6.7



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.

6.7



Mutations

<u>IST-2.E.2</u>

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.

6.7



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.

6.7



Mutations

<u>IST-4.A.2</u>

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

 BDEMO BDEMO and Turner syndrome.

6.7



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.

6.7



Mutations

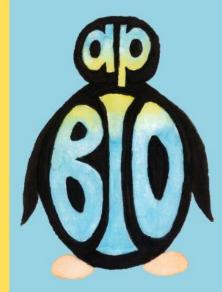
IST-4.B.1

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.

TOPIC

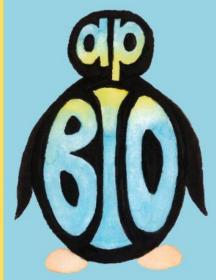
6.7



What are mutations?

TOPIC.

6.7

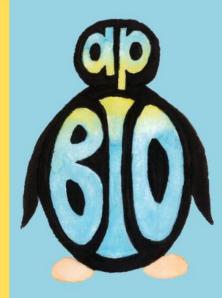


What are mutations?

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

TOPIC

6.7



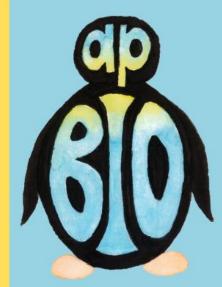
Which mutation results in a change in the amino acid?

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

TOPIC

6.7

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.

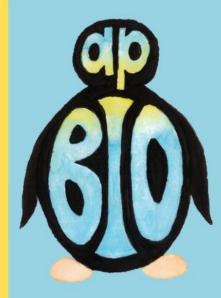
I took my dog for a walk.

I took my fish for a walk.

@APBIOPENGUINS

TOPIC

6.7



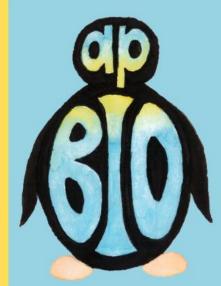
Which mutation results in no change in the amino acid?

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

TOPIC

6.7

Which mutation results in no change in the amino acid?



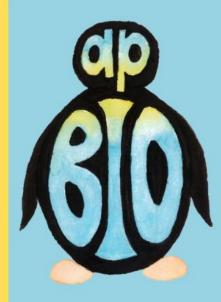
D. Silent

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.

TOPIC

6.7



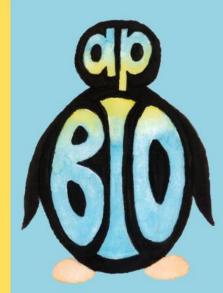
Which mutation results from +/- one nucleotide base?

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

TOPIC

6.7

Which mutation results from +/- one nucleotide base?

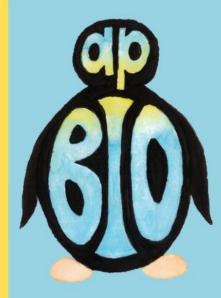


A. Frameshift

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.

TOPIC

6.7



Frameshift mutation in what area causes most damage?

A. Intron

B. Start of exon

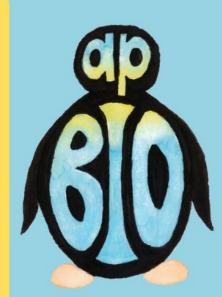
C. Middle of exon

D. End of exon

TOPIC

6.7



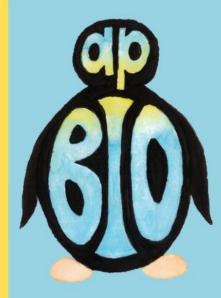


B. Start of exon

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.

TOPIC

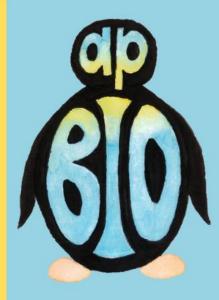
6.7



How does a heterozygote and homozygous dominant produce same phenotype?

TOPIC

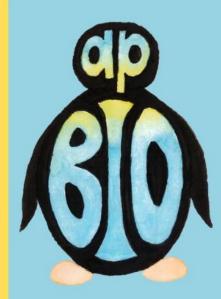
6.7



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.

