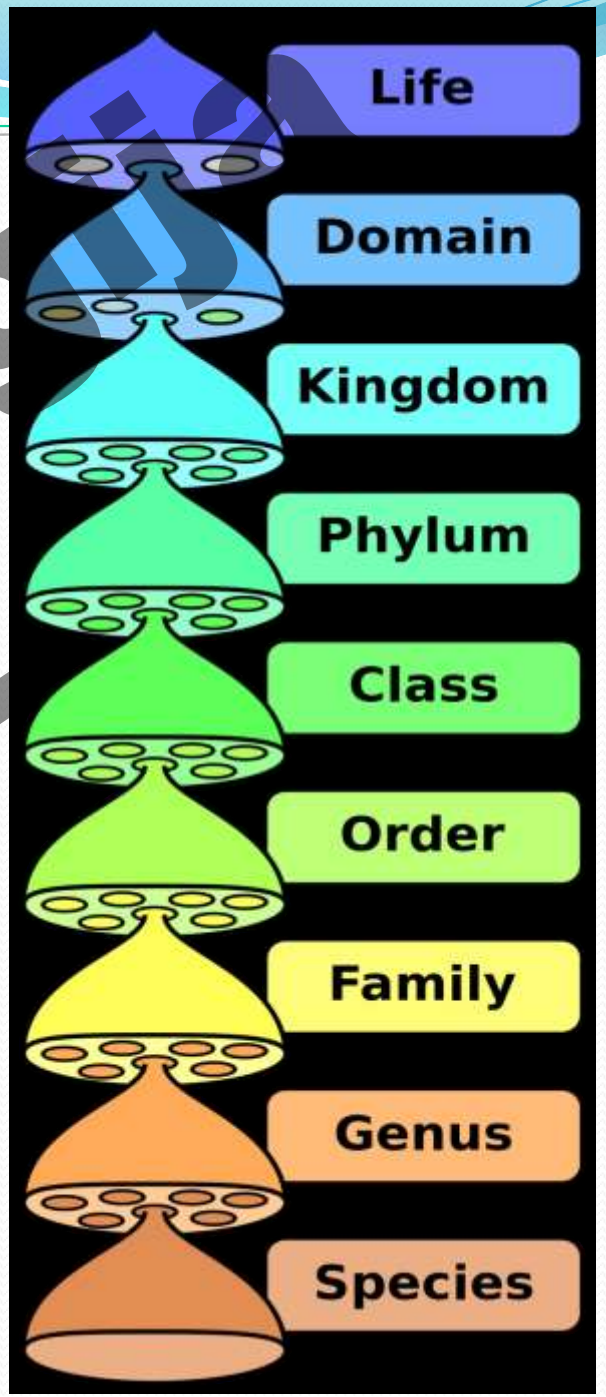
Mikroevolucioni procesi, makroevolucioni režimi

16. 11. 2012.

- Nakon kambrijske eksplozije dolazi do „stabilizacije“ biosfere
- Kako se odvija „normalna“ evolucija?
- I kako se u to uklapaju velike promene fizičkih uslova do kojih povremeno dolazi tokom geološke istorije?

Stari kreacionizam/ esencijalizam

- Line (18. vek): taksonomske kategorije su nepromenljive
- Esencijalizam
- Sa Kivijeom počinje preokret: biološka istorija mnogo dinamičnija nego što se činilo!
- Prva polovina 19. veka: pojava prvih evolucionih teorija

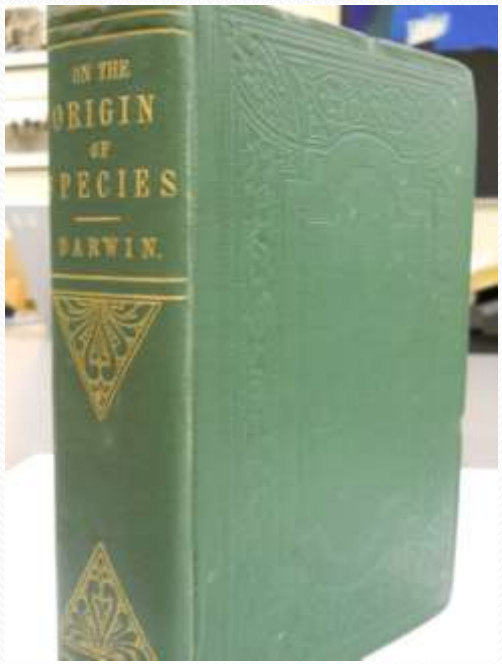


Šta znači reč evolucija?

