Objective Sheet 6

HUMAN VARIATION AND EVOLUTION **HUMAN PERSPECTIVES CHAPTER 14-20**

1. UNDERSTAND THAT HUMANS CAN SHOW MULTIPLE VARIATIONS AND HOW THE CHANGING ENVIRONMENT CAN INFLUENCE THE SURVIVAL OF ORGANISM WITH GENETIC VARIATION.

- In every species there is genetic variation due to sexual reproduction, mutations, meiosis (random assortment of chromosomes, crossing over and non disjunction) and epigenetics
- Thus there are some genes that are advantageous in the gene pools due to the environment in which the species lives in. The organism with the advantageous genes are more likely to survive an the one with out the advantageous gene are more likely die
- There are some mutations that have no advantage or disadvantage but if the environment changes then must of the this mutations can become useful, thus increasing the survival chances of the organism with the gene.

• Random assortment
• Crossing over
• Non-disjunction
• Random fertilisation
• Mutations

2. EXPLAIN HOW GENE POOL ALLELE FREQUENCIES CAN BE ALTERED: MUTATIONS, DIFFERENT SELECTION PRESSURES, RANDOM GENETIC DRIFT, FOUNDER EFFECT AND CHANGES IN GENE FLOW.

MUTATION

- Permanent changes in the DNA of a chromosome and may result in totally new characteristics in an individual. If the mutation occurs in gamete, it can be passed in from generation to generation.
- Can be caused by;

• Chemicals
• High temperatures
• Radiation
• Natural means

- New mutation could be harmful and could be lost
- New mutation could also be advantageous increasing the chance of survival and changing the gene frequency

DIFFERENT SELECTION PRESSURES

- Include environmental, disease, predation and mate selection
- Different environment pressures such as wet/dry ect will favour individuals that suit that environment more than the ones that do not thus the gene pool frequency changes

RANDOM GENETIC DRIFT

- Gene frequency in large population tends to stay the same
- In small populations, there may be by pure chance a random, non directional variation in alleles frequency
- The population needs to be isolated

Eg- An allele that is rare in large populations may become frequent in a small population

FOUNDER EFFECT
A group of individuals may become isolated from a population but the gene frequency in the founder population may not be the same as the original population. If there is no gene flow between the original population and the founder populations, the gene frequencies will be different

CHANGE IN GENE FLOW
- Allows for emigration and immigration which has the effect of adding and deleting alleles to a population and can change gene frequencies

Barrier to gene flow
- Social
- Cultural
- Geographical

Population size
- Larger population act as a buffer for random changes un allele frequency
- Smaller population can allow for change in allele frequencies more easily

Mate selection
- Random mating allows for animals and plants gametes are released in the wind/water
- Assortive mating only certain combination come together at higher frequencies

3. EXPLAIN HOW SURVIVAL CAN BE ENHANCED IF PARTICULAR PHENOTYPES ARE SELECTIVELY ADVANTAGEOUS.

Those organisms that survive because of their favourable phenotypes in a populations will pass on favourable alleles to their offspring. Gradually, the characteristics of a population changes so that is becomes better suited to the environment. Th survival of this with favourable traits is selectively advantageous.

HETEROZYGOUS ADVANTAGE
- In recessive homozygous form the allele can be lethal and organism with that genotype will die off
- Although in heterozygous form the gene can be advantageous to the survival of the organism thus it stays in the population
- An example of this is the sickle cell allele, in recessive homozygous form the gene is lethal although in heterozygous form the gene is said to provide protection against malaria
- Thus is the tropics where malaria in very common the allele stays in the population and is not bread out

4. GIVE EXAMPLES OF GENETIC DISEASES FOUND IN SPECIFIC POPULATIONS

TAY SACHS (TSD)

A Genetic disorder caused by a missing enzyme that results in fatty substances accumulating in the nervous system

- Is a lethal recessive condition
- People with homozygous genes generally die prematurely, often before they can reproduce, so these harmful genes usually disappear from the gene pool.
- High allele frequency in Ashkenazi Jews 1 in 2500 compared to 1 in 500,000 world wide
- This is due to;
- Genetic drift - population of Ashkenazi Jews are small and isolated, which increases the chance of genetic drift
- Heterozygous advantage - those who have heterozygous Tay Sachs appear to have an increased resistance (survival advantage) to Tuberculosis and would be more likely to reprocess and pass their allele on to the next generation
- Isolated - the frequency of the allele is maintained in the population and has not be lost due to its survival advantage

