A Highlight of Ideas discussed in Week 1:

Phenotype:

The observable structural or functional (e.g., behavioral, physiological, biochemical) characteristics of an individual organism; each phenotype is determined to a varying degree by the genotype and environmental factors.

Genotype:

The word genotype is used in two senses-

(a) A genotype is an individual's collection of genes (ie collection of all genes).

(b) When used in reference to a particular gene, the term refers to the two alleles inherited for that particular gene.

The genotype is “expressed” when the information encoded in the genes' DNA is used to make protein and RNA molecules. The expression of the genotype contributes to the individual's observable traits, called the phenotype.

Allele:

One of two or more forms of a gene that reside at the same position on a pair of chromosomes. For example, "A", "B" and "O" are the three alleles present at the ABO Blood group locus.

Locus:

A locus is the specific physical location of a gene on a chromosome, like a genetic street address. The plural of locus is "loci".

Population:

A population is a group of organisms of the same species that live in a particular geographic area at the same time, with the capability of interbreeding.

Darwinian Fitness:

There are multiple definitions of this term. One useful definition is-

"the genetic contribution of an individual to the next generation's gene pool relative to the average for the population, usually measured by the number of offspring or close kin that survive to reproductive age".

In short, Darwinian Fitness is a relative measure of reproductive success of an organism in passing its genes to the next generation's gene pool (ie number of offspring it produces).

In biology, "fitness" refers to a biological condition in which a competing variant is increasing in frequency relative to other competing variants in a population. It is also referred to, in particular, as Darwinian fitness.

Note that Darwinian fitness differs from the "physical fitness", which is more associated with health, muscle tissues, oxygen, and physical effort.

Darwinian fitness is more concerned about reproductive success. Darwinian fitness describes how successful an organism has been at passing on its genes. The more likely that an individual is able to survive longer to reproduce, the higher is the fitness of that individual. Thus, it may indicate the relative measure of reproductive success of an organism in passing its genes to the next generation.

Again, note that Darwinian Fitness denotes the relative ability of an individual to survive, reproduce and propagate genes IN A GIVEN ENVIRONMENT. Thus, when one says an organism is biologically fit, it means that the organism is adapted and suitable to its environment based on its relative reproductive success with respect to others in its population.

There are two ways through which fitness can be measured: absolute fitness and relative fitness.

Fitness Function:

The relationship between a phenotype and Darwinian fitness.

Phenotypic Variance:

Traits within populations are generally variable. This variation can be quantified by calculating "Phenotypic Variance" (VP).

All instances of phenotypic variance (VP) within a population are the result of genetic sources (VG) and/or environmental sources (VE). This relationship can be summarized as follows (Falconer & Mackay, 1996; Lynch & Walsh, 1998):

$VP = VG + VE$

However, in order to determine the values for both VG and VE, researchers must consider a number of additional variables.

Genotype- Phenotype Map:

Simple, one to one genotype-phenotype mapping is extremely rare. However, such characters do exist. For example, the characters typically associated with Mendelian crosses; some human diseases such as Sickle cell anaemia, haemophilia etc.

Most genotype-phenotype mapping is complex. This is because-

Phenotype is a product of both genotype and environment. Therefore, the same genotype can yield different phenotypes.

Each character is usually affected by a large number of genes and different combinations of these genes can still lead to the same phenotype.

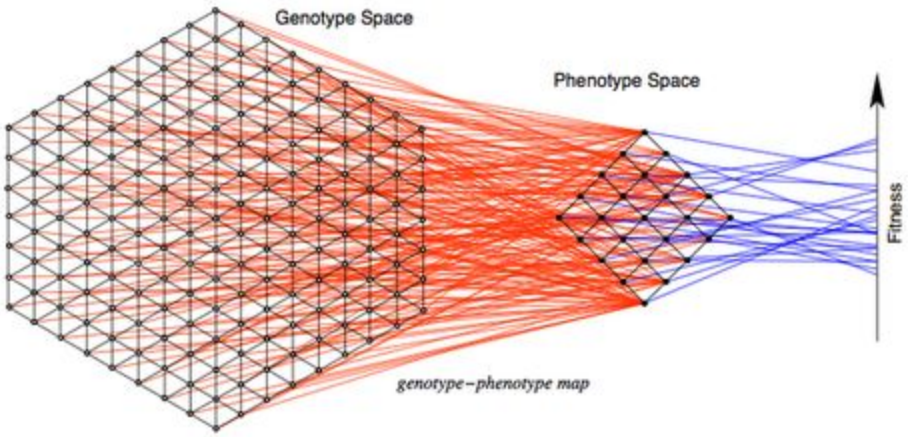


Figure 1. Fitness and Genotype-Phenotype map. Redrawn after [20]

For those of you who want to explore more:

You can create your own Genotype-Phenotype map using AVIDA.

