Name:

Understandings, Applications and Skills (This is what you maybe assessed on)

	Statement	Guidance
3.4.U1	Mendel discovered the principles of inheritance with experiments in which large numbers of pea plants were crossed.	
3.4.U2	Gametes are haploid so contain only one allele of each gene.	
3.4.U3	The two alleles of each gene separate into different haploid daughter nuclei during meiosis.	
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.	
3.4.U5	Dominant alleles mask the effects of recessive alleles but co-dominant alleles have joint effects.	
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.	
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.	Alleles carried on X chromosomes should be shown as superscript letters on an upper case X, such as X^h.
3.4.U8	Many genetic diseases have been identified in humans but most are very rare.	
3.4.U9	Radiation and mutagenic chemicals increase the mutation rate and can cause genetic diseases and cancer.	
3.4.A1	Inheritance of ABO blood groups.	The expected notation for ABO blood group alleles: O = i, A=I^A, B = I^B.
3.4.A2	Red-green colour blindness and hemophilia as examples of sex-linked inheritance.	
3.4.A3	Inheritance of cystic fibrosis and Huntington's disease.	
3.4.A4	Consequences of radiation after nuclear bombing of Hiroshima and accident at Chernobyl.	
3.4.S1	Construction of Punnett grids for predicting the outcomes of monohybrid genetic crosses.	
3.4.S2	Comparison of predicted and actual outcomes of genetic crosses using real data.	
3.4.S3	Analysis of pedigree charts to deduce the pattern of inheritance of genetic diseases.	

Recommended resources:

http://bioknowledgy.weebly.com/34-inheritance.html

Allott, Andrew. *Biology: Course Companion.* S.l.: Oxford UP, 2014. Print.

3.4.U1 Mendel discovered the principles of inheritance with experiments in which large numbers of pea plants were crossed.

1. Mendel is known as the father of genetics for his extensive experimental work with peas. His findings enabled him to form the principles of inheritance. Use the DNA Interactive animations) to find out about Mendel:
    - https://www.dnalc.org/view/16002-Gregor-Mendel-and-pea-plants.html
    - https://www.dnalc.org/view/16170-Animation-3-Gene-s-don-t-blend-.html

    a. State the approximate number of seeds used in each trial.

    b. List three examples of traits Mendel investigated.

    c. Explain what is meant by the term 'pure-bred'.

    d. Describe the key experimental finding that led to the establishment of the principles of inheritance.

Nature of science: Making quantitative measurements with replicates to ensure reliability. Mendel's genetic crosses with pea plants generated numerical data. (3.2)

2. To reach valid conclusions often statistical tests are used to help analyse the data collected. Outline why large sample sizes are preferable to smaller ones.

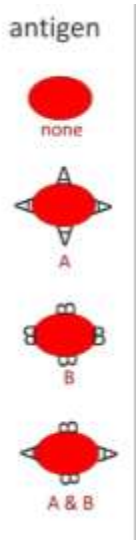
3.4.U2 Gametes are haploid so contain only one allele of each gene.
3.4.U3 The two alleles of each gene separate into different haploid daughter nuclei during meiosis.
3.4.U4 Fusion of gametes results in diploid zygotes with two alleles of each gene that may be the same allele or different alleles.

3. State definitions of the following:

Genotype	
Phenotype	
Dominant allele	
Recessive allele	
Codominant alleles	
Homozygous	
Heterozygous	
Carrier	
Phenotype	
Autosomal genes	
Sex-linked inheritance	

3.4.A1 Inheritance of ABO blood groups. AND 3.4.U5 Dominant alleles mask the effects of recessive alleles but co-dominant alleles have joint effects.

4. Human ABO blood types follow a codominant inheritance pattern.
   a. Describe what is meant by "some genes have multiple alleles."