SICKLE CELL DISEASE

- Sickle cell disease is a blood condition seen most commonly in people of African ancestry.
- The disorder is caused by a point mutation in one of the genes that codes for haemoglobin, the blood protein that carries oxygen.
- Is a lethal recessive condition
- People with sickle cell disease have inherited two copies of the recessive mutant gene.
- This mutation causes the red blood cells to take on a sickle shape, rather than their characteristic donut shape.
- Individuals who suffer from sickle cell disease are chronically anaemic, experience significant damage to their heart, lungs, and kidneys, and often die.
- People who inherit only one copy of the mutant allele have a mix of normal red blood cells and some abnormal cells. These people are said to have the sickle cell trait and lead normal, healthy lives.
- People with sickle cell trait have a greater resistance to malaria than people with the normal genotype (i.e. homozygous dominant).

5. EXPLAIN THE THEORY OF EVOLUTION USING NATURAL SELECTION AS THE KEY PROCESS – VARIATION, STRUGGLE FOR EXISTENCE, ISOLATION AND DIFFERENTIAL SELECTION, SPECIATION.

Dawin's deductions

1) There is a struggle for existence between organism
2) Survival if the fittest occurs (those organisms most suited to their environment wiki survive in large numbers)
3) In nature, the environment limits the size of the population. (Floods, disease, predicates, shelter, water

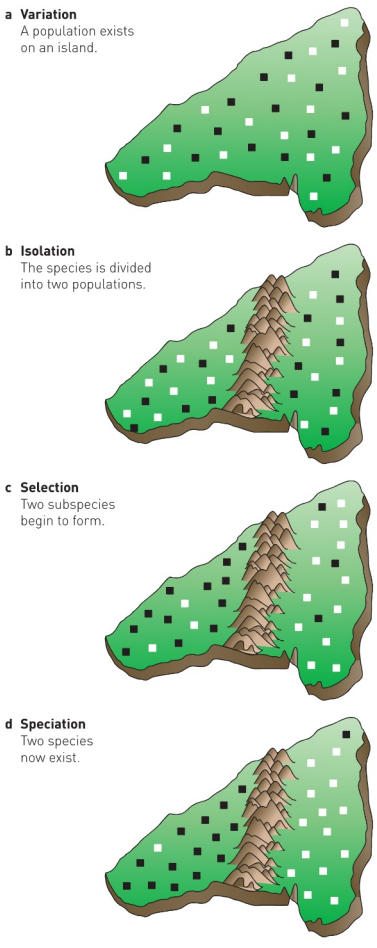
DAWINS THEORY

1) Reproduction produce variation
2) Organism best suited to the environment will survive and reproduce
3) Over time, the structure of the population will change (change in the gene pool)
4) This is called evolution

SPECIATION

1) Variation

A range of variation exists within the population, which shares a common gene pool



Sources of variation

- Sexual reproduction
- Mutation
- Epigenetics

2) Isolation

A barrier has formed, divining the population into two populations. No interbreeding occurs between the two population. Each population has a seperate gene pool

Types of barriers

- Geographical (oceans, rivers, mountains ect.)
- Behavioural
- Reproductive
- Sociocultural (ethic groups, religion, tribal, language)

4) Selection

Different selection pressures act on each of the two populations over a number of generations. This brings about change in the gene frequencies of each gene pool. Such changes lead to the evolution of seperate subspecies

Type of selection pressures

- Environment (rainfall, soil, temperature ect.)
- Disease
- Predation
- Mate selection

5) Speciation

Over a long period of time the changes in the gene frequencies may be great enough to prevent the production of fertile offspring by interbreeding between the two populations from even occurring again. When this happens, two species exist

6. DESCRIBE HOW THE FOLLOWING SUPPORT THE THEORY OF EVOLUTION: FOSSILS AND COMPARATIVE STUDIES (COMPARATIVE ANATOMY, EMBRYOLOGY, PROTEIN STUDIES AND DNA)

