Mutations: Types & Effects

A Level

Part A Point mutations

Point mutations are changes that happen to one or a few nucleotides. They are most likely to occur during DNA replication (i.e. during S phase of the cell cycle).

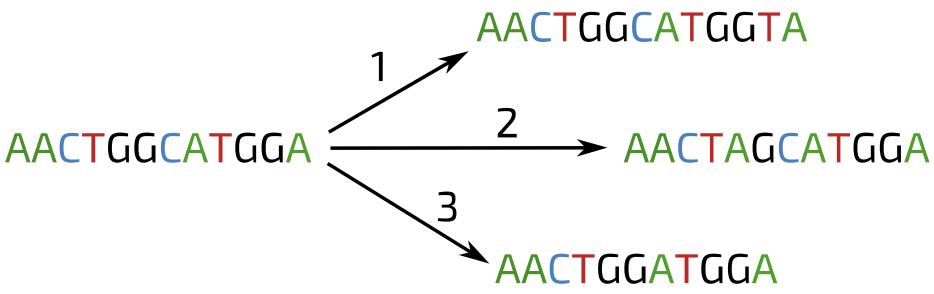
What is the name given to the point mutation in which the correct nucleotide is replaced by an incorrect nucleotide?

What is the name given to the point mutation in which a nucleotide is/a few nucleotides are added into the sequence?

What is the name given to the point mutation in which a nucleotide is/a few nucleotides are lost from the sequence?

Part B Mutation identification

The image below shows a DNA sequence undergoing three different types of point mutation.



Match the mutation to the type.

- Mutation 1: [ ]
- Mutation 2: [ ]
- Mutation 3: [ ]

Items:

insertion	inversion	deletion	translocation	duplication	substitution

Part C Coding sequence mutations

Within a coding region of a gene, a single-nucleotide [ ] is likely to have a large effect on the amino acid sequence, because this type of mutation will change the reading frame (the grouping of the sequence into codons). Therefore, this type of mutation (which can also be called a [ ] mutation) will not only likely change the amino acid at that site, but all subsequent amino acids in the sequence.

Within a coding region of a gene, a single-nucleotide [ ] will only change the amino acid at that site (and it may not change the amino acid, due to the redundancy of the genetic code). However, this type of mutation could produce a premature [ ] in the sequence, which would remove all subsequent amino acids from the amino acid sequence.

A substitution which changes the amino acid is called a [ ] substitution.

A substitution which does not change the amino acid is called a [ ] substitution.

Items:

synonymous | **insertion/deletion** | **stop codon** | **substitution** | **nonsynonymous** | **frameshift**

Part D Chromosomal mutations

Chromosomal mutations are changes that happen to entire regions of a chromosome, and may therefore cause changes to multiple genes. They are most likely to occur during meiosis, as a result of homologous chromosomes not aligning correctly.

What is the name given to the chromosomal mutation in which a region of the chromosome is removed and lost?

What is the name given to the chromosomal mutation in which a region of the chromosome becomes repeated within the same chromosome?

What is the name given to the chromosomal mutation in which a region of the chromosome is removed and becomes part of another chromosome?

What is the name given to the chromosomal mutation in which a region of the chromosome is removed and reinserted in the opposite direction within the same chromosome?

Part E Mutation consequences

Which of the following are possible consequences of a germ cell mutation? Select all that apply.

- [ ] a new advantageous allele of a gene is produced that may be passed on to offspring
- [ ] a new deleterious allele of a gene is produced that may be passed on to offspring
- [ ] a change in the amino acid sequence that the gene codes for
- [ ] no change in the amino acid sequence that the gene codes for
- [ ] a change in the expression of the gene (e.g. the gene is expressed in different contexts, or for different durations)
- [ ] a change to an essential cell process that causes the death of the cell

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Which of the following are possible consequences of a somatic cell (e.g. liver cell) mutation? Select all that apply.

- [ ] a new advantageous allele of a gene is produced that may be passed on to offspring
- [ ] a new deleterious allele of a gene is produced that may be passed on to offspring
- [ ] a change in the amino acid sequence that the gene codes for
- [ ] no change in the amino acid sequence that the gene codes for
- [ ] a change in the expression of the gene (e.g. the gene is expressed in different contexts, or for different durations)
- [ ] a change to an essential cell process that causes the death of the cell

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Genotypes & Phenotypes

A Level

Part A Gene definitions

Match the terms to the definitions in the table below.

