

## **Genetics Skills Pack**

for A Level Edexcel B Biology

Topics 1, 2, 7 and 8

Update v1.1, April 2021

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POD 8095

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### **Contents**

Thank You for Choosing ZigZag Education	ii
Teacher Feedback Opportunity	iii
Terms and Conditions of Use	iv
Teacher's Introduction	
Skills and Specification Topics Covered by this Activity Pack	2
Student Introduction	3
Introduction to Genetics	4
Worksheets	5
DNA: the Code of Life	5
Cell Division	8
Meiosis and Variation	11
Polymerase Chain Reaction	14
DNA Profiling	17
Mutation and Expression	20
Gene Technology	23
Dihybrid Inheritance	28
Codominance and Multiple Alleles	33
Sex Linkage	36
Chi-squared Tests	39
Population Genetics	43
Exam-style Questions	46
Answers	52
APPENDIX 1 – Fundamentals of Genetics: GCSE Recap	70

### **Teacher's Introduction**

This Genetics Skills Pack is designed to help your student develop skills both during class and in an extracurricular setting. The content covers a range of topics across the A Level course, specifically including content from **Topic**1- Biological Molecules, **Topic 2- Cells, Viruses and Reproduction of Living Things, Topic 7- Modern Genetics** and **Topic 8- Origins of Genetic Variation**.

The pack contains 13 skill sections. Although each section does relate to a particular part of the A Level resource, the exercises are primarily skills-focused. These skills are all relevant to the A Level Edexcel B examinations. The competencies which are developed and tested by this resource include:

- Understanding and applying the rules which govern the genetic code, including the effect of mutations
- Recognising the stages of meiosis, and understanding the importance of meiosis in creating genetic diversity
- Drawing and using Punnett squares in a variety of situations, including dihybrid crosses
- Drawing and using genetic cross diagrams in a variety of situations, including codominance, and where multiple alleles are present
- Recognising types of mutation, and understanding extragenomic factors which affect gene expression
- Using percentages and fractions to describe and compare probabilities
- Using ratios to discuss phenotype
- Understanding how sex chromosome dimorphism affects inheritance patterns
- Calculating and interpreting chi-squared values
- Substituting numbers into the Hardy–Weinberg equation, and rearranging the equation to calculate unknown values
- Answering 'short essay' questions relating to biotechnology and genetic engineering
- Interpreting the results of DNA profiling

The resource also includes a 'GCSE Genetics refresher' in the appendix. This section includes a Pop Quiz, which can be given as a homework to prepare for further study of genetics, or used as self-assessment so that students feel confident to progress.

Each section contains a short section of background information, followed by worked examples and then in-depth questions to test students' knowledge (an answers section can be found at the end of the resource). The questions are split into three levels of difficulty, illustrated using symbols. In addition, seven worksheets contain Activity Boxes, which approach the topic in an unconventional way and should stimulate a deeper understanding.

The pack also includes three full exam-style genetics questions, which closely replicate the style of question found in an examination paper.

We hope this resource will be useful to your teaching, and help your students to tackle an area of biology which many find challenging, so that each student gains a deeper, holistic understanding of the subject.

### Remember!

Always check the exam board website for new information, including changes to the specification and sample assessment material.

January 2018

### Update v1.1, April 2021

- Ratio of 4:8:4 corrected to 1:1:1:1 in question 3 on page 31 and in answers on page 59
- Codominance and sex-linked alleles corrected to be in superscript on pages 33 and 35–38, and in answers on pages 60 and 62
- Reminder about how to calculate degrees of freedom added on page 40
- PCR cycler corrected to thermal cycler on pages 14, 16 and 55

### Free Updates!

Register your email address to receive any future free updates\* made to this resource or other Biology resources your school has purchased, and details of any promotions for your subject.

 resulting from minor specification changes, suggestions from teachers and peer reviews, or occasional errors reported by customers

Go to zzed.uk/freeupdates

### **Skills and Specification Topics Covered by**

This table is for either teachers' or students' use. It should help you to quice covered by this pack and which parts of the specification they relate to. As which require improvement and use the appropriate worksheets.

		_	
Topic No.	Page Title	Page	Skills Covered
1	Description of Life	5	Understanding how DNA amino acid sequences ar
2	Cell Division	8	<ul><li>Recognising the stages of the stages</li></ul>
3	Meiosis and Variation	11	<ul><li>Recognising the stages of</li><li>Understanding the important meiosis in creating general</li></ul>
4	Polymerase Chain Reaction	14	<ul><li>Outlining complex proce</li><li>Using diagrams to prese information</li></ul>
5	DNA Profiling	17	Interpreting results from profiling
6	Mutation and Expression	21)	<ul> <li>Cognising types of mu</li> <li>Understanding gene reg complex and multifacete</li> </ul>
7	Technology	23	<ul><li>Answering longer-form of the complex purples of the complex purples are the complex purples.</li></ul>
8	Dihybrid Inheritance	28	<ul><li>Writing about probability fractions and percentage</li><li>Drawing and reading Pur</li></ul>
9	Codominance and Multiple Alleles	33	Using genetic cross diag
10	Sex Linkage	36	Understanding how sex dimorphism affects inhe Using rercentages to expand abolity
11	Chi-squared Tests	2.3	Calculating and interpret chi-squared values
12	ation Genetics	43	Using the Hardy–Weinbe



### **Student Introduction**

If you studied GCSE Biology, you will already be familiar with some aspects talked about Gregor Mendel and the birth of the science of genetics. You and chromosomes, dominant and recessive alleles, generally and phenoty heterozygous individuals. You will have predicted and complete and calculated simpleted and the process which creates severally severally and the process which creates severally severally and the structure of DNA, mutakes chrom was.

