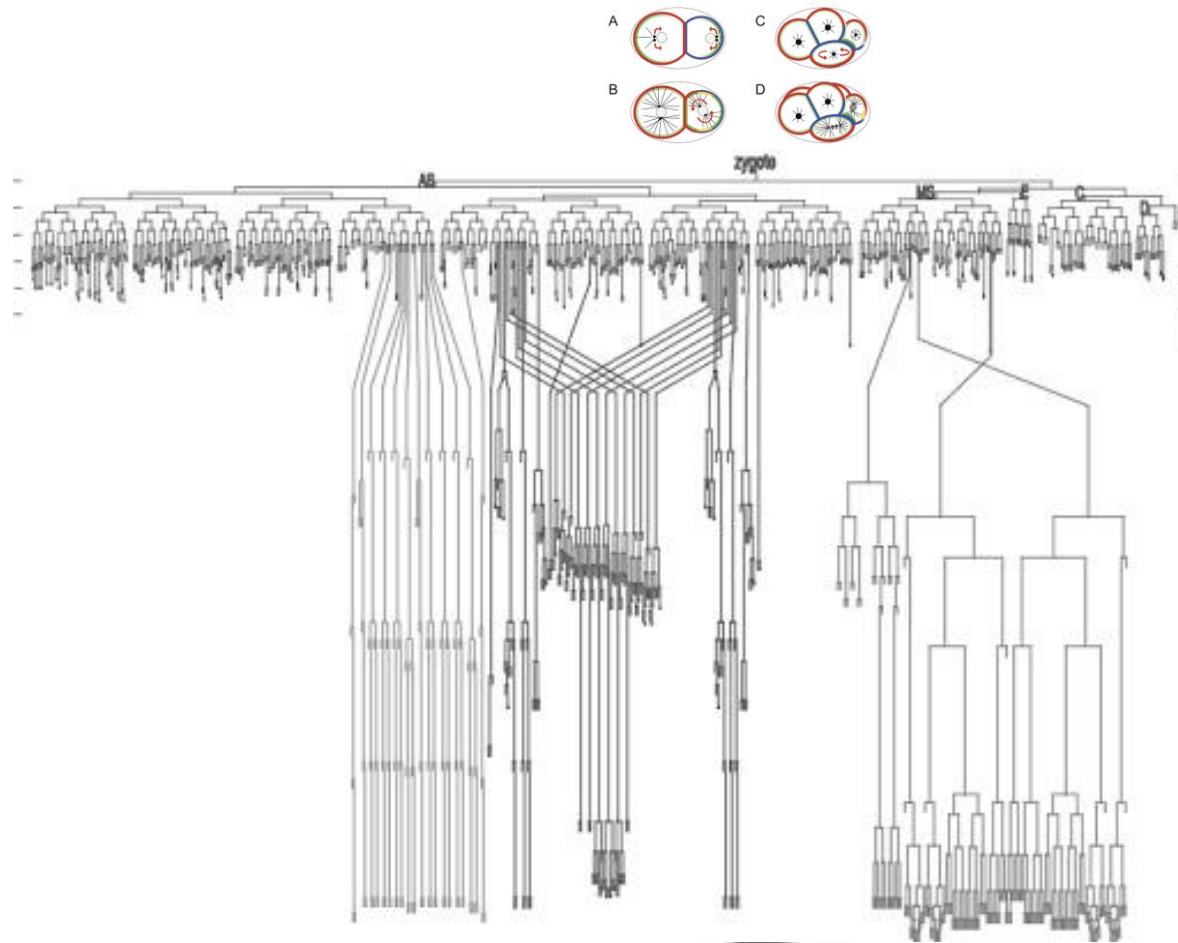
tree shape

genealogies shape



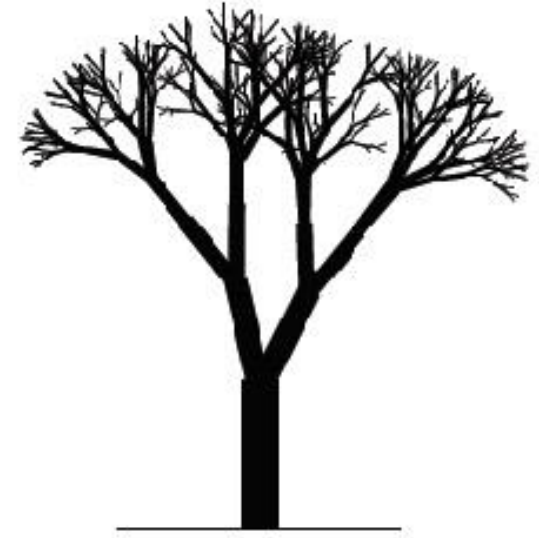
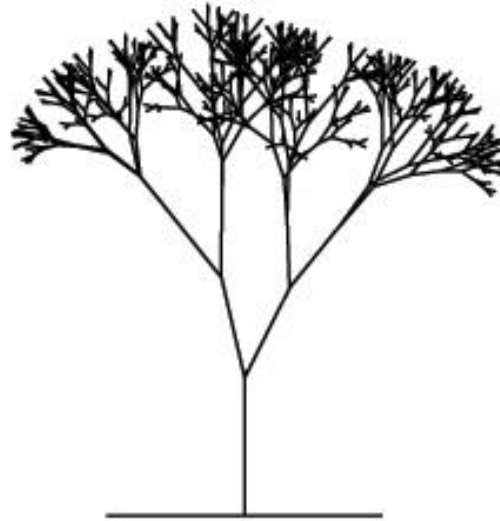
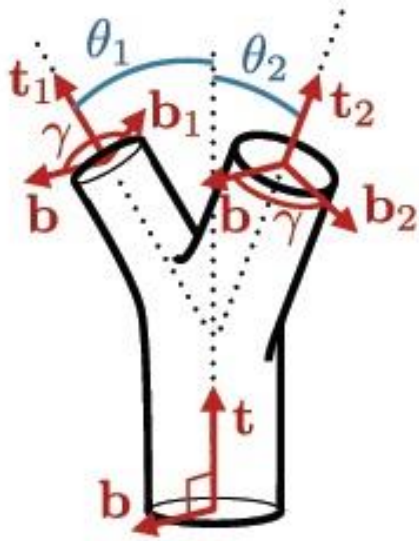
hierarchic levels of multiplication

- **ontogeny** (descent by mitosis) – **tokogeny** (descent by reproduction)
- **phylogeny** (descent by speciation) -



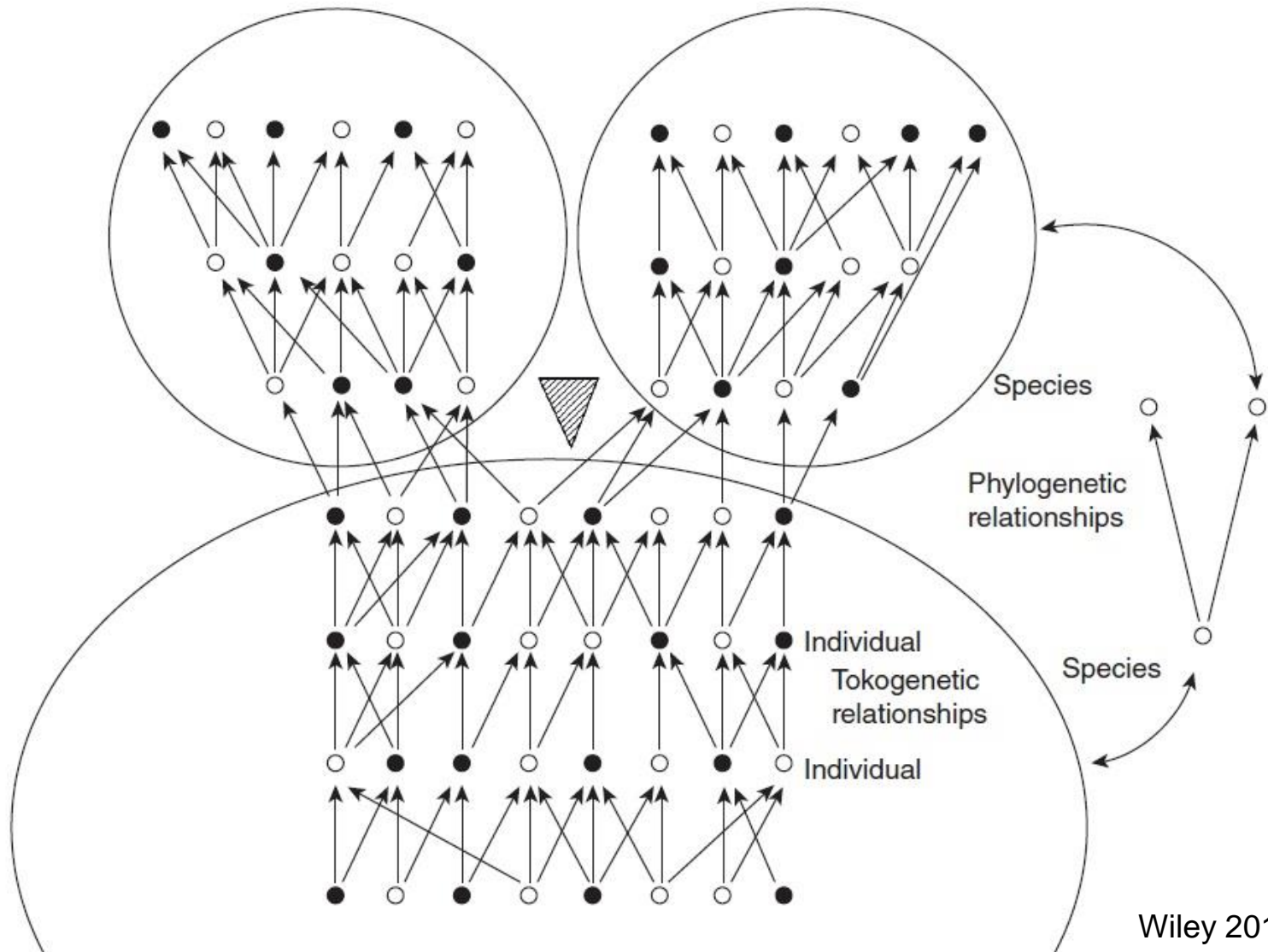
hierarchic levels of multiplication

- **ontogeny** (descent by mitosis) – **tokogeny** (descent by reproduction)
- **phylogeny** (descent by speciation) -



hierarchic levels of multiplication

- **ontogeny** (descent by mitosis) – **tokogeny** (descent by reproduction)
- **phylogeny** (descent by speciation) -



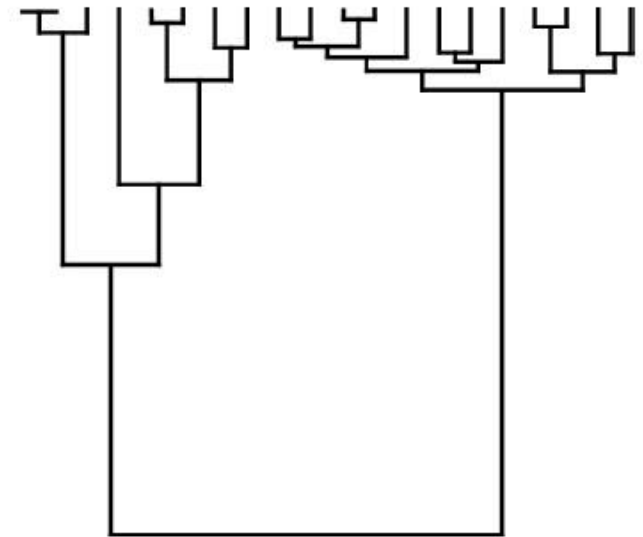
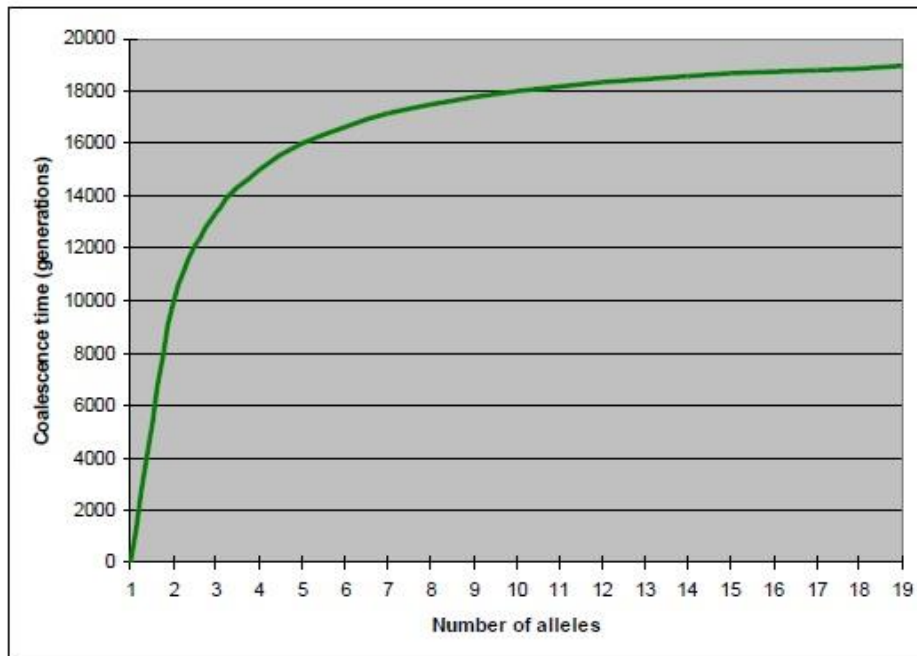
coalescent theory

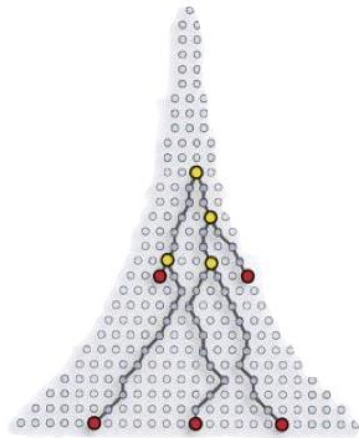
provides the link between phylogenetic models utilizing tree graphs and the background mechanisms of population genetics.

Enables to model sources of gene-tree / species-tree incongruences that arise due to population level processes, and make estimates about demographic parameters and interpret the shape of genealogies.

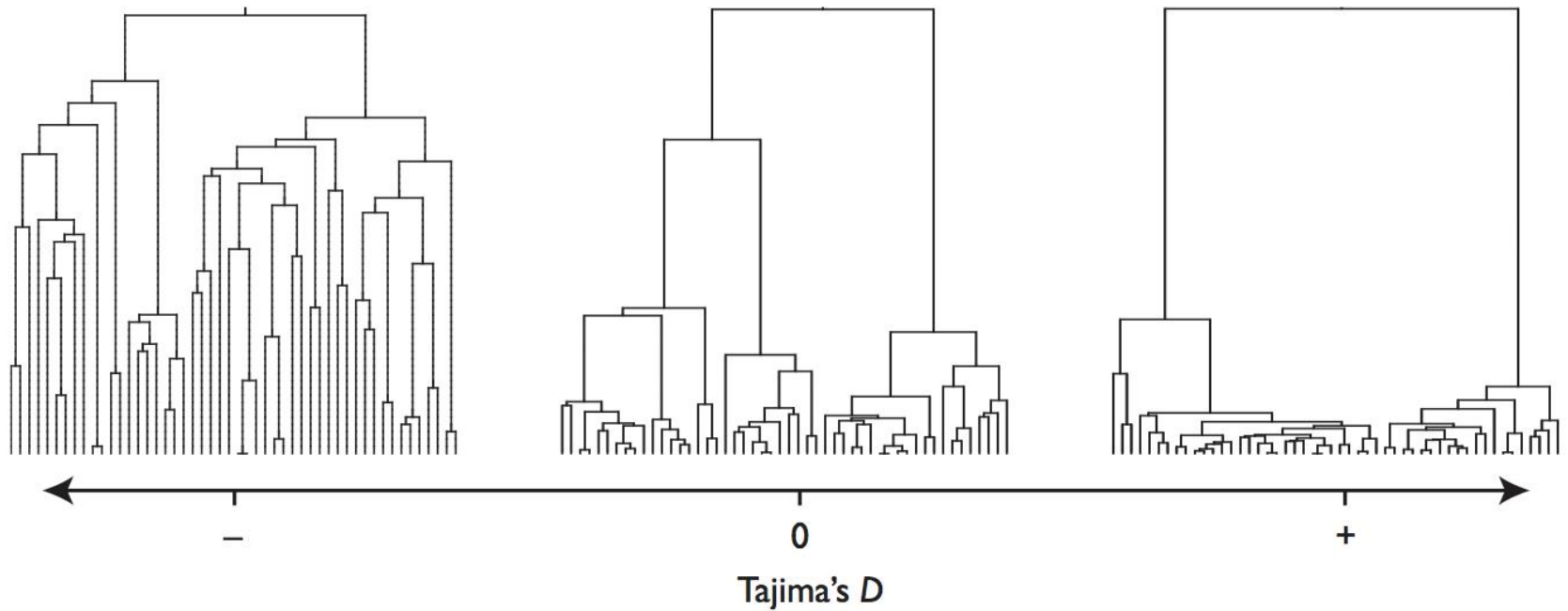
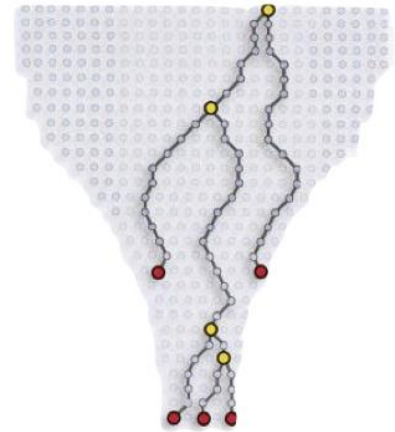
Assumptions – neutral model:

Finite constant population size – coalescent time depends upon $4N$
non-overlapping generations
random mating
no mutations
no selection
without population structure

