

# The Spectrum of Mutation Effects

Visual Guide to Mutation Classification



## Neutral Mutations



No effect on organismal fitness. Most common type of mutation.

**Examples:** Synonymous substitutions, mutations in non-coding regions, changes that don't affect protein function

## Deleterious Mutations



Reduce organismal fitness. Subject to negative selection.

**Examples:** Nonsense mutations, frameshifts, mutations causing genetic disorders, loss-of-function mutations

## Advantageous Mutations



Increase organismal fitness. Basis of evolutionary adaptation.

**Examples:** Antibiotic resistance in bacteria, lactase persistence in humans, pesticide resistance in insects



### Mutation Rate

10<sup>-8</sup> to 10<sup>-11</sup> per base pair per generation



### Evolutionary Impact

Raw material for natural selection



### Frequency Distribution

Most mutations are neutral or slightly deleterious



### Selection Pressure

Removes deleterious,  
preserves advantageous  
mutations

### Mutation Statistics

Neutral Mutations:	~70%
Deleterious Mutations:	~28%
Advantageous Mutations:	~2%
Human Genome Mutations/Generation:	~70
Typical Mutation Rate ( $\mu$ ):	$10^{-5}$ to $10^{-6}$ per locus

### Fitness Consequences



#### Neutral

$$w = 1.0$$

No selection



#### Deleterious

$$w < 1.0$$

Negative selection



#### Advantageous

$$w > 1.0$$

Positive selection

### Key Evolutionary Concepts



#### Molecular Clock

Neutral mutations accumulate at a constant rate,  
used for dating evolutionary events



#### Mutation-Selection Balance

Equilibrium between new deleterious mutations and  
their removal by selection



#### Genetic Load

Reduction in population fitness due to deleterious  
mutations

