

## Genes and Mutations

1. Define:

**Genetics** – Genetics may be defined as the science or study of heredity, and is concerned with the physical and chemical properties of the genetic material (DNA or RNA), how this material is transmitted from one generation to the next, and how the information it contains is expressed.

**Phenotype** – The phenotype of an organism is the observed characteristics of that organism or in the case of bacteria, the observed characteristics of a population of organisms. For example, the phenotype of *E. coli* colonies is variable depending upon the type of medium used (colonies are pink on MAC, yellow on T-7, and dark with a metallic green sheen on EMB ).

**Point mutation** – A point mutation is one involving a change in a single nucleotide (base) within a DNA strand or in the RNA of some viruses. Point mutations may be categorized as substitutions, additions, or deletions.

**Translocation** – A translocation (transposition) is a type of non-point mutation involving the movement of a segment of DNA (one or more genes in length) from one location (locus) to a new location within a chromosome or plasmid, between chromosomes, or between chromosomes and plasmids. These may be caused by transposable elements (transposons) or by crossing over.

**Mutagenic agent (mutagen)** – A mutagen is a factor (chemical or physical) that increases the mutation rate of nucleic acids. Base analogs, alkylating agents and ultra-violet light are examples of mutagens.

2. Genetics/ DNA or RNA in some viruses

3. Locus

4. Gene/ split gene

5. Genome

6. Genotype/ environment

7. Genome/ phenotype **Note** – For this class, genomic DNA is restricted to the chromosome(s) and does not include plasmid, mitochondrial or chloroplast DNA.

8. Phenotype/ environment the organism is exposed to./ The genetic composition of an organism may not be expressed for a variety of reasons. In bacteria, many genes are associated with inducible or repressible operons. If the repressor proteins associated with these operons are active, transcription is blocked and the genetic information cannot be expressed. In diploid organisms certain genes are not expressed because they are recessive. The characteristics they encode are masked by characteristics encoded by dominant genes.

9. Mutation
10. Mutation/ evolution
11. One per 100 million copies of the DNA present/ at least one. The m-concentration for a bacterial culture is usually around  $10^{-9}$  cells/ml of medium (that's 1 billion cells/ml).
12. Substitutions/ The substitution of one base for another within a gene may or may not change the amino acid sequence the gene is encoding because the genetic code is redundant (several different codons encode the same amino acid). A single substitution occurring in the middle part of a gene will influence only one codon, so will potentially change only one amino acid. The addition or deletion of a single base within a gene will result in a frame shift (shift in the codon reading frame) during translation, so will most likely change all of the amino acids being encoded beyond the mutation point. It is unlikely that such a random assortment of amino acids will form a functional protein, although it is potentially possible.
13. Translocations (or transpositions)/ transposons (transposable elements)
14. The addition or the deletion of individual bases within/ codons/ ultra violet light
15. Mutagenic agent or mutagen
16. Base analogs
17. Base analogs/ substitution
18. Missense – The second purine in the sequence shown is Guanine, so the second codon would encode Aspartic acid. If a substitution caused the guanine to be replaced by a pyrimidine (cytosine or thymine), the new codon would encode glutamic acid.
19. Substitutions/ Ultra violet light
20. Ultra violet (UV) light/ deletion
21. Transposons or transposable elements/ translocations (transpositions)
22. Transposons or transposable elements/ transposase
23. Thymine-thymine dimers/ recognizing and breaking the bonds between the thymine molecules thus removing the dimer.