- Slučaj revolucije je lak: *De revolutionibus orbium celestium* (1543) – primer termina "izvezenog" iz astronomije!
- *Evolucija* je suprotan primer – termin formiran u biologiji, koji je potom počeo da se primenjuje i u drugim naukama.
- Latinski: *evolutio* – odmotavanje, odvijanje (recimo svitaka)
- Evolucija se dugo vremena koristila isključivo kao termin za embriološki razvoj jedinke – individualno, a ne kolektivno značenje.
- Albrecht von Haller (1744): *evolutionem theoria* ("teorija evolucije") u razvoju embriona u odraslu jedinku.
- Ovo takođe implicira evoluciju kao progresivni trend ("napredak") što se zadržalo i kada je embriološka interpretacija odbačena!

Ključna godina

- Wallace (1859) se javlja sa Molučkih ostrva...
- Džentlmenski sporazum
- Darwin (1859) i docnija izdanja:
  - Prirodna selekcija
  - Drugi procesi (pluralizam)
- Selekcija na osnovu **adaptivne vrednosti** karaktera
- Mnogo problema u originalnoj verziji, pre svega zbog nerazumevanja načina prenošenja karaktera među generacijama!



Tri temelja klasičnog darvinizma

- Izotropna varijabilnost
- Prirodna selekcija na osnovu adaptacije (adaptacionizam)
- Prirodna selekcija deluje na svim nivoima, mikro- i makro- (ekstrapolacionizam)

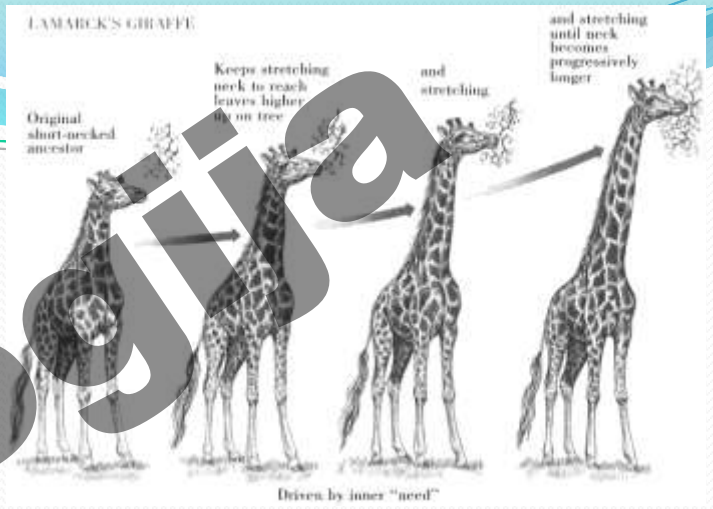
Funkcija vs. forma

- Drevna dilema...
- Iste dileme iz domena umetnosti su igrale ulogu u istoriji nauka u životu!

Oko 1900. godine: mnogo različitih evolucija...

- Funkcionalističke teorije evolucije:
  - darvinizam
  - (neo)lamarkizam
- Formalističke teorije evolucije:
  - ortogeneza
  - saltacionizam (makro-mutacije)

(Neo)lamarkizam



- Nasleđivanje stečenih karaktera
- Funkcionalizam: karakteri se pojavljuju zbog funkcije koju obavljaju
- Daleko brža evolucija (u poređenju sa darvinizmom)
- Motivacija: kratka vremenska skala lorda Kelvina i drugih
- Motivacija: humanistički optimizam viktorijanskog doba

Neobični slučaj žabe-babice

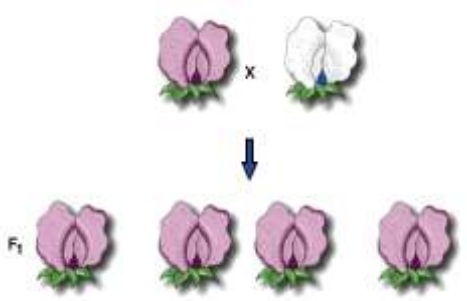
- Pol Kamerer (1880-1926), bečki zoolog, ekspert za vodozemce
- Brojni eksperimenti navodno potvrđuju nasleđivanje stečenih osobina
- Ključni eksperiment: *Alytes obstetricians*
- Bizarna priča sa tragičnim krajem (Noble & Przibram 1926)
- Nakon skandala neolamarkizam brzo nestaje sa scene…
- …sem u SSSR-u (docnije i Kini), gde ima katastrofalne posledice
- Koestler (1971): Kamerer sam žrtva prevare?
- Vargas (2009): otkriće epigenetike?

