**HUMAN BIOLOGY – YEAR 12**

**TASK 9 – GENE POOLS AND MUTATIONS EXTENDED RESPONSE**

**MARKING KEY WEIGHTING: 7.5%**

1. Thalassemia is a condition that affects the indigenous people of Tharu. Explain how the disease is inherited and what effects it has on the individual person. **(2 marks)**

*Recessive disease – inherit recessive allele from each parent (1)*

*Effects – causes production of abnormal haemoglobin, resulting in destruction of red blood cells/anaemia/extreme fatigue (1)*

1. The incident of Thalassemia is much higher in the Tharu people of Terai than the Nepalese that have migrated into the area. Discuss the evolutionary mechanisms that have allowed these differences to occur?

(**5 marks)**

*Natural selection – being a carrier for Thalassemia mean immune to malaria (1)*

*Area high in malaria = heterozygous better chance of survival, breed and pass on alleles (1)*

*Increasing frequency in the gene pool as those without allele die removing alleles (1)*

*Nepalese allele frequency less as never exposed to malaria /just moved into malaria area*

*no selection advantage was present for being a carrier so allele frequency minimal(1)*

1. If malaria was to return to the Terai region, what affect would you expect it to have on the allele frequencies in the gene pool s of that area? **(3 marks)**

Allele frequency increase/maintain in Tharu people(1)

Allele begin to appear in the Nepalese community/start to increase (1)

as those carrying have selection advantage and will survive, breed and pass on alleles increasing the frequency (1)

1. Discuss how the link between the Tay Sachs allele and Tuberculosis led to a change in the allele frequency of the Jewish population living in the Ghettos.

**(4 marks)**

* Carriers of the Tay Sachs allele has an immunity to TB/selection advantage
* TB killed people with two normal alleles removing them from the gene pool
* Those with Tay Sachs died before reproducing removing their alleles from the gene pool
* So frequency of Tay Sachs allele and normal allele remained the same/constant

1. Explain, with reference to evolutionary mechanisms, why the incidence of Tay Sachs is unusually high in the present day Ashkenazi Jewish community.

**(4 marks)**

* Isolated community by sociocultural barrier
* Founder population had high occurrence of the allele
* Chance of heterozygous meeting and having offspring high which maintains the frequency/makes it slow to decline
* Population is small so it takes time to breed out an allele even though no selection advantage

1. Discuss the evolutionary mechanisms that have brought about the increased incidence of the disease on the island.

**(4 marks)**

*First three:*

* Disease and typhoon reduced population to very small gene pool / bottleneck / Founder Effect
* All current population descendants of Ruler who was a carrier for the condition
* Island isolated and small population/no immigration-emigration

*Plus one:*

* so inbreeding occurred causing the allele frequency to be higher than normal
* increase is random/No natural selection occurring as disease does not really provide any advantage or disadvantage

ii. If the island population started to become more dependent on agriculture rather than fishing, what effect would this have on the allele frequency for achromatopsia in the island gene pool? **(2 marks)**

*no effect*

*colour blindness provides no selection advantage for anything related to fishing or agriculture*

4) Discuss the evolutionary mechanisms that brought about the high incidence of Huntington’s disease in the community of Lake Maracaibo. Include an explanation that although the occurrence of Huntington’s disease is much higher on the island than elsewhere, the occurrence of the disease is not totally unexpected in a community.  **(5 marks)**

All three:

* One woman had the allele arrived on island – Founder Effect
* Had lots of children / high incidence of the allele in gene pool due to lots of children
* Island small and isolated population so frequency of allele increased more than normal

Both of the following to explain why not unexpected in a community:

* Because those with the disease breed before they realised
* 50% chance that it would be passed on