FOSSILS

- Fossil record have allowed scientists to build up a sequence of evolution of a particular animal or plant.
- They show common ancestors as fossil evidence can show similarities between species

COMPARATIVE STUDIES

Homologous structures

- Organs that are similar in structure, but have evolved to have differing functions
- Organisms that have homologous structure are likely to have a common ancestor

Vestigial organs

- Structures of reduced size that appear to have no function

Eg- appendix, wisdom teeth, ear moving muscles

Embryology

- Comparing early stages of development. Vertebrates have great similarity between species

Eg- They all have gill pouches/arches early in their development suggesting that all vertebrates started with fish

- Over times, divergence has occurred to give us 5 classes

Protein comparison

- Proteins are sequences of amino acids chains and can be identified
- Animals of the same species have identical amino acids in their proteins. Degree of difference between proteins is an estimate of the amount of evolution that has taken place

<u>Ubiquitous proteins</u>

- There are ones that appear in all species and perform veg basic but essential tasks needed for life, They carry out the same function no matter where they are found

Eg- Cytochrome C is used in evolutionary studies and has changes very little over time. By comparing the cytochromes C in different species. More support of evolution change can be done.

DNA comparison

- All living things use the same DNA code.
- The more similar the DNA sequences, the close the organisms are.

Eg- chimps and man show remarkable similarity (98%)
The DNA difference increase form chimps (98%) to gorillas (lower %) to other primates (much lower)

Endogenous retroviruses (ERV)

- These are non functional DNA (junk DNA)
- A viral sequence that becomes part of an organisms genome
- They store their genetic information as RNA. Copy it's RNA genome into DNA (called reverse transcription)
- More closely related species have more junk sequence in common.

Mitochondria DNA (mtDNA)
- Found in mitochondria in the form of small circular molecules (5-10 in each mitochondria) contain 37 genes - 13 with instructions to make enzymes, 24 for making mtDNA
- Only inherited from mother (egg cell) as sleek mitochondria is used up
- Has a higher rate of mutation than nuclear DNA. Human mtDNA is slowly diverging form our original ancestors
- The amount of mutation directly reacted to the time passed. Similarly between mtDNA can be used to estimate relationships
- If it is identical then closely related (sibilants, mother/child)
- Can be shed right trace migration routes of ancient people

7. EXPLAIN HOW COMPARATIVE GENOMICS, COMPARATIVE BIOCHEMISTRY AND BIOINFORMATICS ARE USED TO PROVIDE FURTHER EVIDENCE OF EVOLUTIONARY MECHANISMS.

COMPARATIVE GENOMICS
- The comparing of genome sequences of different species. Helps to identify regions of similarity and difference.
- Can be used to study evolutionary changes in organisms by measuring the changes in DNA
- Genome is an organism's complete set of DNA
- Allows researchers to identify genes that are preserved among species, as well as genes that give each species its unique characteristics

COMPARATIVE BIOCHEMISTRY
- Shows a common ancestor through comparing the DNA and proteins of organism the more difference in the sequence the more distantly related
- By comparing the sequence of the human genome with genomes of other organisms, researchers are able to identify regions of similarities and differences
- The procedure provides an effective means if studying evolutionary changes among organisms, helping to identify genes that are preserved among species, as well as genes that give each organisms its unique characteristics

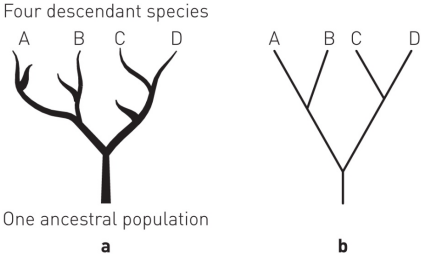
BIOINFORMATICS
- Uses all types of science including: computer science, statistics and applied mathematics to help understand biological processes.
- Uses computers to describe molecular components of living things.

Eg- measuring changes in DNA
- Provides the IT platform for the data collected by genomics.
- This platform capable of storing and managing the large amount of information collected

Annotation
- A process where genes and other features in a DNA sequence are identified. Done by computers because size of an entire genome is huge

8. UNDERSTAND THAT A PHYLOGENETIC TREE CAN BE CONSTRUCTED BY GENOMIC INFORMATION.

- A phylogenetic tree is a branching diagram or tree showing the inferred evolutionary relationships among various biological species
- It is based upon similarities and differences in their physical or genetic characteristics
- The root of the the tree represents the ancestral linage and the tip of the branches represents the descents of that ancestor
- As you move from the root to the tip, you are moving forward in time.



