**Review Worksheet: Intro and Gene Mutations**

**Name: ……………………………………………………………..**

*Do these questions, using your learning resources. Look at the “marks” to give you an idea of the level of detail required in the response (formative only – does not count towards your grade). At the end, mark your work, correct it, and fill in the reflection section. Questions marked \* require you to use reasoning, inferring and application of knowledge, or perhaps extra research to get the answer. It won’t be right there in the text.*

1: Define the term “species”

(3 marks)

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

(4 marks)

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3: What is a “population” of organisms?

(3 marks)

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4: What is a gene pool?

(2 marks)

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5: What is allele frequency?

(1 mark)

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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)

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

(4 marks)

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

(4 marks)

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9: What is a mutagen?

(1 mark)

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

(3 marks)

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

(6 marks)

|  |  |  |
| --- | --- | --- |
|  | **Somatic** | **Germline** |
| **Cell Type involved** |  |  |
| **Effects** |  |  |
| **Heritability** |  |  |

12: List and describe the three types of point mutation

(3 marks)

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13: Which types of point mutation cause frame shift?

(1 mark)

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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 |  |  |
| Nonsense Mutation |  |  |
| Neutral Mutation |  |  |
| Silent Mutation |  |  |

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

(6 marks)

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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 |  |  |
| Duchenne Muscular Dystrophy |  |  |
| Cystic Fibrosis |  |  |

14: What are “lethal recessive” conditions?

(1 mark)

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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)

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

(5 marks)

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Go back and mark your work using the marking key provided. What score did you get? /68

*I included enough detail in my answers.*



*I was able to find information in the text/powerpoint presentation.*

*I was able to reason and infer where the information wasn’t directly in the text (questions with \*).*

*I marked my work and wrote down any answers where I missed marks.*