L27-Molecular Evolution +Evolution in Geological Time
Mutations

Frameshift mutations can cause major amino acid changes, resulting in genetic diseases - anemia due to unbalanced globin protein production (thalassemia)
Mutations

Chromosomal mutations that affect copy number (‘ploidy’) and rearrangements

Arise frequently during meiosis

generally lethal

trisomy 21-Down’s
Gene duplications
Gene duplications - ”supplies raw material for biological evolution”
-Jianzhi Zhang, TREE

-redundant functions facilitate emergence of new functions through the acquisition of mutations (1 gene /100 My in verts.)
–leucine biosynthesis + TCA cycle
–red + green sensitive opsins in humans
Gene duplications and new functions

Subfunctionalization - paralogous 'division of labor'

Neofunctionalization - paralogs evolve novel attributes

RNASE -> RNASE1 + RNASE1B
-facilitates persistence on plant material
-N acquired from cellulolytic gut bacteria
Gene duplications - retention of ancestral functions balanced by acquisition of mutations

Duplicate genes acquire deleterious mutations and are pseudogenized
Evolutionary forces behind gene duplication

Dykhuisen-Hartl theory: random mutations are fixed in one gene under relaxed purifying selection, which is the result of reduced functional constraint provided by the redundancy

>no need for positive selection
Evolutionary forces behind gene duplication

Dykhuisen-Hartl theory: random mutations are fixed in one gene under relaxed purifying selection, which is the result of reduced functional constraint provided by the redundancy.

> no need for positive selection

Multifunctional specialization: positive selection drives a multifunction-encoding gene to specialize, as enabled by the redundancy.
Mutations in nonprotein coding DNA

Transcriptional regulation mutations can interfere with appropriate temporal or conditional expression of genes.

Mobile genetic elements (transposons) likely not under selection, and can change, as well as introduce indel mutations.

(Micro)satellites repetitive sequences dispersed throughout the genome - Huntington’s (>35 CAG repeats) - useful for phylogenetic/forensic analyses.
Measuring genetic change

transitional states are not always available
multiple substitutions can obscure evolutionary history
Measuring genetic change

Models of sequence evolution used to correct for differences between observed and expected species divergence
DNA substitution models of evolution

**Jukes-Cantor (homologous)**
-equal probability of character change

**Kimura Two-Parameter**
-pur->pur changes more likely than pur->pyr

![Diagram showing DNA base pair transitions and transversions](image)
DNA substitution models of evolution

Felsenstein 1981 (F81)
-similar to JC and K2P
-accounts for base composition (25-75%)
-assumes even distribution of base composition

*Thermus* and *Deinococcus* are closely related but trees using base composition may not group them
DNA substitution models of evolution

Felsenstein 1981 (F81)
- similar to JC and K2P
- accounts for base composition (25-75%)
- assumes even distribution of base composition

Hasegawa-Kishino-Yano 1985 (HKY85)
- merges K2P and F81

Likelihood tests are used to evaluate the appropriateness of the model.
Frequencies of substitutions by nucleotide feature

Empirical evidence about rates of change in different regions can parameterized to inform models sequence evolution
Nonuniform nucleotide site variation

Limitations on sites that can vary can impact the rate of sequence divergence

(A) can vary at 80% of sites at a rate of 0.5%/My but (B) varies at 50% of sites at a rate of 2%/My

While site changes *saturate* more rapidly in (B) than (A), one might incorrectly infer that (B) is evolving more quickly.
Models of molecular evolution

Haemoglobin alpha-globin present in most verts

Comparison of a.a. changes between increasing different species shows a proportional increase in seq. distance

Steady rate of change suggests that a-globin behaves in like a molecular clock
Models of molecular evolution

Natural selection acts to remove deleterious mutations (purifying) while fixing those that confer fitness benefits (positive).

Experimental evidence showed that at many loci up to an average of 30% polymorphism exist with an average heterozygosity of 11%.

If most mutations are deleterious, how can so much variation be maintained in populations?
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Models of molecular evolution

*If most mutations are deleterious, how can so much variation be maintained in populations?*

**Neutral mutations** are those that do not confer selective costs
What type of mutation would most likely be neutral?

A. synonymous
B. frameshift
C. nonsynonymous
D. 4 nucleotide indel
Models of molecular evolution

*If most mutations are deleterious, how can so much variation be maintained in populations?*

**Neutral mutations** are those that do not confer selective costs

Motoo Kimura 1st proposed the neutral theory of evolution in 1969.
- deleterious and selectively removed
- neutral and slight chance of fixation
Models of molecular evolution

Functional constraint strongly impacts the frequency of deleterious and neutral mutations and the rate of substitutions. Alpha-globins (essential for hemoglobin function) are under considerable functional constraint.
L27-Molecular Evolution

+Evolution in Geological Time
The Paleogene is divided into four ages, but only two are shown here. What is shown as Calabrian is actually three ages—Calabrian from 1.8 to 0.78 Ma, Middle from 0.78 to 0.13 Ma, and Late from 0.13 to 0.01 Ma. Walker, J.D., Geissman, J.W., Bowring, S.A., and Babcock, L.E., compilers, 2012, Geologic Time Scale v. 4.0: Geological Society of America, doi: 10.1130/2012.CTS004R3C. ©2012 The Geological Society of America. The Cenozoic, Mesozoic, and Paleozoic are the Eras of the Phanerozoic Eon. Names of units and age boundaries follow the Gradstein et al. (2012) and Cohen et al. (2012) compilations. Age estimates and picks of boundaries are rounded to the nearest whole number (1 Ma) for the pre-Cenomanian, and rounded to one decimal place (100 ka) for the Cenomanian to Paleocene interval. The numbered epochs and ages of the Cambrian are provisional. Cohen, K.M., Finney, S., and Gibbard, P.L., 2012, International Chronostratigraphic Chart: International Commission on Stratigraphy, www.stratigraphy.org (last accessed May 2012). (Chart reproduced from the 34th International Geological Congress, Brisbane, Australia, 5–18 August 2012.) Gradstein, F.M., Ogg, J.G., Schmitz, M.D., et al., 2012, The Geologic Time Scale 2012: Boston, USA, Elsevier, DOI: 10.1016/B978-0-444-59425-9.00004-4.
Evolutionary trends
patterns of directional change over time
Cope’s rule—mammals get larger the longer they exist
passive: constraint on size
active: selection for size
Evolutionary trends

Active trends through parallel change or species selection

A
An active trend appears as a general directional shift in trait values within a clade

B
Under species selection, subclades with smaller body size tend to go extinct while those with larger body size tend to speciate.

C
Alternatively, each subclade may undergo parallel evolutionary change toward larger body size.
Phyletic Gradualism

New species arise through gradual transformation of ancestral species

Darwin accepted gradualism as the primary means for species diversification - ‘gaps’ in the fossil record were confounding
Phyletic Gradualism

New species arise through gradual transformation of ancestral species.
Punctuated Equilibria

Niles Eldredge and Stephen J. Gould (1972)

Prolonged stasis of well-adapted ‘forms’ (not to exclusion of less well-adapted ‘forms’) interrupted by rapid shifts from one state to another.

May explain rapid evolutionary changes observed during Cambrian explosion
Punctuated Equilibria

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Punctuated anagenesis: loss of ancestral state
Punctuated Equilibrium

• Pattern of change in the fossil record
  – Long periods of little or no change (stasis) followed by rapid change
  – Stasis is punctuated by rapid change

• A hypothesis about the evolutionary process
  – Evolutionary change accompanied speciation which occurred “off stage” in small (allopatric) populations (i.e., subpopulations of a species).