Forces and mechanisms of evolution

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Why do we care?

- Popular understanding of evolution is rife with misunderstanding
  - people are more likely to avoid or fear what they don't understand – easier to manipulate
- Important implications for health, medicine, ethics, and everyday life – you can be more empowered as individuals and members of society when you understand these basics!

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Before we begin…

Evolution (in biology)

=

a change in allele/gene frequencies in a population over time

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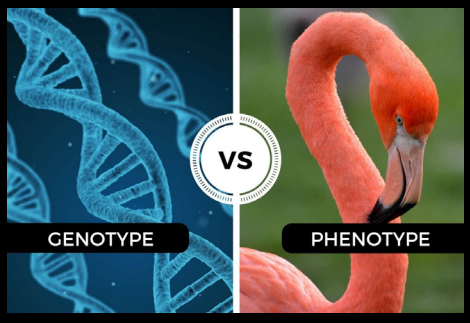
An organism's genotype is the set of genes that it carries.



An organism's phenotype is all its observable characteristics — which are influenced both by its genotype and by the environment.

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Evolution

- Microevolution:

  small changes occurring within species, such as a change in allele frequencies.
- Macroevolution:

  changes produced only after many generations, such as the appearance of a new species.
- The basic evolutionary mechanisms & processes are the same, only our scales of analysis are shifted

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Explaining evolution

- If the Earth's flora & fauna change over time, what is the mechanism by which they change?
- Evolution is a two-stage process
  - Stage 1 - the production and/or redistribution of variation
  - Stage 2 - natural selection acting on this variation.

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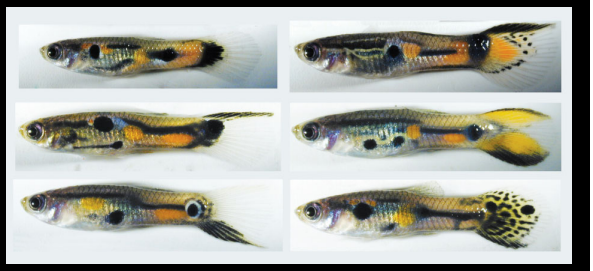
Variation – necessary for evolution by natural selection!



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Four forces that produce or redistribute variation

1) mutation
2) gene flow
3) genetic drift
4) recombination

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1) Mutation

- Change in DNA that MAY result in a change in the physical appearance of an organism
  - error in replication, or other alteration of nucleotide base sequence
- Creates variation for natural selection to act on
  - if it is beneficial, then natural selection favors that individual, making them more likely to survive and pass the mutation (and the advantageous trait) along to their offspring

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Mutation

Mutation is the creative force of evolution, as it is the only force that can produce **novel** variation.

Jean Grey and Peter Parker

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Mutation

- Can have good, bad, or null effect
- Effect depends on where in the genome the mutation is found (i.e. somatic cells vs. gametes, autosomal or on sex chromosomes, introns vs. exons)
- Small scale: point mutation, insertion/deletion

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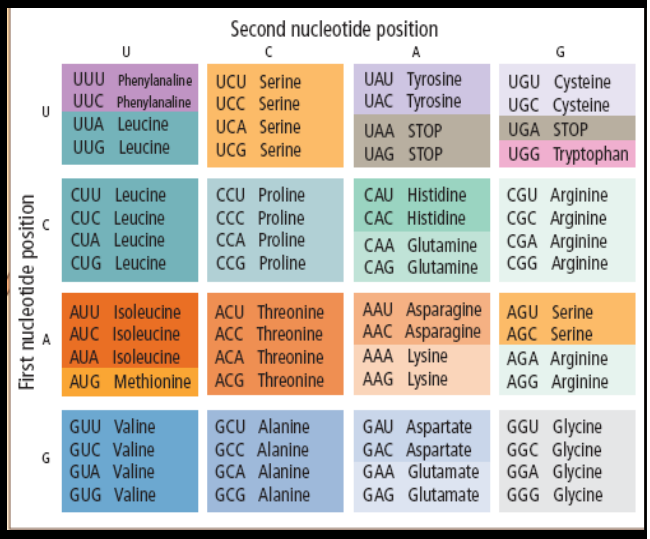
Remember… genetic code