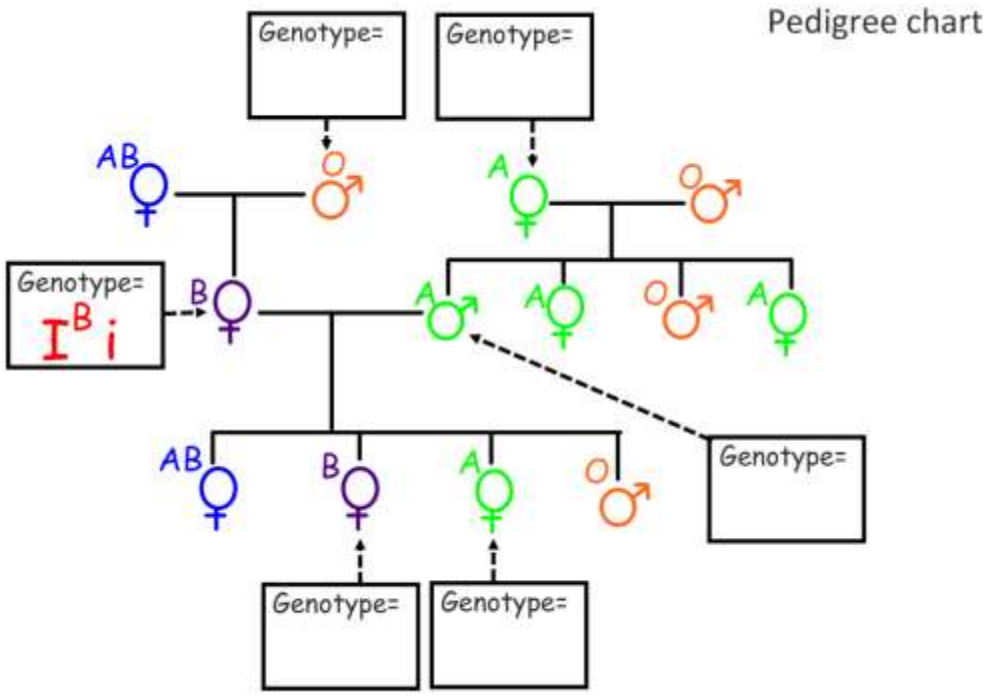
   b. Complete the table (both genotype and phenotype) below to show how blood type is inherited.

alleles	i	I^A	I^B
i			
I^A			
I^B			

5. State the genotype and phenotype which is an example of codominance.

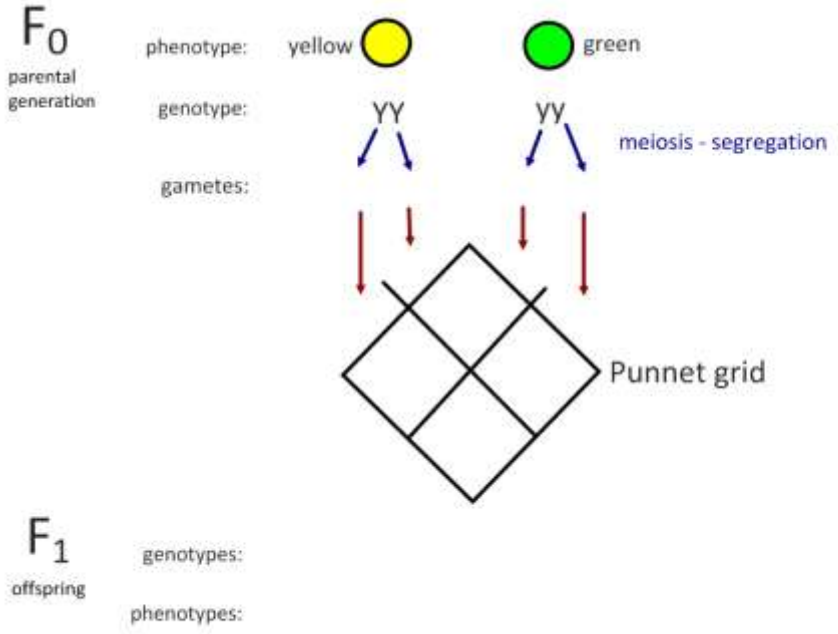
6. Explain why the identified genotype above is an example of codominance.