Ortogeneza

- Haacke, Eimer, Hayatt, Schindewolf, Berg...
- Formalizam: karakteri se pojavljuju kao forma, kojoj se posle pronalazi funkcija
- „Neizbežna sudbina“ svake biološke forme
- Uticaj klasične nemačke filozofije
- Primeri: karakteri koji rastu bez adaptivne vrednosti

Saltacionizam

- Nove vrste nastaju **velikim** skokovima
- *Natura facit saltum?*
- Jako uticajan pre i u Darvinovo doba (polemika sa F. Jenkinom i 5. izdanje *Postanka*)
- Otkriće genetike (Mendel i ponavljači) **podstaklo** saltacionizam
- De Friz (mutacija), Tomas Hant Morgan, brojni docniji veliki darvinisti...

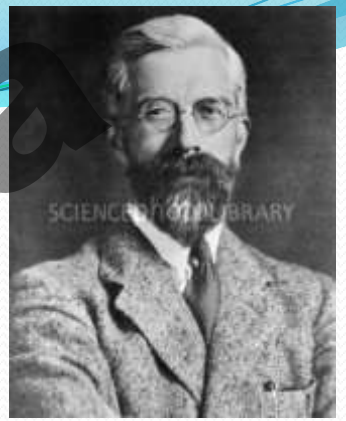


Makro-mutacije

- U 20. veku novo ime: makro-mutacionizam
- Otto Schindewolf: mutacije i supernove
- Richard Goldschmidt (oko 1940): “hopeful monsters”
- Razumevanje molekularne biologije nakon otkrića strukture DNK – zanemarljiva relativna frekvencija neletalnih makro-mutacija...
- ...osim među bakterijama gde je situacija komplikovanija.

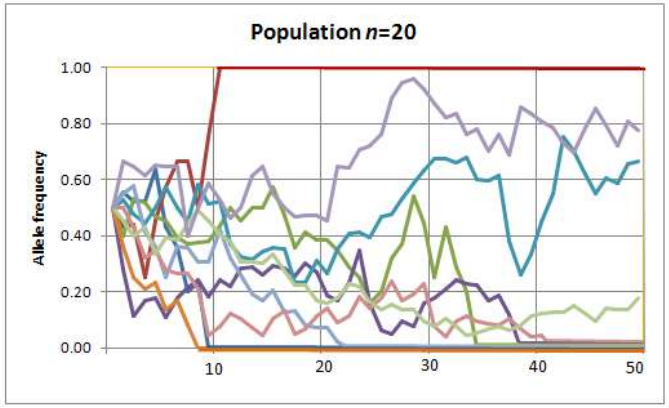
Moderna sinteza

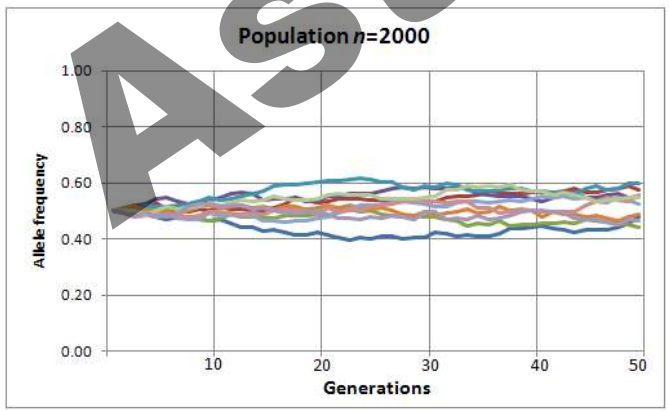
- „Sintetička teorija“, „neodarvinistička sinteza“, itd.
- Populaciona genetika + darvinistička evolucija
- Sveto trojstvo: Fischer, Haldane, Wright (1920-te i 30-te)
- Dobžanski: adaptivni pejzaž
- Od „stogodišnjice“ (1959), Moderna sinteza dominantna…
- …barem do najnovijih vremena, kad se pojavljuju nedoumice