- code used to translate DNA sequences into proteins
- Universal
- Triplets (codon in DNA and mRNA, anti-codon in tRNA)
- Continuous
- Redundant



mRNA codons and corresponding amino acids

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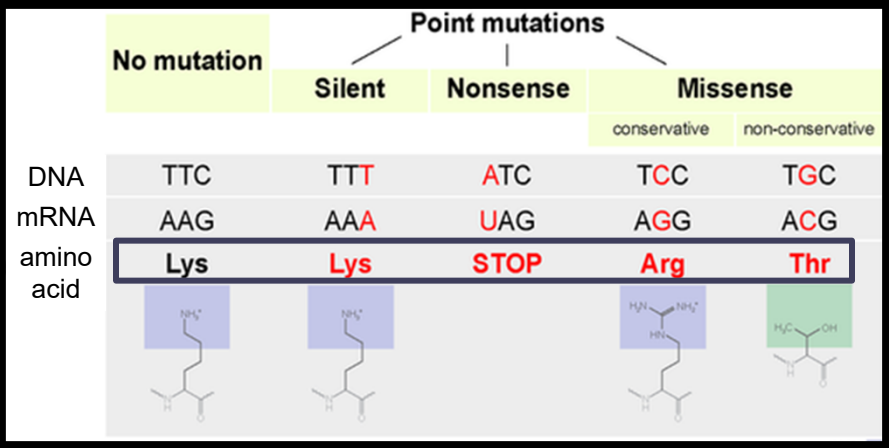
Point mutation

- Mutation of a single nucleotide



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Point mutation: sickle cell anemia

- Red blood cells have a protein called hemoglobin that carries oxygen from lungs to the rest of the body (HbA)
- Sickle cell anemia: a disease caused by a point mutation (missense) in DNA that makes a less efficient version of hemoglobin (HbS)

glutamate

DNA: … TGA GCA CTT CTC TTT …

DNA: … TGA GCA CAT CTC TTT …

valine

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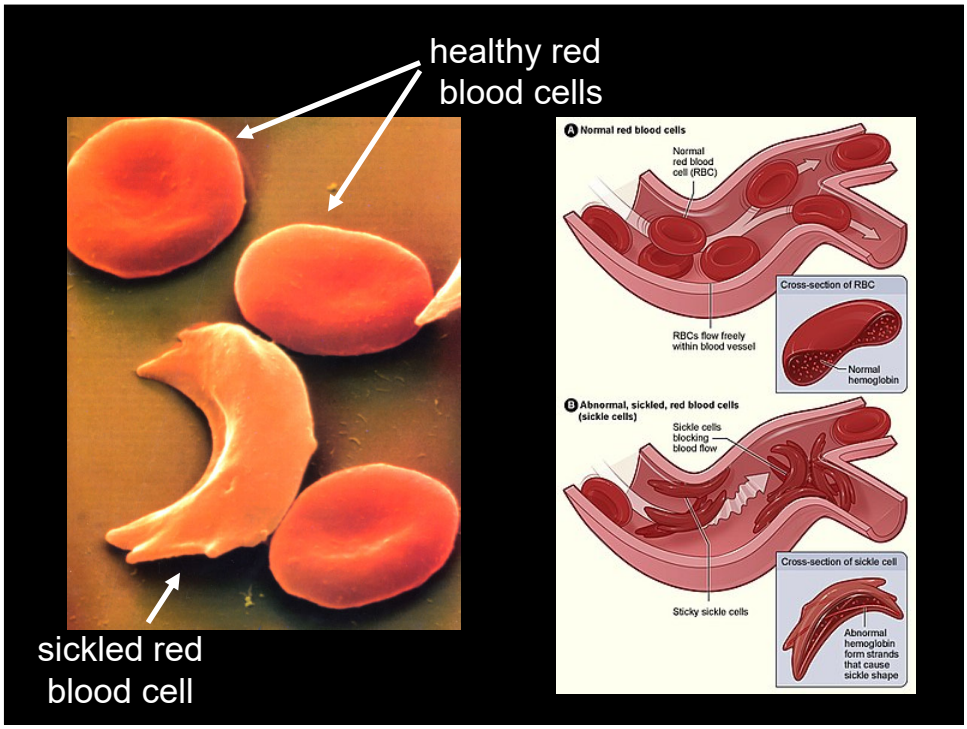

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Insertions/deletions

- Insertion mutation or deletion mutation of several bases

Insertion

Deletion

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Insertions/deletions

- Insertion mutation or deletion mutation of several bases
- common insertions of trinucleotide repeat sequences, linked to many genetic disorders
- CAG – polyglutamine expansion diseases