6.7



What is the primary source of genetic variation?

A. Crossing over

B. Independent assortment

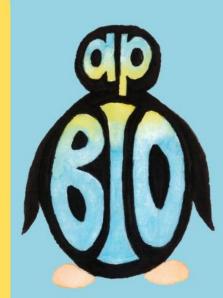
C. Mutations

D. Random fertilization

TOPIC

6.7

What is the primary source of genetic variation?



C. Mutations

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

TOPIC

6.7



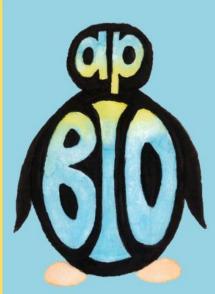
Which of the following is the cause of Trisomy 21?

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

TOPIC

6.7

Which of the following is the cause of Trisomy 21?



C. Nondisjunction

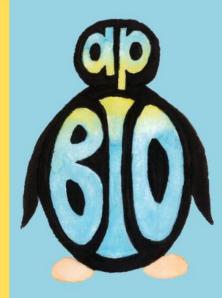
Individuals with Trisomy 21 has an additional chromosome 21.

This results from nondisjunction.

Nondisjunction results from homologous chromosomes or sister chromatids not separating during anaphase I or II.

TOPIC

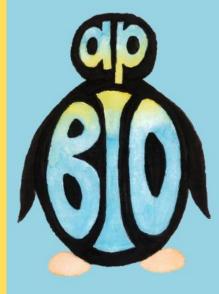
6.7



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

TOPIC

6.7



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.

TOPIC

6.7



Triploid organisms are sterile (unable to reproduce)

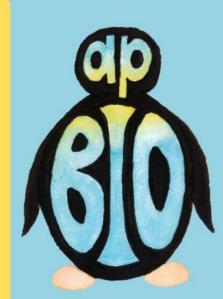
A. True

B. False

TOPIC

6.7

Triploid organisms are sterile (unable to reproduce)

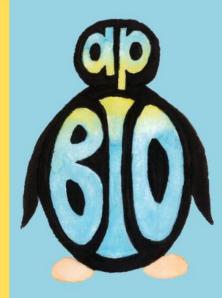


A. True

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.

TOPIC

6.7



Why are triploid organisms sterile?

TOPIC

6.7



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)

TOPIC

6.7



Which enzyme has proofreading capabilities?

A. DNA Polymerase

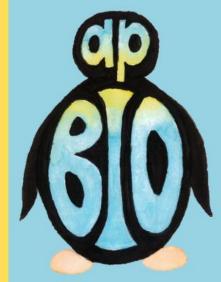
B. Helicase

C. Ligase

D. Topoisomerase

TOPIC

6.7



Which enzyme has proofreading capabilities?

A. DNA Polymerase

PNA polymerase is the enzyme responsible for DNA replication. A 3'→ 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.

TOPIC

6.7



Which is responsible for halting DNA replication because of error?

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

@APBIOPENGUINS

TOPIC

6.7

Which is responsible for halting DNA replication because of error?

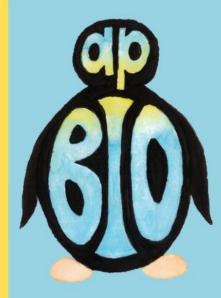


C. p53

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.

TOPIC

6.7



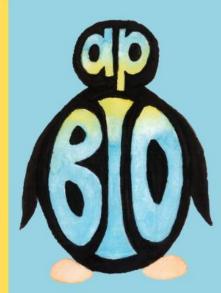
In disease with too many copies of genes (Huntington's)...

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

TOPIC

6.7

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



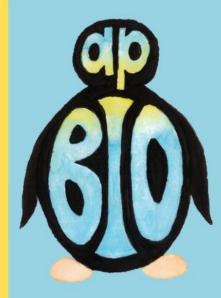
B. Duplication

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.