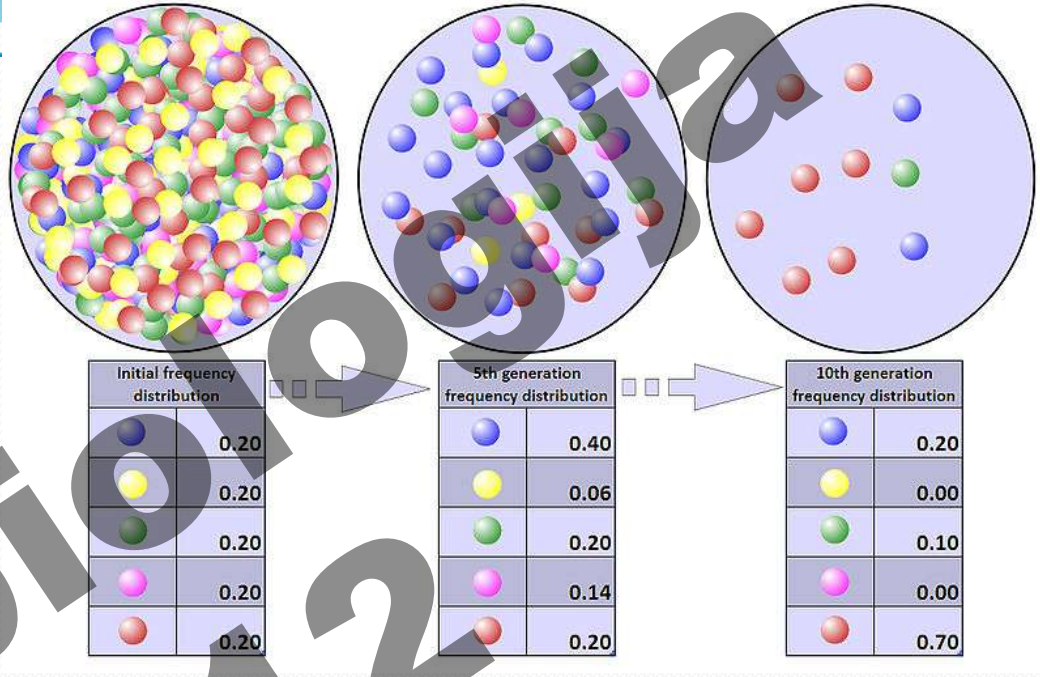


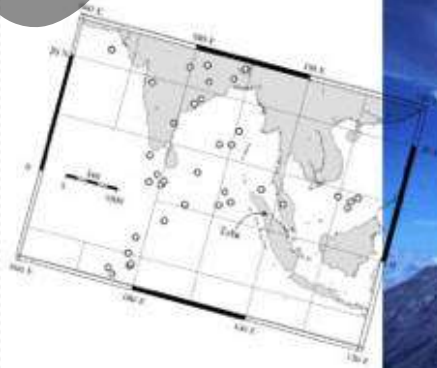
Genetički drift

- „Wrightov efekat“, „greška uzorkovanja“
- Relevantan za male populacije









Genetički drift u evoluciji čoveka?

PNAS

More genes underwent positive selection in chimpanzee evolution than in human evolution

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Observations of numerous dramatic and presumably adaptive phenotypic modifications during human evolution prompt the common belief that more genes have undergone positive Darwinian selection in the human lineage than in the chimpanzee lineage since their evolutionary divergence 6–7 million years ago. Here, we test this hypothesis by analyzing nearly 14,000 genes of humans and chimps. To ensure an accurate and unbiased comparison, we select a proper outgroup, avoid sequencing errors, and verify statistical methods. Our results show that the number of positively selected genes is substantially smaller in humans than in chimps, despite a generally higher nonsynonymous substitution rate in humans. These observations are explainable by the reduced efficacy of natural selection in humans because of their smaller long-term effective population size but refute the anthropocentric view that a grand enhancement in Darwinian selection underlies human origins. Although human and chimp positively selected genes have different molecular functions and participate in different biological processes, the differences do not ostensibly correspond to the widely assumed adaptations of these species, suggesting how little is currently known about which traits have been under positive selection. Our analysis of the identified positively selected genes lends support to the association between human Mendelian diseases and past adaptations but provides no evidence for either the chromosomal speciation hypothesis or the widespread brain-gene acceleration hypothesis of human origins.