  - Huntington's disease (autosomal dominant, variable age of onset)

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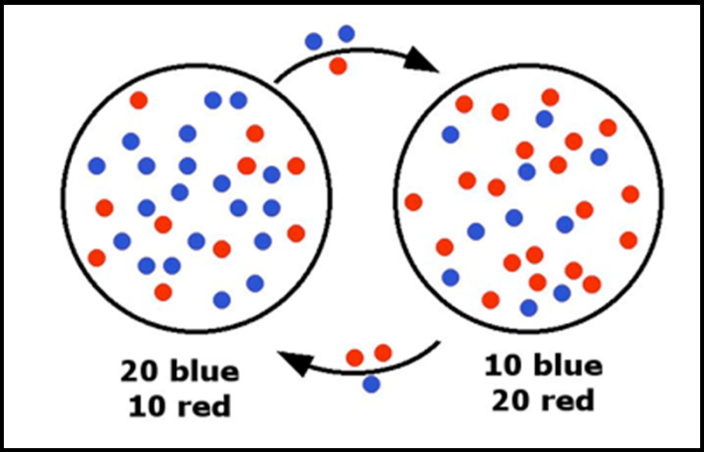
2) Gene flow



Gene flow = transfer of genes between populations (migration AND then mating)

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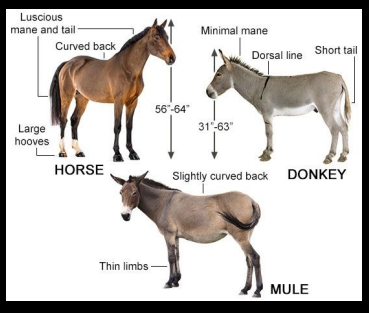
2) Gene flow

- Can affect allele frequencies and introduce new genetic variation into a population (gene pool)
- Gene flow between species – hybridization



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2) Gene flow

- Can affect allele frequencies and introduce new genetic variation into a population (gene pool)
- Gene flow between species – hybridization
- Barriers to gene flow – usually physical, but can also be behavioral, biological, etc.

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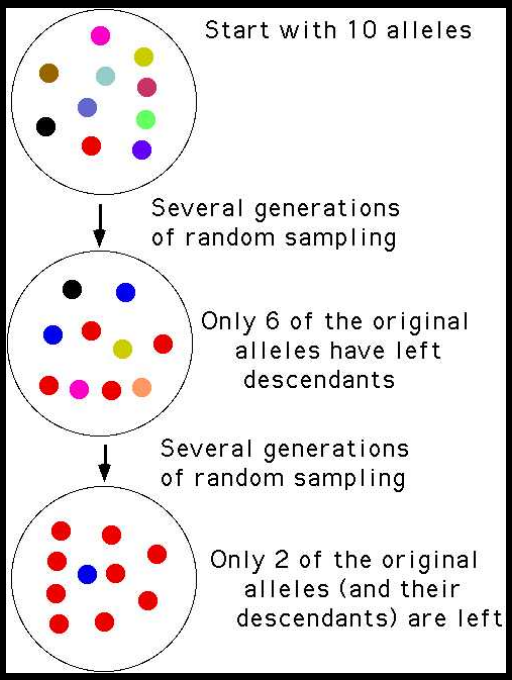
3) Genetic drift

- Changes in allele frequencies produced by random chance
- Small populations drift more rapidly than large ones, as chance events have more of an effect.



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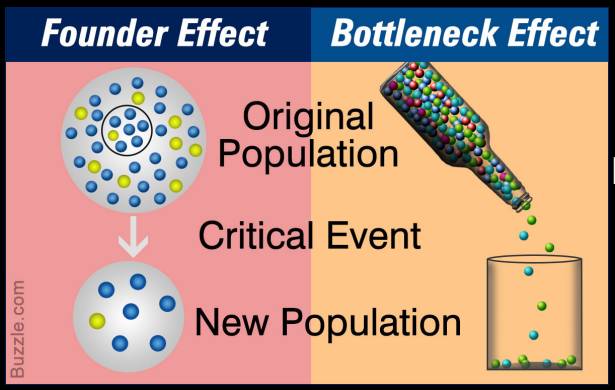
Specific forms of genetic drift: bottleneck and founder effect

