Genetic Variation

Genetic variation *is important for the survival of a species. Greater variation within a species increases the chances of survival for the species because the variant organisms within the species are able to respond differently to the environmental changes.*

Overview

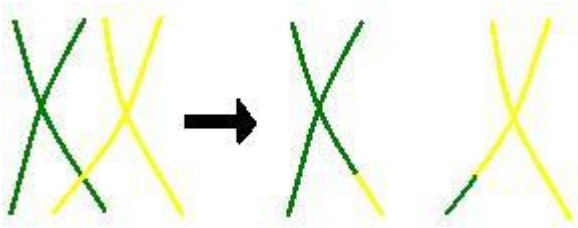
Sexual reproduction results in a great variety of possible gene combinations that can be produced in the offspring of any two parents. This variety is due to the sorting and recombination of genes that occurs during meiosis. Since each offspring receives a different combination of alleles from the parent organisms, both genotypic and phenotypic diversity results.

Although asexual reproduction is faster and requires less energy than sexual reproduction, offspring are almost always genetically identical to their parents; there is little to no genetic variation.

Meiosis & Genetic Diversity

Meiosis is the form of cell division by which unique gametes (sex cells) are produced. Each diploid parent cell divides twice during meiosis and produces four haploid daughter cells.

During meiosis, the process of **crossing over** results in new combinations of alleles due to the fact that genes are located on separate chromosomes. When crossing over occurs, different parts of chromosomes are exchanged, meaning that genes (and their alleles) are transferred to new chromosomes. When meiosis separates these chromosomes, the new combination of alleles is transferred to the offspring, resulting in a new combination of traits.



Errors made during crossing over can also result in genetic variation. For example, genes, or even entire chromosomes, can be deleted or duplicated, resulting in even more genotypic possibilities.

Transposons

Transposons, or **jumping genes**, can independently replicate and insert new copies of themselves within an organism's genome. Since transposons can replicate and insert themselves several times during the formation of gametes, these genes can cause a large amount of genetic variation.

Fertilization & Genetic Variation

Since parents are genetically different, when their gametes and chromosomes are united during **fertilization**, genetic variation results. No two organisms produced by sexual reproduction will share an identical genome unless they come from the same fertilized egg.

Chromosomal Mutations and Genetic Variability

Though all types of mutations contribute to genetic variability, only chromosomal mutations are covered in this section.

Deletion

Many diseases can be caused by chromosomal abnormalities. For example, the disease Cri du chat, which causes children to have a cat-like cry, is caused by the deletion of part of chromosome 5.

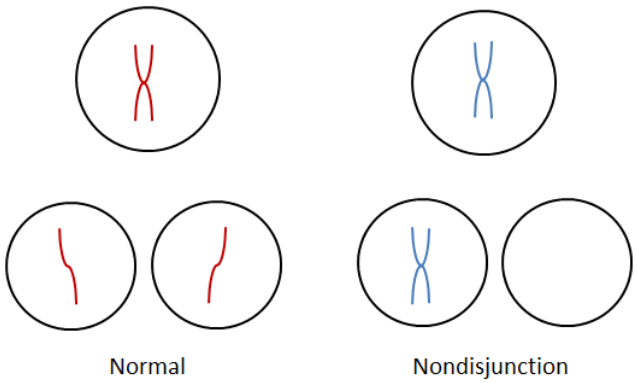
Translocation

Chromosome translocation occurs when material is exchanged between two chromosomes, or part of one chromosome becomes fused onto another chromosome.

Some human disorders are caused by chromosome translocation, such as cancer, infertility, and translocation Down syndrome. Translocation Down syndrome, for example, occurs because a section of chromosome 21 becomes fused onto another chromosome. It accounts for less than 5% of the total cases of Down syndrome reported.

Nondisjunction

Nondisjunction occurs when chromosomes do not separate correctly during cell division. The resulting daughter cells will either be missing or have extra copies of chromosomes. The picture below compares normal cell division to cell division in which nondisjunction has occurred.



Notice the abnormal number of chromosomes in the daughter cells on the right. One daughter cell has 2 copies of the chromosome whereas the second cell is missing a copy.

Some chromosomal abnormalities occur in the sex chromosomes. Trisomy X, for example, is a type of chromosomal abnormality in which a female has three X chromosomes (XXX). Females with trisomy X often have learning disabilities and may be taller than normal, but they do typically undergo normal sexual development and are able to conceive children. Turner syndrome, on the other hand, is caused by a female only receiving one complete X chromosome and can result in infertility and other health problems. Both Trisomy X and Turner syndrome are examples of genetic diseases caused by nondisjunction.

Inversion

Inversion occurs when part of a chromosome breaks off and reattaches in the reverse direction.

Mutations

*A change in the sequence of nucleotides in an organism's genetic material is known as a **mutation**.*

Background

Mutations can occur randomly during DNA replication when base pairs are added, deleted, or substituted, or they can be caused by environmental factors, such as overexposure to radiation or toxic chemicals.