Fortuna MA, Zaman L, Ofria C, Wagner A (2017) The genotype-phenotype map of an evolving digital organism. PLoS Comput Biol 13(2): e1005414. doi:10.1371/journal.pcbi.1005414

An interesting exposition on the properties of Genotype-Phenotype maps:

Ahnert SE. 2017 Structural properties of genotype – phenotype maps. J. R. Soc. Interface 14: 20170275. http://dx.doi.org/10.1098/rsif.2017.0275

Mutation:

A relatively permanent change in hereditary material that involves either a change in chromosome structure or number or a change in the nucleotide sequence of a gene's codons and that occurs either in germ cells or in somatic cells but with only those in germ cells being capable of perpetuation by sexual reproduction.

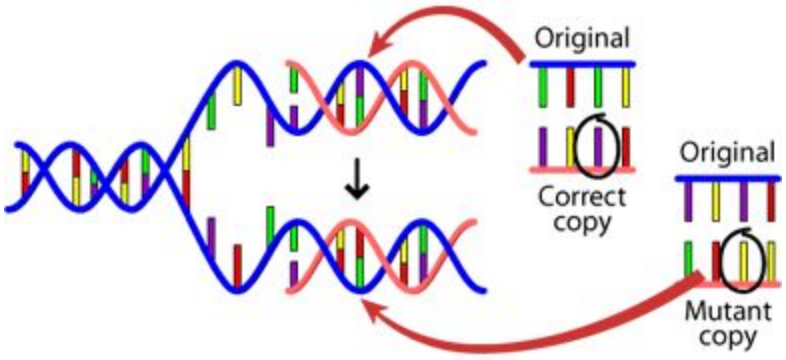
Mutation is the ultimate source of all genetic variation.

Causes of Mutation:

Mutations happen for several reasons.

DNA fails to copy accurately: This is the most common cause of Mutations.

Most of the mutations that we think matter to evolution are "naturally-occurring." For example, when a cell divides, it makes a copy of its DNA — and sometimes the copy is not quite perfect. That small difference from the original DNA sequence is a mutation.



External influences can create mutations

Mutations can also be caused by exposure to specific chemicals or radiation. These agents cause the DNA to break down. This is not necessarily unnatural — even in the most isolated and pristine environments, DNA breaks down. Nevertheless, when the cell repairs the DNA, it might not do a perfect job of the repair. So the cell would end up with DNA slightly different than the original DNA and hence, a mutation.

Types of Mutation:

I. Based on Structure

1. Point Mutation
2. Multiple bases
3. Large areas of a chromosome
4. An entire chromosome

5. An entire set of chromosomes.

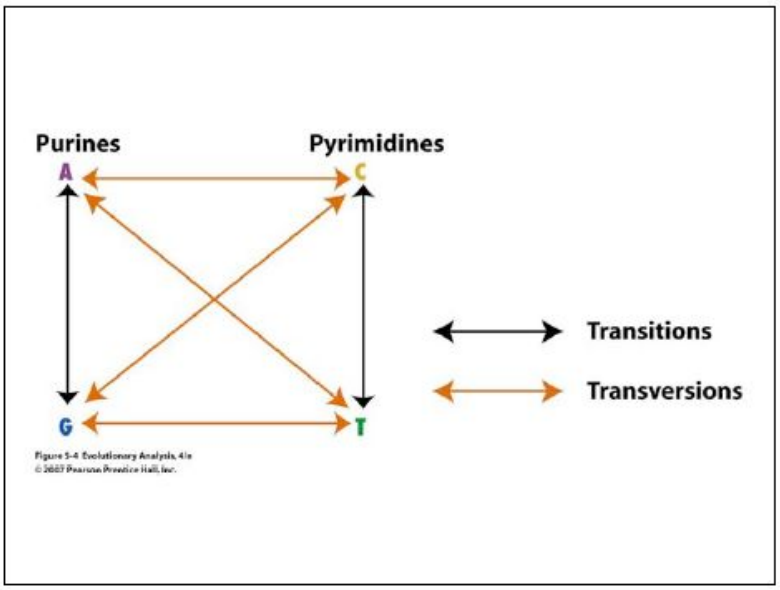
1. Point Mutation: Only one base is mutated. This is the most common type of mutation.

1.a. Base Substitutions

One base substitutes another base.

Transition: this occurs when a purine is substituted with another purine or when a pyrimidine is substituted with another pyrimidine.

