**Tay Sach’s disease**

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| **Description** |
| Population - Ashkenazi Jews/Jewish people |
| Cause – Missing enzyme which is essential for fat metabolism |
| Symptoms – * Build-up of fatty acids in the nervous system
* From a few months of age mental and physical disabilities develop quickly
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| Inheritance – Recessive trait is passed from the 2 carrier parents |
| Effect on gene pool – * Affected individuals die in childhood/before reproductive age
* Carrier couples choose to not reproduce
* Heterozygous individuals are resistant to tuberculosis
* Increase Tay-Sachs allele frequency in population
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**Thalassaemia**

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| **Description** |
| Population – Mediterranean, Greece/Italy |
| Cause – Mutations of the gene responsible for haemoglobin production |
| Symptoms – * Defects in the formation of haemoglobin
* Sufferers have fewer functioning red blood cells
* Sufferers can have anaemia and be iron deficient
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| Inheritance – Recessive trait is passed from carrier parents to offspring  |
| Effect on gene pool – * More mutations found in gene pool greatly increases mortality rate
* Increases thalassemia allele frequency in population
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**Sickle-cell anaemia**

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| **Description** |
| Population - Black African population |
| Cause – Mutations of the gene responsible for haemoglobin production |
| Symptoms – * Red blood cells have a sickle shape (crescent)
* Reduces oxygen carrying ability
* Fatigue/shortness of breath
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| Inheritance – Recessive trait is from affected parent to offspring |
| Effect on gene pool – * Individuals who are homozygous usually die early, disease can be fatal
* Individuals who are heterozygous are called ‘sickler’s’ and have the sickle trait
* Heterozygous individuals are resistant to malaria/ increases sickle cell allele frequency in population
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