Term	Definition
	a region of DNA that codes for a protein
	one of several (two or more) variants of a gene that exist in a population
	the allele(s) that an individual has
	an observable trait that an individual has
	containing two copies of the same allele
	containing two different alleles

Items:

gene | recessive | phenotype | allosome | autosome | allele | homozygous | dominant | heterozygous | genotype

Part B Allelic interactions

Match the terms to the definitions in the table below.

Term	Definition
	an allele that will produce a phenotype regardless of the presence of other alleles for that gene
	an allele that will only produce a phenotype if it is the only allele present
	when two different alleles combine to produce a different phenotype from either homozygous phenotype
	when the phenotype produced by a genotype for one gene depends on the genotype for another gene

Items:

heterozygous **recessive** **homozygous** **epistasis** **sex-linked** **codominance** **dominant** **autosomal-liked**

Part C Chromosomes & linkage

Each human [ ] cell contains [ ] pairs of chromosomes. One of these pairs is responsible for sex determination in the early embryo. These two chromosomes are called [ ] (also called "allosomes"). The other chromosomes (non-sex chromosomes) are called [ ].

Genes found on the sex chromosomes are described as "[ ]". This means that the inheritance patterns of these genes differs between males and females.

Genes that are located on the **same** non-sex chromosome are described as "[ ]". This means that the alleles for these genes will be inherited together (i.e. not recombine with alleles found on the homologous chromosome) unless crossing over occurs.

Items:

autosomal-linked **gametic** 46 **sex-linked** **autosomes** **sex chromosomes** 23 **somatic**

Part D Examples

Match the terms to the descriptions in the table below.

Description	Example of
Two plants are both homozygous for a gene involved in flower colour. One plant produces red flowers, and the other plant produces white flowers. When the two plants are bred together, all of the offspring produce pink flowers.	
Two flies are both heterozygous for two genes: one involved in body colour and one involved in wing length. When the two flies mate, almost all of the offspring either have brown bodies & long wings **or** black bodies & short wings. Very few offspring have brown bodies & short wings or black bodies & long wings.	
A female mouse is homozygous for a gene ("gene A"). A male mouse only has one copy of gene A, and contains a different allele to the female. When the two mice mate, none of the male offspring contain the father's allele for gene A.	
Two dogs are both heterozygous for two genes ("gene A" and "gene B"). Gene A codes for a pigment, and is therefore involved in body colour. When the two dogs mate, the offspring that have the same genotype for gene A are different colours because they have different genotypes for gene B.	

Items:

epistasis **codominance** **autosomal-linkage** **sex-linkage**

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Gameboard:
STEM SMART Biology Week 18

Blood Types

A Level

Part A The ABO gene

The ABO gene codes for an enzyme involved in modifying cell-surface glycoproteins. Once modified, these glycoproteins act as antigens (cell-surface molecules that the immune system can recognise as self or non-self). There are multiple alleles for this gene, and these can be split into three main types: $\mathbf{I^A}$, $\mathbf{I^B}$, and **i** (alternatively named $\mathbf{I^O}$). The $\mathbf{I^A}$ allele produces type A antigens and the $\mathbf{I^B}$ allele produces type B antigens. The **i** allele produces a non-functional enzyme, and therefore does not produce type A or type B antigens.

The table below shows how these alleles relate to blood type.

Blood Type	Antigens produced	Genotype
AB	type A and type B	I^AI^B
A	type A only	I^AI^A or I^Ai
B	type B only	I^BI^B or I^Bi
O	neither	ii

Fill in the blanks below.

- Allele $\mathbf{I^A}$ is [ ] allele $\mathbf{I^B}$ and [ ] allele **i**.
- Allele $\mathbf{I^B}$ is [ ] allele $\mathbf{I^A}$ and [ ] allele **i**.
- Allele **i** is [ ] allele $\mathbf{I^A}$ and [ ] allele $\mathbf{I^B}$.

Items:

dominant to **recessive to** **codominant with**

Part B The Rh genes

The Rh genes are a group of genes that encode red blood cell membrane transporters. One of these genes, the RhD gene, encodes a membrane transporter that can act as [ ] (a cell-surface molecule that the immune system can recognise as self or non-self). There are two main types of alleles for this gene: RhD positive (+) and RhD negative (-). The RhD positive allele produces the RhD antigen, while the RhD negative allele does not. Individuals with one positive allele and one negative allele produce RhD antigens. Therefore, the RhD positive allele is [ ] and the RhD negative allele is [ ].

