**10 SCIENCE GENETICS REVISION**

**SC1 Define the following terms:**

DNA – **genetic code for a living organism**

Chromosome – **very long strand of DNA, found in all cells of the body**

Gene – **a section of a chromosome or DNA that codes for (is responsible for the making of) a protein**

Trait – **physical appearance, the phenotype of a genetically inherited condition**

Heredity – **the study of genetics, or how traits are inherited**

Karyotype – **the number and visual appearance of the chromosomes in the cell nuclei of an organism or species.**

**SC2 Explain the double helical structure of DNA**

**The double helix of DNA is a pair of vertical strands of sugar and phosphate molecules, being held together by base units joining them together like rungs on a twisted ladder. The base units occurs in pairs of bases. Adenosine and Thymine bond together to make one rung, while cytosine and guanine join together to form the other rung.**

**SC3 Describe the role of DNA in living things**

**DNA provides the blueprint for living organisms. The sequence of the bases within the DNA cause certain proteins to be made. The proteins control the growth and development of the organism and its metabolism and functioning.**

**SC4 Define the following:**

Punnet square **- A technique for predicting the outcomes of a monohybrid cross**

Genotype **- the two alleles within an organism**

Phenotype **- The physical appearance of a trait in an organism**

Recessive allele **- the allele which is “masked”, and only expressed of two recessive alleles are present.**

Dominant allele **- the allele which is always expressed if present.**

Offspring **- the children or next generation of parents mating.**

Homozygous **- is when an organism has two identical alleles – both recessive, or both dominant.**

Purebred **- as above. It means that the organism gives a repeatable inheritance pattern.**

Heterozygous  **- is when an organism has two different alleles – one recessive, one dominant.**

Monohybrid **- Is the cross (mating) involving the inheritance of a single gene.**

**SC5 Use punnet squares for monohybrid crosses to predict genotypes and phenotypes of offspring**

Practice Question: Dark hair is dominant over light hair in pigs. Cross a Homozygous dark haired male with a heterozygous female to find their genotype and phenotype.

**Not sex-linked so use “normal” punnet square and alleles, D and d.**

|  |  |  |
| --- | --- | --- |
|  | **D** | **D** |
| **D** | **DD****Dark hair** | **DD****Dark hair** |
| **d** | **Dd****Dark hair** | **Dd****Dark hair** |

**Genotype: 50%DD, 50% Dd**

**Phenotype: 100% dark hair (all four outcomes have a dominant allele)**

**SC6 Use punnet squares for sex-linked crosses to predict genotypes and phenotypes of offspring**

Practice question: In humans, diabetes is a sex-linked recessive trait. If a homozygous normal woman and a diabetic man have children, predict the phenotypes and genotypes of their children.

**Is sex-linked so use XY for males, XX for females, and males only get one allele. Use D and d**

|  |  |  |
| --- | --- | --- |
|  | **Xd** | **Y** |
| **XD** | **XDXd “normal”** | **XDY****“normal”** |
| **xD** | **XDXd “normal”** | **XDY****“normal”** |

**Genotype: MALES – 100% XDY; FEMALES – 100% XDXd**

**Phenotype: MALES/FEMALES – 100% normal (all have a dominant allele)**

**SC7 Use a pedigree chart to represent patterns of inheritance**

**MALE**

**FEMALE**

**Offspring**

**Unaffected (“notmal”)nmFEMALE**

**Affected MALE**

**SC8 Analyse inheritance patterns to predict genotypes and phenotypes of parents and offspring**

**The following pedigree shows a condition “Faulty tooth enamel” which is inherited by having a dominant allele**



1. State how many generations are shown in the pedigree **Three**
2. Identify the phenotype of individual II2, II4 and II8

**II2 – normal tooth enamel, II4 – normal II8 - normal**

1. Identify the relationship between individual II5 and III7 **Siblings (brother sister)**
2. Write down the genotypes for all even numbered individuals. Answer uses F and f as the alleles