9. EXPLAIN THE TERM FOSSIL AND BE AWARE OF THE PROBLEMS WITH THE FOSSIL RECORD.

FOSSIL

Fossils are the preserved remains, or traces, of once-living organisms. They include footprints, burrows, shells or faeces.

PROBLEMS WITH FOSSIL RECORD

Few organism become fossilised

- Poor probability of becoming fossilised
- Fossil conditions must be suitable
- Soil layers must have minimal disturbance
- More likely for hard parts

Incomplete fossils

- Fossils need to be found
- Earth movements/rock cycling
- may have destroyed fossils
- Inaccessible sites

Classification of species

- Difficult to classify as a species unless you can observe “interbreeding”

Different interpretations

- Scientists may make different assumptions from the same evidence.

Eg- The debate of Neanderthal position in the human evolutionary tree

10. DESCRIBE THE TERM ABSOLUTE DATING AND USE AS EXAMPLES: POTASSIUM-ARGON AND RADIOCARBON DATING. KNOW THEIR LIMITATIONS.

ABSOLUTE DATING

- Absolute dating tells you the chronological age of an object (e.g. 500 yr).
- Absolute, or chronological dating, is based on any event which occurs at a constant rate or is repeated at regular intervals and is measurable.

Eg- Tree ring dating (dendrochonology), Radiometric/radioisotope and Luminescence

Radiometric/radioisotope dating

- Radiometric (also known as radioisotope or radioactive) dating is an absolute dating method for determining the chronological age of a rock or mineral by measuring the proportions of an original radioactive material and its decay product.
- Radioisotopes are unstable and break down or decay to form more stable isotopes of another element (the daughter product).
- Radioisotopes are radioactive, emitting radiation as they undergo decay, which can be measured. (Eg- Carbon-14 and Nitrogen-14)

Carbon 14 dating

A method of calculating the age of a fossil or artefact using the known rate of decay of radioactive carbon

- All living things contain carbon in a known ratio.
- C-14 is formed in the atmosphere by the action of cosmic radiation.
- Some of this C14 is taken in by plants during photosynthesis.
- When an organism dies, the C14 decays to C12, and the ratio of C14 to C12 changes
- This ratio can be measured and an estimate of age can be made
- The half life of C14 is 5730 yrs

Limitations:
- Can only use organic materials (with C)
- Need at least 3g of sample to use C14
- Amount of CO_2 in air can vary
- Only useful up to 50000 yrs

Potassium argon dating
A method of calculating the age of a fossil or artefact using the known rate of decay of radioactive potassium

- K-40 is a naturally occurring isotope.
- It decays to form Ar-40 and Ca-40
- Half life is 1.3 billion years.
- Used in dating volcanic rocks (igneous) and minerals older than 300,000 yrs

Limitations:
- Only used for volcanic rocks/ash
- Must be older than 200,000 yrs to get a reading

Luminescence
- Luminescence is a fairly recent technique used for dating artifacts such as stone tools and pottery.
- When crystals of thorium and potassium in the soil are irradiated, part of the radiation is released in the form of light and the rest is trapped in the crystal lattice of the material to be dated (e.g. pottery or stone).
- When the material is heated, the stored energy is released as light, the so called thermoluminescence effect.
- Thermoluminescence dating can be used to determine how much time has elapsed since the last time the object was heated. The older the object, the more light will be released.

11. DESCRIBE THE TERM RELATIVE DATING AND USE AS EXAMPLES: PRINCIPLE OF SUPERPOSITION, CORRELATION OF ROCK STRATA AND INDEX FOSSILS. KNOW THEIR LIMITATIONS.

