



Mutations

IST-2.E.1

Changes in genotype can result in changes in phenotype—

- a. The function and amount of gene products determine the phenotype of organisms.
 - i. The normal function of the genes and gene products collectively comprises the normal function of organisms.
 - ii. Disruptions in genes and gene products cause new phenotypes.



Mutations

IST-2.E.2

Alterations in a DNA sequence can lead to changes in the type or amount of the protein produced and the consequent phenotype. DNA mutations can be positive, negative, or neutral based on the effect or the lack of effect they have on the resulting nucleic acid or protein and the phenotypes that are conferred by the protein.



Mutations

IST-4.A.1

Errors in DNA replication or DNA repair mechanisms, and external factors, including radiation and reactive chemicals, can cause random mutations in the DNA—

- a. Whether a mutation is detrimental, beneficial, or neutral depends on the environmental context.
- b. Mutations are the primary source of genetic variation.



Mutations

IST-4.A.2

Errors in mitosis or meiosis can result in changes in phenotype—

- a. Changes in chromosome number often result in new phenotypes, including sterility caused by triploidy, and increased vigor of other polyploids.
- b. Changes in chromosome number often result in human disorders with developmental limitations, including Down syndrome/ Trisomy 21 and Turner syndrome.



Mutations

IST-4.B.1

Changes in genotype may affect phenotypes that are subject to natural selection. Genetic changes that enhance survival and reproduction can be selected for by environmental conditions—

- a. The horizontal acquisitions of genetic information primarily in prokaryotes via transformation (uptake of naked DNA), transduction (viral transmission of genetic information), conjugation (cell-to-cell transfer of DNA), and transposition (movement of DNA segments within and between DNA molecules) increase variation.**



Mutations

IST-4.B.1

Changes in genotype may affect phenotypes that are subject to natural selection. Genetic changes that enhance survival and reproduction can be selected for by environmental conditions—

- b. Related viruses can combine/recombine genetic information if they infect the same host cell.**
- c. Reproduction processes that increase genetic variation are evolutionarily conserved and are shared by various organisms.**

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What are mutations?

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What are mutations?



**Changes in the DNA sequence or
the DNA quantity in a cell.**

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Which mutation results in a change in the amino acid?

- A. Frameshift**
- B. Missense**
- C. Nonsense**
- D. Silent**

Which mutation results in a change in the amino acid?

B. Missense



A missense mutation is a substitution point mutation. One nucleotide is substituted with another. This substitution causes **ONE amino acid to be substituted for **ANOTHER** amino acid.**

This is similar to a word being substituted in a sentence.

I took my dog for a walk.

I took my fish for a walk.



Which mutation results in no change in the amino acid?

- A. Frameshift**
- B. Missense**
- C. Nonsense**
- D. Silent**



Which mutation results in no change in the amino acid?

D. Silent

A silent mutation is a substitution point mutation. One nucleotide is substituted with another. This substitution causes ONE amino acid to be substituted for the SAME amino acid.

Since you have the same amino acid, it is not obvious that there has been a mutation so it is SILENT.



**Which mutation results from
+/- one nucleotide base?**

- A. Frameshift**
- B. Missense**
- C. Nonsense**
- D. Silent**

Which mutation results from +/- one nucleotide base?

A. Frameshift



If ONE nucleotide has been inserted or deleted, this will shift the reading frame. Recall, the reading frame is the group of triplets read as codons. If there is a shift by 1 or 2, the ribosome will be pairing the wrong codons. This will cause a change to all amino acids downstream from the insertion or deletion.



Frameshift mutation in what area causes most damage?

- A. Intron**
- B. Start of exon**
- C. Middle of exon**
- D. End of exon**

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Frameshift mutation in what area causes most damage?

B. Start of exon



If the frameshift mutation occurs at the start of the expressed exons, all of the codons downstream will be affected. This will cause the most damage of the options.

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How does a heterozygote and homozygous dominant produce same phenotype?

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How does a heterozygote and homozygous dominant produce same phenotype?

The single dominant allele produces enough enzyme/protein products to produce the desired phenotypic result.



What is the primary source of genetic variation?

- A. Crossing over**
- B. Independent assortment**
- C. Mutations**
- D. Random fertilization**

What is the primary source of genetic variation?

C. Mutations



Mutations will add NEW variation to the population. This will increase genetic variation.



Which of the following is the cause of Trisomy 21?

- A. Duplication**
- B. Frameshift mutation**
- C. Nondisjunction**
- D. Point mutation**



Which of the following is the cause of Trisomy 21?

C. Nondisjunction

Individuals with Trisomy 21 has an additional chromosome 21. This results from nondisjunction. Nondisjunction results from homologous chromosomes or sister chromatids not separating during anaphase I or II.

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What phase of meiosis is cause of nondisjunction? Describe nondisjunction.