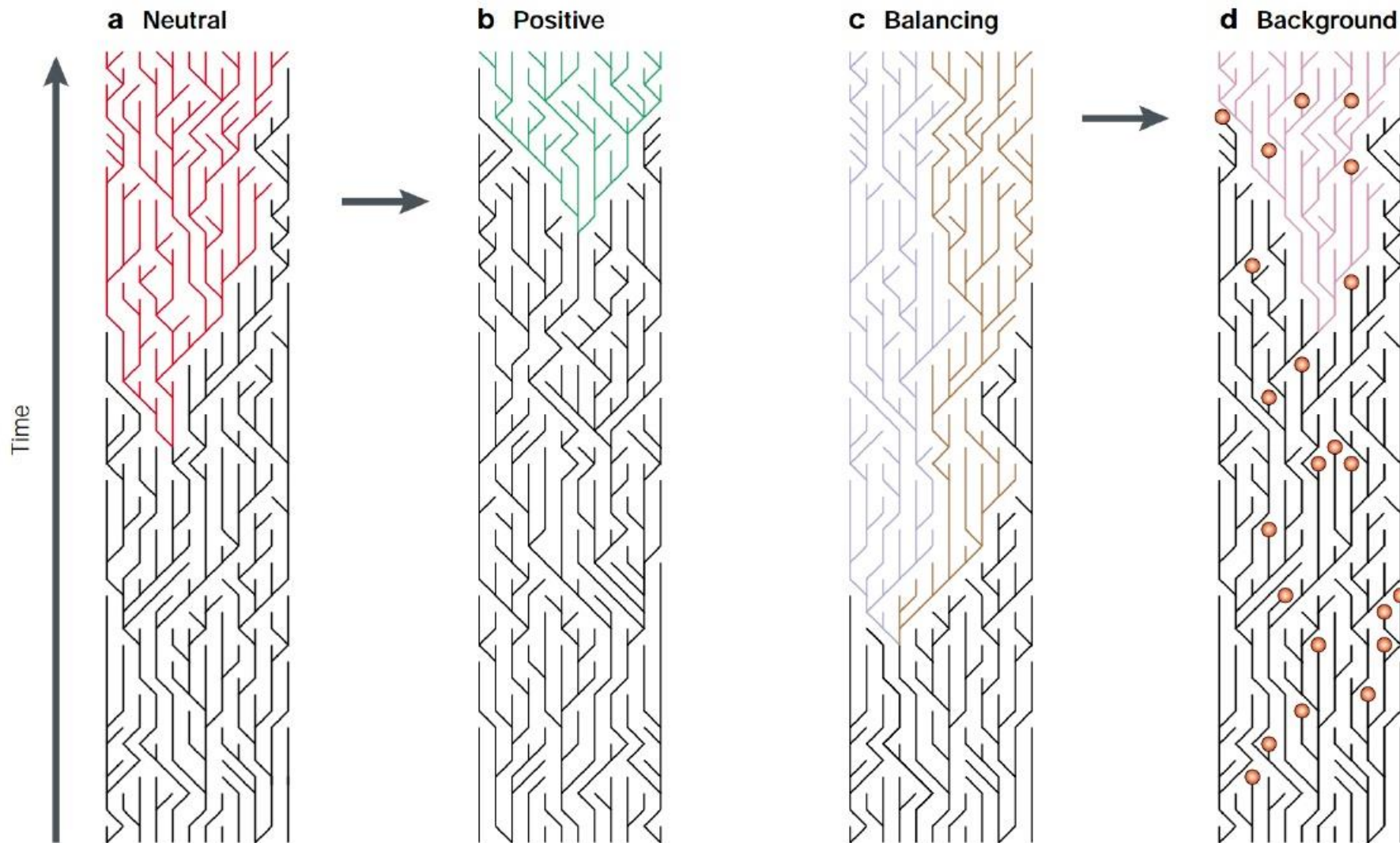


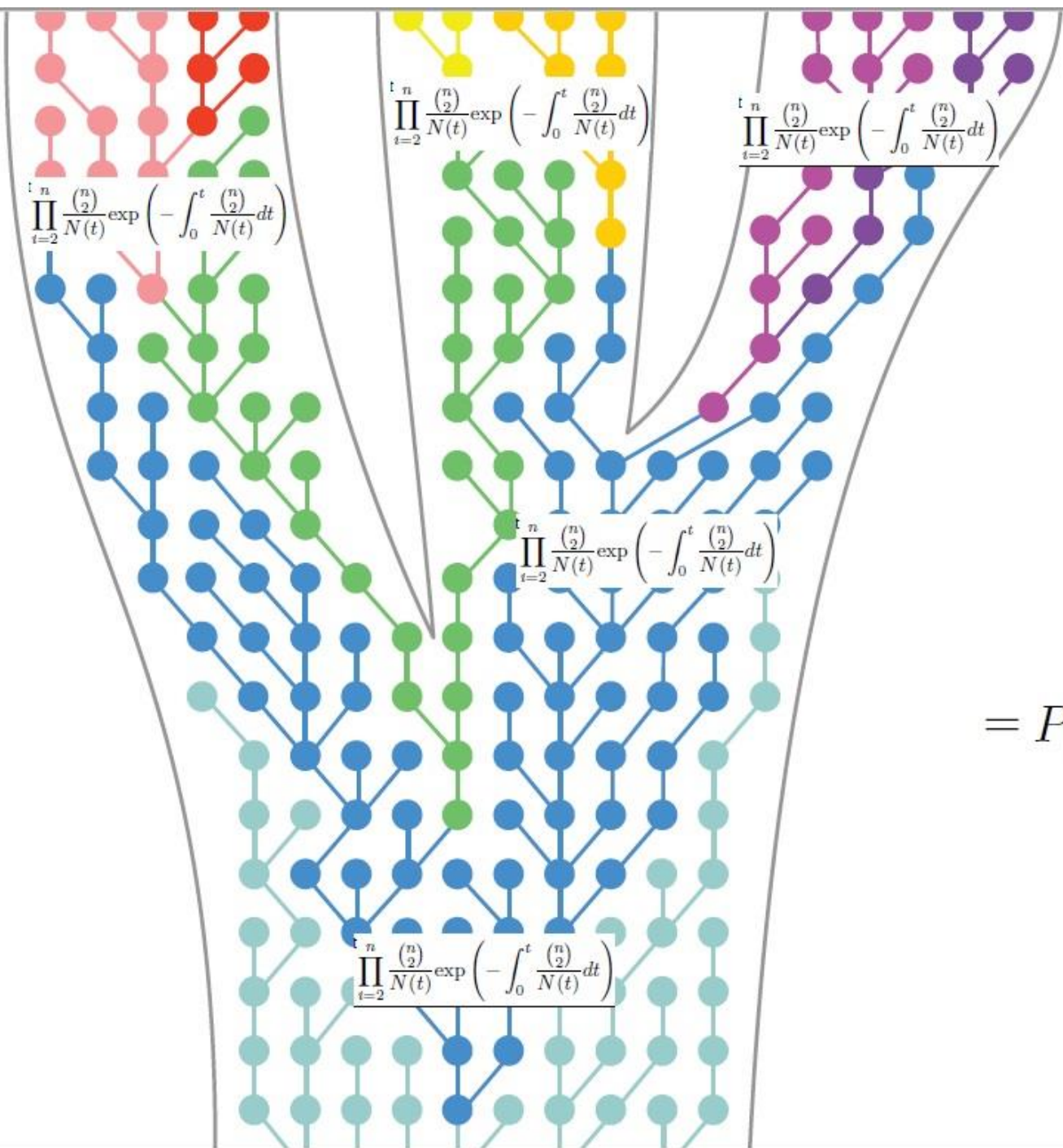


Compares singleton mutations
to shared diversity



Coalescent models usually assumes neutrality,
but can also be used to detect selection.





$$\prod_{t=2}^n \frac{\binom{n}{2}}{N(t)} \exp\left(-\int_0^t \frac{\binom{n}{2}}{N(t)} dt\right)$$

$$\prod_{t=2}^n \frac{\binom{n}{2}}{N(t)} \exp\left(-\int_0^t \frac{\binom{n}{2}}{N(t)} dt\right)$$

$$\prod_{t=2}^n \frac{\binom{n}{2}}{N(t)} \exp\left(-\int_0^t \frac{\binom{n}{2}}{N(t)} dt\right)$$

$$\prod_{t=2}^n \frac{\binom{n}{2}}{N(t)} \exp\left(-\int_0^t \frac{\binom{n}{2}}{N(t)} dt\right)$$

$$\prod_{t=2}^n \frac{\binom{n}{2}}{N(t)} \exp\left(-\int_0^t \frac{\binom{n}{2}}{N(t)} dt\right)$$

$$= P(g_i | S)$$

mitochondrial Eve

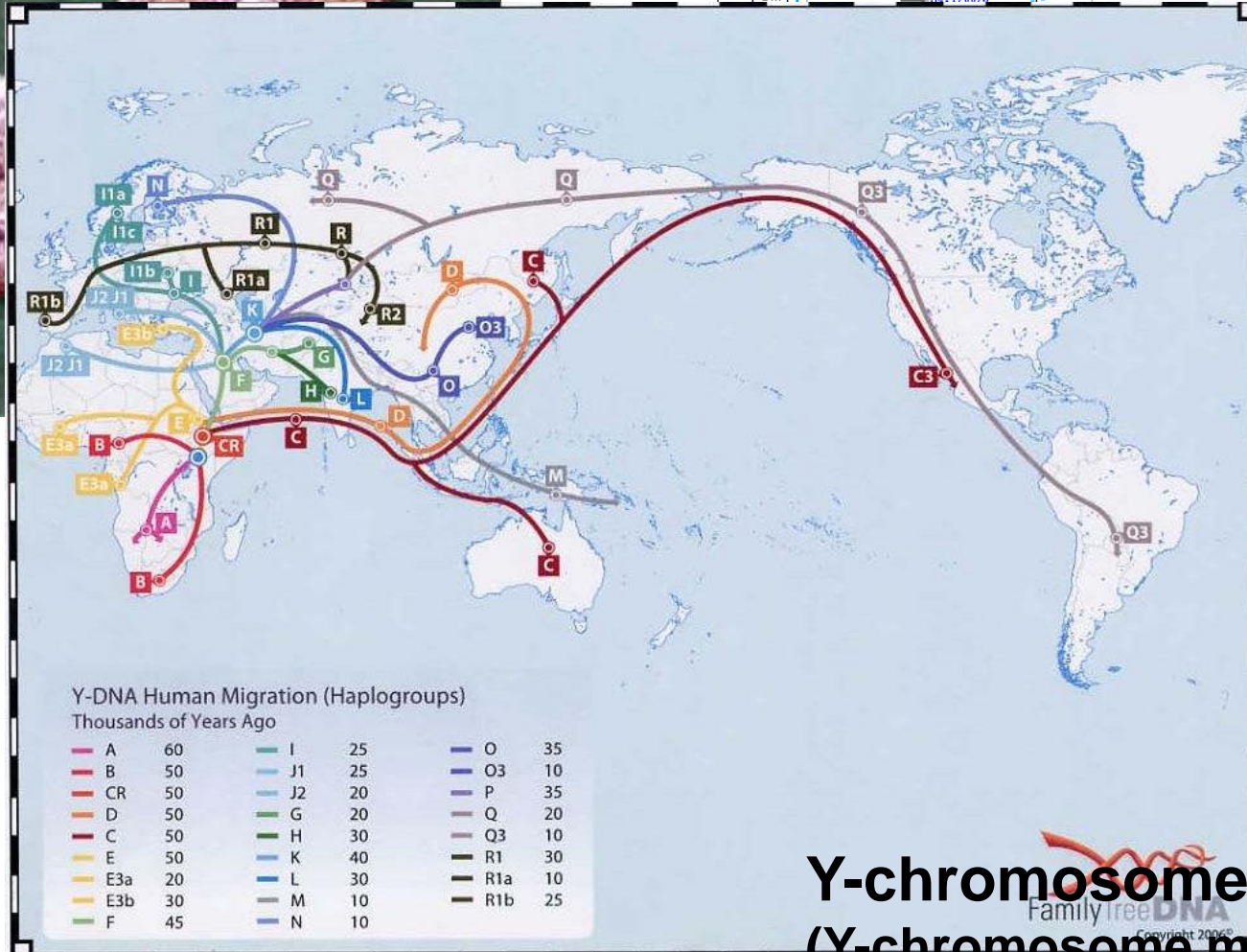
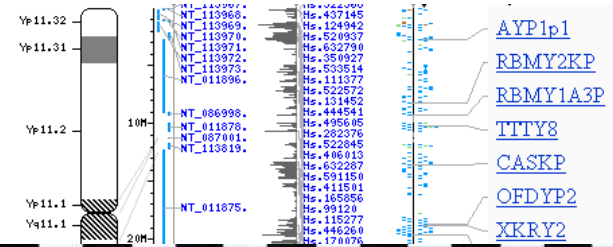
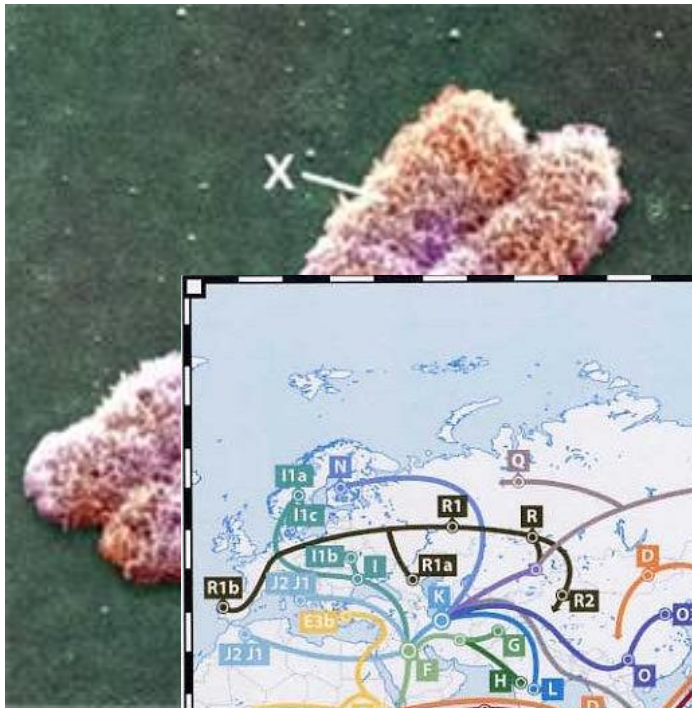
(mitochondrial mrca
– most recent common ancestor)



Brown et al. (1980) – small divergence in mtDNA in recent populations of *Homo sapiens* ($p=0.0036$) – divergence 180 – 360 000 years, bottleneck (Garden of Eden, Noah's Ark)

Cann et al. (1987) – hypothesis out of Africa





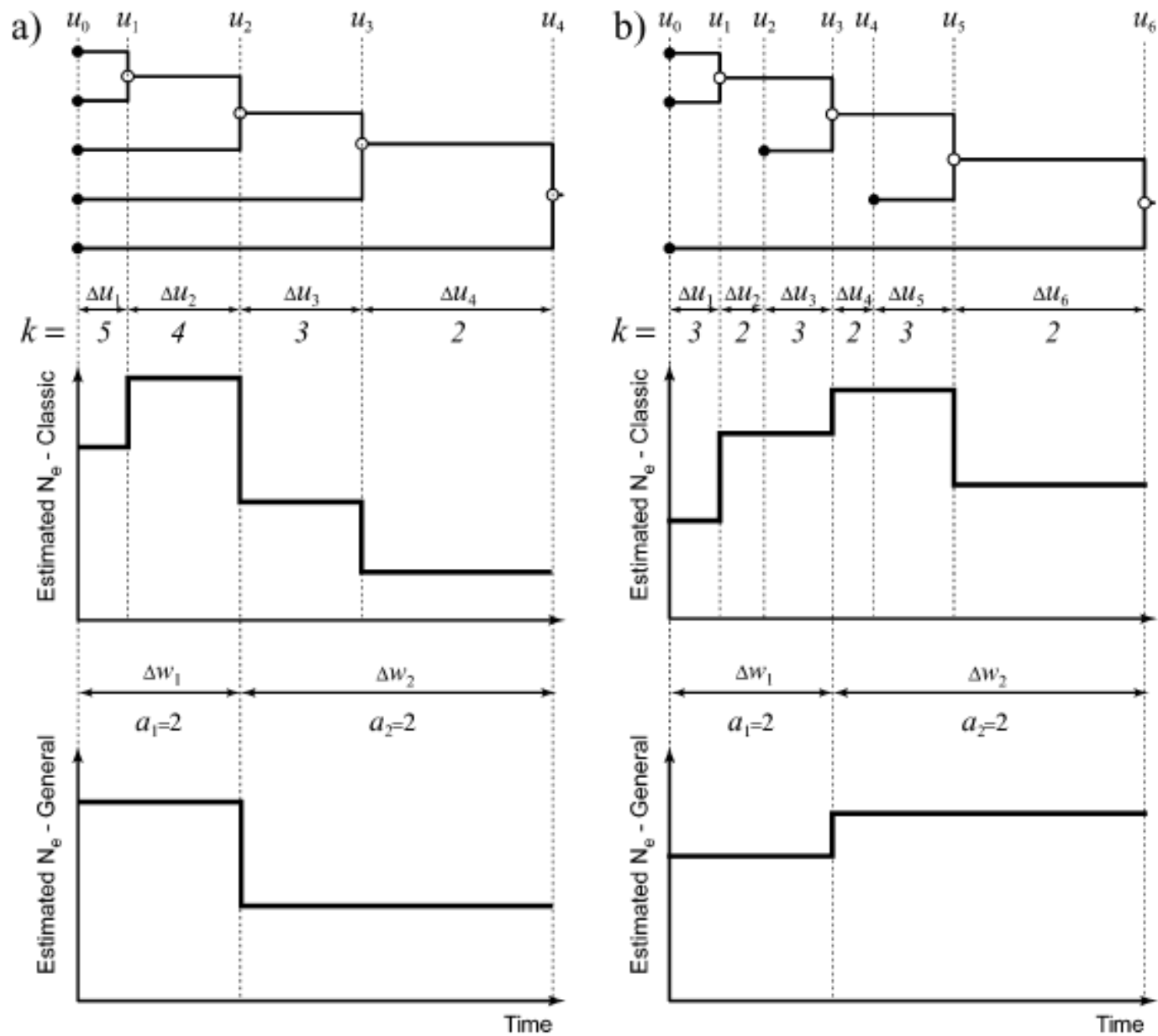
Y-chromosome Adam
(Y-chromosome mrca)

