Contents

1 Evolutionary Thinking

Introduction (Lecture)

- Essential questions of evolutionary biology:
  - Why do organisms look so different?
  - Why develop elaborate sexual traits?
  - Why do organisms senesce?
- Evolution is mainly an historical science and thus must relay on other methods of reconstructing the past or making inferences about evolutionary forces.
- **Proximate**: a question about a mechanistic cause; provides an immediate explanation about how a mechanistic cause functions.
- **Ultimate**: why, or the reason, a trait or organism is the way it is; an evolutionary explanation.
- Example of proximate vs ultimate in Galapagos finches:
  - Proxmate: developmental growth factor is increased/decreased in some birds.
  - Ultimate: different habits are selected on breaks that maxmize food gathering ability.
- Evolutionary biology's approach to answering questions:
  - **Empirical data**: observation studies, experiments; the *comparative method*.
  - **Theory**: predictions that use models and mathematical reasoning which can be be tested with empirical data.
- Overview of the components of evolution by natural selection:
  - Genetic variation exists, via mutations.
  - Mutations are heritable.
  - The is an advantage to survival and/or reproduction from the mutation.
  - Individuals with the advantage in survival/reproduction are selected for.

10 Studying Adaptation

Hypothesis Testing: Oxpeckers Reconsidered

- **Adaption**: a trait, or a suite of traits, that increases the fitness of its possessor.
- No hypothesis for the adaptive value of a trait should be accepted simply because of its plausibility.
- Oxpeckers and impalas traditionally were thought to have a mutally beneficial existence; oxpeckers ate ticks and impalas provided a safe environment.
- Experiments on cattle were done to test whether this observation was true:
  - Results show red-billed oxpeckers have no effect tick loads of cattle.
  - Red-billeld oxpeckers maintained open wounds, even enlarging existing wounds to feed on the cattle's blood.
  - Red-billed oxpeckers removed hosts' earwax; whether this is good of bad is unclear.
  - Even these results must remain in question, as cattle are not the native host for the birds.
- Other important points to remember:
  - Differences among populations or species are not always adaptive.
  - Not every trait is adaptive.
  - Not every adaptation is perfect, often the adaption just happened to work well enough or by chance better than other adaptations.

Experimental Design

- Defining and testing effective control groups is critical.
- Treatments of controls and experimental measures must be handled as close to exactly alike as possible.
- Randomization is a key technique for equalizing miscellaneous effects and a tool to avoid bias.
- Reproduction is essential in order to help remove potential outlier effects.
  - Allows for greater understanding of precision, accuracy, and variation by providing more data for statistical tests.

4 Evolutionary Trees

How to Read an Evolutionary Tree

- **Phylogeny**: aka evolutionary tree or phylogenetic tree, is a diaggram showing the history of divergence and evolutionary change. Essentially, it's the genealogical relationships of organisms based on descent with modification.
  - **Taxa**: the units you are analyzing, e.g. certain species or DNA sequences.
    - **Character**: a feature or trait present among the taxa of interest, e.g. teeth of mammals or nucleotides of DNA sequences.
      - *Character state(s)*: an alternative condition of a character, which are able to evolve one to another, e.g. pointed/flat teeth of mammals.
    - *Ancestral character*: a trait that was possessed by the common ancestor.
    - *Derived character*: a trait the was not possessed by the common ancestor and instead evolved in at least one of the descendants.
      - **Synapomorphy**: derived character state shared by two or more taxa and used to define a clade of taxa.
      - **Autapomorphy**: derived character state in only one taxon.
    - **Outgroup**: a taxon or taxa that are used to root the phylogeny or determine ancestral character states.
    - **Ingroup**: the set of taxa that are the focus of the phylogeny.
  - **Nodes**: points at which the tree splits; represents mutations, speciation events, or character changes.
  - **Anagensis**: descent with modification, but no speciation.
  - **Cladogenesis**: speciation, origin of clades.
    - **Clade**: also known as a monophyletic group, an ancestor and all of its descendants.
    - **Paraphyletic group**: a group of organisms consisting of an ancestor and some of its descendants.
  - **Sister**: a taxa or clade that are most closely related to each other; they share the most recent common ancestor.
- **Homology**: similarity due to common descent; continuity of a trait, character, or character state through time.