**What phase of meiosis is cause of nondisjunction?
Describe nondisjunction.**



Anaphase I or II

Nondisjunction involves the chromatids or homologous chromosomes not separating during anaphase. This results in a gamete with an additional chromosome or missing a chromosome.

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**Triploid organisms are sterile
(unable to reproduce)**

- A. True**
- B. False**

Triploid organisms are sterile (unable to reproduce)

A. True



Triploid organisms lack homologous chromosomes. They have THREE of each chromosome thus cannot pair during prophase I, line up during metaphase I, nor segregate evenly during anaphase I.

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**Why are triploid organisms
sterile?**

Why are triploid organisms sterile?



Unable for homologous chromosomes to pair up during meiosis.

(Triploid is three sets, how can you get paired off with three)



Which enzyme has proofreading capabilities?

- A. DNA Polymerase**
- B. Helicase**
- C. Ligase**
- D. Topoisomerase**

Which enzyme has proofreading capabilities?

A. DNA Polymerase



DNA polymerase is the enzyme responsible for DNA replication. A $3' \rightarrow 5'$ proofreading exonuclease domain is intrinsic to most DNA polymerases. It allows the enzyme to check each nucleotide during DNA synthesis and excise mismatched nucleotides in the $3'$ to $5'$ direction.



**Which is responsible for halting
DNA replication because of
error?**

- A. DNA Polymerase**
- B. Helicase**
- C. p53**
- D. Ras**

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Which is responsible for halting DNA replication because of error?

C. **p53**



p53 is a tumor suppressor gene/protein. **p53** is a transcription factor that activates many genes involved in essential maintenance of genetic stability. This gene will inhibit the cell cycle in the presence of DNA damage.



In disease with too many copies of genes (Huntington's)...

- A. Deletion**
- B. Duplication**
- C. Inversion**
- D. Translocation**

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In disease with too many
copies of genes
(Huntington's)...

B. Duplication



Duplication results when a gene
is duplicated resulting in
multiple copies of the gene.

Huntington's Disease is an
autosomal dominant trait that
results from duplication.

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**Involved in cri-du-chat where
chromosome is shorter**

- A. Deletion**
- B. Duplication**
- C. Inversion**
- D. Translocation**

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Involved in cri-du-chat
where chromosome is
shorter

A. Deletion



Deletion results in a region of
the chromosome being removed.

Cri-du-chat ("cry of the cat")
is a genetic disorder resulting
from a piece of chromosome 5 is
missing.



**Involved in down syndrome
without nondisjunction**

- A. Deletion**
- B. Duplication**
- C. Inversion**
- D. Translocation**

**Involved in down
syndrome without
nondisjunction**

D. Translocation



**Translocation results from
chromosome segments changing
positions.**

**This sometimes takes place in
Down Syndrome and Patau
syndrome resulting an additional
chromosome in the offspring.**

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What is horizontal transmission of traits?

**What is horizontal
transmission of traits?**



**Transmission of traits NOT from
parent-progeny**



Cell-to-cell transmission?

- A. Conjugation**
- B. Transduction**
- C. Transformation**
- D. Transposition**



Cell-to-cell transmission?

A. Conjugation

Prokaryotes are able to undergo cell-to-cell transmission by passing genetic information through sex pili in a process called conjugation.



Movement of DNA between or within segments?

- A. Conjugation**
- B. Transduction**
- C. Transformation**
- D. Transposition**

**Movement of DNA
between or within
segments**

D. Transposition



Transposition involves the DNA moving to a new location so it transfers position when it undergoes transposition.



Viral transmission of traits

- A. Conjugation**
- B. Transduction**
- C. Transformation**
- D. Transposition**

**Viral transmission of
traits**

B. Transduction



**As seen during the Hershey and
Chase experiment,
bacteriophages traditionally will
transduce their genetic
information into the host
nucleus.**



Uptake naked DNA

- A. Conjugation**
- B. Transduction**
- C. Transformation**
- D. Transposition**

Uptake naked DNA

C. Transformation



During the transformation experiment, bacteria will take up genetic information (plasmids) from the environment to become antibiotic resistant bacteria/green glowing bacteria.

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What type of virus is able to insert its genome into host genome?



What type of virus is able to insert its genome into host genome?

Retrovirus

Viruses like HIV have an RNA genome. They can use reverse transcriptase to make a complement DNA strand then inserts in genome to become a pro virus.



What is the genotype?

- A. Genes in a genetic library for transformation**
- B. Physical characteristics due to gene expression**
- C. Specific genes an organism has (ex: heterozygous)**
- D. The type of genetic material the organism has**

What is the genotype?

C. Specific genes an organism has (ex: heterozygous)



Genotype is the genetic information within the DNA. This is traditionally discussed as homozygous dominant, heterozygous, or homozygous recessive.

For example, Pp.



What is the phenotype?

- A. Genes in an organism causing physical traits**
- B. Good genes that increase reproductive fitness**
- C. Physical characteristics of an organism**
- D. Showing types of genes (ex. homozygous dominant)**

What is the phenotype?