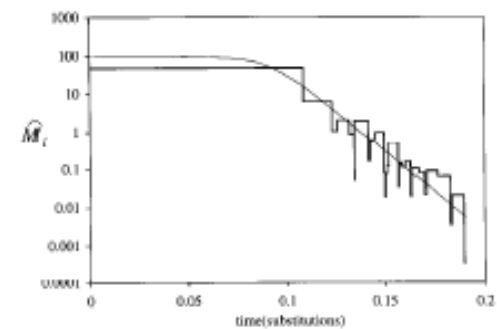
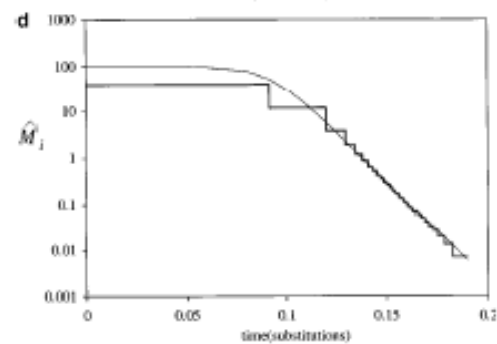
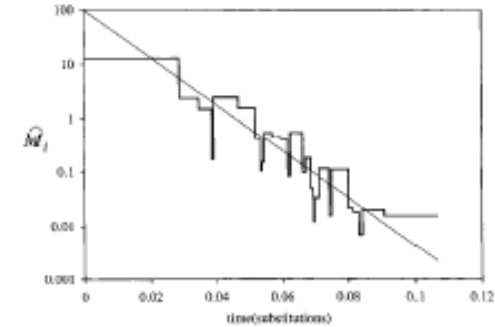
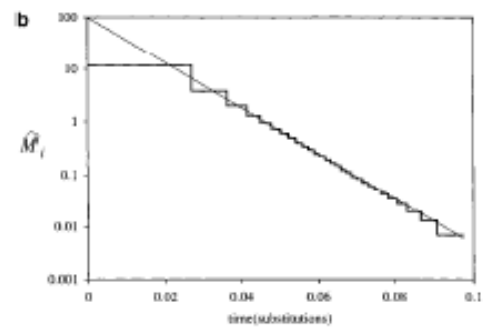
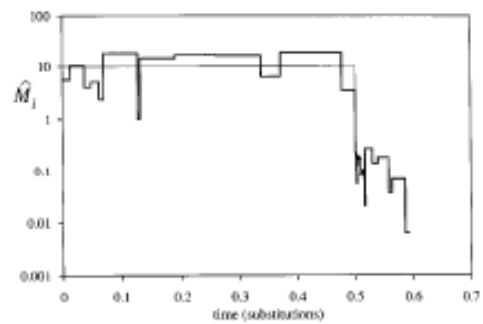
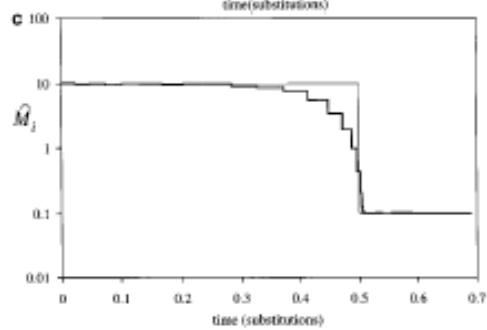
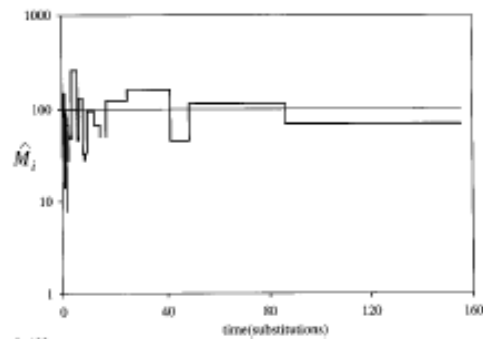
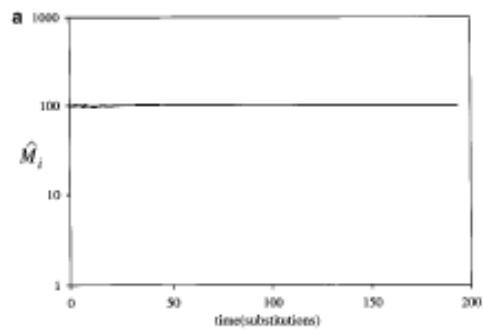
FamilyTreeDNA
Copyright 2006

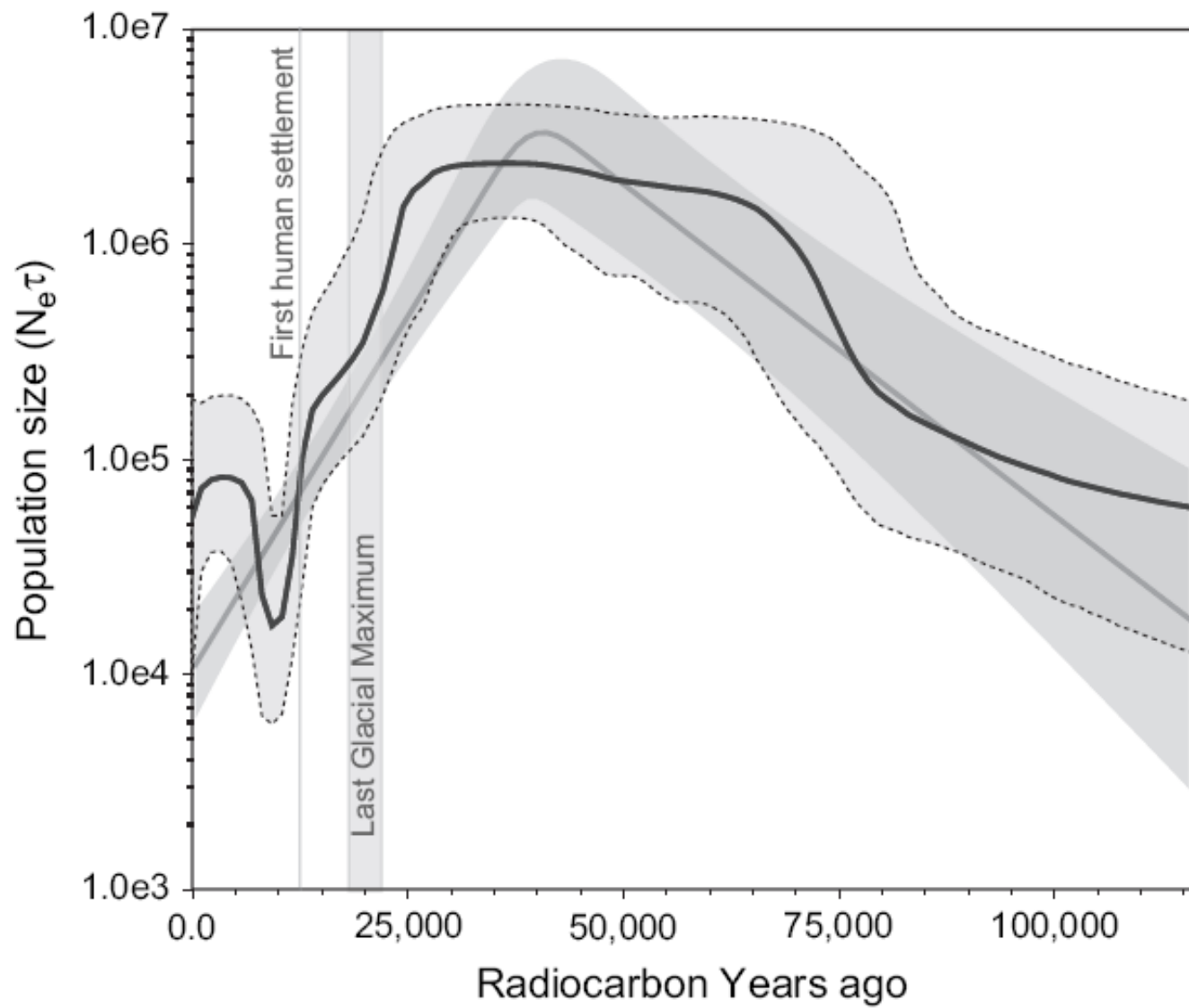
molecular demography

skyline plots









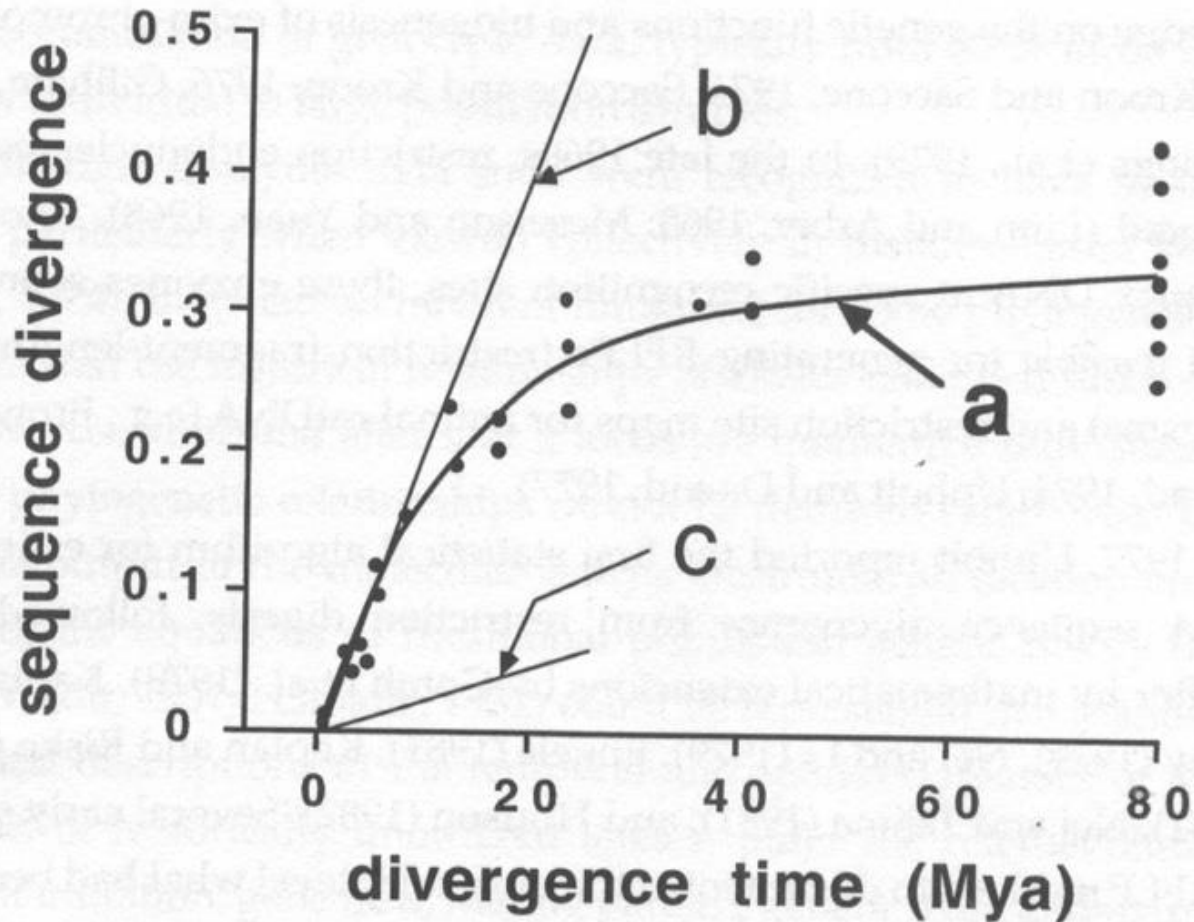
molecular clock

Brown and Wright, 1975 – parthenogenetic lizards

Brown et al., 1979 – primates

calibration: 2% between taxa per 10^6 years

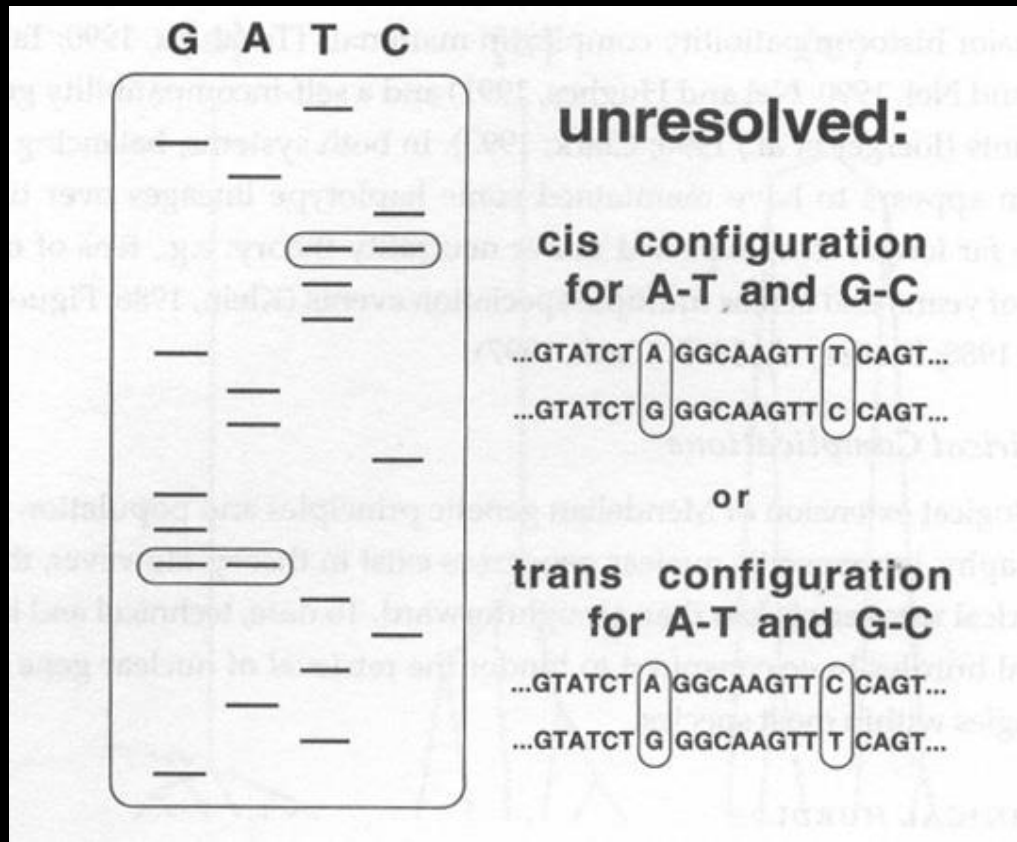
1% per lineage per 10^6 years



nDNA

methodological complications – diploid (problems with haplotype isolation in heterozygotes, gene families – crossamplifications

biological properties – relatively slow evolution, recombinations



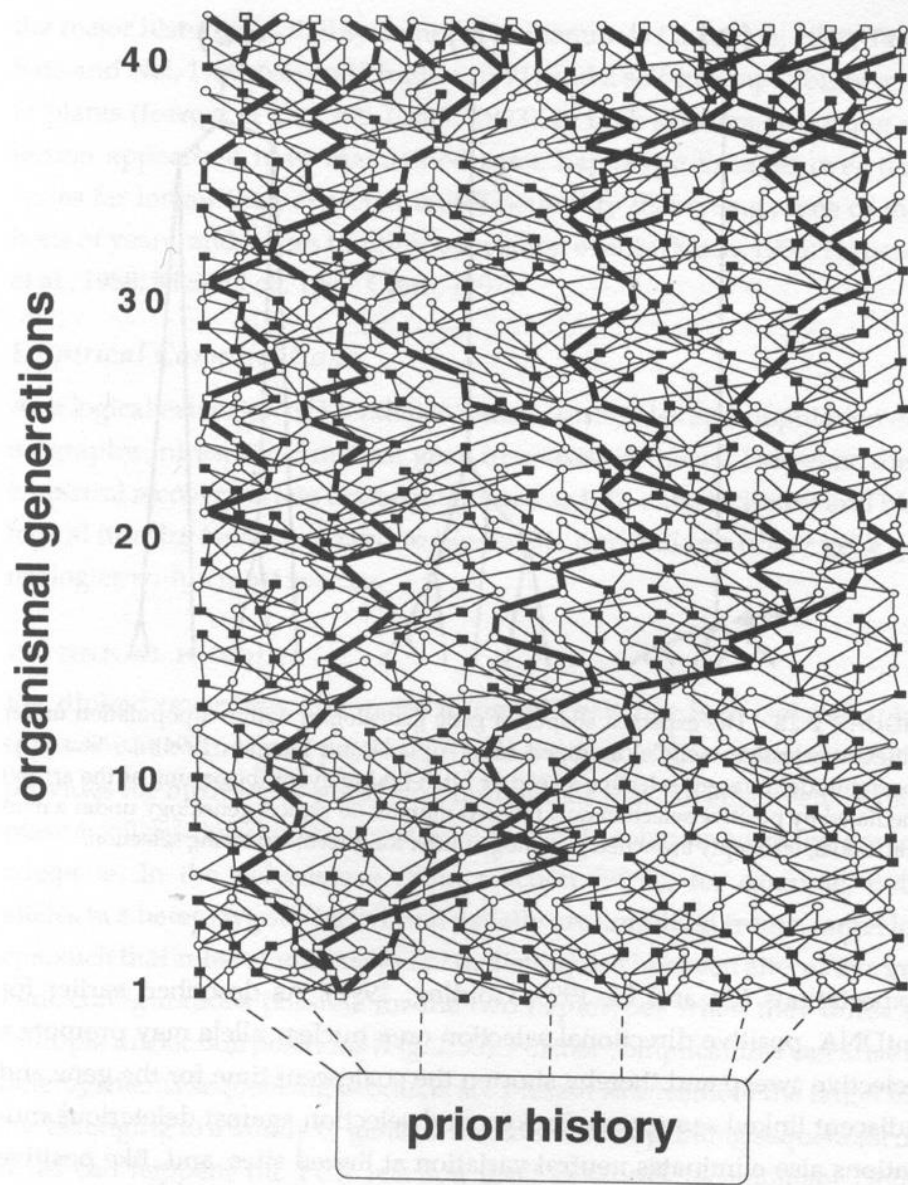
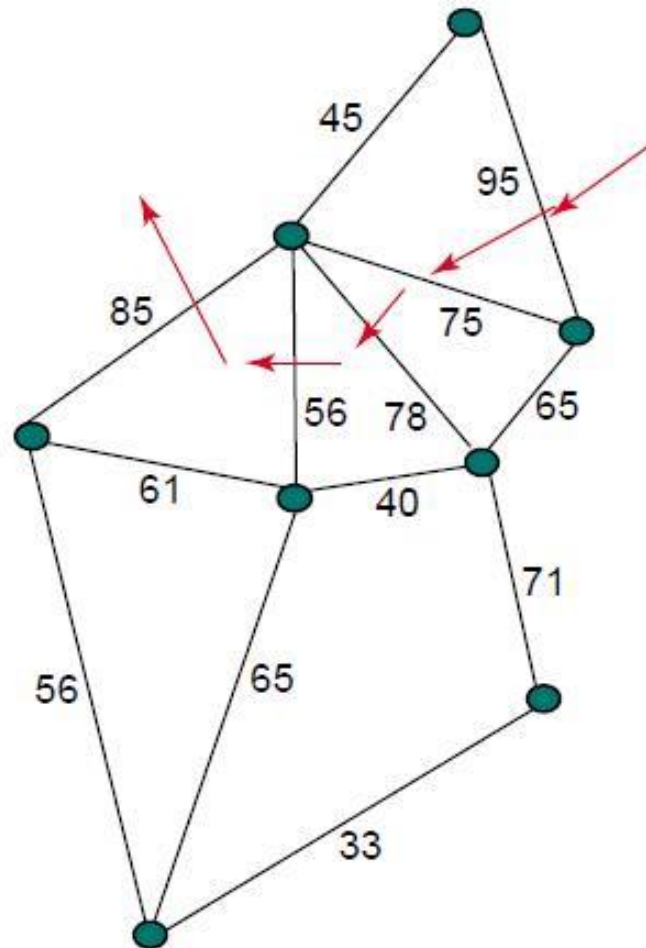
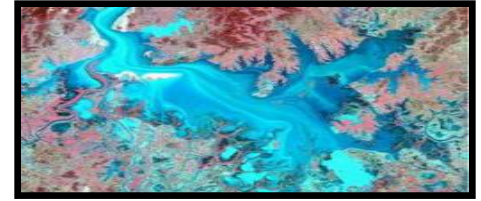