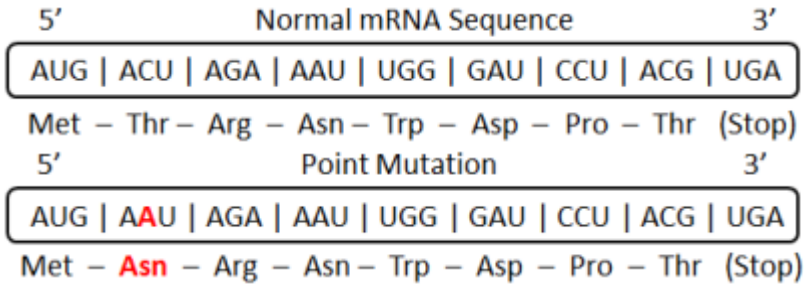
Mutations can occur in any cell in an organism's body, but they will be passed on to an organism's offspring only if the mutation occurs in the sex cells (gametes) of the organism. If a mutation occurs in the body cells of an organism, such as skin cells, bone cells, muscle cells, and nerve cells, it cannot be passed on to potential offspring. These mutations can only be passed on to the mutant body cells' daughter cells (cells that are produced when the mutant cell divides).

Sometimes mutations create changes in an organism's appearance or behavior. Some of these changes may be beneficial; other changes may be detrimental. And sometimes mutations have no effect on an organism at all.

Point Mutations

A **point mutation** is a mutation in a single base pair in a strand of DNA. Some genetic diseases, like cystic fibrosis, color blindness, hemophilia, and sickle cell anemia, can occur as a result of a point mutation.

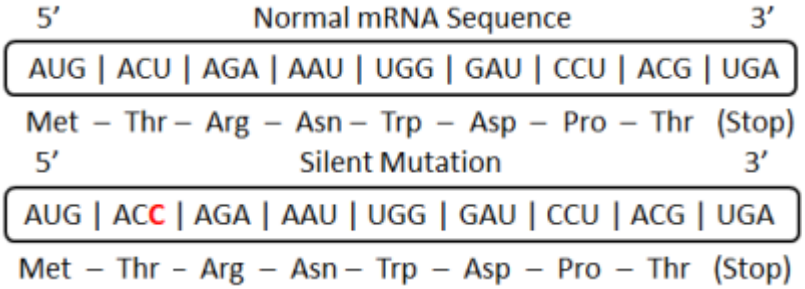
The image below demonstrates an example of a point mutation. Notice how the substitution of a single base pair results in a different amino acid. The remainder of the protein sequence remains unchanged.



Silent Mutation

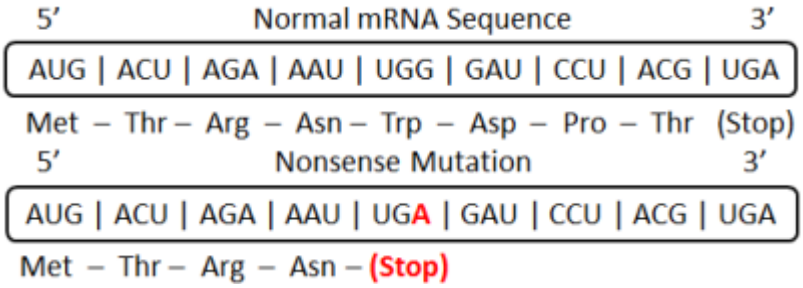
A **silent mutation** is a specific type of point mutation. Because many amino acids have more than one codon, it is possible for a mutation in a single base pair to have no effect on the polypeptide sequence.

The example below shows how some mutations are silent, meaning they have no effect on the translated protein. Though the second codon, or triplet code, has changed, the amino acid sequence of the resulting protein is the same as that coded for by the normal mRNA strand.



Nonsense Mutation

A **nonsense mutation** changes an amino acid codon into a stop codon. This causes the normal polypeptide sequence to be shorter.

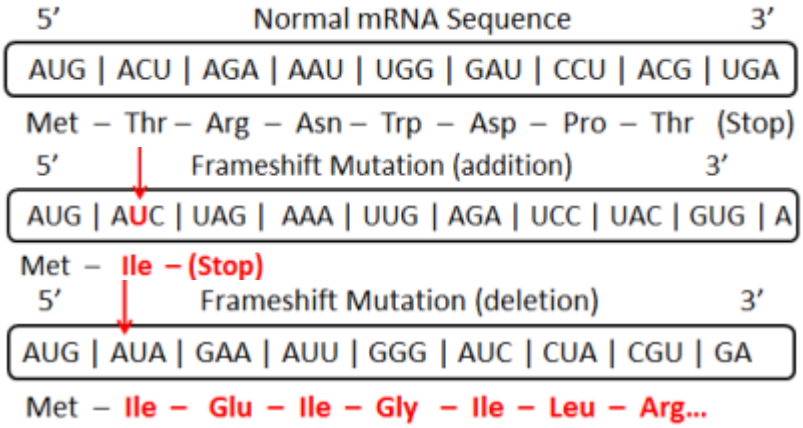


The image above shows how proteins are truncated by nonsense mutations.

Frameshift Mutations

A **frameshift mutation** is a mutation that causes the reading frame of a codon sequence to be shifted. Since a codon is a sequence of three nucleotides that code for a specific amino acid, any insertion or deletion of nucleotide base pairs that are not in multiples of three will cause a frame shift mutation. Insertions or deletions in multiples of three will cause a protein to be shorter or longer than normal, but the entire sequence of the amino acids will not be shifted.

The image below shows how insertions and deletions affect the polypeptide sequence.



Chromosome Translocation

Chromosome translocation is caused when material is exchanged between two chromosomes or part of one chromosome becomes fused onto another chromosome. Some human disorders, such as cancer, infertility, and translocation Down syndrome, are caused by chromosome translocation.