Variation

• Selection acts on variation

• Where does variation come from?

• How it is maintained?

• What processes affect this variation?
Mechanisms that affect variation

• Mutation
  – Generates new variation directly

• Recombination
  – Generates variation by shuffling of genes

• Genetic drift
  – Decreases variation within a population
  – Increases variation between populations

• Migration
  – Tends to decrease variation between populations
Variation: Mutation
Many kinds of mutation

- Single Point Mutations
- Insertions / deletions
- Inversions
- Duplications
- Chromosome abnormalities
Many sources of mutations

- DNA replication
- Misseggregation of chromosomes
- Physical Damage
  - UV light
Mutations occur before selection: The Lederberg experiment

Colonies grow on a plate. NO penicilin

Plate is replicated. Penicilin is added.

Some colonies survive. They carry a resistance mutation

Add Penicilin to the original plate. Did the same colonies survive?
Mutations are random with respect to selection

- Mutations do not happen in a directed fashion
Luria-Delbruck Experiment

(A) Induced mutation
(B) Spontaneous mutation
The effects of mutations

• The effects of new mutations:
  – No effect (neutral)
  – Beneficial
  – Deleterious (bad)

• The relative frequency and magnitude of each of these classes is an open question in evolutionary biology
The effects of mutations

Most people believe that most mutations are deleterious
Mutation Rates

Mutation rates per base pair are small.
Mutation rate per genome are roughly constant
An argument for small effect beneficial mutations: Fisher’s geometric model
Deleterious Mutations can be maintained under mutation selection balance

• Deleterious mutations are eliminated by selection

• But are generated by mutation
The replicator equation: introducing mutations

\[ W_a = 1 \]

\[ W_A = 1 + s \]

\[ \Delta p_{t+1} = \frac{\Delta W}{W} p_t (1 - p_t) \]
The replicator equation: introducing mutations

\[ \Delta p_{t+1} = \frac{\Delta W}{W} p_t (1 - p_t) \]  \hspace{1cm} \text{Change due to selection}

Change due to mutation:

\[
\begin{align*}
p_A(t+1) &= p_A(t) - \mu p_A(t) \\
p_a(t+1) &= p_a(t) + \mu (1 - p_a(t))
\end{align*}
\]
The replicator equation: introducing mutations

\[ \Delta p_t^m = -\mu p_t \]

\[ \Delta p_t = \frac{\Delta W}{W} p_t (1 - p_t) - \mu p_t \]

\[ \Delta p_t = 0 \Rightarrow \hat{p}_t \approx 1 - \frac{\mu}{s} \]
The replicator equation: introducing mutations

\[ \Delta p_t = \frac{\Delta W}{W} p_t (1 - p_t) - \mu p_t \]

\[ \hat{p} \approx 1 - \frac{\mu}{s} \]
Mutation Selection Balance

• Deleterious ("bad") mutations can be maintained by mutation

• Mutational Load

• Maintains genetic variation
Example: sickle cell anemia
Sickle cell anemia

Consequences from the molecular, to the cellular and to the organismal level
Sickle cell anemia

Confers protection from Malaria.

Distribution of the sickle cell trait

Distribution malaria
Summary

• Mutations are the basis for variation

• Mutations are mostly deleterious

• Deleterious mutations can be maintained by mutation-selection balance

• Beneficial mutations are expected to be of small effect