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| **Topic 3: Genetics (18 hours)** | | | |
| **Essential idea:** Every living organism inherits a blueprint for life from its parents. | | | |
| **3.1 Genes** | | | |
| **Nature of science:**  **3.1.NOS1** Developments in scientific research follow improvements in technology—gene sequencers are used for the sequencing of genes. (1.8) | | | Pg.148 |
| **Understandings:** | | **International-mindedness:**  • Sequencing of the human genome shows that all humans share the vast majority of their base sequences but also that there are many single nucleotide polymorphisms that contribute to human diversity.  **Theory of knowledge:**  • There is a link between sickle cell anemia and prevalence of malaria. How can we know whether there is a causal link in such cases or simply a correlation?  **Aims:**  • **Aim 7:** The use of a database to compare DNA base sequences.  • **Aim 8:** Ethics of patenting human genes. | |
| **3.1.U1** A gene is a heritable factor that consists of a length of DNA and influences a specific characteristic. | Pg.142 |
| **3.1.U2** A gene occupies a specific position on a chromosome. | Pg.143 |
| **3.1.U3** The various specific forms of a gene are alleles. | Pg.143-144 |
| **3.1.U4** Alleles differ from each other by one or only a few bases. | Pg.144 |
| **3.1.U5** New alleles are formed by mutation. | Pg.145 |
| **3.1.U6** The genome is the whole of the genetic information of an organism. | Pg.147 |
| **3.1.U7** The entire base sequence of human genes was sequenced in the Human Genome Project. | Pg.147 |
| **Applications and skills:** | |
| **3.1.A1** Application: The causes of sickle cell anemia, including a base substitution mutation, a change to the base sequence of mRNA transcribed from it and a change to the sequence of a polypeptide in hemoglobin. | Pg. 146 |
| **3.1.A2** Application: Comparison of the number of genes in humans with other species. | Pg.142 |
| **3.1.S1** Skill: Use of a database to determine differences in the base sequence of a gene in two species. | Pg.144 |
| **Guidance:**  • Students should be able to recall one specific base substitution that causes glutamic acid to be substituted by valine as the sixth amino acid in the hemoglobin polypeptide.  • The number of genes in a species should not be referred to as genome size as this term is used for the total amount of DNA. At least one plant and one bacterium should be included in the comparison and at least one species with more genes and one with fewer genes than a human.  The Genbank® database can be used to search for DNA base sequences. The cytochrome C gene sequence is available for many different organisms and is of particular interest because of its use in reclassifying organisms into three domains.  • Deletions, insertions and frame shift mutations do not need to be included. |  |

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| **Topic 3: Genetics (18 hours)** | | | |
| **Essential idea:** Chromosomes carry genes in a linear sequence that is shared by members of a species. | | | |
| **3.2 Chromosomes** | | | |
| **Nature of science:**  **3.2.NOS1** Developments in research follow improvements in techniques—autoradiography was used to establish the length of DNA molecules in chromosomes. (1.8) | | | Pg.150-151 |
| **Understandings:** | | **International-mindedness:**  • Sequencing of the rice genome involved cooperation between biologists in 10 countries.  **Utilization:**  Syllabus and cross-curricular links:  Biology  Topic 1.6 Cell division  **Aims:**  • **Aim 6:** Staining root tip squashes and microscope examination of chromosomes is recommended but not obligatory.  • **Aim 7:** Use of databases to identify gene loci and protein products of genes. | |
| **3.2.U1** Prokaryotes have one chromosome consisting of a circular DNA molecule. | Pg.149-150 |
| **3.2.U2** Some prokaryotes also have plasmids but eukaryotes do not. | Pg.150 |
| **3.2.U3** Eukaryote chromosomes are linear DNA molecules associated with histone proteins. | Pg.151-152 |
| **3.2.U4** In a eukaryote species there are different chromosomes that carry different genes. | Pg.152 |
