HSC Biology – Module 5: Heredity – Inheritance Patterns in a Population Study Notes

❖ Inheritance Patterns in a Population

Inquiry question: Can population genetic patterns be predicted with any accuracy?

- ❖ *investigate the use of technologies to determine inheritance patterns in a population using, for example: (ACSBL064, ACSBL085)*
  - ➢ *DNA sequencing and profiling (ACSBL086)*

DNA sequencing & DNA profiling

Each individual (except for identical twins) has a unique genetic code. Therefore, these genetic sequences can be compared in order to determine inheritance patterns in a population.

DNA profiling or DNA fingerprinting is a process widely used in forensics to compare the DNA of suspects with that found at the crime scene although it can also be used to determine paternity and therefore population patterns. Human DNA contains particular genes which are more likely to vary between individuals. These 'highly variable genes' are compared as they will almost certainly be unique enough to identify an individual without having to sequence their entire genome.

1. DNA samples are collected, and restriction enzymes are used to cut the DNA into small pieces at specific places
2. These DNA fragments will be different sizes and are able to be separated via gel electrophoresis:
   a. The fragments are placed on a gel surface and an electric current is applied
   b. As DNA is negatively charged, theses fragments move towards the positive electrode. Small fragments have less mass but the same charge, so they move faster through the gel.
   c. The gel is then stained with dye and the fragments of DNA are visible as bands representing similar sized pieces.
3. The differences between the patterns of the bands is compared

Sanger sequencing involves making many copies of the target region of DNA in order to sequence it:

1. DNA sample is mixed with DNA polymerase, primer (DNA which lets the polymerase know where to start) and nucleotides including a tiny amount of tagged dideoxy nucleotides (chain-ending versions) colour coded depending on base type (A, T, C, G)
2. After heating (to denature the sample DNA) and cooling (to allow the primer to bind to the sample DNA), the polymerase begins to make copies using the added nucleotides
3. Once a dideoxy nucleotide is used, the strand ends, and the process repeats until each nucleotide in the sequence serves as the ending point of a strand and is therefore colour coded
4. A variation of gel electrophoresis (capillary electrophoresis) is used on the new strands where a lazer records the colour and therefore base type of the dideoxy nucleotide as each different sized strand passes it (small strands pass first)
5. This allows the nucleotide sequence to be recorded

- *investigate the use of data analysis from a large-scale collaborative project to identify trends, patterns and relationships, for example: (ACSBL064, ACSBL073)*
  - *the use of population genetics data in conservation management*
  - *population genetics studies used to determine the inheritance of a disease or disorder*
  - *population genetics relating to human evolution*

- *the use of population genetics data in conservation management*

Population Genetics in Conservation Management

Conservation genetics is used in order to ensure that the genetic makeup of a population will allow it to adapt to changing selection pressures in its environment over time. The idea is that populations displaying large amounts of genetic diversity are more likely to continue to reproduce.

Researchers from the National Herbarium of New South Wales and Deakin University are currently collaborating on a large-scale project to map the genome of the Wollemi Pine. Known only from fossils, a tiny surviving population of Wollemi Pines was discovered near Sydney in 1994.

Initial gene mapping found 0% genetic diversity between each individual in the population, a real anomaly in the natural world. This caused huge conservation issues as having an identical genetic makeup means that a single pathogen would wipe out the entire species. The aim of this project is to better understand why there is no genetic diversity in the wild population and to find a way to help conserve this living fossil.

- *population genetics studies used to determine the inheritance of a disease or disorder*

Population Genetics and Inheritance of Disease/Disorders

By understanding population genetics, scientists can better recognise, predict and therefore treat inherited diseases and disorders.

A large-scale collaborative project known as the International Genomics of Alzheimer's Project was set up in 2009 by 4 Alzheimer's research consortia from across the world. Their aim was to try to determine the location of genes responsible for causing late-development Alzheimer's Disease.

Nine loci (positions of genes) were identified as possible causes of Alzheimer's. These genes will continue to be studied in order to determine whether or not they are actually responsible, and if so, how they can be restored to normal function.

- *population genetics relating to human evolution*

Population Genetics and Human Evolution

Population genetics assesses the differences and similarities between the genetic makeup of ancient and modern populations in order to model the evolutionary and migratory paths of modern cultures.

The Human Genome Project (1990-2003) was an extremely large-scale collaborative project which sought to identify the position of all the genes in the human chromosome and create a database for that information for public use. It gave scientists a much clearer understanding of the causes of genetic disorders as well as the patterns of evolution.

The HGP allowed us to compare our modern genome with the genetic makeup of ancient common ancestors and to see why similar species such as *Homo neanderthalensis* (Neanderthals) diverged. It also accounted for slight genetic differences that have appeared in different cultures in our more recent history.