molecular evolution | population size

Although humans and their closest living relatives, chimpanzees, are highly similar at the genetic level (1–6), they differ in many morphological, physiological, and behavioral traits (7). Phenotypically, modern humans appear to have changed considerably more than modern chimps from their common ancestors (7–10). Many of these evolutionary modifications in humans, such as the origins of bipedalism, speech and language, and other high-order cognitive functions, are widely thought to be adaptive (11–13). These observations led to a common belief that more genes underwent positive Darwinian selection in the human lineage than in the chimpanzee lineage. Indeed, there are more reports of positively selected genes (PSGs) in humans than in chimps (12, 13). Nonetheless, this difference may be largely due to a lack of study in chimps. To avoid such a bias, one could identify and compare all PSGs from the human and chimp genomes. Positive selection acting on a protein-coding gene may be detected by various population genetic and molecular evolutionary methods that use intraspecific polymorphism data, interspecific divergence data, or a combination of the two (14–16). However, because of the paucity of polymorphism data from chimps, a fair comparison between the two species would have to be limited to the divergence data. Such data can be used to estimate the ratio of nonsynonymous to synonymous substitution rates (ω). An ω value significantly >1 indicates the action of positive selection, whereas an ω significantly <1 indicates negative (or purifying) selection. Using this approach, two earlier studies (17, 18) pioneered the identification of human and chimp PSGs at the genomic scale, although no comparison was made between the numbers of human and chimp PSGs. In fact, the studies' results would be unsuitable for the comparison, owing to a number of deficiencies. First, both studies used the mouse as an outgroup, to distinguish between human-specific and chimp-specific nucleotide substitutions, because of the unavailability of genome sequences from any closer outgroups at that time. Because mouse is distantly related to human and chimp, this practice introduces errors. Second, one of the studies (17) was based on less reliable statistical methods and assumptions (19), whereas the other (18) used the draft chimp genome sequence (1) known to contain many more errors than the finished human genome sequence (20, 21). Because the majority of genes in a genome have $\omega < 1$, and sequencing errors have an expected ω of 1, the errors inflate ω and the false detection of positive selection. In this work, we first design a protocol to rectify these problems and then use the protocol to identify and compare human and chimp PSGs. Our results show substantively more PSGs in chimpanzee evolution than in human evolution.

Results and Discussion

Study Design. To compare human and chimp PSGs impartially, we made three improvements in the design of the analysis. First, to distinguish nucleotide substitutions that occurred in the human lineage from those that occurred in the chimp lineage, we used the macaque monkey as the outgroup. Because the divergence time between the macaque and human/chimp is approximately a quarter of that between the mouse and human/chimp (22–24), the reliability of our analysis was expected to increase significantly. Gene ortholog determination and sequence alignment among the more closely related human–chimp–macaque gene trios is also more reliable than among human–chimp–mouse trios.

Second, we applied an improved branch-site likelihood method for identifying PSGs (25), which has been shown by computer simulation to produce good results even when some of the assumptions are violated (25). The method requires that the branches in a phylogenetic tree be separated into foreground and background branches *a priori*, where foreground branches are tested for the occurrence of positive selection. The method assumes that two classes of codons, either negatively selected (class 0) or neutral (class 1), exist in the background branches. This null model is compared with an alternative model in which a proportion of class 0 codons, and the same proportion of class 1 codons, become positively selected in the foreground branches. Positive selection in foreground branches is inferred for a gene if the likelihood of the observation of the gene sequences is significantly higher under the alternative model than under the null model. To further verify the suitability of the method in the present context, we conducted additional computer simulations specifically designed to mimic the

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The authors declare no conflict of interest.

Abbreviation: PSG, positively selected gene.