7. Complete this pedigree chart to show the inheritance of blood types in this family.

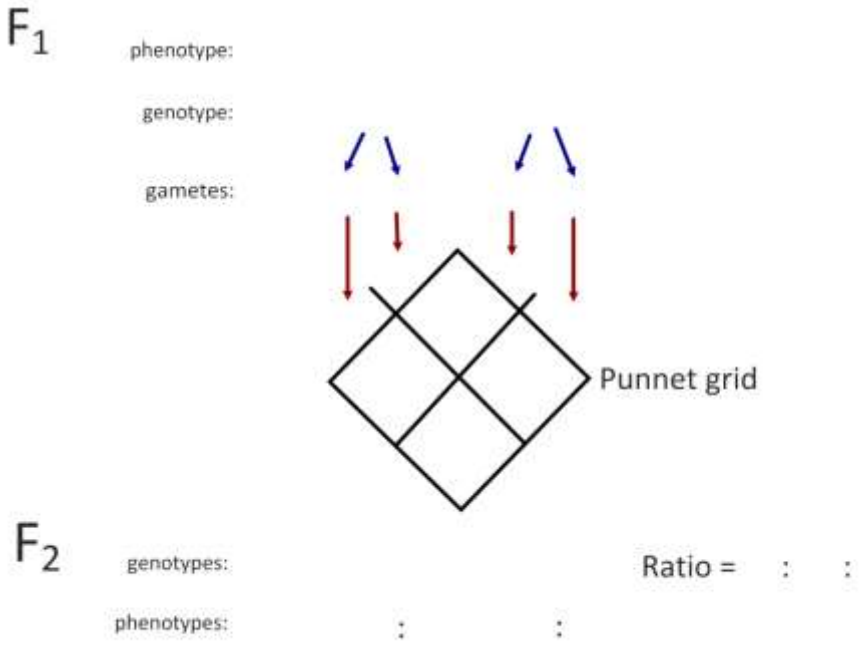
8. 

3.4.S1 Construction of Punnett grids for predicting the outcomes of monohybrid genetic crosses.

9. One of Mendel's experiments looked at the trait for pea colour.
   a. Complete the punnet grid below to show the outcome of the monohybrid cross that results in peas of different colours.



   b. Complete the punnet grid below to show the possible outcomes of a cross between **two members of the F1 generation**. Describe all genotypes produced.



c. A **test cross** is where an unknown genotype is breed with a **homozygous recessive individual**. Explain how a test cross could be used to determine the genotype of a yellow pea.

3.4.S2 Comparison of predicted and actual outcomes of genetic crosses using real data.

10. Cat genetics - do the inherited traits match what we know about cat genes?
    a. View the presentation on inheritance (http://www.slideshare.net/diverzippy/bioknowledgy-presentation-on-34-inheritance) and use the cat genetics slide to predict the **expected ratio of genotypes and phenotype of the piebald trait** a based on a cross of the phenotypes shown by the parents.

Parents	male	female
Phenotype		
Genotype		
Gametes		
Punnett grid		

Offspring

genotype (and ratio)

phenotype (and ratio)

b. Complete the table to compare expected genotype ratio with the observed outcomes from the cross.

genotype	observed	expected
No white (ss)		
Some White (Ss)		
Mostly White (SS)		

c. Use the chi squared formula and the critical values table to determine whether the actual outcome matches the predicted cross.

Chi-square value = $\sum_{i=1}^{N} \frac{(obs_i - exp_i)^2}{exp_i}$ =

Degrees of freedom (df) = Number of classes – 1 =

Is the hypothesis outlined by the theory of pielbald genetics and the expected cross outcomes supported by the data?
(is Chi-square value < critical value)

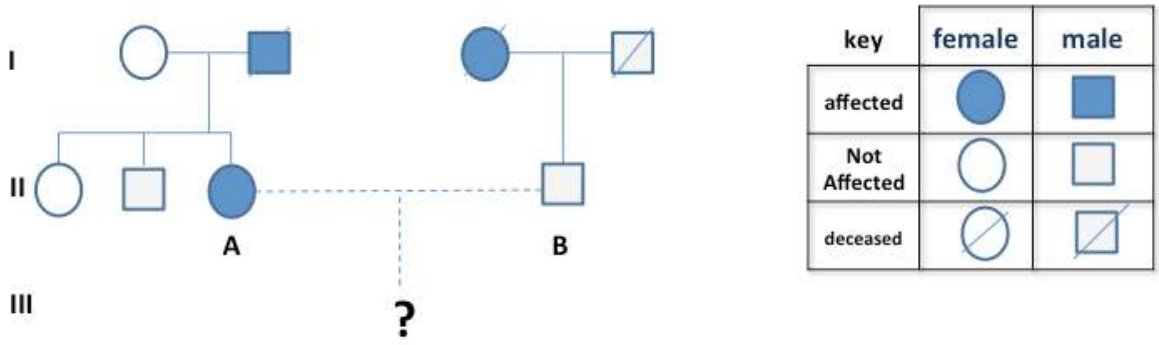
df	critical values at 5%
1	3.84
2	5.99
3	7.82
4	9.49
5	11.07

3.4.U8 Many genetic diseases have been identified in humans but most are very rare.