- *Homologous trait*: found in a taxa that inherited the trait from a common ancestor.

▷ **Homoplasy**: or analogous, similarity in the characters or traits in different taxa due to convergent evolution, parallelism, or reversal, but not due to common descent.

- **Convergent evolution**: similar traits due to selective forces and not shared ancestry.
  - **Parallelism**: convergent evolution in recently diverged taxa.
- **Reversal**: derived traits or character states that revert to the ancestral form.

Inferring Phylogenetic Trees

▷ **Parsimony**: relationships that require the smallest number of character changes are most likely to be correct.

- Based on shared and derived traits(synapomorphies).
- Reconstruction using parsimony:
  1. Code characters.
  2. Make up a taxon×character matrix.
  3. Search for synapomorphies, and the shortest tree.
  - Outgroups can help polarize (ancestral vs derived) the characters.
- **Treelength**: a measure of evolutionary change using parsimony.
  - Shortest tree length produces most parsimonious tree.
  - Length determined by number of synapomorphies.
  - Homoplasious characters increase tree length.

▷ **Distance Methods**: converts a sequence alignment to genetic distances between pairs of sequences.

- Branch length is proportional to genetic differences.

▷ **Maximum likelihood**: a method of estimating the parameters of a probability distribution by maximizing a *likelihood function*.

- One of the more dominant means of statistical inference.
- **Likelihood**: measure of goodness of fit of a statistical model to a sample of data for given values of the unknown parameters.
- $P(D|H)$; probability(P), Data(D), Hypothesis(H)

- **Bayesian**: uses the likelihood function to create a quantity called the *posterior probability* of trees using a model of evolution based on prior probabilities in order to produce the most likely tree.
- **Bootstraping**: creating a value that indicates how many times out of 100 (normally) that the same branch was observed when repeating the phylogenetic reconstruction on re-sampled (pseudoreplicated) set of dat.

▷ **Molecular clocks**: the average rate at which species' genomes accumulates neutral mutations over time.

- Generally a linear rate.
- Used to measure evolutionary divergence.

6 Mendelian Genetics I

Hardy-Weinberg Equilibrium

- **Population**: a group of interbreeding individuals and their offsring.
- **Gene pool**: the set of all genes, or genetic information, in any population.
- **Genotypic frequency**: number of individuals with a given genotype divided by the total number of individuals in the population.
  - The proportion (i.e., $0 < f < 1$) of genotypes in a population.
- **Allele frequencies**: relative frequency of an allele at a particular locus in a population.
  - **Locus**: a fixed position on a chromosome where a particular gene of genetic marker is.
  - Monoploids: frequency of an allele is the result of the number of copies of the allele divided by sample size.
    - $p = i/N$
    - p: frequency | i: copies of alleles | N: sample size
  - Diploids: frequency of alleles within three possbile genotypes at a locus with two alleles.
    - $p = f(AA) + \frac{1}{2}f(AB)$ frequency of A-allele
    - $q = f(BB) + \frac{1}{2}f(AB)$ frequency of B-allele
  - Allele frequency can always be calculated from genotype frequency, whereas the reverse requires the *Hardy-Weinberg principle* of random mating apply.
- **Hardy-Weinberg principle**: allele and genotype frequencies in a population will remain constant in the absence of evolutionary influences.
  - Allele frequencies do not change from one generation to the next.
  - Genotypic frequencies after one generation of random mating: $p^2 + 2pq + q^2$
  - Evolutionary influences: genetic drift, mate choice, assortative mating, natural selection, sexual selection, mutation, gene flow, meiotic drive, genetic hitchhicking, population bottleneck, founder effect, and inbreeding.
    - *Most of these influences will be discussed later.*

Selection

- **Fitness**: success at which a organism produces fertile offspring.
- **Competition**: an interaction between organism in which the fitness of one is lowered by the presence of another.
- **Selection**: the act on a heritable phenotypic trait due to competition.
  - Can be members of the same of different species.
  - Not always directional and adaptive, instead selection pressure is applied and removes the less fit variants.
  - Can be classified in different ways, such as effect on a trait, on genetic diversity, by life cycle, by unit of selection, or by the resource in competition.