**C. Physical characteristics
of an organism**



**Phenotype is the physical
characteristics that result from
the genotype.**

For example, purple flowers.



Mutation resulting in a different amino acid

- A. Chromosomal**
- B. Missense**
- C. Nonsense**
- D. Silent**

**Mutation resulting in a
different amino acid**

B. Missense



**Missense mutation results in a
DIFFERENT amino acid. This is
due to a single nucleotide
substitution.**

**Example: Sickle Cell,
Cystic Fibrosis**



Mutation resulting in the same amino acid

- A. Chromosomal**
- B. Missense**
- C. Nonsense**
- D. Silent**

**Mutation resulting in
the same amino acid**

D. Silent



**Silent mutations have no
observable difference in the
amino acid sequence. The codon
before and after will code for
the same amino acid.**



**Mutation resulting in a stop
codon (premature stop)**

- A. Chromosomal**
- B. Missense**
- C. Nonsense**
- D. Silent**

Mutation resulting in a stop codon (premature stop)



C. Nonsense

A nonsense mutation results in a stop codon for a substitution of codon that codes for an amino acid which leads to a shorter protein product.



If a nucleotide base pair was deleted in an intron, what would be the result?

- A. Frameshift mutation**
- B. Longer protein product**
- C. No change in synthesized protein**
- D. Shorter protein product**

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If a nucleotide base pair was deleted in an intron, what would be the result?

C. No change in synthesized protein



Recall, the introns are removed during post-transcriptional modification. If a mutation takes place in the intron, then it will not be observed as the introns are removed.



What happens if a nucleotide base pair inserted into an exon?

- A. Frameshift mutation**
- B. Longer protein product**
- C. No change in synthesized protein**
- D. Shorter protein product**

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What happens if a nucleotide base pair inserted into an exon?

A. Frameshift mutation



If a single nucleotide base pair is inserted, this will shift the reading frame resulting in a frameshift mutation.



What is the result of a mutation?

- A. Gain new trait**
- B. Loss of trait**
- C. No change in trait**
- D. All of the above**

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What is the result of a mutation?

**D. All of the above
(Gain, Loss, or No change
in trait)**



The mutation could cause a different ligand to bind which could result in a new function.

The mutation could inhibit binding of a ligand which could result in the absence of a function.

The mutation could have no effect on the binding site or on the folding of the protein resulting in no change in function.



Which results in new genetic variability?

- A. Crossing over**
- B. Independent assortment**
- C. Mutations**
- D. Random fertilization**

Which results in new genetic variability?

C. Mutations



Mutations results in new traits.

If there are more traits available, the genetic variability has increased.



What does it mean to be triploid?

- A. Three units of chromosomes**
- B. Three sets of a chromosome**
- C. Three sets of organisms born**
- D. Three sets of population**

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What does it mean to be triploid?

A. Three units of chromosomes



The prefix of "tri" means **THREE**.

The suffix of "ploid" means having **CHROMOSOME SETS**

Triploid has **THREE SETS** of **CHROMOSOMES**

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**Why are triploid organisms
sterile?**

Why are triploid organisms sterile?



They are unable to pair up the homologous chromosomes during meiosis to synthesize the gametes

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**Natural selection acts on
genotype.**

- A. True**
- B. False**

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Natural selection acts on genotype.

B. False



Natural selection cannot act on what it cannot observe. Natural selection acts on phenotype, but it does affect the genotype resulting is an increase in allele frequency.

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**Natural selection affects
genotype.**

A. True

B. False

Natural selection affects genotype.

A. True



Natural selection will select individuals with favorable traits that align with the environment. As these traits are selected, the allele frequency will change which affects the genotype of the organisms.



Taking up DNA from the environment

- A. Conjugation**
- B. Transduction**
- C. Transformation**
- D. Transposition**

Taking up DNA from the environment

C. Transformation



Transformation was observed in the Griffith experiment. The living R cells are transformed into living S cells as they took up DNA from the heat-killed S cells.



**Insertion of DNA by
bacteriophage**

- A. Conjugation**
- B. Transduction**
- C. Transformation**
- D. Transposition**

**Insertion of DNA by
bacteriophage**

B. Transduction



Transduction is the process of a bacteriophage injecting the generic information which transduces the host cell.



DNA moves to new position in genome

- A. Conjugation**
- B. Transduction**
- C. Transformation**
- D. Transposition**

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**DNA moves to new
position in genome**

D. Transposition



**Genetic information is able to be
transferred to another position
through transposition.**



**Genetic exchange between
bacteria**

- A. Conjugation**
- B. Transduction**
- C. Transformation**
- D. Transposition**

**Genetic exchange between
bacteria**

A. Conjugation



Conjugation is the attachment of two prokaryotes then transferring genetic information between the two which increases the genetic variability.