RELATIVE DATING
Compares the age of a fossil or artefact relative to another fossil or artefact (ie whether they are older or younger)

Superposition
- Unless disturbed, the lower beds in a sedimentary sequence are older than the rocks above.
- For single location/artefact

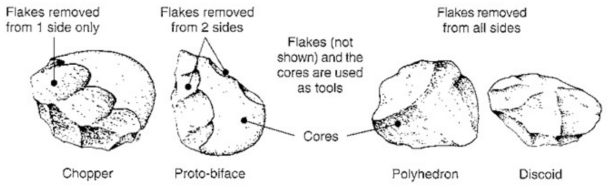
Comparative stratigraphy
- Stratigraphy is the study of sedimentary rocks.
- If the sequence of sedimentary rocks in different areas is similar it is likely that they are of the same age.

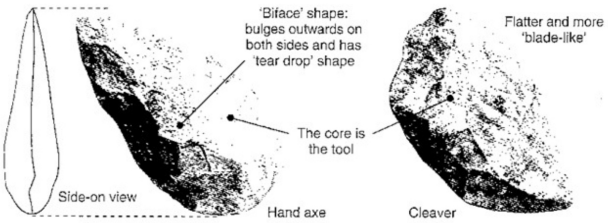
Index fossils
Fossil or organism that were on the earth for only a short period of time and therefore are useful in the relative dating if rock strata

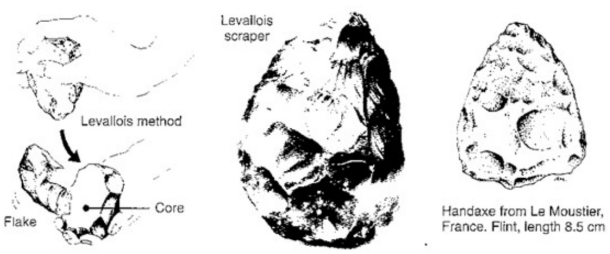
- Index fossils are of distinctive appearance, have a short time span and have a broad geographical distribution.
- If rocks in different locations contain the same index fossils, it is likely that both areas are of the same age.

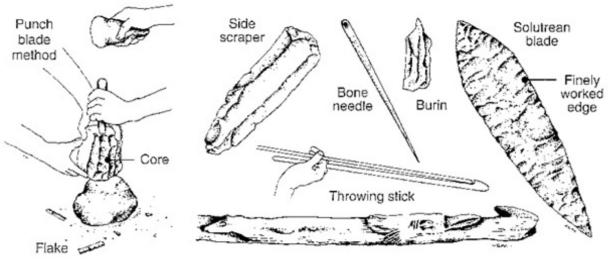
14. UNDERSTAND THAT THE STUDY OF THE MANUFACTURING OF, AND TRENDS. IN TOOL USE PROVIDE ANOTHER INSIGHT INTO THE LIFESTYLES OF;

	Description
OLDOWAN TOOLS Homo Habilis	- First appeared 2.5 million years ago - These tools were simple rive worn pebbles created with a minimum number of flakes being removing - Had a core and a cutting edge - Used for cutting
ACHEULIAN TOOLS Homo Erectus	- First appeared 1.5 million years ago - Used many more blows to make than oldowan tools - Tear drop shaped and contained a bi-face - They had a standard design but differed greatly in size
MOUSTERIAN TOOLS Homo Neanderthals	- First appeared 15,000 years ago - Flint became a preferred material to produce stone tools as it was more predictable in the way it would chip - Much finer workmanship was possible - The **levallios** tools making method involves preparing a core then striking off a large over flake which was retouched in one surface only
UPPER PALAEOLITHIC Homo Sapien and Homo Neanderthaleneis	- First appeared 40,000 years ago - **Punch blade** technique used to create tools - Long thin flakes were removed and shaped into tools - Many materials used such as bone, ivory and antler **Aurginacian** - 40,000 - 28,000 years ago - Associate with both Homo Sapien and Homo Neanderthalensis **Solutrean** - 22,000 - 19,000 years ago - Elegant tool designs made by heating and sudden cooling flint stone **Magdelanian** - 18,000 - 12,000 years ago - Delicate flaking stone for arrows and spears - Many new tools invented









TRENDS IN TOOL CULTURE

- Increase variety of materials used
- pebbles, stone, bone, antler, ivory, blade.
- Tools were made for specific tasks.
- Tools took longer to make.
- Increase in complexity and refinement of delicate tools.
- Made by increase in number of flakes/blows to core