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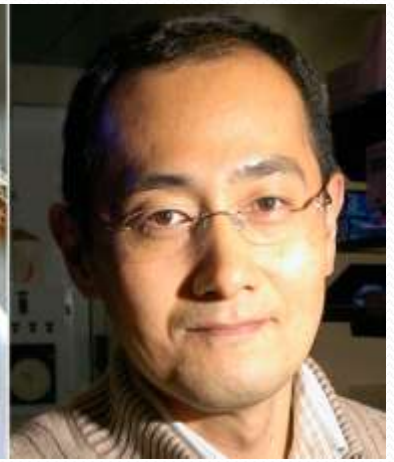
Epigenetika

- Veoma velika tema…
- Nobelova nagrada za medicinu 2012. (Gurdon & Yamanaka)
- Kog su pola šarene mačke?
- *Hongerwinter*
- Da li će se deo genoma uključiti ili isključiti zavisi od hemijskih reakcija van samog genoma
- Npr. metilacija (dodavanje CH_3^+) mRNK reguliše ekspresiju FTO gena, jednog od nosilaca nasledne gojaznosti



Rezime:
šta su (mikro)evolucioni procesi?

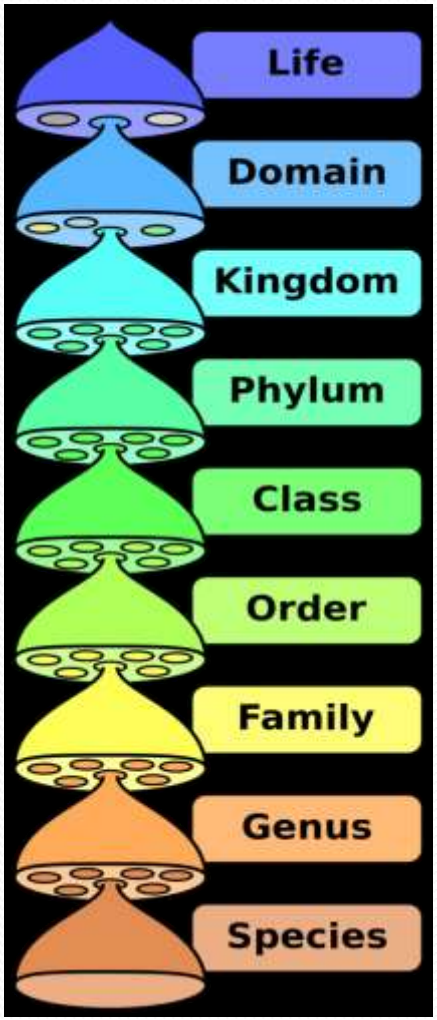
- **Prirodna selekcija**
- **Genetički drift**
- Ekološka ograničenja
- Epigenetski procesi
- [Usmerene mutacije/horizontalni transfer gena/nasleđivanje stečenih karaktera – samo mikroorganizmi i/ili posebni uslovi]

Izazovi Modernoj sintezi

- Jedinica selekcije?
- Relativni značaj genetskog drifta?
- Evo-devo (ili, još novije, eko-evo-devo)
- Novi formalizam na nivou molekularne biologije (npr. prioni)
- Cela oblast **makroevolucije**

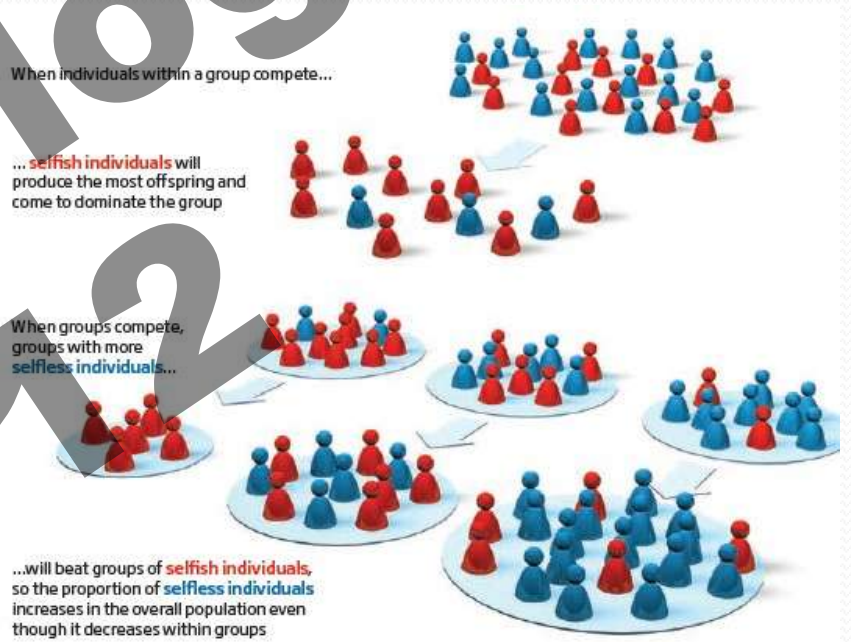
Šta je **makro**evolucija?