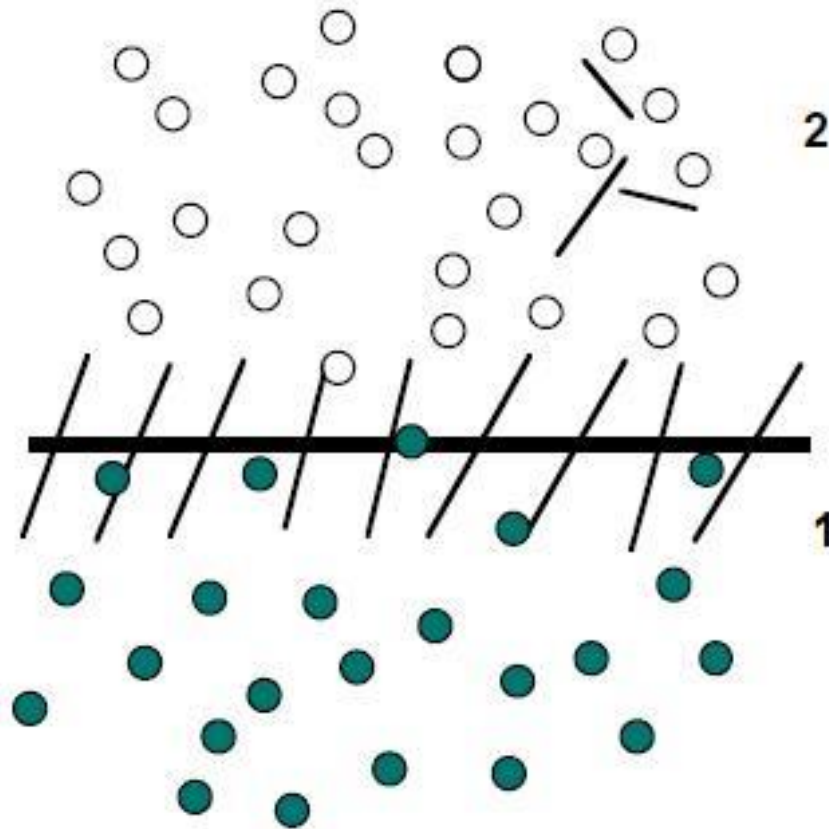
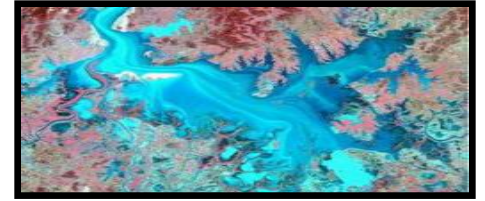
FIGURE 2.28 Coalescent process (heavy lines) for an autosomal gene within an organismal pedigree spanning more than 40 generations. This pedigree is an amalgamation of its matrilineal and patrilineal components as presented separately in Figs. 2.1 and 2.26, with additional pathways connecting each daughter to her father and each son to his mother.

landscape genetics

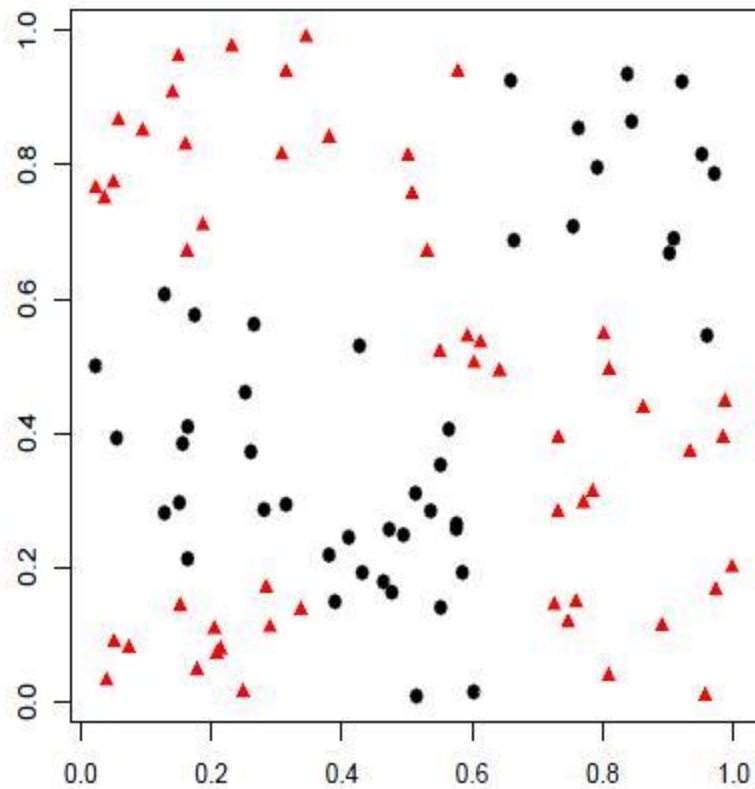
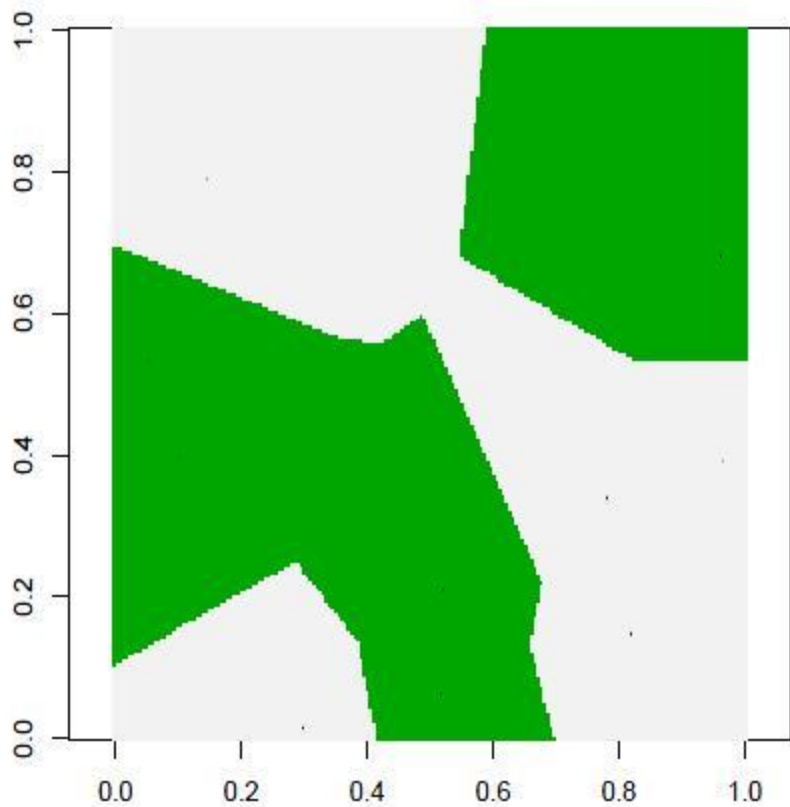
maximum difference Monmonier algorithm



The Womble approach



Population membership



ALLOPATRY AND DIVERGENCE

examples of divergent evolution in isolation - islands

Table 2.1 A simple classification of island types distinguishing: (1) classic types of 'real' island, being land surrounded by open water, from (2) habitat islands, for which the contrast between the 'island' and the surrounding matrix is less stark but still sufficient to represent a barrier or filter to population movements. Australia, given its huge size, is essentially continental in character and in practice is not treated as an island in the present work

Type of island	Examples
Land surrounded by water	
Island continent	Australia
Oceanic islands	Hawaii, Canaries
Continental fragments	Madagascar, New Caledonia
Continental shelf islands	British Isles, Newfoundland
Islands in lakes or rivers	Isle Royale (Lake Superior), Barro Colorado island (Lake Gatún), Gurupá island (River Amazon)
Habitat islands	
Patches of a distinct terrestrial habitat isolated by a hostile matrix	Great-Basin (USA) mountain tops surrounded by desert Woodland fragments surrounded by agricultural land Thistle heads in a field Continental lake (Baikal, Titicaca)
Marine habitat islands	The fringing reef around an isolated oceanic island Coral reefs separated from other reefs by stretches of seawater Seamounts (submerged or not yet emerged mountains below sea level) Guyots (submerged flat-topped former islands, i.e. a type of seamount)

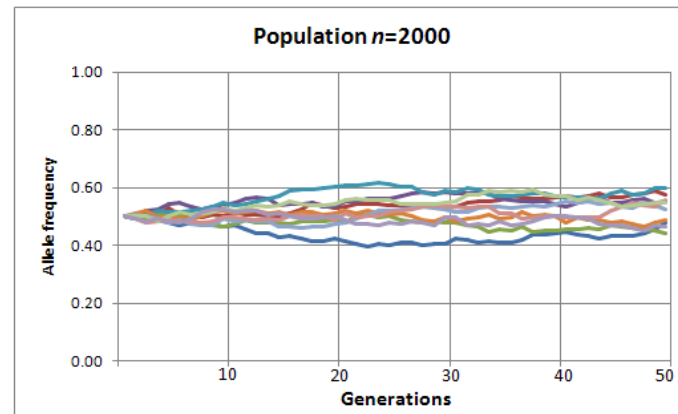
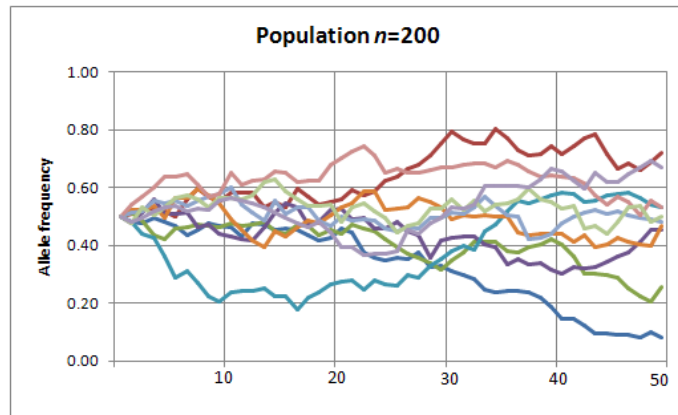
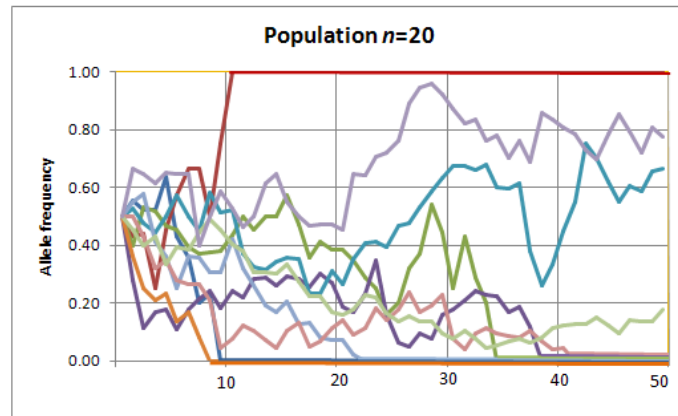
insular evolution – proximate mechanisms

genetics of small isolated populations

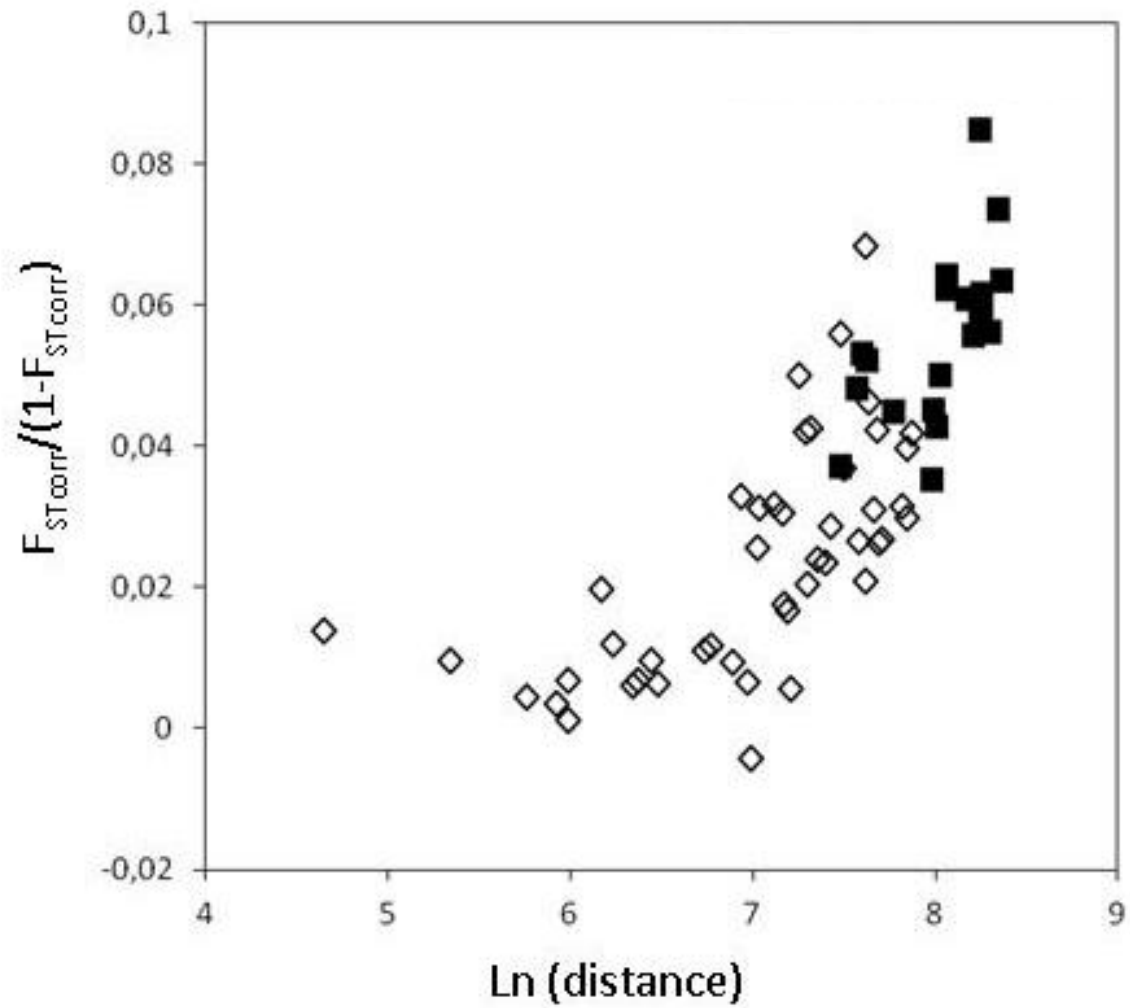


- neutral processes** – often connected with demographic changes (bottlenecks) during island colonization, sudden speciation scenarios
- **founder effect** (Mayr 1954) – partial transfer of ancestral polymorphism in course of colonization event
 - **founder-flush model** (Carson 1968) – founder effect followed by population expansion with reduced selection
 - **transilience model** (Templeton 1980) – rapid shift to different peak in adaptive landscape during short period of inbreeding following colonization event, without substantial reduction of genetic variability

- genetic drift



isolation by distance



ISOLATION BY DISTANCE*

SEWALL WRIGHT

The University of Chicago

Received November 9, 1942

STUDY of statistical differences among local populations is an important line of attack on the evolutionary problem. While such differences can only rarely represent first steps toward speciation in the sense of the splitting of the species, they are important for the evolution of the species as a whole. They provide a possible basis for intergroup selection of genetic systems, a process that provides a more effective mechanism for adaptive advance of the species as a whole than does the mass selection which is all that can occur under panmixia.

RANDOM DIFFERENTIATION UNDER THE ISLAND MODEL

Mathematical consideration requires the use of simple models of population structure. The simplest model is that in which the total population is assumed to be divided into subgroups, each breeding at random within itself, except for a certain proportion of migrants drawn at random from the whole. Since this situation is likely to be approximated in a group of islands, we shall refer to it as the island model.

The gene frequency (q) of a subgroup tends to vary about a certain equilibrium point (\bar{q}) in a distribution curve ($\phi(q)$) determined by the net systematic pressure (measured by Δq , the net rate of change of gene frequency per generation from recurrent mutation, immigration, and selection) in conjunction with the cumulative effects of accidents of sampling (random deviation δq , variance per generation $\sigma_{\delta q}^2$) (WRIGHT 1929, 1931, 1942).