An individual's blood type is based on their ABO alleles and their Rh alleles. For example, an individual with one I^A allele, one **i** allele, one RhD positive allele, and one RhD negative allele, would have the blood type [ ].

Items:

A+	**an antigen**	**A-**	**O-**	**dominant**	**codominant**	**an antibody**	**recessive**	**O+**

Part C Blood types and blood transfusion

Individuals will produce antibodies against antigens that their own cells [ ]. If, during a blood transfusion, an individual receives blood cells of an incompatible type, an immune response will be triggered against these blood cells. Antibodies binding to blood cells can cause the blood cells to clump together, which can cause blood vessel blockage and rupture.

O- individuals [ ] antibodies against A antigens, B antigens, and RhD antigens. Therefore, they can receive [ ] during a blood transfusion. Their blood cells can be given to any other blood type, because they lacks antigens. Because of this, O- individuals are called "universal donors".

AB+ individuals [ ] antibodies against A antigens, B antigens, and RhD antigens. Therefore, they can receive [ ] during a blood transfusion. Because of this, AB+ individuals are called "universal recipients".

Items:

produce	**do not produce**	**only O- blood cells**	**blood cells of any type**	**only AB+ blood cells**

Part D Offspring blood type(s)

In one family, the following blood types are present:

- Mother: A+
- Father: O-

Which of the following blood types could their children have? Select all that apply.

Assume no mutations occur.

- [ ] AB+
- [ ] AB-
- [ ] A+
- [ ] A-
- [ ] B+
- [ ] B-
- [ ] O+
- [ ] O-

Part E Maternal blood type(s)

In another family, the following blood types are present:

- Father: AB-
- Child 1: A-
- Child 2: B-

Which of the following blood types could the mother have? Select all that apply.

Assume no mutations occur.

- [ ] AB+
- [ ] AB-
- [ ] A+
- [ ] A-
- [ ] B+
- [ ] B-
- [ ] O+
- [ ] O-

Part F Paternal blood type(s)

In another family, the following blood types are present:

- Mother: O-
- Child 1: B-
- Child 2: A+

Which of the following blood types could the father have? Select all that apply.

Assume no mutations occur.

- [ ] AB+
- [ ] AB-
- [ ] A+
- [ ] A-
- [ ] B+
- [ ] B-
- [ ] O+
- [ ] O-

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Genetic Differences

A Level

Part A Gene sequences

A student analysed a gene sequence that had been identified in four different types of organism. The gene codes for a functional protein. A section of the gene's DNA is shown below. The rest of the DNA from this gene (not shown) is identical in all four different types of organism.

organism	DNA sequence					
human	ACG	CCT	CGT	CAC	GCT	AAA
oak tree	ACG	GAA	TAT	GTA	GCT	AAA
mushroom	ACG	GAA	CTC	TTA	GCT	AAA
E. coli bacterium	ACG	TAC	GAT	GGG	GCT	AAA

Which of the following statements are correct? Select all that apply.

- [ ] This gene may code for chlorophyll.
- [ ] This gene may code for a ribosomal protein.
- [ ] A single-base deletion in the first codon would only change the first amino acid of the protein.
- [ ] This gene may be found in the nucleus or in the cytoplasm.
- [ ] The protein that this gene codes for is likely to be more similar in plants and fungi than in other organisms.
- [ ] The human version of this gene only differs by three codons from the *E. coli* version of this gene.

Part B Mitosis & mutation

A diploid plant cell divides by mitosis.

After mitosis of this cell, a mutation occurs that changes the genotype of **one** of the daughter cells. This mutant daughter cell produces a non-functional enzyme instead of the functional enzyme produced by the other daughter cell. This mutation has no effect on the phenotype of the plant or the number or length of chromosomes in the plant cell.

Which of the following statements are correct? Select all that apply.

- [ ] The sequence of bases along each allele will be the same in each daughter cell.
- [ ] The chromosomes in the nucleus of each daughter cell will contain the same genes.
- [ ] Both daughter cells are haploid.
- [ ] Both daughter cells will contain the same alleles for every gene.
- [ ] The mutation may have changed the amino acids in the active site of the enzyme.

