**Question 41 (20 marks)**

1. A minute sample of DNA has been obtained from an individual for paternity testing and to establish any possible link with a rare genetic disorder.
2. Outline two **named** techniques in biotechnology that are carried out before proceeding to DNA sequencing of the genome.
3. Rapid developments in genomic testing methods have made the sequencing of a person’s DNA faster and cost-efficient. List the ethical issues involved in the use of the genomic information as a `lifetime health resource’. [14]

|  |  |
| --- | --- |
| Polymerase Chain Reaction - named | 1 |
| Denaturation – double-stranded DNA is separated into single strands by heating to 96° C.  Annealing – use of Taq polymerase, primers, free nucleotides to complete DNA replication whilst cooled to 72°C.  Elongation– Process is repeated with newly formed DNA strands – thermos-cycling of heating and cooling to produce 2ⁿ copies for n cycles.  End result – Amplification of DNA | 4 |
| Gel Electrophoresis - named | 1 |
| DNA is cut by restriction enzymes to fragments of varying size (kilobases)  DNA is micro-pipetted into wells on a gel plate & subject to an electric current in a GE cell  Being negatively charged, DNA moved towards the positive electrode  Shorter, lighter DNA fragments travel faster than the longer, heavier DNA fragments.  A pattern of DNA bands can be detected when DNA treated with chemicals fluoresce under ultra violet light and photographed to show the DNA profile. | 5 |
| Ethical Issues (any 3 or suitable responses) | 3 |
| Genetic information is hereditary so knowledge of an individual’s own genome has implications for members of the family  Legislation is needed to ensure that there is no discrimination on the basis of genetic information  Genetic procedures as such are still costly – not accessible to all -, and there is no easy answer as to who should pay for them.  Our ability to interpret genomic information is still at an early stage. Even for those whom we know, with certainty, have a genetic condition, it can be challenging to identify a specific disease-causing gene change. We do carry many different types of genetic variants. So genomic testing can lead to incorrect or over-diagnosis.  Genomic information has the potential to influence the direction of human evolution because human intervention determines which embryos to implant and which to discard after identifying the genome. Sometimes the stem cells of healthy embryos are used as donor cells to generate tissue to treat individuals with genetic abnormalities.  When this occurs, the affected individual, who might otherwise die, may lead a relatively healthy life, even reaching reproductive age—then the defective alleles may be passed on to offspring and the genetic disorder is perpetuated.  Other issues surround the privacy and disclosure of genetic information and the storage or future use of test samples and data etc |  |

1. Scientists trying to establish the evolutionary relationship between primates decided to complete a comparative study using nuclear DNA.
2. Describe how this would be conducted. (2 marks)

**Any 2**

* **Work out the DNA sequence for each primate species in question**
* **Compare the DNA sequences for each species**
* **Comparison of viral sequences (ERVs) / explanation of hybridisation process**

1. Explain how it could show evolutionary relationships between primates. (1 mark)

**More differences in DNA sequences means longer time since they diverged from a common ancestor / vice versa**

1. Palaeontologists are yet to find a common ancestor for chimpanzees and humans in the fossil record. It is thought to have lived between 5 and 7 million years ago.
2. Suggest **three** reasons why such a fossil has been hard to find. (3 marks)

**Any 3**

* **Conditions for fossilisation has not been met (no fossil)**
* **Inaccessible location / buried too deep in the ground**
* **Destroyed by human / geological activity (eg folding, volcanic)**
* **Unable to date fossil as it does not meet limitations**
* **Fossil too fragmented**
* **Scientists disagree with interpretation of fossils**

1. A scientist who is completing excavations in Eastern Africa unearths a fossil he believes is the ‘missing link’ between chimpanzees and humans. If this is true, explain why C-14 dating would be of little use in this scenario. (2 marks)

**Fossil is older than 60 000 years / too old**

**Not enough C-14 remaining in fossil to provide accurate date**

**Question 41**

The HEXA gene codes for an enzyme that breaks down lipids found within cells, mostly in the central nervous system. If both alleles for this gene are mutated, the individual will suffer from a fatal disease called Tay-Sachs.

1. In Australia, the frequency of the mutated allele is very low, about 1 in 300. In the Ashkenazi Jew population, however, the frequency is about 1 in 26.

Explain **two** evolutionary mechanisms that scientists reason could be the cause for such different gene pools.

(7 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| Random Genetic Drift | 1-7 |
| * Small population |
| * Increase in frequency due to chance |
| * Likely as Ashkenazi Jews are (a small and) reproductively isolated population |
| Natural Selection |
| * Carrier / heterozygote survival advantage |
| * Increased resistance to tuberculosis |
| * Survivors more likely reproduce and pass allele to offspring |
| * High selection pressure for allele as Jews subject to higher tuberculosis risk (in overcrowded ghettos) |
|  | **Total 7** |

1. Researchers are hoping to start human clinical trials for Tay-Sachs disease using gene therapy. They hope to achieve this by using modified viruses that are infused into cerebrospinal fluid.

Describe the process of how the viruses would be genetically engineered and then how it could lead to a treatment for Tay-Sachs. (13 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| * The normal (HEXA) gene is removed | 1-11 |
| * by cutting it at a recognition site |
| * With a restriction enzyme |
| * Which makes a staggered cut made on either side of gene |
| * Creating sticky ends from (overhanging) unpaired nucleotides |
| * DNA / genetic material removed from virus |
| * Viral DNA cut with same restriction enzyme |
| * To create sticky ends that are complementary to gene |
| * DNA ligase is an enzyme |
| * That joins sticky ends of gene and viral DNA |
| * This amalgamation is called recombinant DNA |
| * Which is inserted back into virus |
| * Virus is now termed a vector |
| * Once inserted in the body, the virus would insert correct gene into body cells / replace faulty gene | 1-2 |
| * Gene could then function to produce the correct protein / enzyme |
|  | **Total 13** |