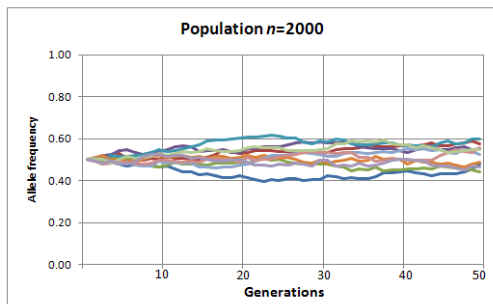
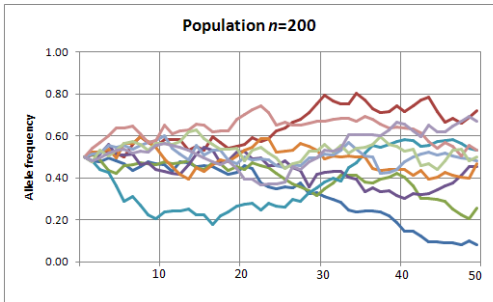
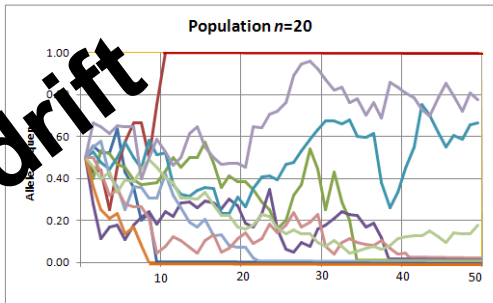
$$(1) \quad \phi(q) = (C/\sigma_{\delta q}^2) \exp \left[2 \int (\Delta q/\sigma_{\delta q}^2) dq \right].$$

Let N be the effective size of the subgroup, m the effective proportion of its population replaced in each generation by migrants, and q_1 the gene frequency in the total population. The rate of change of gene frequency per generation in a subgroup, taking account only of immigration pressure, is $\Delta q = -m(q - q_1)$. In a random breeding population $\sigma_{\delta q}^2 = q(1 - q)/2N$. Substitution in (1) gives the following, choosing C so that $\int_0^1 \phi(q) dq = 1$ (WRIGHT 1931, 1942).

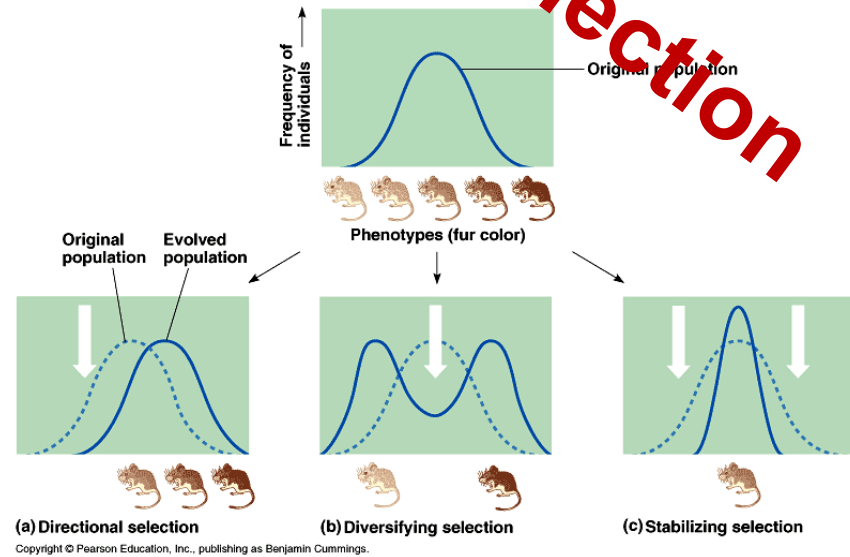
$$(2) \quad \phi(q) = \frac{\Gamma(4Nm)}{\Gamma(4Nm q_1) \Gamma[4Nm(1 - q_1)]} q^{4Nm q_1 - 1} (1 - q)^{4Nm(1 - q_1) - 1}$$

$$(3) \quad \bar{q} = \int_0^1 q \phi(q) dq = q_1$$

drift

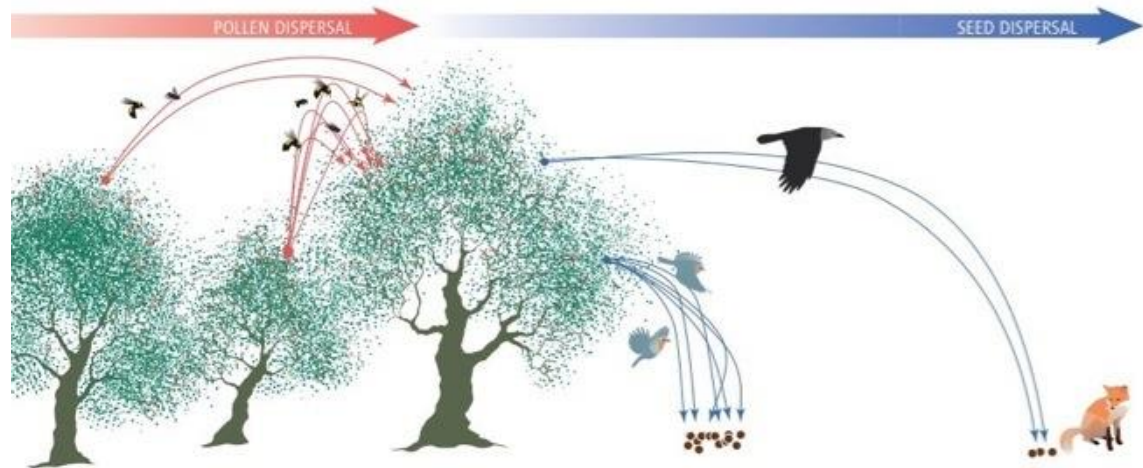


selection



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flow



Eco-evolutionary problems connected with islands:



natural selection, adaptive radiation - Charles Darwin, A.R. Wallace
insular model of population structure – Sewall Wright
allopatric speciation, founder effect – Ernst Mayr
small population paradigm in conservation biology – Graeme Caughley
metapopulation – Ilka Hanski, Michael Gilpin
taxon cycle – Edward O. Wilson
community assembly rules – Jared Diamond
Island biogeography theory, r - K strategy – Robert MacArthur, E.O. Wilson

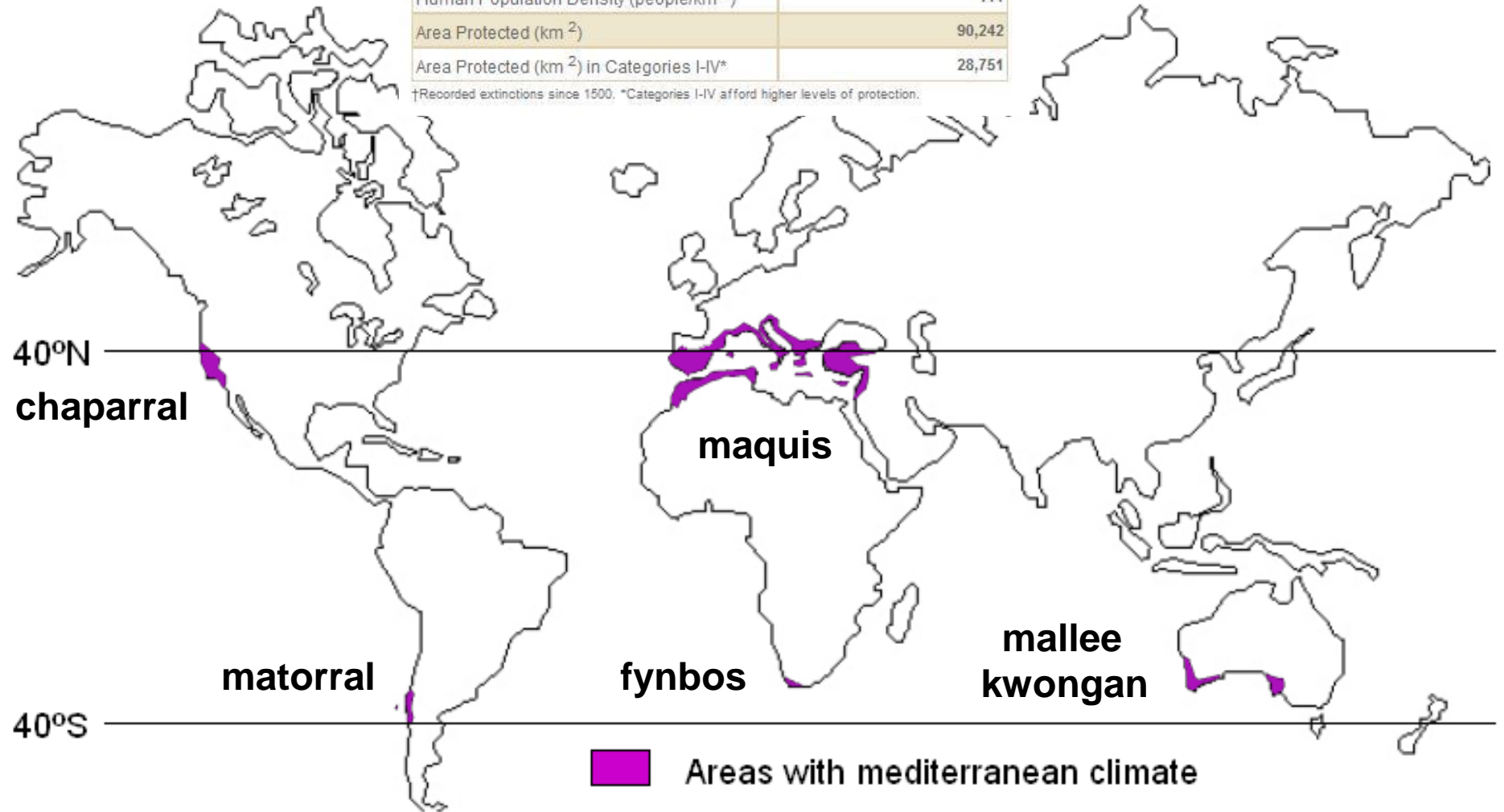
Mediterranean



VITAL SIGNS

Hotspot Original Extent (km ²)	2,085,292
Hotspot Vegetation Remaining (km ²)	98,009
Endemic Plant Species	11,700
Endemic Threatened Birds	9
Endemic Threatened Mammals	11
Endemic Threatened Amphibians	14
Extinct Species†	5
Human Population Density (people/km ²)	111
Area Protected (km ²)	90,242
Area Protected (km ²) in Categories I-IV*	28,751

†Recorded extinctions since 1500. *Categories I-IV afford higher levels of protection.



— BIODIVERSITY —
HOTSPOTS

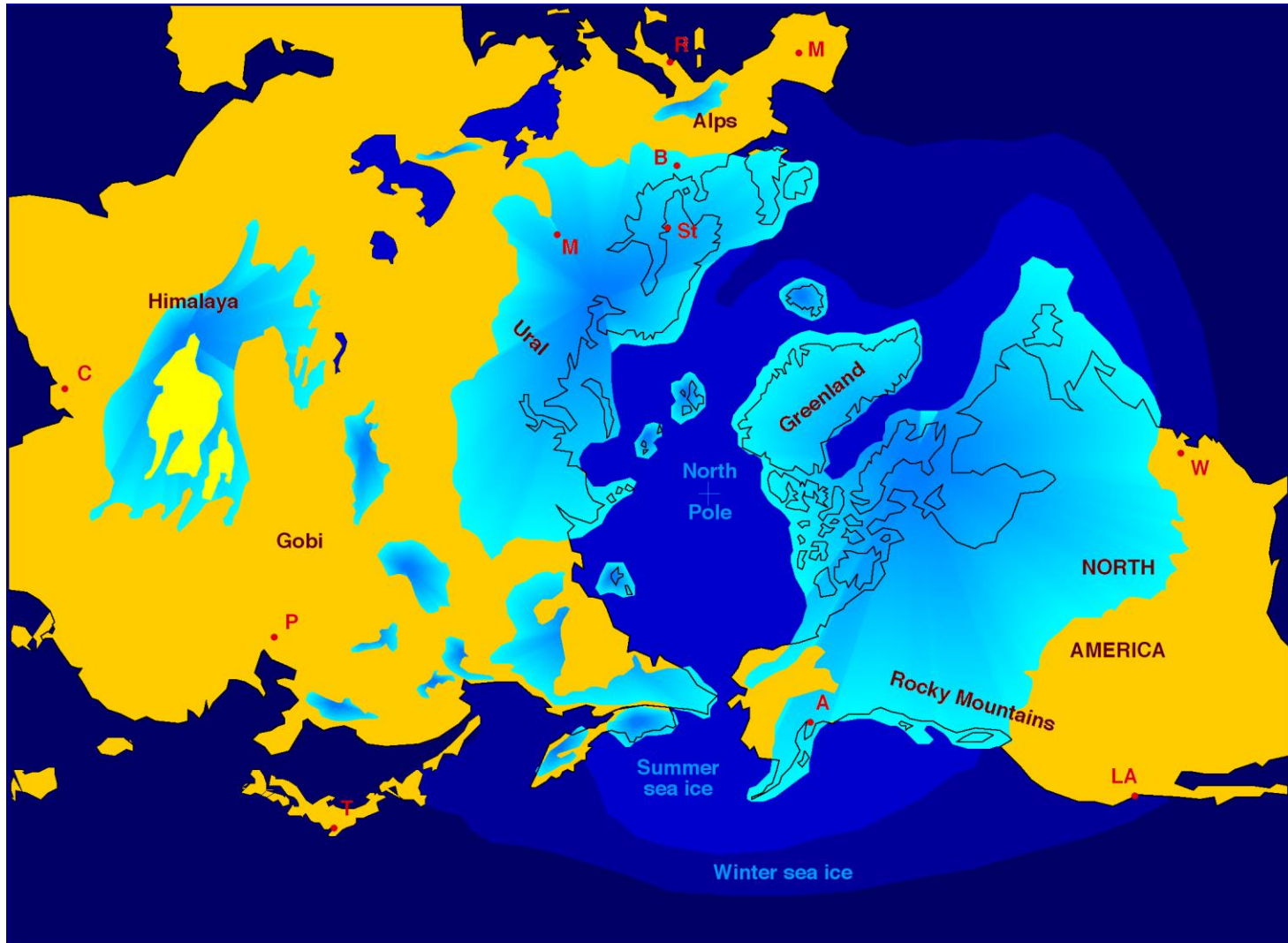
(Use the menu to see all the Hotspots by name.)

DIVERSITY AND ENDEMISM

Taxonomic Group	Species	Endemic Species	Percent Endemism
Plants	22,500	11,700	52.0
Mammals	226	25	11.1
Birds	489	25	5.1
Reptiles	230	77	33.5
Amphibians	79	27	34.2
Freshwater Fishes	216	63	29.2

Ice ages

since Pliocene epoch, 2.56 Mya





periglacial steppe

Role of glaciations in distribution of genealogical lineages and speciation

Traditional paradigm of European phylogeography

- Glaciations pushed temperate components of interglacial communities into the south
- Owing to geomorphology of southern Europe, they survived in Mediterranean refuges (with **divergent evolution** and **possible allopatric speciation**)
- Recent (interglacial) northern ranges are outcome of postglacial recolonization from mediterranean refugia
- In case of secondary contacts of allopatrically evolved lineages (suture zones) – situation reflecting degree of evolution of reproductive isolating mechanisms (**reinforcement, hybrid zones, introgression**)
- cf. distribution of European sister taxa

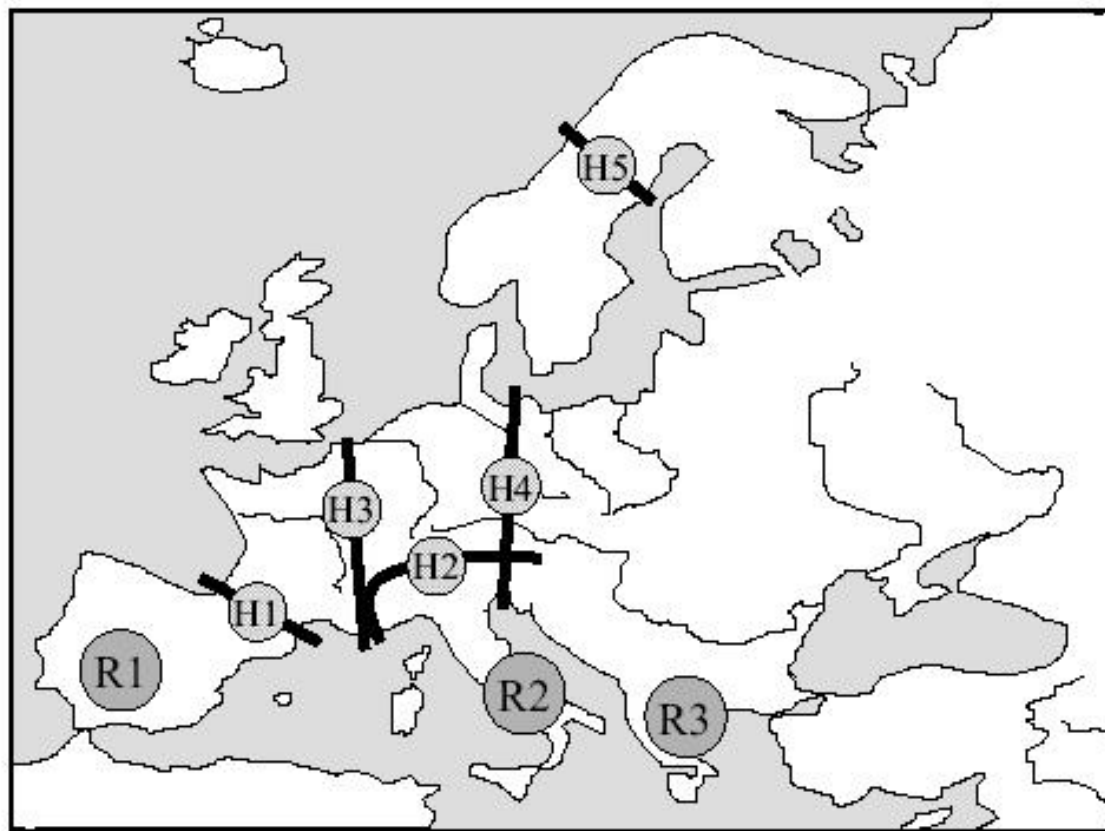


Figure 1

The three major Mediterranean refugial and differentiation centres of Southern Europe during the last ice-age (R1: Atlantic-Mediterranean, R2: Adriatic-Mediterranean, R3: Pontic-Mediterranean) and the geographic location of the five most important contact and hybridisation areas where different biota got into secondary contact during the postglacial range expansion processes (H1: Pyrenees, H2: Alps, H3: western Central Europe, H4: eastern Central Europe, H5: Central Scandinavia. (Based on Taberlet et al. [9] and Hewitt [5]).