By Effect on a Trait

- **Stablizing selection**: the simplies case in which selection acts to hold a trait at a stable optimum.
- **Directional selection**: favours extreme values of a trait.
- **Disruptive selection**: acts during transition periods when current mode is sub-optimal, but alters trait in more than one direction.
  - **Univariate**: when the trait is both quantitatively favoured in either direction and can lead to speciation.

By Effect on Genetic Diversity

- **Purifying selection**: aka negative selection; acts to remove genetic variation from the population.
- ***de novo* mutation**: introduces new variation and opposes negative selection.
- **Balancing selection**: acts to maintain genetic variation, even in absence of *de novo* mutation by frequency-dependent selection.
  - **Frequency-dependent selection**: fitness that depends of the phenotypic or genotypic composition of a population.
    - Positive: fitness increases as frequency of the trait increases.
    - Negative: fitness decreases as the frequency of the trait increases.

- **Overdominance**, *aka heterozygote advantage*: when a combination of alleles confers a selective advantage over individuals with one allele.
- **Underdominance**, *aka heterozygote disadvantage*: when the heterozygote has lower fitness than either homozygote.

By Life Cycle Stage

- **Viability selection**: *aka survival selection*: increases probability of survival.
  - Can act to improve probability of survival before and after reproduction.
- **Fecundity selection**: increases the rate of reproduction given survival.
  - May be split into sub-components including sexual selection, gametic selection, gamete viability, compatability selection, and zygote formation.

Mutation

- **Mutation**: alteration in the nucleotide sequence of the genome of an organism.
  - May not produce discernible phenotypic changes.
  - The ultimate source of genetic variation.
  - Have several types of changes, from no effect, to small changes, or complete loss of function.

Large-Scale Structural Mutations

- **Gene duplications**, *aka amplifications*: repetition of a chromosomal segment or attachment of extra piece of chromosome to another, leading to multiple copies of chromosomal regions.
- Deletions of large chromosmal regions.
- **Fusion genes**: mutations that join previously separated genes into one new distinct gene.
- **Chromosmal rearrangement**: large scale changes in structure of chromosomes, leading to speciation in isolated, inbred populations. Includes:
  - **Chromosomal translocations**: interchange of genetic parts from nonhomologous chromosomes.

    - **Chromosomal inversions**: reversing the orientation of a chromosomal segment.
    - Non-homologous chromosomal crossover.
    - **Interstitial deletions**: inverse of fusion genes; removes a segment of DNA joining distant genes.
  - **Loss of heterozygosity**: loss of one allele, by deletion or genetic recombination, in a organism that previously had two different alleles.

Small-Scale Mutations

- **Point mutation**: a single nucleotide base change, that can result in a variety of effects.
- **Insertions**: add one or more extra nucleotides into the DNA.
  - Usually caused by transposable elements, or errors during replication or repeating elements.
  - Can cause*reading frame shift*, possibly effecting how many codons are read, and thus altering the gene product.
- **Deletions**: remove one or more nucleotides from the DNA.
  - Also can cause a reading frame shift like insertions.
  - Generally irreversible.
- **Substitutions**: exchange of a single nucleotide for another.
  - Often classified as transitions or transversions.
  - Generally a purine (A-G) for a purine, or a pyrimidine (C-T) for a pyrimidine.
  - Can be reversed by another point mutation.

Impact on Protein Sequence

- Effect of mutation depends heavily on where it occurs, particularly in a coding or non-coding region.
- Regulator sequences, e.g. promoters, enhancers, silencers, can alter gene expression but are less likely to alter protein sequence.
- **Frameshift mutation**: caused by insertion or deletion of nucleotides that is not divisible by three, resulting in a different translation from the original.