Migration to a new habitat



Natural or human-made disaster

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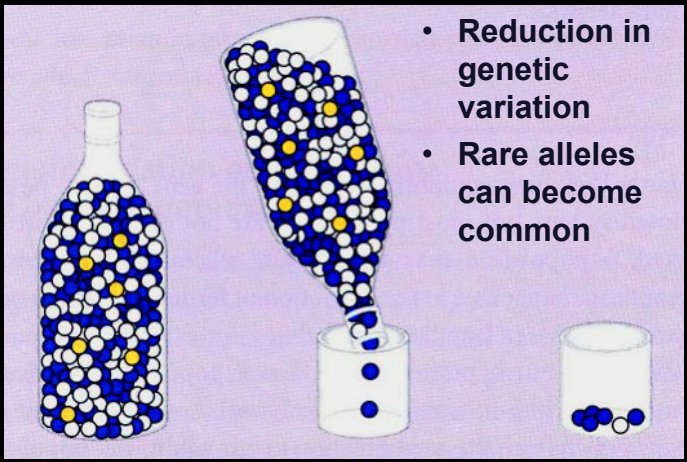
Forms of drift: Bottleneck



- **Reduction in genetic variation**
- **Rare alleles can become common**

Bottleneck = Loss of genetic diversity due to drift, usually linked to population decline

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Forms of drift: Founder effect

- Founder effect = change in allele frequencies in small populations that become separated from parent populations
- new population carries only the genetic diversity of the founders of the new group
- rare alleles, if present in any of the founders, can become very common
- e.g., Tay-Sachs in Pennsylvania Dutch

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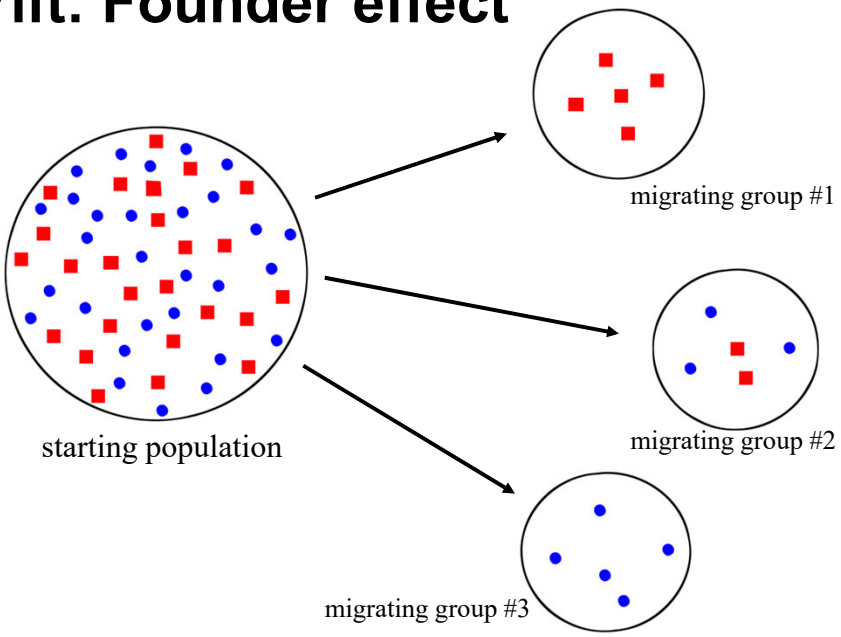
Drift: Founder effect



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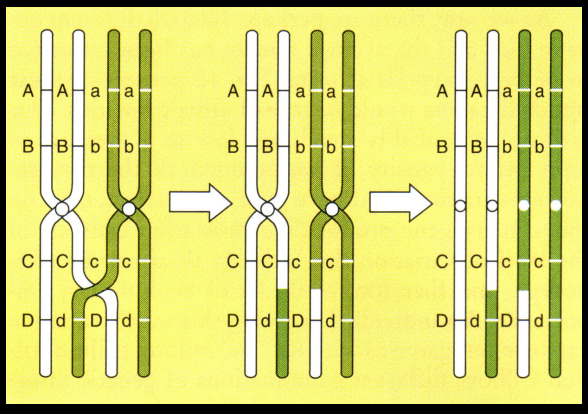
4) Recombination (crossing-over)



When homologous chromosomes line up together just prior to the first meiotic division, they can exchange genetic material – increases the number of possible genetic combinations another 10 billion times!