| **3.2.U5** Homologous chromosomes carry the same sequence of genes but not necessarily the same alleles of those genes. | Pg.152 |
| **3.2.U6** Diploid nuclei have pairs of homologous chromosomes. | Pg.155 |
| **3.2.U7** Haploid nuclei have one chromosome of each pair. | Pg.154 |
| **3.2.U8** The number of chromosomes is a characteristic feature of members of a species. | Pg.155 |
| **3.2.U9** A karyogram shows the chromosomes of an organism in homologous pairs of decreasing length. | Pg.157-158 |
| **3.2.U10** Sex is determined by sex chromosomes and autosomes are chromosomes that do not determine sex. | Pg.157 |
| **Applications and skills:** | |
| **3.1.A1** Application: Cairns’ technique for measuring the length of DNA molecules by autoradiography. | Pg.151 |
| **3.1.A2** Application: Comparison of genome size in T2 phage, *Escherichia coli*, *Drosophila melanogaster*, *Homo sapiens* and *Paris japonica.* | Pg.153-154 |
| **3.1.A3** Application: Comparison of diploid chromosome numbers of *Homo sapiens*, *Pan troglodytes*, *Canis familiaris*, *Oryza sativa*, *Parascaris equorum.* | Pg.155-156 |
| **3.1.A4** Application: Use of karyograms to deduce sex and diagnose Down syndrome in humans. | Pg.158 |
| **3.1.S1** Skill: Use of databases to identify the locus of a human gene and its polypeptide product. | Pg.154 |
| **Guidance:**  • The terms karyotype and karyogram have different meanings. Karyotype is a property of a cell—the number and type of chromosomes present in the nucleus, not a photograph or diagram of them.  • Genome size is the total length of DNA in an organism. The examples of genome and chromosome number have been selected to allow points of interest to be raised.  • The two DNA molecules formed by DNA replication prior to cell division are considered to be sister chromatids until the splitting of the centromere at the start of anaphase. After this, they are individual chromosomes |  |

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| **Topic 3: Genetics (18 hours)** | | | |
| **Essential idea:** Alleles segregate during meiosis allowing new combinations to be formed by the fusion of gametes. | | | |
| **3.3 Meiosis** | | | |
| **Nature of science:**  **3.3.NOS1** Making careful observations—meiosis was discovered by microscope examination of dividing germ-line cells. (1.8) | | | Pg.160 |
| **Understandings:** | | **Theory of knowledge:**  • In 1922 the number of chromosomes counted in a human cell was 48. This remained the established number for 30 years, even though a review of photographic evidence from the time clearly showed that there were 46. For what reasons do existing beliefs carry a certain inertia?  **Utilization:**  • An understanding of karyotypes has allowed diagnoses to be made for the purposes of genetic counselling.  Syllabus and cross-curricular links:  Biology  Topic 1.6 Cell division  Topic 10.1 Meiosis  Topic 11.4 Sexual reproduction  **Aims:**  • **Aim 8:** Pre-natal screening for chromosome abnormalities gives an indication of the sex of the fetus and raises ethical issues over selective abortion of female fetuses in some countries. | |
| **3.3.U1** One diploid nucleus divides by meiosis to produce four haploid nuclei. | Pg.160-161 |
| **3.3.U2** The halving of the chromosome number allows a sexual life cycle with fusion of gametes. | Pg.161 |
| **3.3.U3** DNA is replicated before meiosis so that all chromosomes consist of two sister chromatids. | Pg.162 |
| **3.3.U4** The early stages of meiosis involve pairing of homologous chromosomes and crossing over followed by condensation. | Pg.162 |
| **3.3.U5** Orientation of pairs of homologous chromosomes prior to separation is random. | Pg.162-163 |
| **3.3.U6** Separation of pairs of homologous chromosomes in the first division of meiosis halves the chromosome number. | Pg.163 |
| **3.3.U7** Crossing over and random orientation promotes genetic variation. | Pg.165-166 |
| **3.3.U8** Fusion of gametes from different parents promotes genetic variation. | Pg.166-167 |
| **Applications and skills:** | |
| **3.3.A1** Application: Non-disjunction can cause Down syndrome and other chromosome abnormalities. | Pg.167 |
| **3.3.A2** Application: Studies showing age of parents influences chances of nondisjunction. | Pg. 167 (dbqs) |