TOPIC

6.7



Involved in cri-du-chat where chromosome is shorter

A. Deletion

B. Duplication

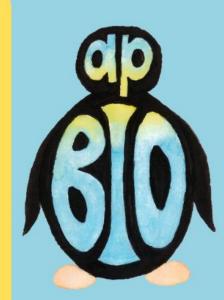
C. Inversion

D. Translocation

TOPIC

6.7

Involved in cri-du-chat where chromosome is shorter



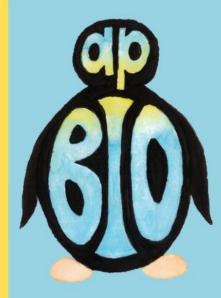
A. Deletion

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.

TOPIC

6.7



Involved in down syndrome without nondisjunction

A. Deletion

B. Duplication

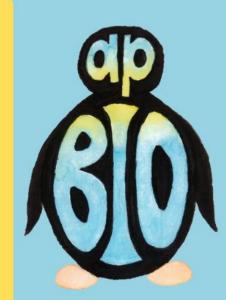
C. Inversion

D. Translocation

TOPIC

6.7

Involved in down syndrome without nondisjunction



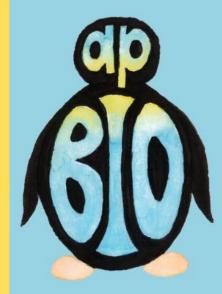
D. Translocation

Translocation results from chromosome segments changing positions.

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

TOPIC

6.7



What is horizontal transmission of traits?

TOPIC

6.7



What is horizontal transmission of traits?

Transmission of traits NOT from parent-progeny

TOPIC

6.7

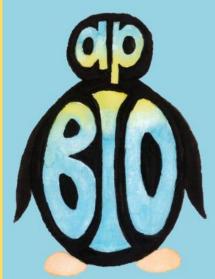


Cell-to-cell transmission?

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

TOPIC

6.7



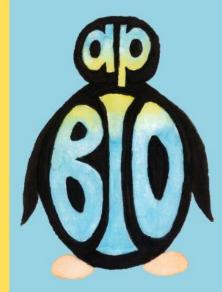
Cell-to-cell transmission?

A. Conjugation

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

TOPIC

6.7



Movement of DNA between or within segments?

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

TOPIC

6.7



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.

TOPIC

6.7

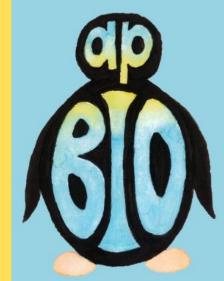


Viral transmission of traits

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

TOPIC

6.7



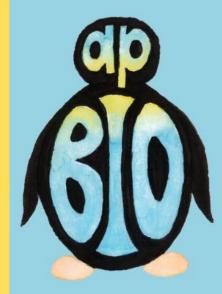
Viral transmission of traits

B. Transduction

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

TOPIC

6.7

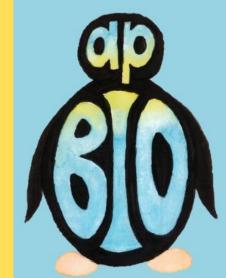


Uptake naked DNA

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

TOPIC

6.7



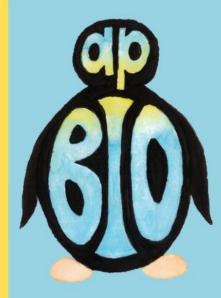
Uptake naked DNA

C. Transformation

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

TOPIC

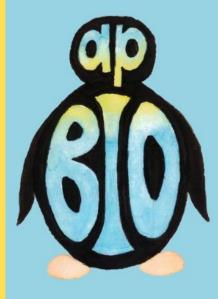
6.7



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

TOPIC

6.7



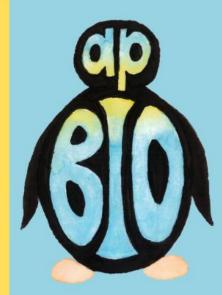
What type of virus is able to insert its genome into host genome?

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.

TOPIC

6.7



What is the genotype?