- **Synonymous substitution**: a condon replacement with another that codes for same amino acid.
  - **Silent substitution**: no phenotypic difference after a synonymous substitution.
- **Nonsynonymous substitution**: a codon replacement that codes for a different amino acid.
  - **Missense mutation**: codon replacement that renders the resulting protein nonfunctional.
  - **Nonsense mutation**: codon replacement that results in a premature stop codon that produces a truncated and often nunfunctional protein.

7 Mendelian Genetics II

Migration

- **Gene flow**: movement of alleles, or genetic variation, between populations.
  - If the rate of gene flow is high enough, then two populations are considered to have equivalent allele frequencies and thus a single population.
  - Constrains speciation by combining gene pools of the groups.
  - May result in the addition of novel genetic variants in the gene pool.

Factors of Gene Flow

- Gene flow is expected to be lower in species that:
  - have low mobility or dispersal.
  - occur in fragmanted habits.
  - have long distances between populations.
  - have small population sizes.
- **Allopatric speciation**: when gene flow is blocked by physical barriers that inhibit gene flow.
- **Sympatric speciation**: result of gene flow that is blocked due to non-physical barriers that inhibit gene flow.
  - Often reproduction barriers are the main factors.

Genetic Drift

- **Genetic drift**: the change in the allele frequencies in a population due to random sampling.
  - Not influenced by environmental factors.
- May cause certain gene variants to become fixed or lost by chance.
- Generally drives populations towards genetic uniformity over time.
- Only mutation or gene flow can introduce new alleles, which acts against genetic drift.
- **Founder effect**: result of sampling error which has an increased likelyhood on populations with low numbers.

- By chance certain alleles can be dominant when they otherwise wouldn't be in a new founding population.
- Often acts to increase rate of genetic drift.

▷ **Genetic bottleneck**: a sharp reduction in the size of population due to environmental events.

- Can essentially cause a founder effect, though it's not a new population.

Molecular Evolution

▷ **Molecular evolution**: the process of change in the sequence composision of cellular molecules across generations.

▷ **Neutral theory of molecular evolution**: most evolutionary changes occur at the molecular level.

- Most variation is due to random genetic drift of mutant alleles that are selectively neutral.
- Compatable with phenotypic evolution, as phenotypes are driven by molecular changes.

▷ **Recombination**: the process that results in genetic exchange between chromosomes or chromosomal regions.

- Can also cause mutations due to misalignment after recombination.
- **Gene repair**: a type of recombination that is the product of DNA repair that corrects damage using a homologous template.
  - Often responsbile for homogenizing sequences of duplicate genes over long periods of time, which reduces nucleotide divergence.

▷ **Genetic hithicking**: change in allele frequency not because of natural selection, but due to proximity to a gene undering selective sweep.

- **Selective sweep**: a beneficial mutation that increases frequency and generally becomes fixed.

Nonrandom Mating

▷ **Inbreeding**: production of offspring from closely genetically related individuals.

- Results in homozygosity, which can increase chances chances of offspring being affected by deleterious or recessive traits.
- **Inbreeding depression**: the reduced fitness in a given population due to inbreeding.

    - Usually caused by population bottlenecks or the founder effect.
  - Can also result in purging of deleterious allels through purifying selection.
  - Can allow for the expression of advantageous phenotypes, which if outweighs the disadvantages, then could potentially lead to speciation.
- **Coefficient of inbreeding**: the probability that two alleles at any locus in an individual are identical by descent.
- Nonrandom mating does not alter allele frequencies and not a mechanism of evolution.
  - Can alter the frequencies of genotypes, changing the distribution of phenotypes in a population, which can alter patters of natural selection.
- **Assortative mating**: mating based on phenotypic factors.
  - Can play a role in sympatric speciation.
  - A form of sexual selection.
  - Can be either positive of negative, selecting for similar or different phenotypes respectively.

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