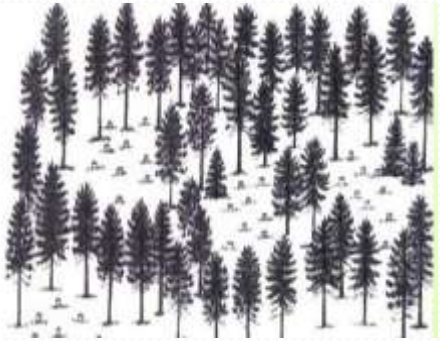
- Jednostavan odgovor: sve što se dešava u biosferi iznad nivoa vrste
- Pravi (i nejasan) odgovor: promene na skali razdvojenih genskih fondova
- Primeri problema:
  - Promene u biodiverzitetu
  - Adaptivne radijacije (poput Kambrijske eksplozije)
  - Masovna izumiranja
  - Biogeografija i ekologija



Postoji li selekcija **među** vrstama?

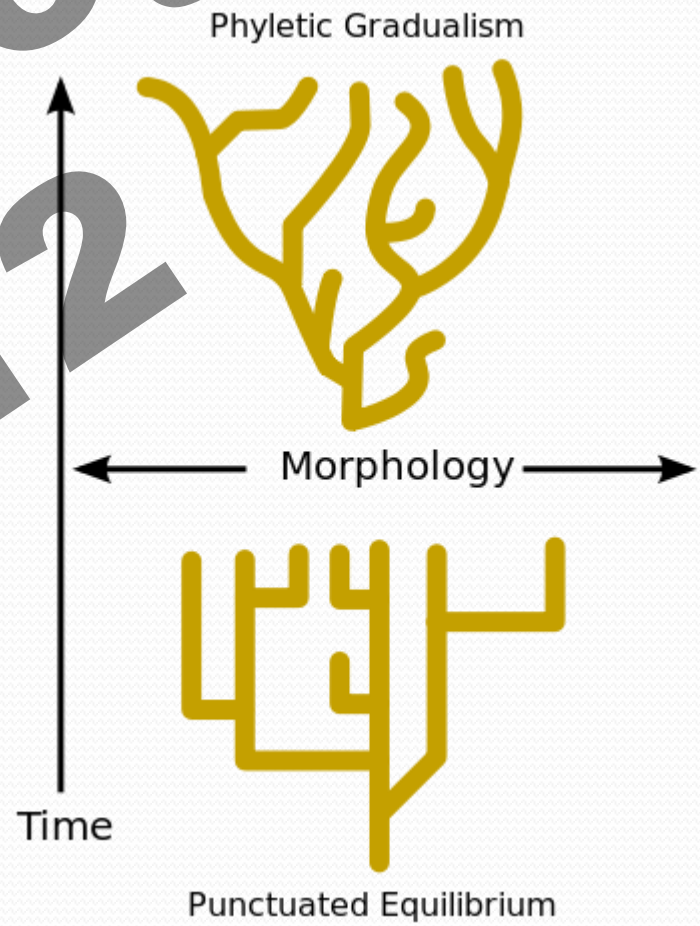
- Grupna selekcija – VRLO kontroverzna tema
- Haldane: rođačka selekcija (*kin selection*)
- Teorijski, grupna selekcija deluje nasuprot individualnoj i može da objasni složene karaktere poput altruizma...
- U praksi, ekstremno teško testirati hipoteze vezane za grupnu selekciju!
- Sociobiologija i sl.