Part C Mutation origins

A child is affected by a dominant genetic condition. All of his cells have the same genotype. The allele associated with this condition is not present in either of his parents.

Which of the following statements are correct? Select all that apply.

- [ ] The mutation may have occurred during meiosis in his father.
- [ ] The mutation may have occurred in the child's DNA during the zygote stage.
- [ ] Both of his grandmothers had the condition.
- [ ] All of the child's future offspring will have the condition.
- [ ] The mutation may have occurred in the child's DNA after he was born.

Adapted with permission from NSAA 2018 Section 1 Q69 & NSAA 2018 Section 1 Q72 & NSAA 2019 Section 1 Q66

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Allele Population Frequencies

A Level

A recessive condition is found within a human population. There were 5000 births in this population within one year. Of these births, 8% had the condition and 32% were homozygous dominant.

One healthy cheek cell is analysed from each person born in this year.

Part A Recessive allele numbers

How many recessive alleles are present in the sample?

Part B Dominant allele numbers

How many dominant alleles are present in the sample?

Adapted with permission from NSAA 2020 Section 2 Q44

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Mutation Probabilities

A Level

The figure below shows how each possible DNA codon is translated.

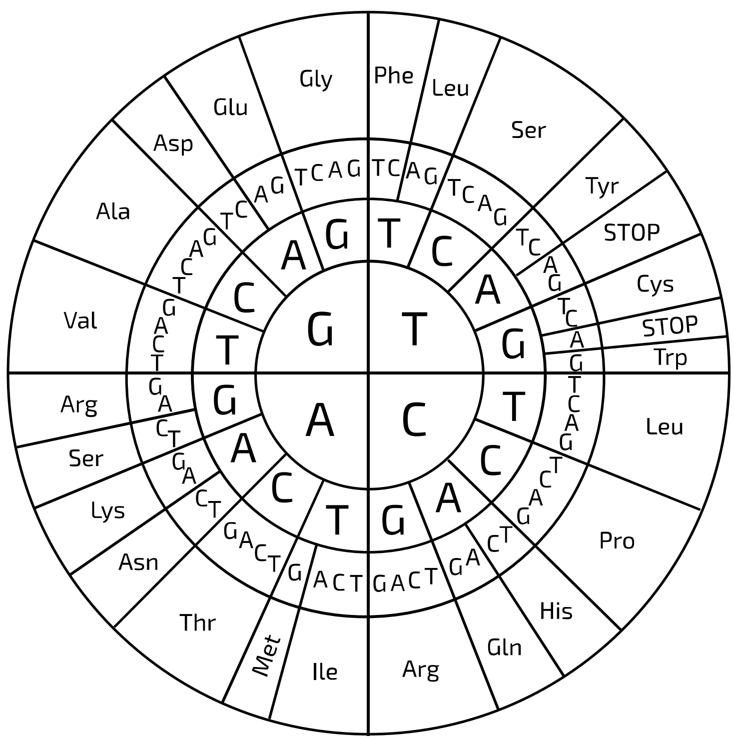

Figure 1: DNA codon wheel. The diagram is read from inside to out, with amino acid names given as abbreviations. For example, the base triplets CAT and CAC both code for the amino acid histidine (His).

In the questions below, assume that it is equally likely that mutations can change any base to any other base, and that the probability of this resulting in a change in any particular base during one cell division is 2×10^{-9}.

Part A Methionine to proline

What is the probability that a triplet that codes for methionine (Met) changes to code for proline (Pro) in one round of division?

Give your answer to 3 significant figures.

Part B Phenylalanine to glutamine

What is the probability that a triplet that codes for phenylalanine (Phe) changes to code for glutamine (Glu) in one round of division?

Give your answer to 3 significant figures.

Part C Start to stop

What is the probability that a start codon (Met) changes to a stop codon (STOP) in one round of division?

Give your answer to 3 significant figures.

Part D Redundancy

Based on **Figure 1**, which of the following amino acids display redundancy/degeneracy in how they are encoded? Select all that apply.

- [ ] tryptophan (Trp)
- [ ] proline (Pro)
- [ ] glutamine (Glu)
- [ ] methionine (Met)
- [ ] phenylalanine (Phe)

Adapted with permission from NSAA 2020 Section 2 Q60