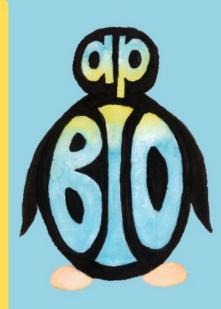
- A. Genes in a genetic library for transformation
- B. Physical characteristics due to gene expression
- C. Specific genes an organism has (ex: heterozygous)
- D. The type of genetic material the organism has

TOPIC

6.7

What is the genotype?

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

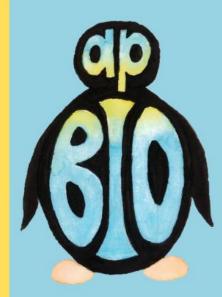


Genotype is the genetic information within the DNA. This is traditionally discussed as homozygous dominant, heterozygous, or homozygous recessive.

For example, Pp.

TOPIC

6.7

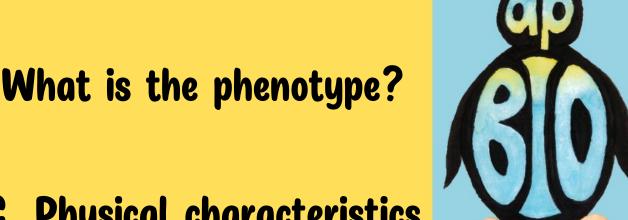


What is the phenotype?

- 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)

TOPIC

6.7



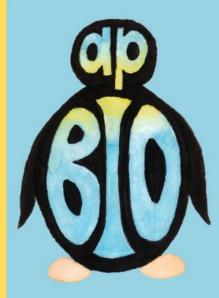
C. Physical characteristics of an organism

Phenotype is the physical characteristics that result from the genotype.

For example, purple flowers.

TOPIC

6.7



Mutation resulting in a different amino acid

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

TOPIC

6.7

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

TOPIC

6.7



Mutation resulting in the same amino acid

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

TOPIC

6.7



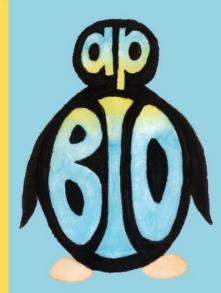
Mutation resulting in the same amino acid

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.

TOPIC

6.7



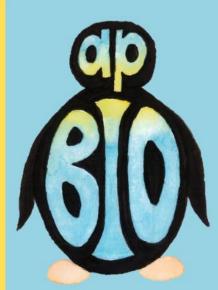
Mutation resulting in a stop codon (premature stop)

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

TOPIC

6.7

Mutation resulting in a stop codon (premature stop)

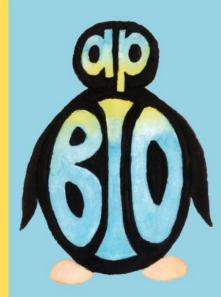


C. Nonsense

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.

TOPIC

6.7



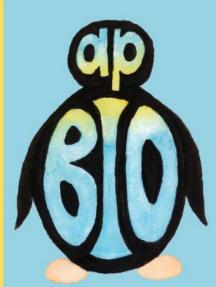
If a nucleotide base pair was deleted in an intron, what would be the result?

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

TOPIC

6.7

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

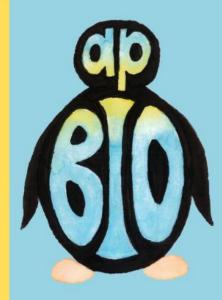


C. No change in synthesized protein

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.

TOPIC

6.7



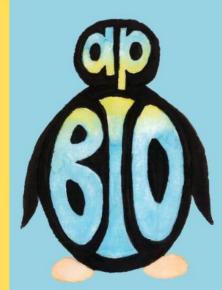
What happens if a nucleotide base pair inserted into an exon?

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

TOPIC

6.7

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.

TOPIC

6.7



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

TOPIC

6.7

What is the result of a mutation?

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

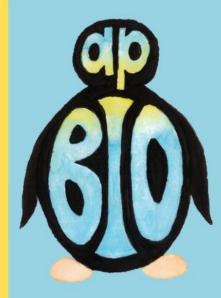
The mutation could cause a different ligand to bind which could result in a new function.

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.

TOPIC

6.7



Which results in new genetic variability?

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

TOPIC

6.7

Which results in new genetic variability?

