**Review Worksheet Answers: Intro and Gene Mutations**

1: Define the term “species”

(3 marks)

*A species is a group of individuals that share many characteristics (1) and are able to breed under natural conditions (1) to produce fertile offspring (1).*

2: What is the difference between a gene and an allele?\*

(4 marks)

*A gene is a base sequence coding for production of protein that performs a specific function in the body (1). An allele is a variant of a given gene (1). For each gene, a person inherits two alleles (1) – one from their mother and the other from their father (1).*

3: What is a “population” of organisms?

(3 marks)

*A population is a group of organisms of the same species (1), living together in the same location (1) at the same time (1).*

4: What is a gene pool?

(2 marks)

*A gene pool is the sum of all of the alleles (1) present in a given population (1).*

5: What is allele frequency?

(1 mark)

*Allele frequency is how often an allele occurs in a given population (1)*

6: The gene mutation that causes Cystic Fibrosis first originated in a person in Western Europe around 2000 years ago. Which population would you expect to have a higher allele frequency for Cystic Fibrosis – the population of Switzerland, or the population of Papua New Guinea? Explain your answer.\*

(5 marks)

*Switzerland would be expected to have the higher allele frequency for Cystic Fibrosis (1). The individual carrying the original mutation would have the vast majority of their descendants living in Europe until very recently (1). Therefore, the allele would have been passed down via those descendants through the European population (1), leading to a higher frequency in that population (1). The allele may only have recently reached Papua New Guinea via European colonists/invaders in the past few centuries and so would be less frequent in that population. (1)*

7: List the FOUR main ways that frequency and type of alleles in a population can change over time.

(4 marks)

*Mutation*

*Gene flow due to interbreeding*

*Genetic Drift*

*Natural Selection*

8: Define the term “mutation” and list ways mutations can be classified.

(4 marks)

*A mutation is a change to a gene sequence or chromosome (1) that occurs during DNA replication and cell division (1). They can be classified by:*

*What causes them to occur (0.5)*

*Whether or not they are heritable (0.5)*

*Their effect on amino acid sequence and protein production (0.5)*

*How much DNA is affected (0.5)*

9: What is a mutagen?

(1 mark)

*A mutagen is a substance or agent that affects the rate of mutation of DNA. (1)*

10: List the two broad types of mutagens and give an example of each.

(3 marks)

*Ionising Radiation (1) – eg UV light / X-rays / Nuclear radiation (0.5 for one example)*

*Chemicals (1) – eg mustard gas / formaldehyde / some drugs (0.5 for one example)*

11: Describe differences between somatic and germline mutations and their effects.

(6 marks)

|  |  |  |
| --- | --- | --- |
|  | **Somatic** | **Germline** |
| **Cell Type involved** | *non reproductive cells (0.5)* | *Gametes (0.5) (sperm and egg) (0.5)* |
| **Effects** | *May have no effect, (0.5)*  *May cause cell death, (0.5)*  *May cause uncontrolled cell growth (cancer)(0.5)* | *May have no obvious effect (0.5)*  *May result in a non-viable gamete (0.5)*  *May cause abnormalities or inherited disease (0.5)*  *May cause an advantage (0.5)* |
| **Heritability** | *Cannot be passed on to offspring (0.5)* | *Can be passed on to offspring (0.5)* |

12: List and describe the three types of point mutation

(3 marks)

*Insertion (0.5) - an additional nucleotide is added to the base sequence (0.5)*

*Deletion (0.5) - a nucleotide is removed from the base sequence (0.5)*

*Substitution (0.5) – a nucleotide in the base sequence is replaced with a different nucleotide (0.5)*

13: Which types of point mutation cause frame shift?

(1 mark)

*Insertion (0.5) and Deletion (0.5)*

14: Fill in the table to show the possible effects point mutations may have on amino acid sequence and protein structure.

(4 marks)

|  |  |  |
| --- | --- | --- |
| **Name of Effect** | **Effect on Amino Acid sequence** | **Effect on Protein Structure** |
| Missense Mutation | *Change amino acid (0.5)* | *Change protein structure and function (0.5)* |
| Nonsense Mutation | *Stop amino acid addition (0.5)* | *Loss of protein structure and function (0.5)* |
| Neutral Mutation | *Change amino acid (0.5)* | *No change to protein structure (0.5)* |
| Silent Mutation | *Change in base sequence but no change in amino acid sequence (0.5)* | *No change to protein structure (0.5)* |

12: Which type of point mutation would be most likely to produce a silent or neutral effect? Explain your answer.\*

(6 marks)

*Base* *substitution (1) would be most likely to produce a silent or neutral effect. This is because substitution only causes a change to a single triplet and therefore amino acid (1), and does not cause frame shift (1). Insertion and Deletion cause frame shift (1) which means all subsequent triplets are affected (1), having a much greater effect on amino acid sequence and protein structure (1).*

13: Fill in the table to show type of inheritance and effects of the single-gene conditions listed.

(6 marks)

|  |  |  |
| --- | --- | --- |
| **Condition** | **Inheritance Type** | **Effects** |
| Albinism | *Autosomal (0.5) Recessive (0.5)* | *Allele for pigment production is faulty(0.5), resulting in white blond hair, very pale skin, pale blue eyes. (0.5)* |
| Duchenne Muscular Dystrophy | *X-linked (1)* | *Allele for muscle development is faulty (0.5) progressive wasting of muscles during childhood, eventual respiratory paralysis and death (0.5)* |
| Cystic Fibrosis | *Autosomal (0.5) Recessive (0.5)* | *Allele for chloride ion movement is faulty (0.5) causing sticky mucus which causes problems in the respiratory and digestive tracts (0.5)* |

14: What are “lethal recessive” conditions?

(1 mark)

*These are mutations to alleles which will cause death in very early childhood (0.5) if two alleles are inherited (recessive inheritance). (0.5)*

15: Tay Sachs Disease is a lethal recessive condition. Explain why the allele frequency for Tay Sachs Disease is much higher in the Ashkenazi Jewish religious population than in the general population.

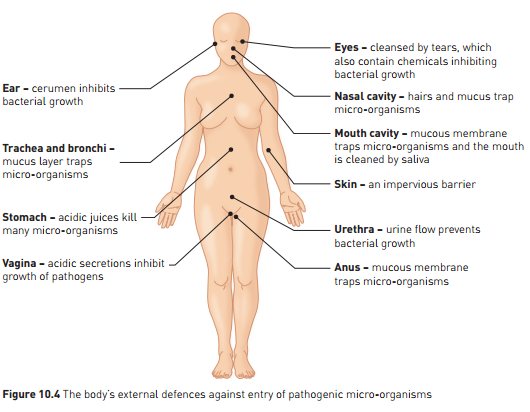
(6 marks)

*Lethal recessive alleles will usually decline in the population (1) as they result in death before reproduction (1) in affected individuals. In small populations, the allele frequency is higher (1) because the likelihood of two carriers meeting and reproducing is much higher (1) so the likelihood of affected or carrier offspring is much higher (1). The Ashkenazi Jewish religious population is small, and generally marry within their religion, so the allele frequency is higher (1).*

 17: List and briefly explain 5 of the body’s **external** defences against disease.

(5 marks)

*Any five of the following with explanation:*

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