| **3.3.A3** Application: Description of methods used to obtain cells for karyotype analysis e.g. chorionic villus sampling and amniocentesis and the associated risks. | Pg.163 |
| **3.3.S1** Skill: Drawing diagrams to show the stages of meiosis resulting in the formation of four haploid cells. | Pg.164 |
| **Guidance:**  • Preparation of microscope slides showing meiosis is challenging and permanent slides should be available in case no cells in meiosis are visible in temporary mounts.  • Drawings of the stages of meiosis do not need to include chiasmata.  • The process of chiasmata formation need not be explained. |  |

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| **Topic 3: Genetics (18 hours)** | | | |
| **Essential idea:** Meiosis leads to independent assortment of chromosomes and unique composition of alleles in daughter cells. | | | |
| **10.1 Meiosis** | | | |
| **Nature of science:**  **10.1NOS1** Making careful observations—careful observation and record keeping turned up anomalous data that Mendel’s law of independent assortment could not account for. Thomas Hunt Morgan developed the notion of linked genes to account for the anomalies. (1.8) | | | Pg.441 |
| **Understandings:** | | **Utilization:**  Syllabus and cross-curricular links:  Biology  Topic 1.6 Cell division  Topic 3.3 Meiosis  Topic 11.4 Sexual reproduction  **Aims:**  • **Aim 6:** Staining of lily anthers or other tissue containing germ-line cells and microscope examination to observe cells in meiosis are possible activities. | |
| **10.1U1** Chromosomes replicate in interphase before meiosis. | Pg.440 |
| **10.1U2** Crossing over is the exchange of DNA material between non-sister homologous chromatids. | Pg.440 |
| **10.1U3** Crossing over produces new combinations of alleles on the chromosomes of the haploid cells. | Pg.441-442 |
| **10.1U4** Chiasmata formation between non-sister chromatids can result in an exchange of alleles. | Pg.440 |
| **10.1U5** Homologous chromosomes separate in meiosis I. | Pg.443-444 |
| **10.1U6** Sister chromatids separate in meiosis II. | Pg.444 |
| **10.1U7** Independent assortment of genes is due to the random orientation of pairs of homologous chromosomes in meiosis I. | Pg.444 |
| **Applications and skills:** | |
| **10.1.S1** Skill: Drawing diagrams to show chiasmata formed by crossing over. | Pg.442 |
| **Guidance:**  • Diagrams of chiasmata should show sister chromatids still closely aligned, except at the point where crossing over occurred and a chiasma was formed. |  |

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| **Topic 3: Genetics (18 hours)** | | | |
| **Essential idea:** The inheritance of genes follows patterns. | | | |
| **3.4 Inheritance** | | | |
| **Nature of science:**  **3.4.NOS1** Making quantitative measurements with replicates to ensure reliability. Mendel’s genetic crosses with pea plants generated numerical data. (3.2) | | | Pg.169-170 |
| **Understandings:** | | **Theory of knowledge:**  • Mendel’s theories were not accepted by the scientific community for a long time. What factors would encourage the acceptance of new ideas by the scientific community?  **Utilization:**  Syllabus and cross-curricular links:  Biology  Topic 1.6 Cell division  **Aims:**  • **Aim 8:** Social implications of diagnosis of mutations, including the effects on the family and stigmatization. | |
| **3.4.U1** Mendel discovered the principles of inheritance with experiments in which large numbers of pea plants were crossed. | Pg.169 |
| **3.4.U2** Gametes are haploid so contain only one allele of each gene. | Pg.171 |
| **3.4.U3** The two alleles of each gene separate into different haploid daughter nuclei during meiosis. | Pg.171-172 |
| **3.4.U4** Fusion of gametes results in diploid zygotes with two alleles of each gene that may be the same allele or different alleles. | Pg.171 |
| **3.4.U5** Dominant alleles mask the effects of recessive alleles but co-dominant alleles have joint effects. | Pg.172 |
| **3.4.U6** Many genetic diseases in humans are due to recessive alleles of autosomal genes, although some genetic diseases are due to dominant or co-dominant alleles. | Pg.177-178 |