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4) Recombination

- **Does not alter allele frequencies**
  - NOT a force of evolution, though it produces variation
- Reshuffles genetic material and produces different combinations of genes that forces of evolution can act on

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Four forces of evolution

1) mutation
2) gene flow
3) genetic drift

} Not directional

4) natural selection

provides more directional change in allele frequencies due to specific environmental factors (adaptation)

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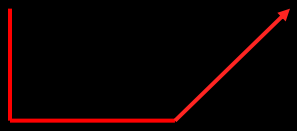
Remember!

- *Natural selection* operates on *individuals* (favorably or unfavorably)
- But, it is the *population* that *evolves*.



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Evolution by natural selection

- Natural selection acts on variation between individuals and leads to differential reproductive success between individuals
- This changes allele frequencies in a population over time
- Evolutionary fitness: measured by an individual's genetic contribution to the next generation, compared with other individuals at that time
  - "survival of the fittest" is a misnomer: it's not about physical fitness!

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Natural selection

- Drives evolutionary change.
- Can maintain the absence of change; i.e., stasis.
- Generates adaptation.

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Types of natural selection

- Directional selection
  - Selecting for greater or lesser frequency of a given trait in a population
- Stabilizing selection
  - Maintains a phenotype by selecting against deviations from it
- Disruptive selection
  - Selects for phenotypes at two extremes, and against intermediate phenotypes

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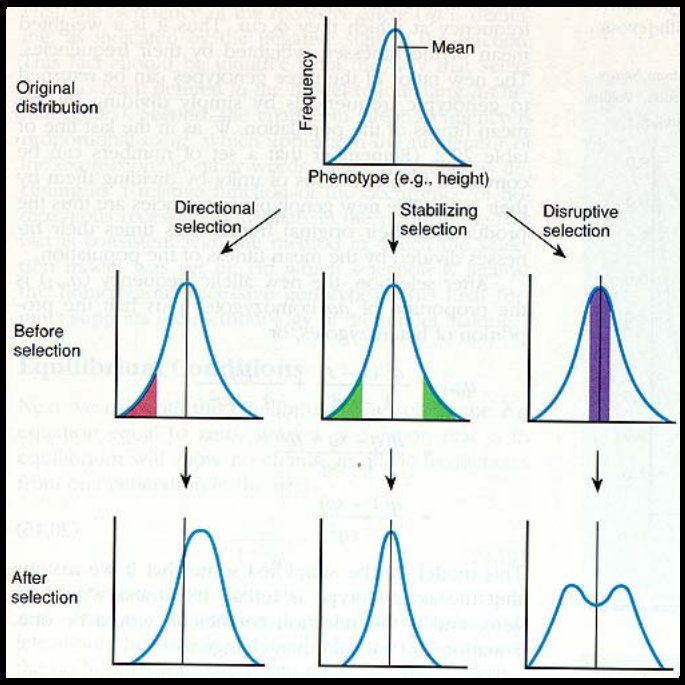

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Natural selection & the sickle cell mutation

- According to natural selection, the sickle cell mutation *should be selected against* because individuals with it have a less efficient version of hemoglobin (harmful individual variation)
- So, why does this NOT happen?

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Sickle cell anemia

- Sickle cell mutation: autosomal recessive
- If the offspring:
  - Inherits two unaffected alleles = unaffected
    - (homozygous dominant - AA)
  - Inherits the affected allele from both parents = sickle cell anemia (fatal)
    - (homozygous recessive - aa)
  - Inherits an affected allele from one parent = mild anemia, not fatal
    - (heterozygous - Aa)

