**Review Worksheet Answers: Heterozygote Advantage**

1: Fill in the table below to provide information about heterozygote advantage for the following diseases.

(38 marks)

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| --- | --- | --- | --- | --- | --- | --- |
| **Disease Name** | **Cause and inheritance** | **Symptoms** | **Selective Agent for Heterozygote Advantage** | **Genotype advantage when agent present and why** | **Effect on allele frequency when agent present** | **Effect on allele frequency when agent absent** |
| Sickle Cell Anaemia | *Faulty allele causing “sickle” shaped blood cells (0.5)*  *Autosomal recessive inheritance (0.5)* | **Dominant (no faulty alleles)***:*  *No sickling, blood cells normal, no symptoms (1)*  **Heterozygous (one faulty allele)**  *Some blood cells sickled (0.5), very mild symptoms under low oxygen conditions (0.5)*  **Recessive (two faulty alleles)**  *All blood cells sickled (0.5), severe anaemia (0.5), death in childhood (0.5)* | *Malaria (1)* | *Heterozygote advantage when malaria present: (1)*  *Dominant: normal blood (0.5), but more likely to get malaria and die.(0.5)*  *Heterozygous: resistant to malaria (1)*  *Recessive: die in childhood (0.5)* | *Carriers of the faulty allele (0.5) are more likely to survive (0.5) in areas with malaria present.(0.5) so allele frequency for the faulty allele will slowly rise (0.5)* | *When there is no malaria, carrying the faulty gene is a slight disadvantage (0.5) due to mild symptoms (0.5), and having two faulty genes results in death (0.5). The allele frequency will slowly decrease in the population. (0.5)* |
| Alpha Thalassemia | *Faulty allele causing defects of alphahaemoglobin (0.5)*  *Autosomal recessive inheritance*  *(0.5)* | **Dominant (no faulty alleles):**  *Normal haemoglobin (0.5), no thalassemia (0.5)*  **Heterozygous (one faulty allele):**  *Mild symptoms of anaemia. (0.5)*  **Recessive (two faulty alleles)**  *Severe anaemia (0.5), death shortly after birth (0.5)* | *Malaria (1)* | *Heterozygote advantage when malaria present: (1)*  *Dominant: normal blood (0.5), but more likely to get malaria and die.(0.5)*  *Heterozygous: resistant to malaria (1)*  *Recessive: die shortly after birth (0.5)* | *Carriers of the faulty allele (0.5) are more likely to survive (0.5) in areas with malaria present.(0.5) so allele frequency for the faulty allele will slowly rise (0.5)* | *When there is no malaria, carrying the faulty gene is a slight disadvantage (0.5) due to mild symptoms (0.5), and having two faulty genes results in death (0.5). The allele frequency will slowly decrease in the population (0.5)* |
| Tay-Sachs Disease | *Faulty allele causing problems with lipid metabolism in the brain. (0.5)  Autosomal recessive inheritance (0.5)* | **Dominant (no faulty alleles):**  *Normal lipid metabolism (0.5), no symptoms (0.5)*  **Heterozygous (one faulty allele)**  *No symptoms, carrier only (0.5)*  **Recessive (two faulty alleles)**  *Progressive nervous system disease (0.5), death in early childhood (0.5)* | *Tuberculosis (1)* | *Heterozygote advantage when tuberculosis present.*  *Dominant: no symptoms of Tay Sachs, (0.5) but more likely to get tuberculosis (0.5)*  *Heterozygous:*  *No symptoms of Tay Sachs (0.5), but resistant to tuberculosis (0.5)*  *Recessive:*  *Death in early childhood (0.5)* | *Carriers of the faulty allele (0.5) are more likely to survive (0.5) in areas with tuberculosis present.(0.5) so allele frequency for the faulty allele will slowly rise (0.5)* | *When there is no tuberculosis, having no alleles for Tay Sachs is an advantage (0.5) because the individual does not have the disease (0.5) AND has a lower chance of passing the allele to offspring. (0.5)  Being a carrier for Tay-Sachs is not a disadvantage to the individual (0.5), but increases the risk of having a child with the disease (0.5), making reproduction less successful. (0.5)*  *Having two faulty alleles results in death in early childhood. (0.5)   If tuberculosis is not present, the allele will slowly decrease in the population.(0.5)* |

2: Ashkenazi Jewish people have a higher allele frequency for Tay-Sachs disease. In part this may be because during World War II they were placed in ghettos and concentration camps with cramped conditions and a high prevalence of tuberculosis, so that the allele rose in the population due to heterozygote advantage.

What other reasons may have caused increased allele frequency for Tay-Sachs disease in Ashkenazi Jews during the second world war, and why is the allele frequency still higher in this population today?

(6 marks)

*The Bottleneck Effect (0.5), a form of random genetic drift (0.5), may have contributed. Killing of Jews during the Holocaust led to a much smaller population (1). The gene frequency for Tay-Sachs may have been higher in the surviving population (0.5) by chance (0.5), and would have remained high (0.5) in the small population due to socio-cultural barriers (0.5) to gene flow (0.5). Ashkenazi Jews tend to marry within their religious and ethnic group (1), so the higher frequency of the allele persists over time (0.5) due to the smaller gene pool (0.5).*

3: Alleles for beta thalassemia are more frequent around the Mediterranean, but there is no evidence that heterozygote advantage is occurring. Suggest another reason why the allele frequency for beta thalassemia may have remained high.

(6 marks)

*The mutation for beta thalassemia initially arose in the Mediterranean (1). Traditionally cousin marriage was a feature in cultures around the Mediterranean (1), to consolidate wealth and power (1). This was a socio-cultural barrier (1) to gene flow (1), so the allele frequency would remain high over time due to a small gene pool with in-breeding (1).*

4: Describe how steroid hormones have their effect on the target cell.

(7 marks)

*Steroid hormones are lipid-soluble (1), so they are able to pass through the cell membrane (1). When they reach the target cell, they diffuse across the cell membrane (1), and bind to receptors (0.5) on the nucleus or other organelles (0.5). The hormone-receptor complex activates the genes (0.5) controlling the formation of particular proteins (0.5), altering transcription (0.5) and therefore hormone synthesis (0.5). These hormones are slow to have an effect (0.5), but the effect is long lasting (0.5).*