Grasshopper
(*Chorthippus parallelus*)



Hedgehogs
(*Erinaceus spp.*)



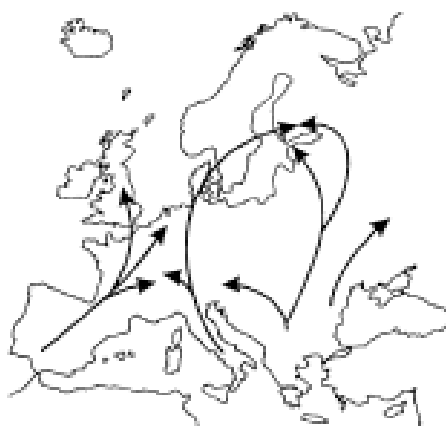
Bear
(*Ursus arctos*)



Alder
(*Alnus glutinosa*)



Oaks
(*Quercus spp.*)



Shrew
(*Sorex araneus*)



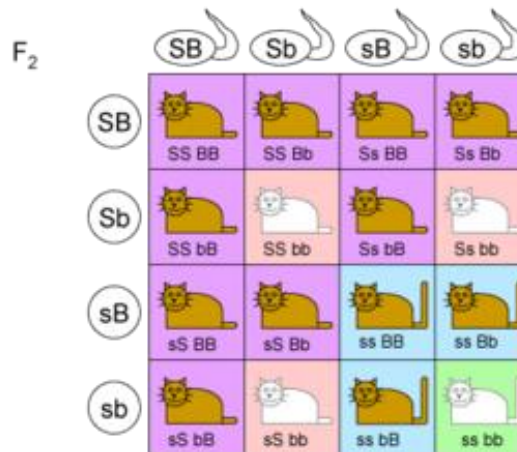
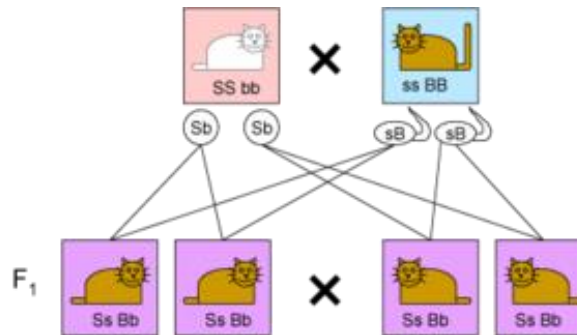
Fig. 2 Three paradigm European postglacial colonization patterns, the grasshopper, the hedgehog and the bear, and three similar species patterns, the alder, the oak and the shrew (see Hewitt 1999 for detailed discussion and references).

SYMPATRY AND HYBRIDIZATION

hybrid – descendant of mating between heterospecifics

integrate – descendant of mating between conspecifics

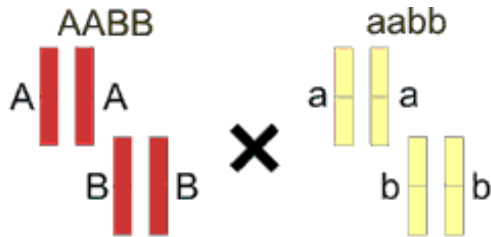
- Definition depends upon species concept used. Recently usually general term for descendant of mating between different lineages



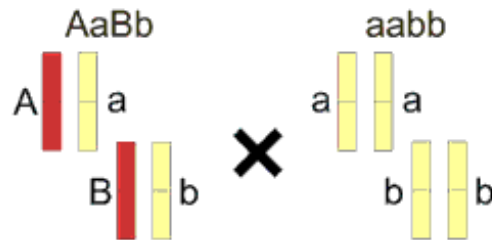
introgression

backcrossing

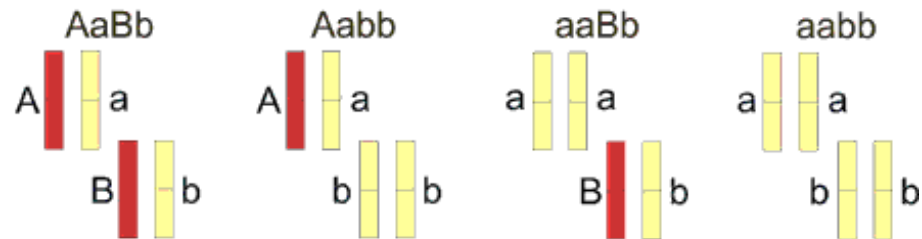
parental



F₁ x P



backcross



phenotype

AB

Ab

aB

ab

odds

1

1

1

1

frequency

25%

25%

25%

25%

introgression – exchange of genes between two species mediated via hybridization and backcrossing

two possible directions

In later generations of unidirectional backcrossing the proportion of latter genome decreases (mtDNA capture, ghost of hybrid past)

different intensity in different loci – islands of genomic differentiation

hybrid fitness

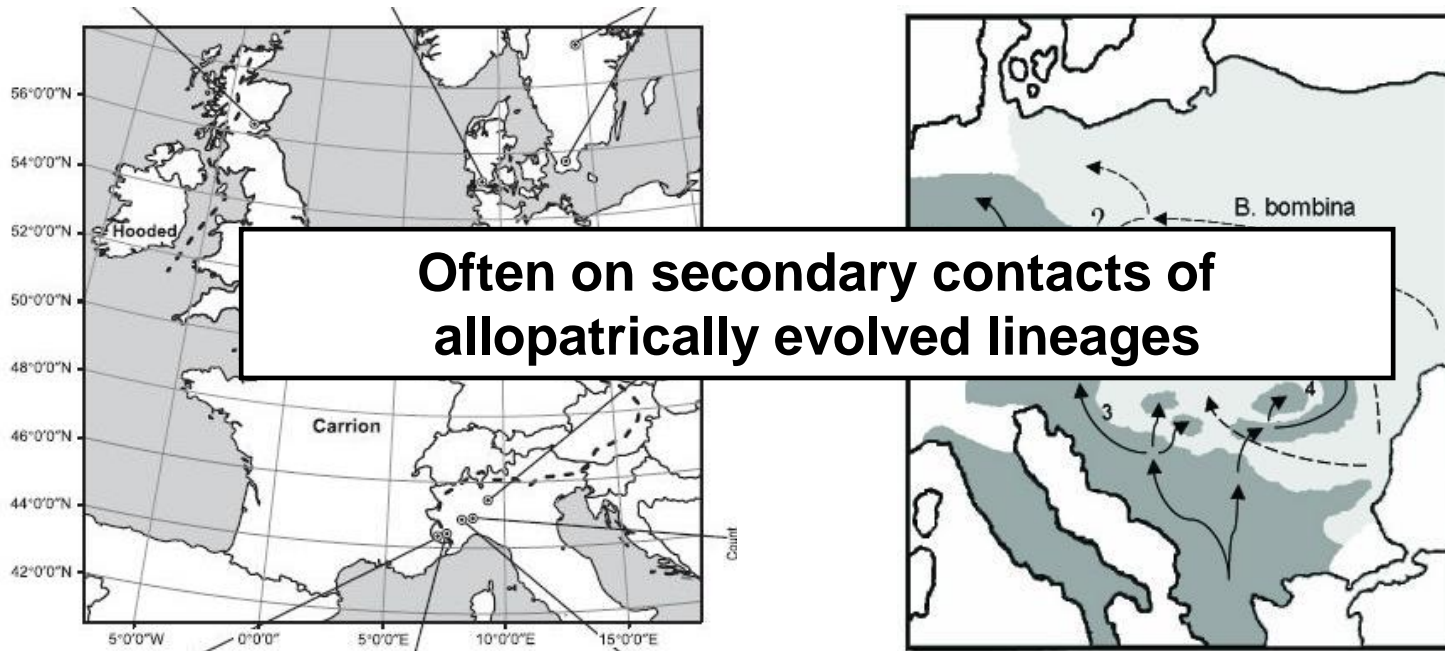
hybrid inferiority – incompatibilities of chromosomes, disruption of epistatic equilibria etc.

hybridní superiority – novel gene combinations, exchange of adaptive loci between taxa, hybrid speciation

Example: *Salvelinus* – sterile F1 hybrids, *Balaenoptera* – fertile F1 hybrids

hybrid zones

Region (linear structure) of contact of genetically unique lineages (species) with production of hybrids. Various composition (A, B, F1, F2, F1A, F1B etc.). Various shape, dynamic in time.

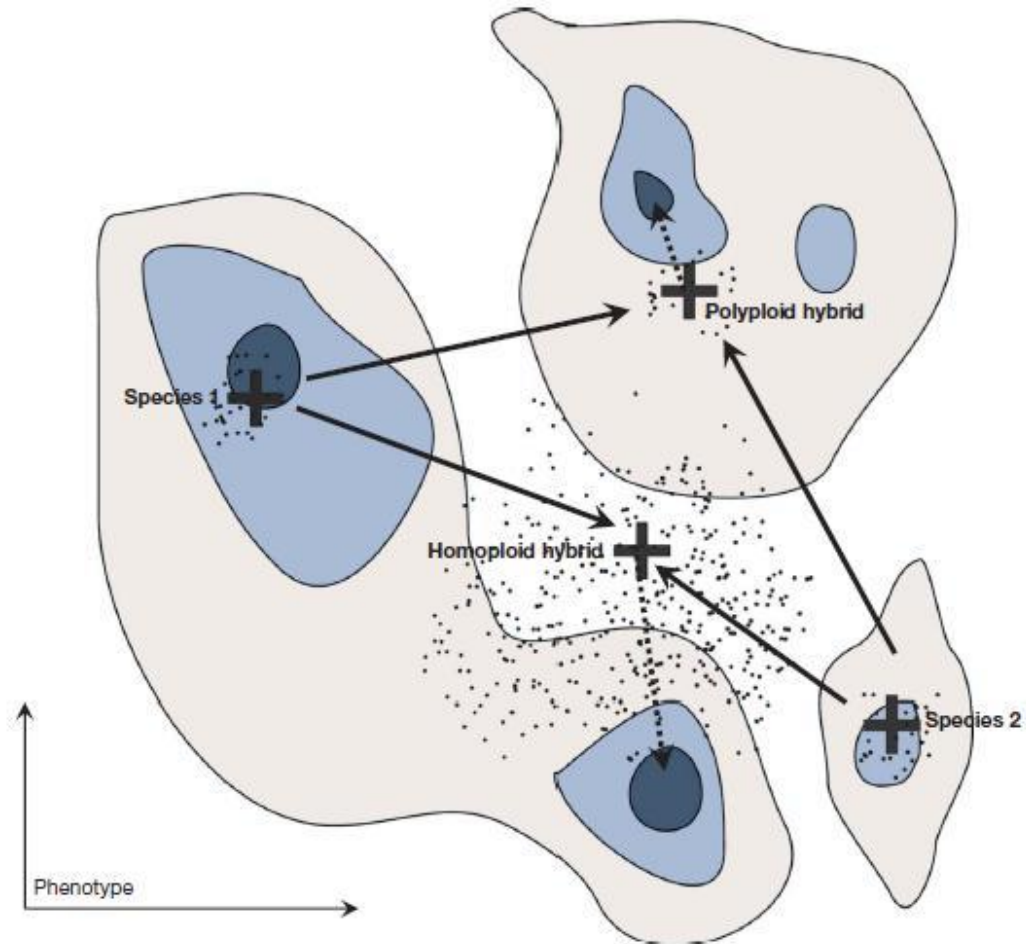


linear

mosaic

hybrid speciation

homoploid x allopolyploid



EXAMPLES

Erinaceus europaeus

E. concolor s.l. (recently *E. roumanicus* + *E. concolor*)



Santucci et al., 1998

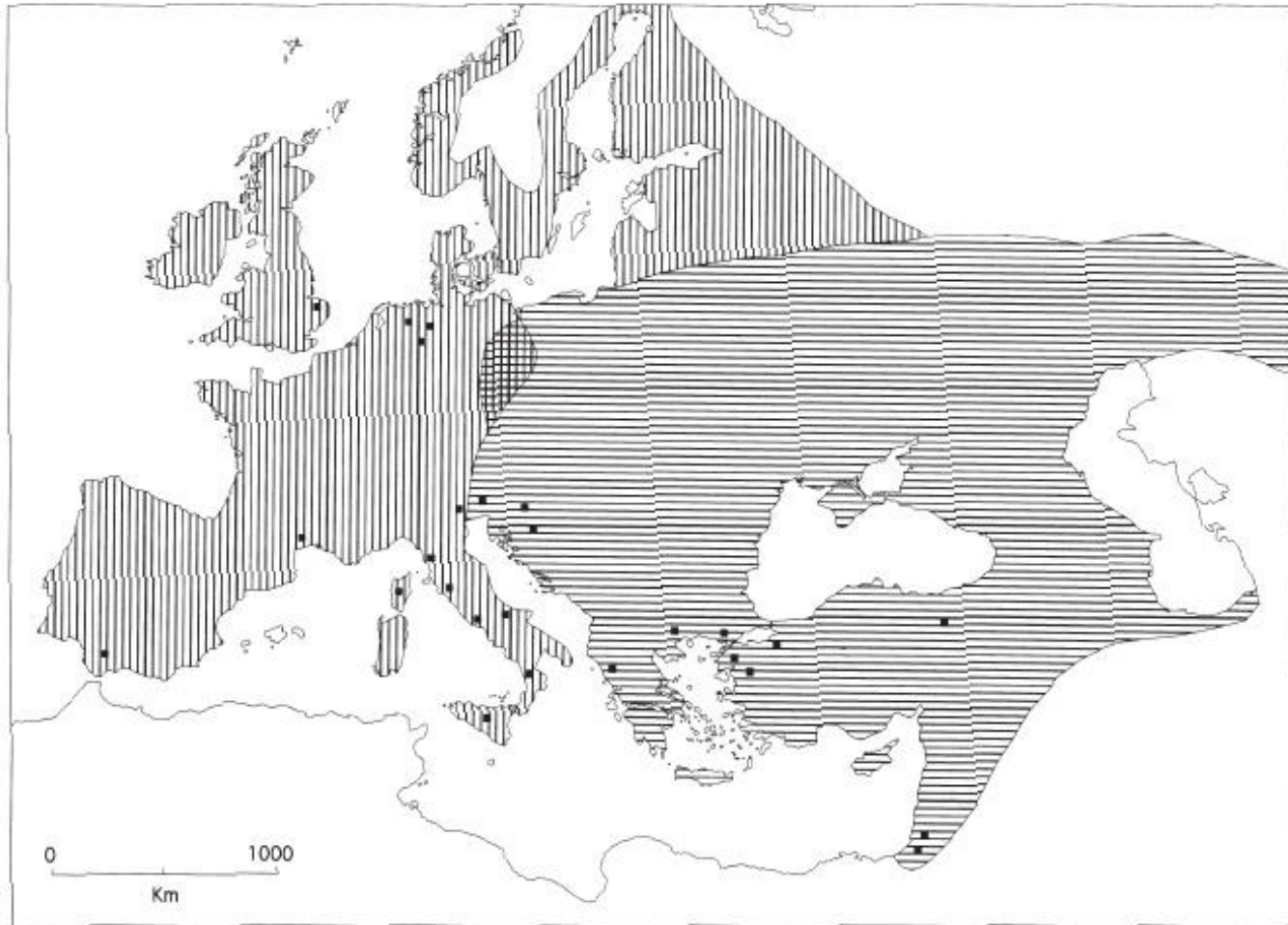


Fig. 1 Map of the geographical distribution of *Erinaceus europaeus* (vertical lines) and *E. concolor* (horizontal lines) showing the locations studied.