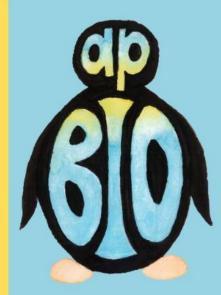
C. Mutations

Mutations results in new traits.

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

TOPIC

6.7



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

TOPIC

6.7

What does it mean to be triploid?

A. Three units of chromosomes

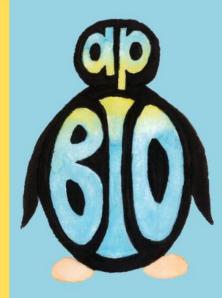
The prefix of "tri" means THREE.

The suffix of "ploid" means having CHROMOSOME SETS

Triploid has THREE SETS of CHROMOSOMES

TOPIC

6.7



Why are triploid organisms sterile?

TOPIC

6.7

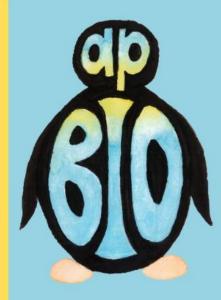


Why are triploid organisms sterile?

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

TOPIC

6.7



Natural selection acts on genotype.

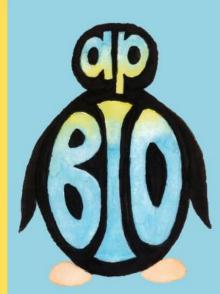
A. True

B. False

TOPIC

6.7

Natural selection acts on genotype.

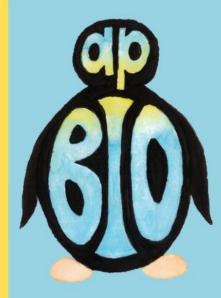


B. False

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.

TOPIC

6.7



Natural selection affects genotype.

A. True

B. False

TOPIC

6.7

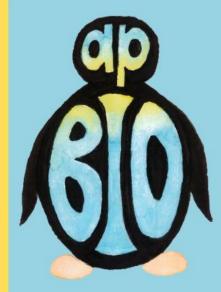
Natural selection affects genotype.

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.

TOPIC

6.7

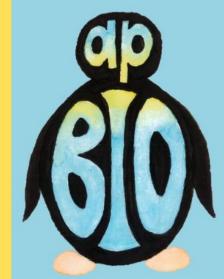


Taking up DNA from the environment

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

TOPIC

6.7



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.

TOPIC

6.7

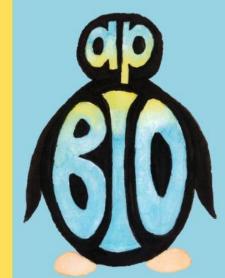


Insertion of DNA by bacteriophage

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

TOPIC

6.7



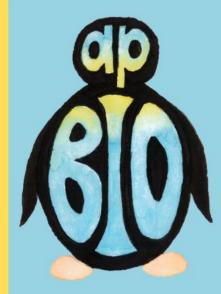
Insertion of DNA by bacteriophage

B. Transduction

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

TOPIC

6.7

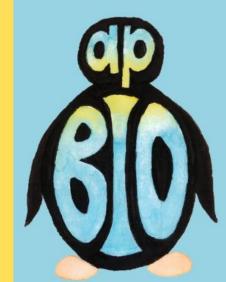


DNA moves to new position in genome

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

TOPIC

6.7



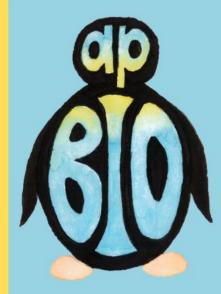
DNA moves to new position in genome

D. Transposition

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

TOPIC

6.7

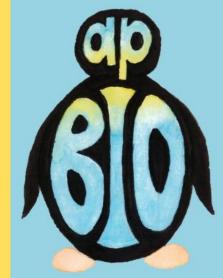


Genetic exchange between bacteria

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

TOPIC

6.7



Genetic exchange between bacteria

A. Conjugation

Conjugation is the attachement of two prokaryotes then transferring genetic infomration between the two which increases the genetic variability.