Transversion: when a purine is substituted for a pyrimidine or a pyrimidine replaces a purine.



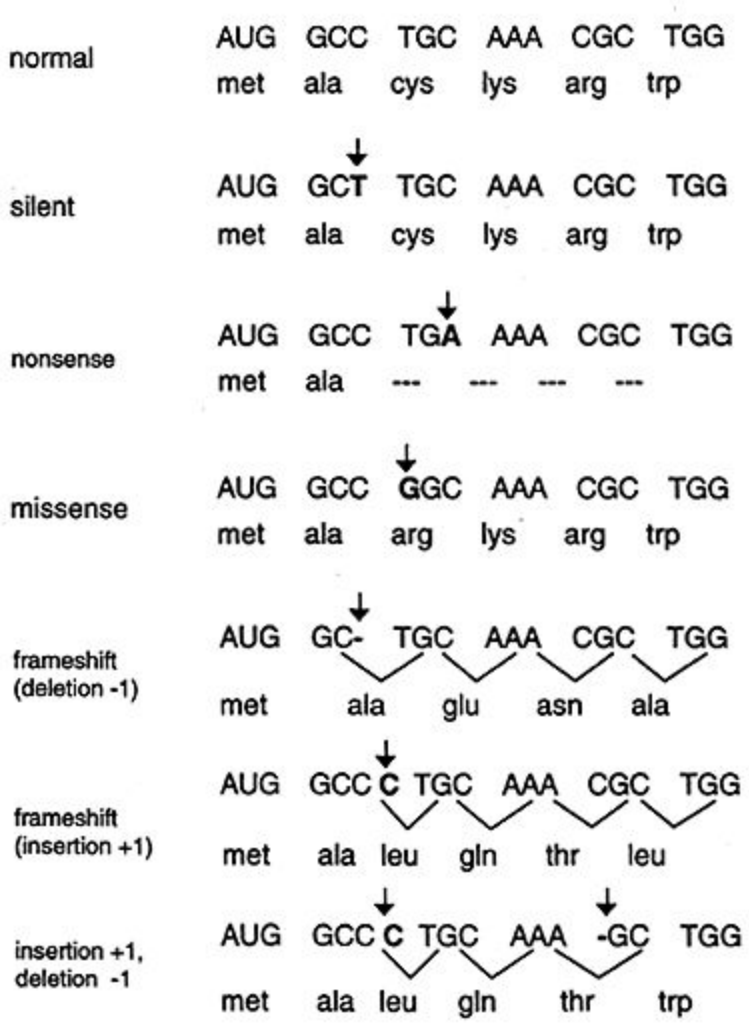
Point mutations that occur in DNA sequences encoding proteins are either Synonymous (silent) or non-synonymous (replacement) mutations. Non- synonymous can be missense or nonsense mutation.

Substitutions do not cause shift in reading frame (non-frame-shift mutation).

Silent: Generally, if a base substitution occurs in the third position of the codon there is a good chance that a synonymous codon will be generated. Thus the amino acid sequence encoded by the gene is not changed and the mutation is said to be silent.

Missence: When base substitution results in the generation of a codon that specifies a different amino acid and hence leads to a different polypeptide sequence

Nonsense: When a base substitution results in a stop codon ultimately truncating translation and most likely leading to a nonfunctional protein.



2. Deletions

A deletion, resulting in a frameshift, results when one or more base pairs are lost from the DNA (see Figure above). If one or two bases are deleted the translational frame is altered resulting in a garbled message and nonfunctional product. A deletion of three or more bases leave the reading frame intact. A deletion of one or more codons results in a protein missing one or more amino acids. This may be deleterious (or not).

3. Insertions

The insertion of additional base pairs may lead to frameshifts depending on whether or not multiples of three base pairs are inserted. Combinations of insertions and deletions leading to a variety of outcomes are also possible. This can again be deleterious (or not).

Definitions and Explanations are taken from the following sources:

National Human Genome Research Institute

NCBI Genetics Glossary

Biology Dictionary

Biology Online Dictionary

Scitable by Nature Education

Merriam Webster Dictionary

Understanding Evolution: Berkeley.edu

http://www2.csudh.edu/nsturm/CHEMXL153/DNAMutationRepair.htm

[20] A testable genotype-phenotype map: Modeling evolution of RNA molecules. In: Lässig, M. and Valleriani, A., editors, Biological Evolution and Statistical Physics, pp. 56–83. Springer-Verlag, Berlin, 2002.

You can study Mutation from the following-

Campbell 9th Edition:
Chapter 17. Page 344-346
Chapter 21. Page 438-439

Life
Chapter 12. Page 250-255
Chapter 9. Page 182-184