**I2 – Ff; II2 – ff, II4 – ff, II6 – Ff, II8 – ff; III2 – Ff, III4 – ff, III6 – ff, III\* - ff, III10 - Ff**

1. Identify if this a dominant or a recessive disorder, and explain how you know this. **Dominant inheritance pattern, as the trait turns up in every generation and does not does not skip a generation anywhere in the pedigree.** However, the is NOT PROOF, and the pedigree could be recessive, but it is very difficult to prove this as the common signs of a recessive pedigree (see answer 11a) are not in this one
2. Does individual I1 have a heterozygous or a homozygous genotype? Justify your answer. **Individual I1 would be homozygous recessive (ff)**



Colour blindness is a sex linked trait in humans.

Predict the Genotype and phenotype of individuals in this pedigree

**I1 = XbY; I2 = XBXB Generation II: 1= XBY; 2= XBY; 3 = XBXB or XBXb 4 = XBXb; 5 = XBY; 6 = XBXb**

**Generation III: 1= XBXB or XBXb; 2= XBY; 3= XBXB or XBXb; 4= XbY; 5= XBXB or XBXb**

Infer if colour blindness is a dominant or recessive trait – justify your response

**Recessive – it skipped a generation. It is present as a phenotype in Gen 1, but not in gen 2, then turns up in Gen3.**

**SC9 Explain the types of mutations and how they occur**

**All mutations are a change in the normal pattern of bases within a gene. They can be caused by:**

* **Errors in the copying of DNA within the production of the sex cells**
* **Ultra violet radiation**
* **Radioactivity**
* **Exposure to chemicals known to induce DNA changes such as tobacco products.**

**SC10 Outline at least 2 factors that cause mutations and the effect these may have on living things**

**See answer above for four factors that can cause mutations. Germline mutations occur in every cell and cause chronic conditions such as haemophilia. Somatic mutaions occur during a lifetime and affect specific areas of the body usually in the form of cancer and tumours**

**SC11 Define the following:**

Evolution – **is the gradual change in a species over a long period of time (many generations)**

Natural selection – **organisms better adapted to their environment are more likely to survive and reproduce, thus passing their adaptations onto their offspring**.

Species – **unique type of organism that can successfully breed with others of its species**

Variation – **differences between organisms within the same species**.

Isolation -

Biodiversity – **refers to the number of different species in a habitat**

**SC12 Outline the processes involved in natural selection including**

Variation – **offspring are born different to each other**

Isolation – **the habitat of a species become unique to them and there is separation from other habitats**

Selection – **the differences between the offspring mean some are more adapted to their environment and these organism are more able to survive and reproduce.**

**SC13 Describe biodiversity as a function of evolution**

The longer a habitat exists, or the more stable it is (unchanging), the more likely it is that organisms will be born with differences which make that habitat suitable for them. This means stable habitats can have a large number of species in them (biodiversity)

**SC15 Explain how genetic characteristics relate to survival and reproductive rates**

Offspring are born genetically different to each other. This means the offspring are physically different to each other. Some of the differences will prove to be an advantage in their habitat, and animals with these differences are more likely to survive. The other, less adapted individuals are more likely to die, leaving the successful organisms to reproduce.die,

**SC16 Define the following:**

Evidence **this are directly from notes – consult those for answers**

Fossil Record

Homologous Structures

Geographical distribution

**SC17 Explain how the following provide evidence for evolution:**

Fossil record - **Scientists can compare the older fossils of a species (or simila species) with younger fossils. This allows scientists to show how there have been changes in species (evolution) over time**

DNA profile - **Scientists can now look at how similar the DNA is in two separate species. This can show how closely they are related. The less alike organisms are the less similarity in their DNA, suggesting that evolution from a common ancestor occurred a very long time ago, but that very similar species share more recent common ancestors**.

Embryology -**The embryos of most vertebrates look very similar and have similar structures This suggests that all animals have a similar origon or ancient ancestor**

Anatomical similarities - **Many animals have body parts that are similar in both structure and function - these are called homologous structures. This suggests that the anatomy of animals is inherited from a common ancestor and evolved over time in different species**