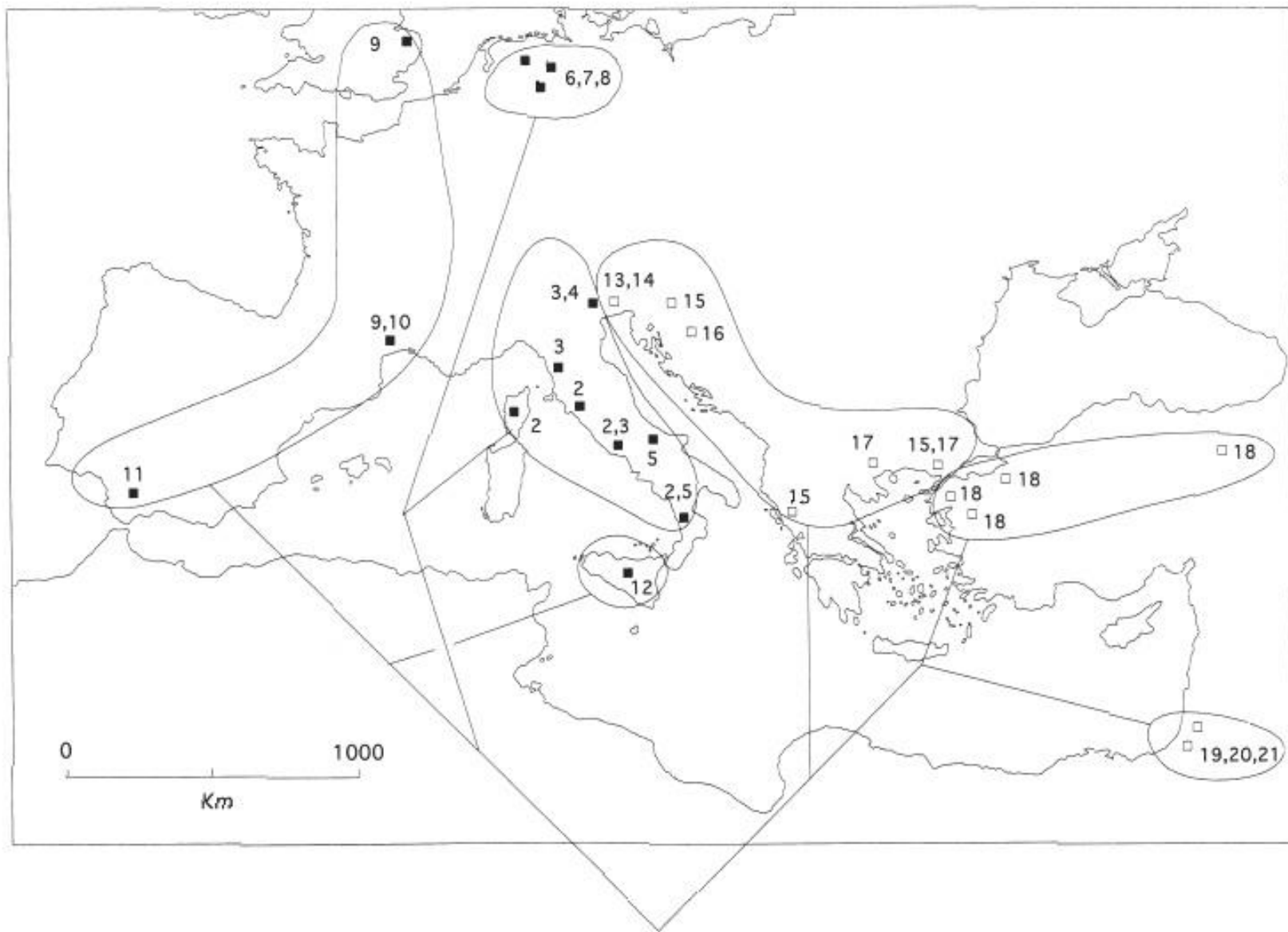
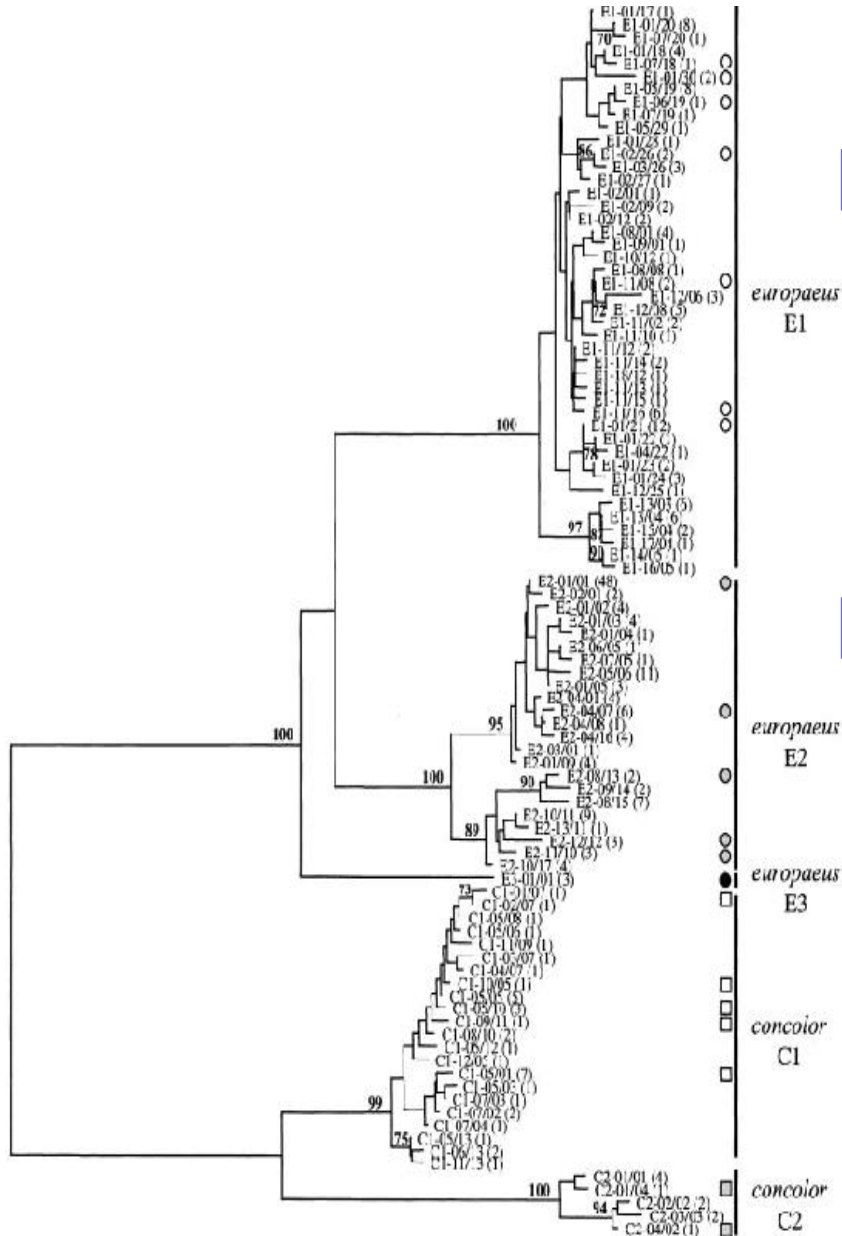


Fig. 6 Phylogeography of the different haplotypes emphasizing the east/ west division of *Erinaceus europaeus* and *E. concolor* and the more recent dichotomies in each of them.

Seddon et al., 2001



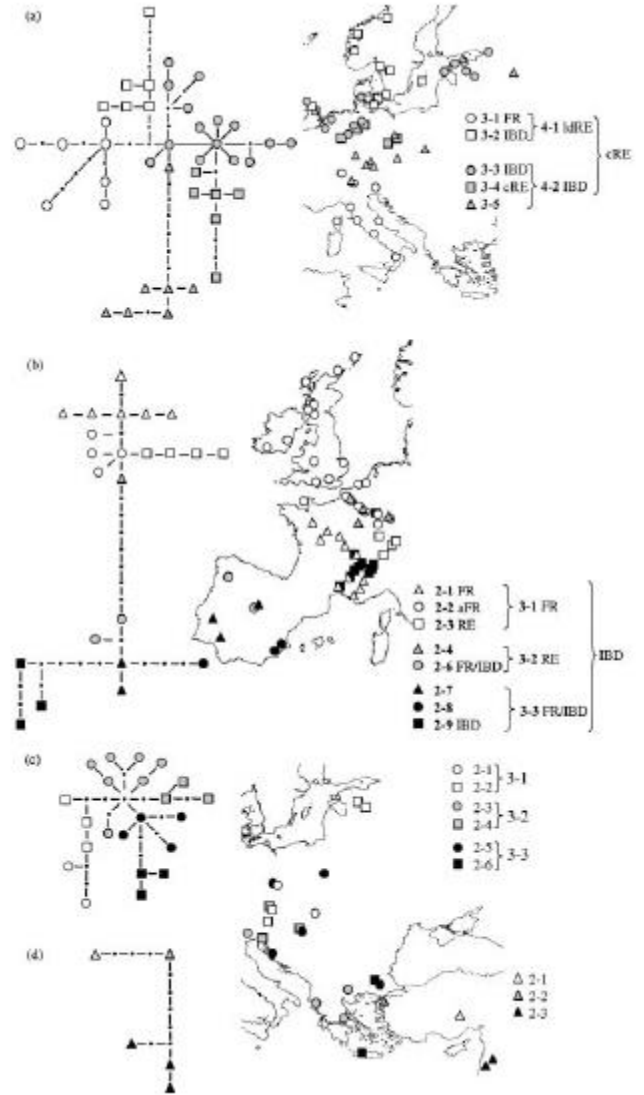
E1

E2

E3

C1

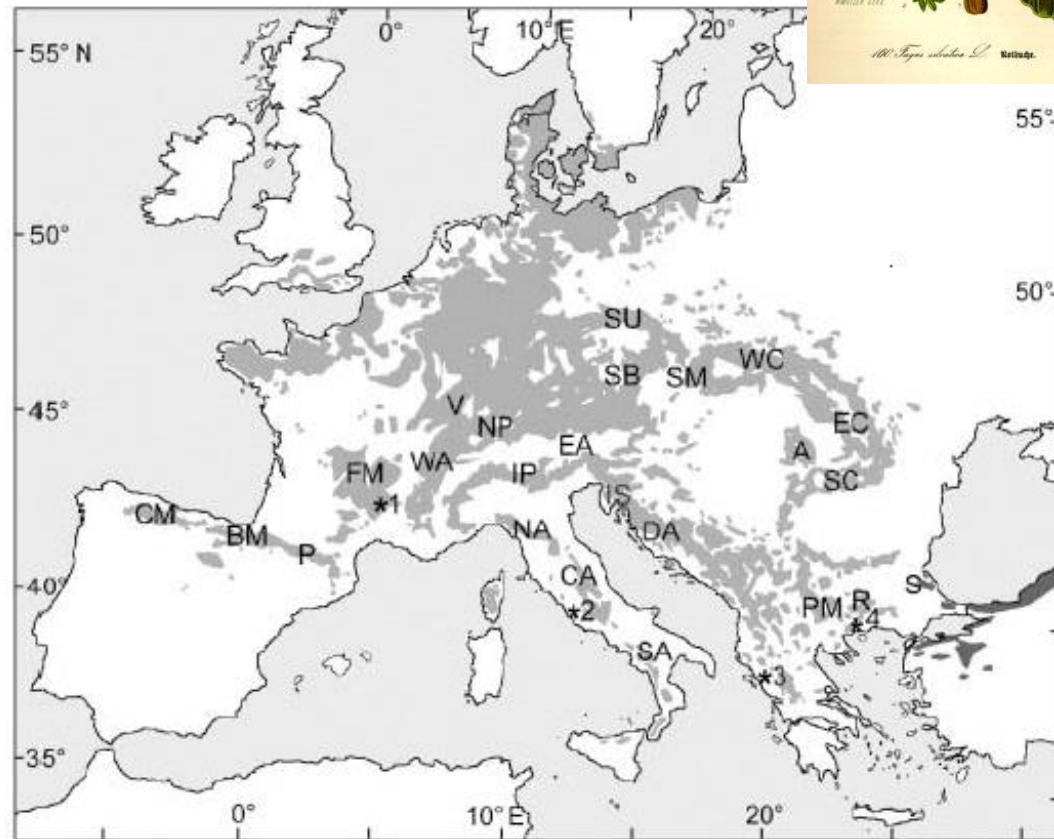
C2



Fagus sylvatica (Magri et al., 2006)



Fig. 1 Modern distribution of *Fagus sylvatica* (light grey; modified from Pott, 1997) and *Fagus orientalis* (dark grey; redrawn from Atalay, 1994) in Europe. Mountain ranges mentioned in the text: Cantabrian Mountains (CM), Basque Mountains (BM), Pyrenees (P), French Massif Central (FM), western Alps (WA), Vosges (V), northern Prealps (NP), Italian Prealps (IP), eastern Alps (EA), southern Bohemia (SB), southern Moravia (SM), Sudety (SU), western Carpathians (WC), eastern Carpathians (EC), Apuseni (A), southern Carpathians (SC), Dinaric Alps (DA), Pirin Mountains (PM), Rhodopes (R), Strandza Mountains (S), southern Apennines (SA), central Apennines (CA), and northern Apennines (NA). Asterisks indicate the location of the four longest European pollen records: (1) Bouchets/Praclaux, (2) Valle di Castiglione, (3) Ioannina and (4) Tenaghi Philippon.



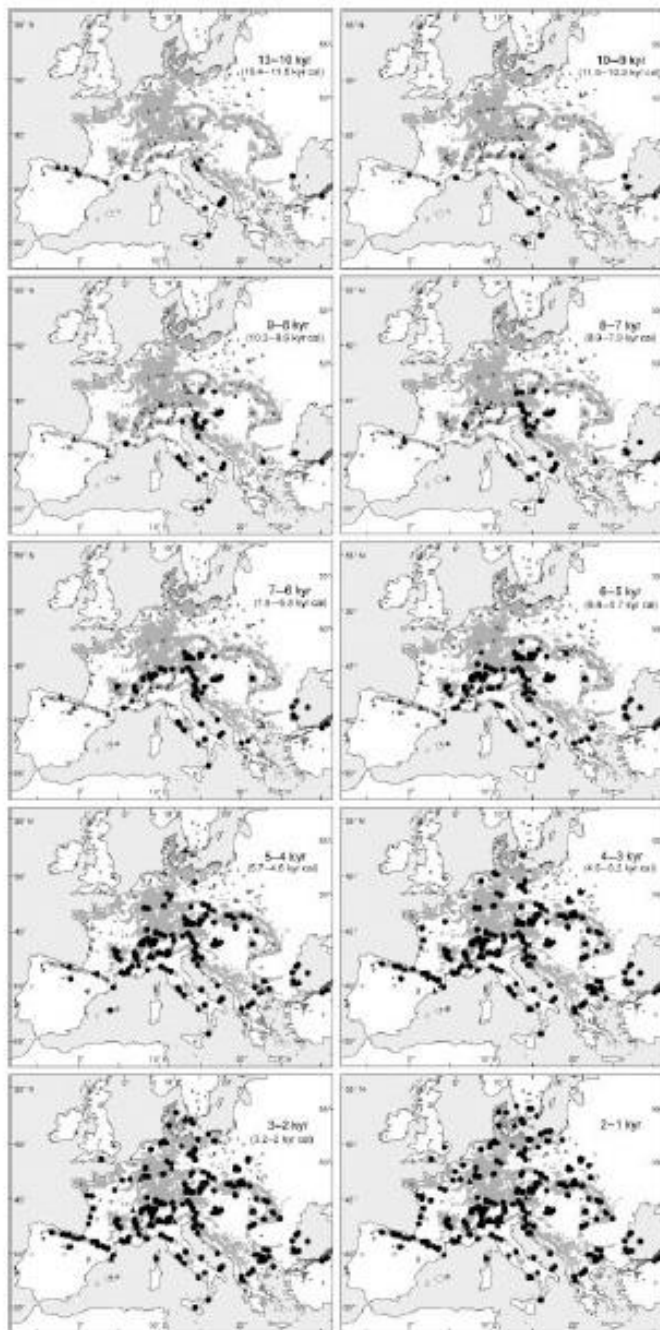


Fig. 4 Geographical distribution of the late glacial and postglacial records of beech in Europe. ○, *Fagus* pollen < 2% or absent; ●, *Fagus* pollen > 2%; ▲, *Fagus* macrofossil. The grey area corresponds to the modern beech distribution (light grey, *Fagus sylvatica*; dark grey, *Fagus orientalis*).

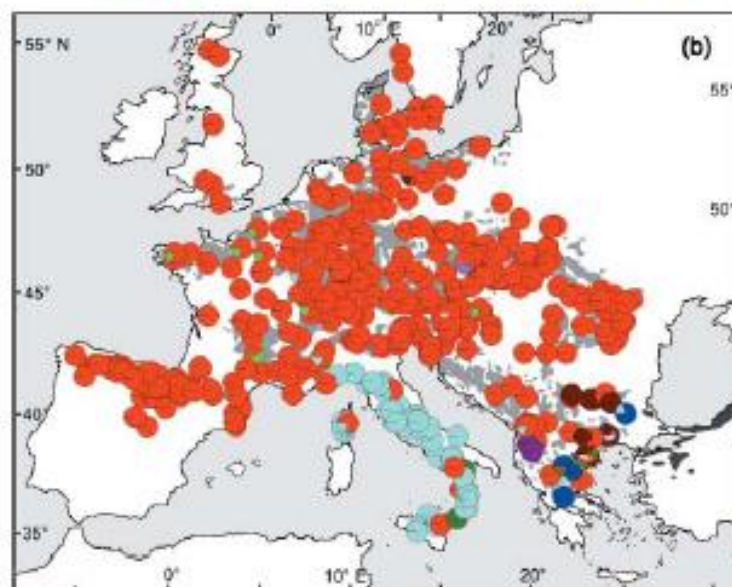
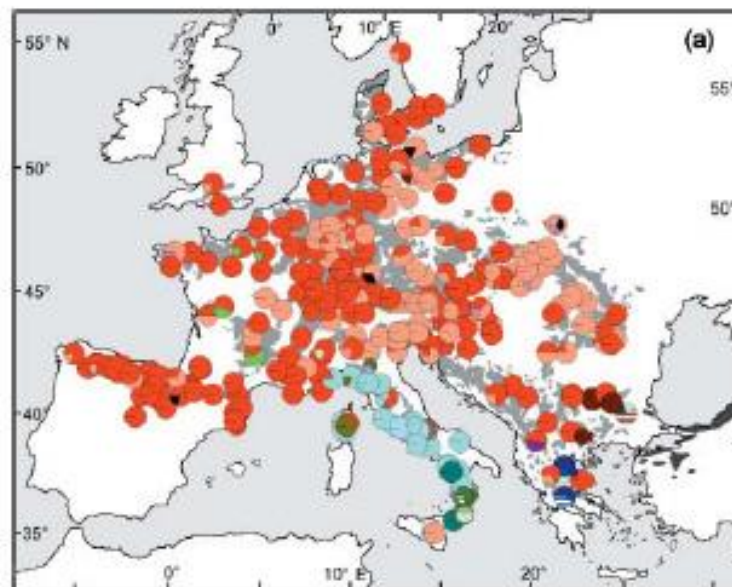


Fig. 5 Geographical distribution of (a) chloroplast haplotypes detected using polymerase chain reaction–restriction fragment length polymorphism (PCR-RFLP) and (b) microsatellites (data for the Italian Peninsula were taken from Vettori *et al.*, 2004).

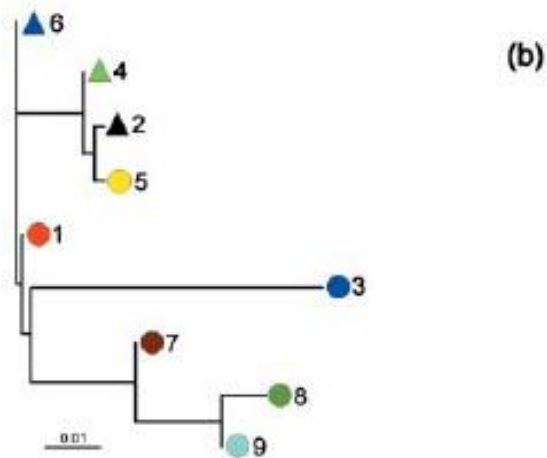
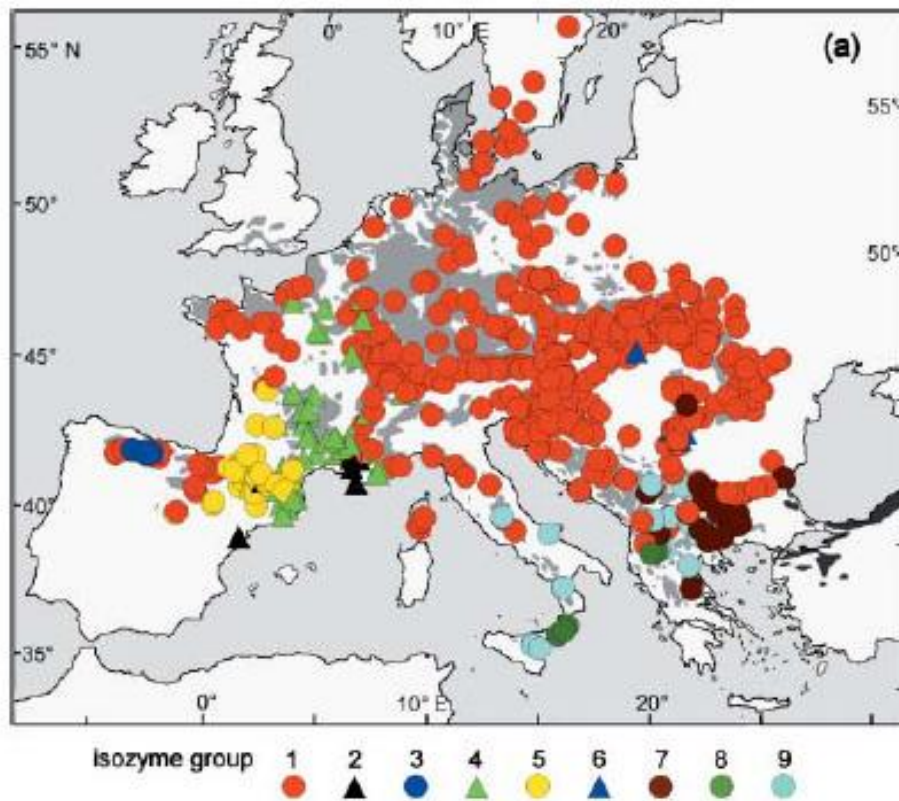


Fig. 6 Spatial analysis of variance (SAMOVA) on isozyme data. (a) geographical distribution of groups; (b) phylogenetic tree obtained using the neighbour-joining approach on the F_{CT} distance matrix for SAMOVA isozyme groups.