11. Explain why genetic diseases are very rare in humans.

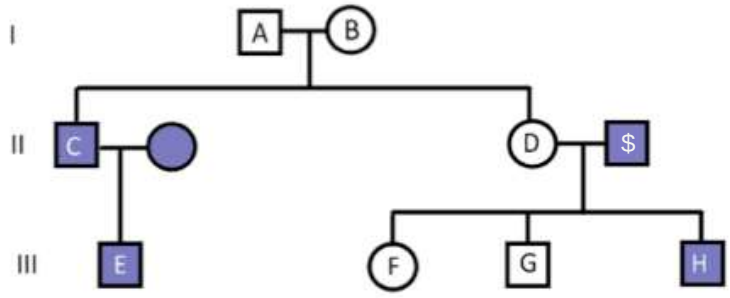
3.4.U6 Many genetic diseases in humans are due to recessive alleles of autosomal genes, although some genetic diseases are due to dominant or co-dominant alleles. AND 3.4.A3 Inheritance of cystic fibrosis and Huntington's disease.

12. Cystic fibrosis (CF) is caused by a mutation in the CFTR gene. Secretions (e.g. mucus, sweat and digestive juices) which are usually thin instead become thick. The secretions block tubes, ducts and passageways. Lung problems in most CF sufferers leads to a early death.
    a. Analyse the pedigree chart below and deduce whether CF is a recessive, dominant or codominant condition. Quote your evidence in your answer.



    b. What is the probability of two parents who are both carriers of (one copy of) the recessive allele producing children affected by CF? Show your workings.

    c. Deduce the genotypes of the selected individuals.



Individual	Genotype
A	
B	
C	
D	

d. D and $ are planning to have another child. Using the information in the pedigree chart in the last question to calculate the % chance that the child will suffer from CF.

13. Huntington's Disease (HD) is a brain disorder that affects a person's ability to think, talk, and move. HD is caused by a mutation in a gene on chromosome 4.
14. Is this a dominant or recessive condition?

15. Is this disorder autosomal or sex-linked

16. Produce a punnett grid to explain the inheritance pattern seen in the offspring of a normal mother and a heterozygous affected father. Don't forget to use a key to explain the genotypes and allele symbols used.

17. Sickle cell disease is another example of codominant inheritance.

a. State the genotypes description, phenotypes and malaria protection of these individuals.

genotype	Hb^AHb^A	Hb^AHb^s	Hb^sHb^s
description			
phenotype			
Malaria protection?			

Allele key: Hb^A produces normal haemoglobin, Hb^S produces fibrous haemoglobin that causes red blood cells to sickle.

b. Predict the phenotype ratios of offspring in the following crosses. Show all your working, and set it out as expected. Take care with notation.

i. Carrier mother with affected father.

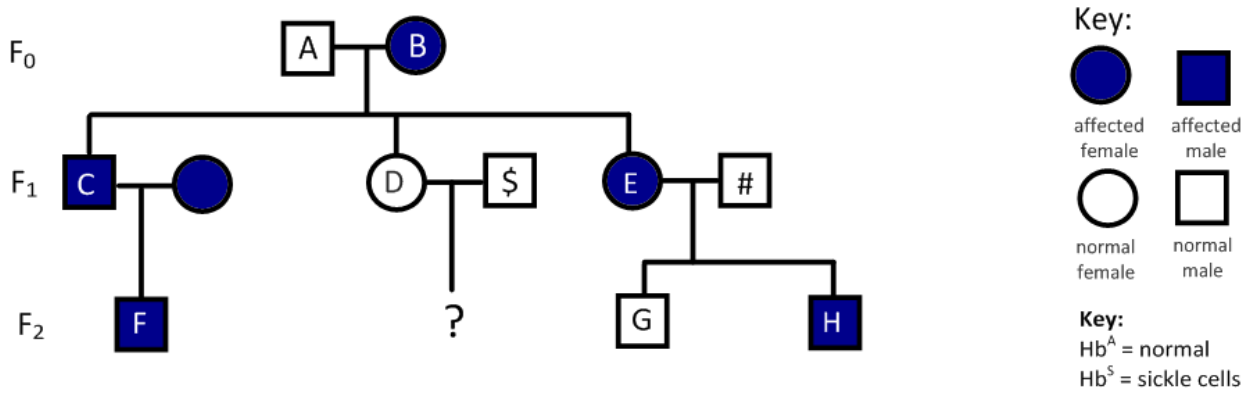
ii. Affected father with unaffected mother.

iii. Carrier mother with carrier father.