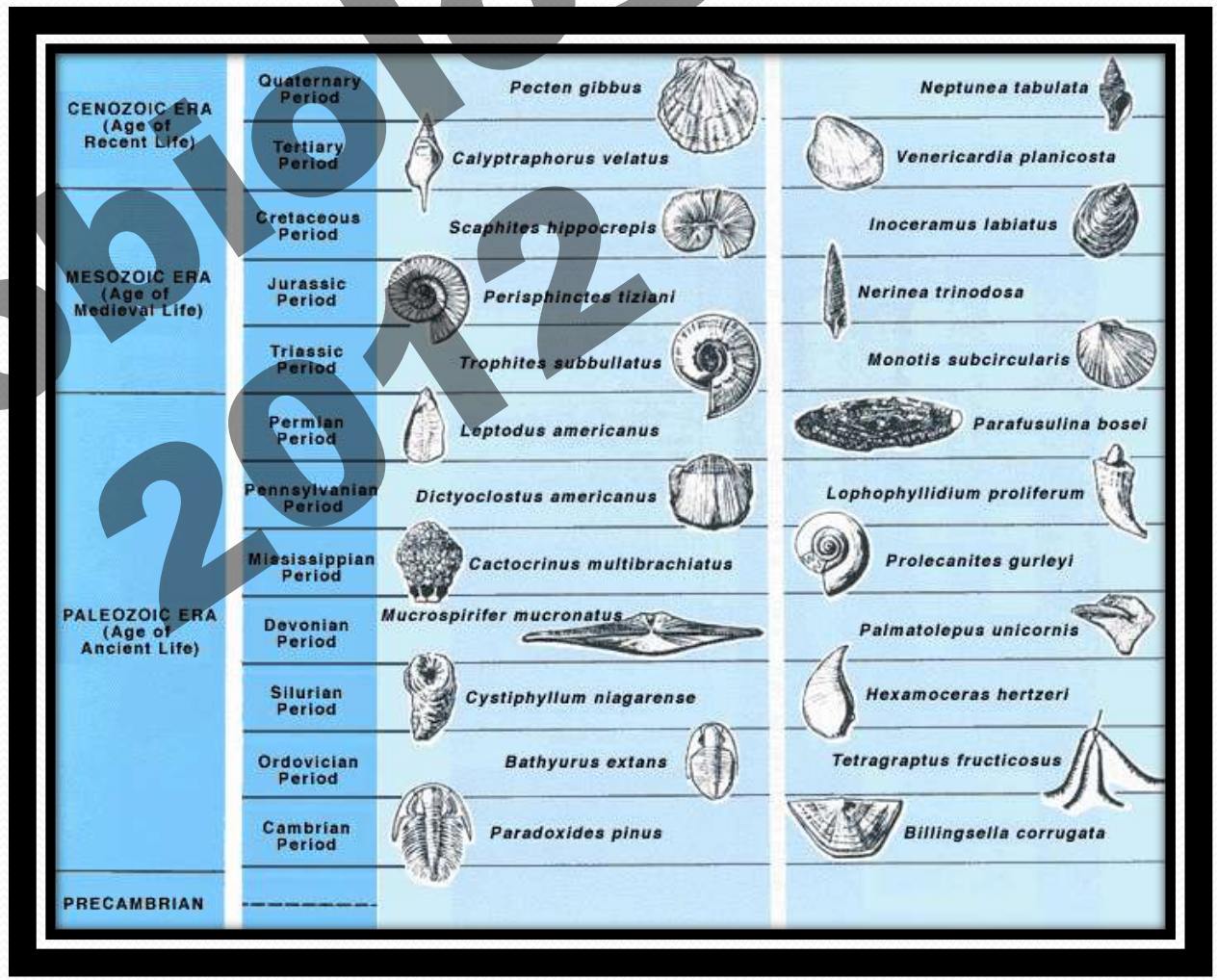
Punktuirana ravnoteža

- Eldredge & Gould (1972)
- Ključni uvid iz paleontologije: **stazis** !
- Oscilacije oko fenotipske srednje vrednosti
- (Stazis je osnova biološkog datiranja!)
- Evolucija kao vojnički život: „dugi periodi dosade razdvojeni epizodama drame“
- Ovo se odnosi na većinu specijacija – komplement diskontinuitetu izumiranja



Indeksni fosili (zonski, indikatori)

- Da indeksni fosili nisu fenotipski stabilni – kako bi datiranje uopšte bilo moguće?
- (samo nemojte da gradualisti čuju…)



Makroevolucioni režimi

- „Smena fauna“ (*faunal overturn*)
- Da li je fauna A „bolja“ od faune B?
- Barnosky (2001): hipoteza dvorske lude *(Court Jester hypothesis*)
- David Jablonski, Andrew Knoll: „promena pravila igre“