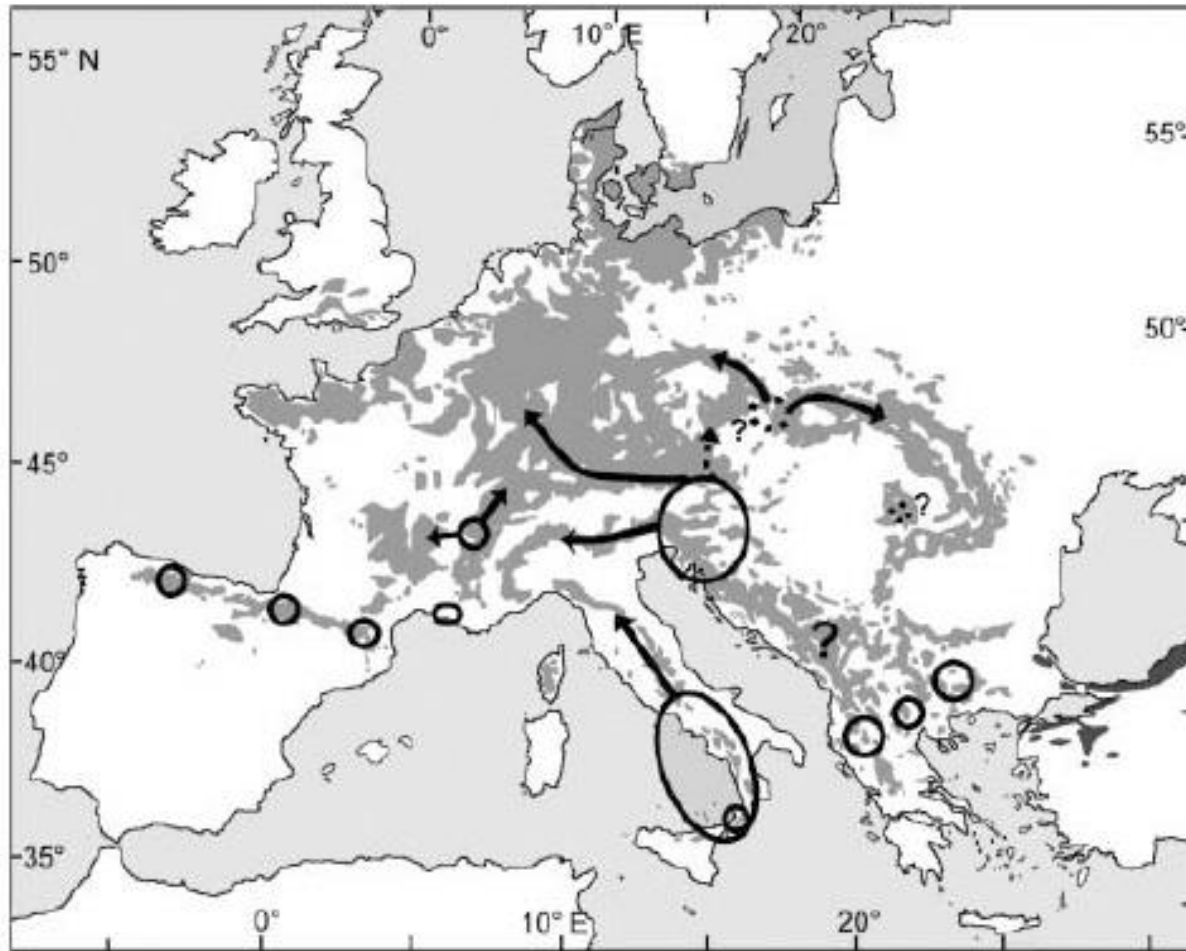


Fig. 9 Tentative location of refuge areas for *Fagus sylvatica* during the last glacial maximum and main colonization routes during the postglacial period.

microrefugia x cryptic refugia

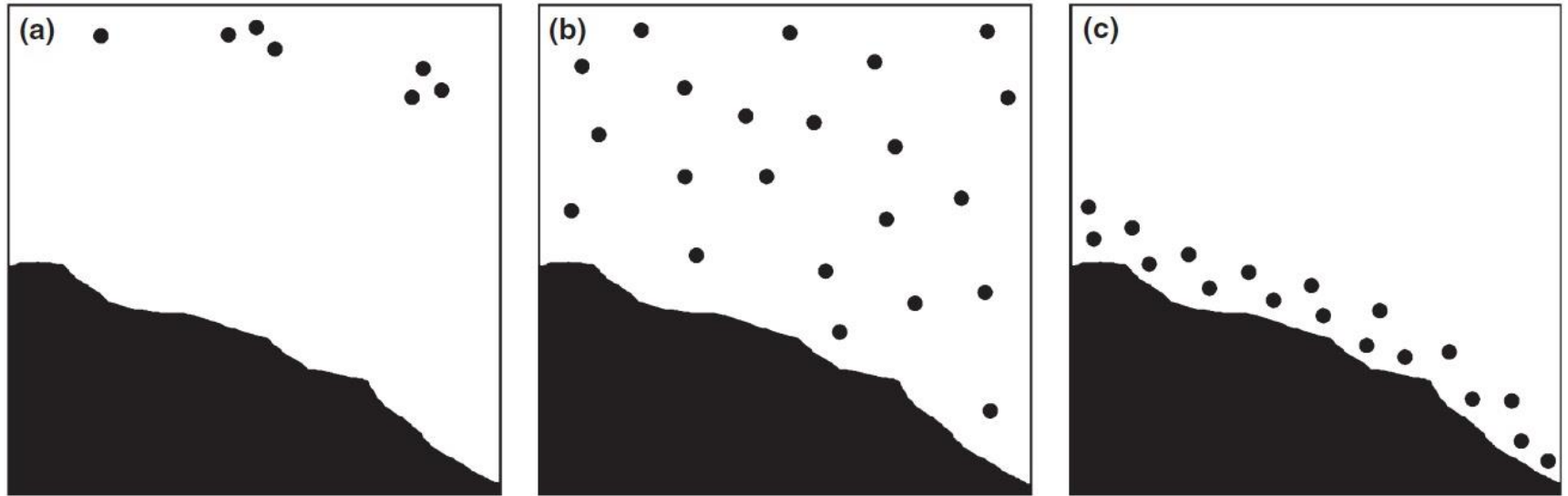
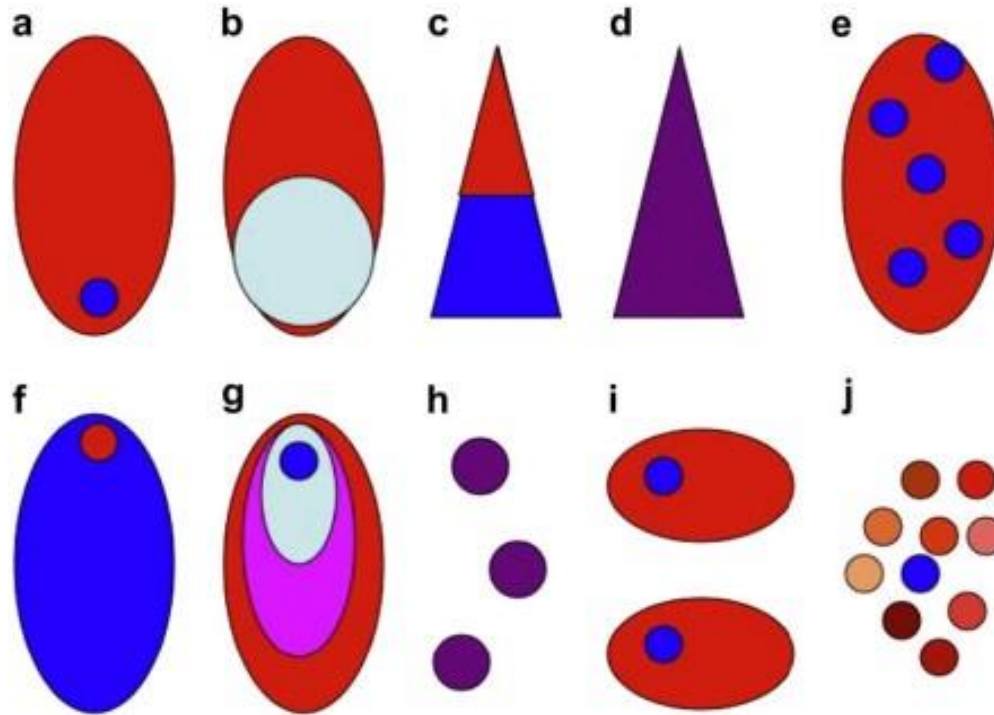


Figure 1 Three types of microrefugia, according to the relative position with respect to the macrorefugium: (a) distal or remote, (b) widespread or diffuse, and (c) proximal or ecotonal.

Types of refugia



Bennet and Provan 2008

Diagrammatic representation of selected species responses to glacial–interglacial climatic changes, in terms of distribution and abundance. Distributions (except where otherwise indicated): blue, glacial; red, interglacial. (a) Classical refugia. (b) Cryptic refugia. Distributions: pale blue, glacial (low abundance, not necessarily continuous). (c, d) Altitudinal shift. Purple: continuous. (e) Tropical refugia. Early-Holocene refugia have a similar pattern, but within the Holocene. (f) Interglacial refugia. (g) Stepwise expansions through successive glacial–interglacial oscillations. Four stages indicated: blue, to light blue, to pink, to red. (h) Stasis, with separated populations. Purple: continuous. (i) Separated into two disjunct populations by ancient vicariance, each of which expands after the LGM. (j) Speciation from one LGM species (blue) into multiple Holocene species (other colours).

Dryas octopetala

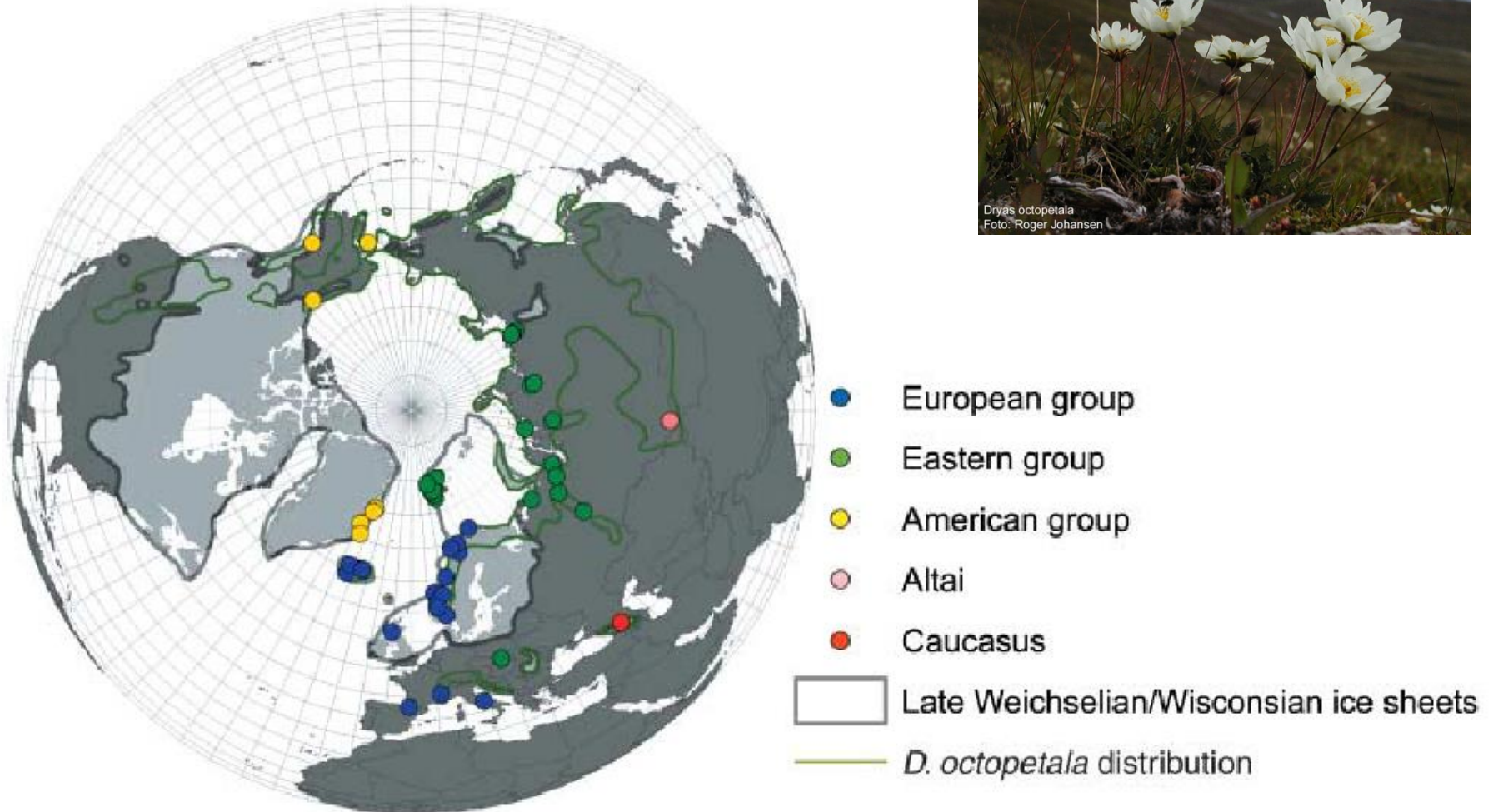


Fig. 1 Geographic origin of the 52 *Dryas* populations analysed and their grouping according to the Bayesian clustering analyses (STRUCTURE) based on 155 amplified fragment length polymorphism (AFLP) markers. The geographical distribution of *Dryas octopetala* is redrawn from Hultén & Fries (1986), and the maximum limits of the late Weichselian/Wisconsinian ice sheets are redrawn from Abbott & Brochmann (2003) and Brochmann *et al.* (2004).

Skrede et al., 2006

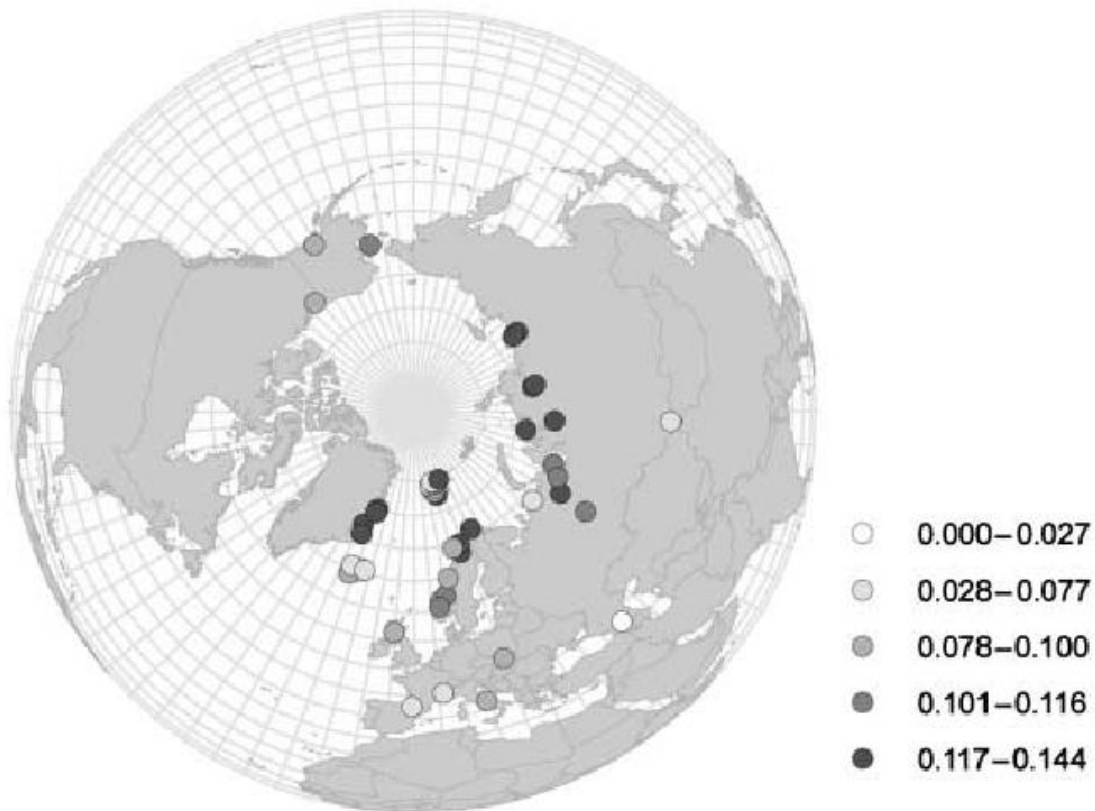


Fig. 2 Intrapopulation variation in *Dryas* measured as Nei's unbiased diversity based on 155 amplified fragment length polymorphism (AFLP) markers. Populations with less than five individuals are excluded.

inverse dynamics of refugia



Schematic map showing some types of refugia for Europe and western Asia. Interglacial refugia for cold-adapted species are shown in blue, glacial refugia for temperate species in red. Long-term refugia, indicated by dark blue/red, are a subset of all refugia that are inhabited throughout at least one full glacial/interglacial cycle. The areas shown in paler colour are refugia in the sense that they are inhabited during the contraction phase, but are not inhabited during the expansion phase owing to the spread of ice sheets during glacials (cold-adapted species), or excessive temperatures and/or too high aridity during interglacials (temperate species). Also shown, in yellow, are interglacial refugia along the oceanic/continental gradient, with a continental refugium in the east and cryptic refugia further west. The ice sheet for the Last Glacial Maximum is taken from Ehlers & Gibbard (2004). The diagram is schematic; not all of the refugia would have been occupied simultaneously, but the ranges are based on real examples.