| **3.4.U7** Some genetic diseases are sex-linked. The pattern of inheritance is different with sex-linked genes due to their location on sex chromosomes. | Pg.178-180 |
| **3.4.U8** Many genetic diseases have been identified in humans but most are very rare. | Pg.183 |
| **3.4.U9** Radiation and mutagenic chemicals increase the mutation rate and can cause genetic diseases and cancer. | Pg.184-185 |
| **Applications and skills:** | |
| **3.4.A1** Application: Inheritance of ABO blood groups. | Pg.174 |
| **3.4.A2** Application: Red-green colour blindness and hemophilia as examples of sexlinked inheritance. | Pg.180-181 |
| **3.4.A3** Application: Inheritance of cystic fibrosis and Huntington’s disease. | Pg.178-179 |
| **3.4.A4** Application: Consequences of radiation after nuclear bombing of Hiroshima and accident at Chernobyl. | Pg.184-185 |
| **3.4.S1** Skill: Construction of Punnett grids for predicting the outcomes of monohybrid genetic crosses. | Pg.173 |
| **3.4.S2** Skill: Comparison of predicted and actual outcomes of genetic crosses using real data. | Pg.175-176 |
| **3.4.S3** Skill: Analysis of pedigree charts to deduce the pattern of inheritance of genetic diseases. | Pg.181-182 |
| **Guidance:**  • Alleles carried on X chromosomes should be shown as superscript letters on an upper case X, such as Xh.  • The expected notation for ABO blood group alleles is:  *Phenotype* O *Genotype* ii  A IAIA or IAi  B IBIB or IBi  AB IAIB |  |

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| **Topic 3: Genetics (18 hours)** | | | |
| **Essential idea:** Genes may be linked or unlinked and are inherited accordingly. | | | |
| **10.2 Inheritance** | | | |
| **Nature of science:**  **10.2.NOS1** Looking for patterns, trends and discrepancies—Mendel used observations of the natural world to find and explain patterns and trends. Since then, scientists have looked for discrepancies and asked questions based on further observations to show exceptions to the rules. For example, Morgan discovered non-Mendelian ratios in his experiments with *Drosophila*. (3.1) | | | Pg.447 |
| **Understandings:** | | **Theory of knowledge:**  • The law of independent assortment was soon found to have exceptions when looking at linked genes. What is the difference between a law and a theory in science?  **Utilization:**  • An understanding of inheritance allowed farmers to selectively breed their livestock for specific characteristics.  Syllabus and cross-curricular links:  Biology  Topic 3.4 Inheritance  **Aims:**  • **Aim 4:** Use analytical skills to solve genetic crosses.  • **Aim 8:** Ethical issues arise in the prevention of the inheritance of genetic disorders. | |
| **10.2.U1** Gene loci are said to be linked if on the same chromosome. | Pg.448-449 |
| **10.2.U2** Unlinked genes segregate independently as a result of meiosis. | Pg.445 |
| **10.2.U3** Variation can be discrete or continuous. | Pg.449 |
| **10.2.U4** The phenotypes of polygenic characteristics tend to show continuous variation. | Pg.450 |
| **10.2.U5** Chi-squared tests are used to determine whether the difference between an observed and expected frequency distribution is statistically significant. | Pg.453 |
| **Applications and skills:** | |
| **10.2.A1** Application: Morgan’s discovery of non-Mendelian ratios in *Drosophila.* | Pg.448 |
| **10.2.A2** Application: Completion and analysis of Punnett squares for dihybrid traits. | Pg.446 |
| **10.2.A3** Application: Polygenic traits such as human height may also be influenced by environmental factors. | Pg.450-451 |
| **10.2.S1** Skill: Calculation of the predicted genotypic and phenotypic ratio of offspring of dihybrid crosses involving unlinked autosomal genes. | Pg.447 |
| **10.2.S2** Skill: Identification of recombinants in crosses involving two linked genes. | Pg.451-452 |
| **10.2.S4** Skill: Use of a chi-squared test on data from dihybrid crosses. | Pg.454 (dbqs) |
| **Guidance:**  • Alleles are usually shown side by side in dihybrid crosses, for example, TtBb. In representing crosses involving linkage, it is more common to show them as vertical pairs, for example:  T B  \_\_\_\_\_  t b  • This format will be used in examination papers, or students will be given sufficient information to allow them to deduce which alleles are linked. |  |