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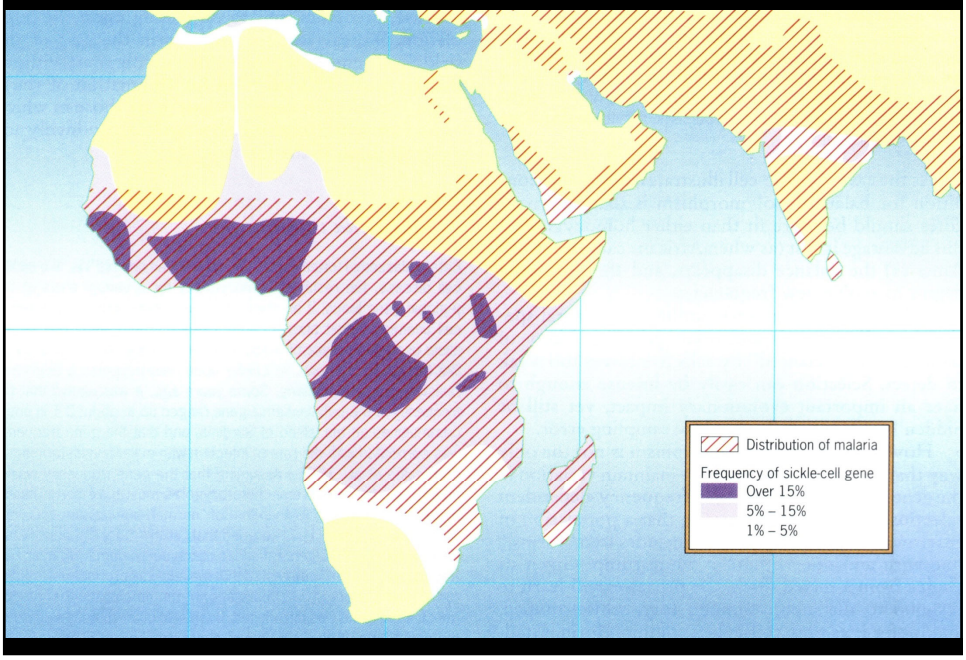
Distribution of sickle cell anemia and malaria



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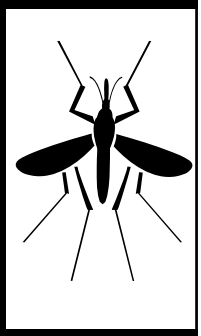
Malaria



- Caused by *Plasmodium* parasites spread by Anopheles mosquito
- Kills 1-3 million annually
- Prevalent in tropical areas
- *Plasmodium* parasites multiply in red blood cells

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How can this pattern be explained?

	Malaria	Anemia
Two HbS alleles	NO	YES
Two HbA alleles	YES	NO
Heterozygote	NO	MILD

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Natural selection & sickle cell

- Why is the mutation maintained in the population if it can cause a fatal disease?
- Because natural selection favors individuals with *one* copy of the mutation (in malaria regions), thus keeping the mutation in the gene pool even if it means that sickle cell anemia may occur in some individuals.
- This is called balancing selection, and such trait is called a balanced polymorphism

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Balancing selection

- Maintains a genetic polymorphism within a population at an intermediate frequency
  - Heterozygote advantage (e.g., sickle-cell)
  - Frequency-dependent balanced polymorphism (i.e. rare prey forms have advantage over typical forms that predators focus on)

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Epigenetics

- Heritable changes in gene expression – how, when, and for how long genes are "switched" on or off (e.g. differentiation of cells)
- Changes in phenotype WITHOUT changes in genotype!
- Reversible!

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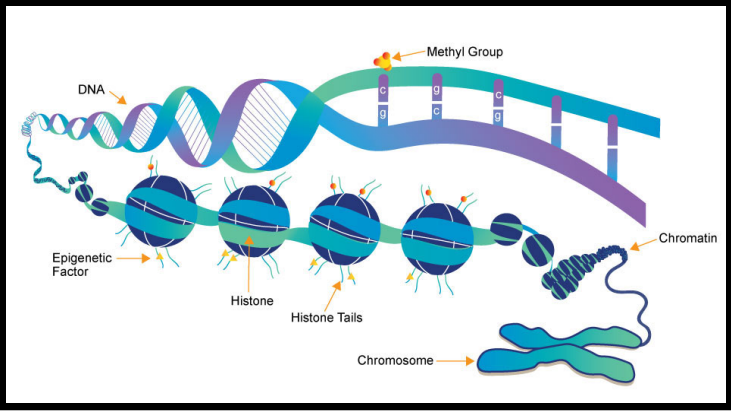
Epigenetic mechanisms

- DNA methylation, histone modification, chromatin remodeling, non-coding RNA (ncRNA)-associated gene silencing etc.



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Epigenetics

- Changes often induced by environment
- Can be transgenerational
- ≠ Lamarckian evolution!
  - Epigenetic changes often non-adaptive
  - reversible
- But… a way for effects of environment—natural, social etc.—to be embodied (Thayer & Non, 2015)

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