3.4.S3 Analysis of pedigree charts to deduce the pattern of inheritance of genetic diseases.

18. The pedigree chart below shows a family affected by sickle cell:
    a. Deduce the genotype of each individual with a letter.

A		F	
B		G	
C		H	
D		$	
E		#	



    b. Calculate the likelihood of any further children produced by E and her # having sickle cell anemia.

    c. Male $ is healthy but of unknown genotype. Calculate the likelihood of any children produced with female D having sickle cell anemia. Show all working.

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. AND 3.4.A2 Red-green colour blindness and hemophilia as examples of sex-linked inheritance.

19. Some inherited disorders are associated with gender.
    a. State two examples of sex-linked genetic disorders.

    b. Explain why sex-linked disorders are more common in males than females.

    c. Explain why human females can be homozygous or heterozygous for sex-linked genes, where males cannot.

    d. The allele for colour blindness (n) is recessive to the allele for normal vision (N). This gene is carried in a non-homologous region on the X chromosome. Complete the table below to show the genotypes and phenotypes of individuals with regard to colour blindness.

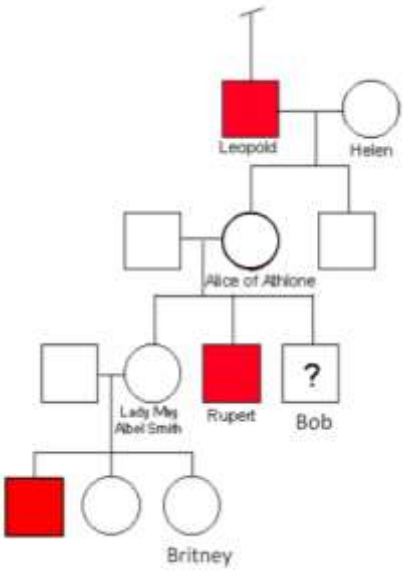
	Female	Male
Normal	$X^N X^N$	
Affected		
Carrier		Not possible! Why?

    e. In the space below, complete a punnet grid to show a cross between a normal male and a carrier female. What is the expected ratio of phenotypes?

20. Hemophilia is a blood-clotting disorder that is also recessive and sex-linked.
    a. State the normal function of the gene associated with hemophilia.

    b. Describe the signs and symptoms of hemophilia.

    c. Use the pedigree chart to deduce the possible genotype(s) of the named individuals.



Leopold
Helen
Alice
Mary
Rubert
Bob
Britney

    d. Suggest reasons why the frequency of some disease-related alleles might be increasing in the population.

<u>3.4.U9 Radiation and mutagenic chemicals increase the mutation rate and can cause genetic diseases and cancer.</u>

21. State the definition of a mutation.

22. Mutations can cause a change in a gene allele, which can be harmful. Occasionally mutations can be beneficial. Some changes however are 'silent', i.e. they don't cause a change in the trait. Explain how this is possible.

23. Mutagens are agents that cause gene mutations. List three types of mutagen.

24. Distinguish between mutations that can affect an individual during their lifetime and those which can lead to genetic diseases.

3.4.A4 Consequences of radiation after nuclear bombing of Hiroshima and accident at Chernobyl.

25. Radiation releases into the environment by humans can causes major problems. Radiation pollution is commonly the result of an accident at a nuclear power station (Chernobyl) or a deliberate after affect caused by the release of a nuclear bomb (Hiroshima). Outline the impacts and evidence of them caused by each incident plus make notes on the limitations of evidence.

	Accident at Chernobyl nuclear power station	**Release of a nuclear bomb at Hiroshima**
Impacts and supporting evidence		
Limitations of the evidence / what cannot be concluded*		

*Not being able to reach a conclusion due to lack of or limitations in the evidence is not the same as saying there no link between the variables. It simply means that the current data is insufficient to allow a conclusion to be drawn.

Citations:

Allott, Andrew. *Biology: Course Companion.* S.l.: Oxford UP, 2014. Print.

Taylor, Stephen. "Essential Biology 4.3 Theoretical Genetics.docx" Web. 7 Sep. 2015. <http://www.slideshare.net/gurustip/essential-biology-43-theoretical-genetics>.