You will build on all of these topics during your A Level Edexcel B course, is make life much easier for you if you're familiar with what you learnt at GCS a quick summary to jog your memory and give you a firmer knowledge of not feeling confident, you should go slowly, and spend some time building so that you can successfully tackle your A Level.

Each worksheet in this pack covers a separate topic within genetics. Your text exercises to do. If you're tackling a worksheet on your own:

- Read the information on the worksheet.
- Read the part of your textbook which covers this topic. If you don't have unfamiliar words or concepts online.
- Look through the worked examples you could the printing answers to then comparing your answers with what's write below.
- Try the activities or questions. If you we have answer sheet, answer as looking at it.
- Check your answor കുട്ടു പ്രാത്രം answer sheet, or hand your work in to







### **Introduction to Genetic**

Genetics is the study of genes, which are the biological code used to produce living organisms. It is also the study of how those genes interact with each other, how they affect the characteristics of the organisms that produce, and how they are parameters of produce, and how they are parameters of DNA in the same of genetics has expanded whickly – the Human Genome Project, completed in 2003, was a colossal multinational effort, involving geneticists from six countries and costing \$2.7 billion.

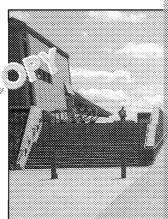


Figure 1: the Sanger major participant in

Because genes control the development, growth and characteristics of all more than any other field of biology) has a thousand practical applications genetic evidence to understand disease, and agronomists (farm scientists) yields. Embryologists use genes to predict how embryos will develop, and genes to connect criminals to crime scenes.

In your A Level course, Topic 1 discusses the structure, replication and read and 8 focus entirely on genetics. An understanding of interitance is also coprocess of cell division, which is covered in Topic 2. Gut top marks in general understanding of probability and ratio of the principles behind some cutting technology.

This pack v you to develop several core skills relating to genetics are inheritance, including:

- understanding how DNA, mRNA and amino acid sequences are relate
- recognising the products and purpose of meiosis
- drawing and reading Punnett squares
- writing about probability using fractions and percentages
- using genetic cross diagrams to show inheritance
- understanding and using pedigree diagrams
- understanding how to use and interpret ratios
- calculating and interpreting chi-squared values
- using the Hardy–Weinberg equation
- identifying different types of mutation
- answering longer-form questions
- understanding complex processive semi-conservative replication, biotechnology)
- recog stages of a complex process in an unfamiliar setting
- interpression interpression in interpr





### **DNA: the Code of Life**

### Background - key points

A gene is a section of DNA which codes for a single peptide or RNA molecule. This fact is fundamental to modern and again and again. However, the genetic material contains in thou carrierent nucleotides: two larger purines (G), and guanine (G), and two smaller pyrimical and two smaller pyrimical and two smaller pyrimical and thymine (T). Purines ar e း ေလစို nitrogenous (nitrogen-containing)

rings; pyrin sare made up of a single ring.

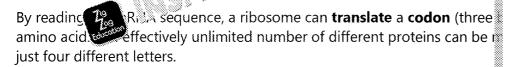
The DNA sequence is **transcribed** into an mRNA sequence using specialised enzymes, including **helicase** which separates the two strands of DNA, and RNA polymerase which uses the DNA to produce a single-stranded mRNA molecule. This process follows similar rules to complementary base pairing, but instead of thymine, mRNA has a base called uracil (U). The table below shows the differences between DNA and mRNA.

Properties	DNA	RNA
Bases	Adenine, cytosine,	Adenine, cytosine,
Dases	guanine, thymine	guanine, uracil
Size	Very large (millions	Small (+h a rd_of
Size	of base pairs)	င်္ကေ pairs)
Strands	Double-st aec	Single-stranded

Compl® always 🖹 while cyto

**Underst** 

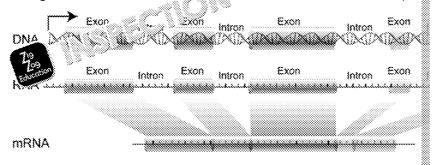
amino a



The triplet code of DNA has a number of features:

- It is **universal** the code essentially reads the same in all organisms, w
- It is **degenerate** because there are 64 different groups of three bases only 20 different amino acids, more than one triplet may code for the
- It is **non-overlapping** the ribosome reads the sequence ACCTGA as ACC / TGA – not as four overlapping triplets – ACC / CCT / CTG / TGA

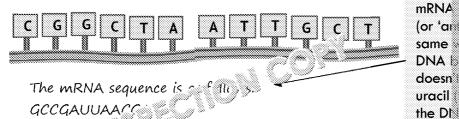
Not all of a gene is used to synthesise proteins, however. In eukaryotes, a ty called exons, separated by sections called introns. Both are transcribed to preare spliced out (removed) before the mRNA mologonal avals to the ribosom sections of the gene are not translated as a final amino acid seque





### **Worked examples**

 a) The sequence below is from the anti-sense strand of a DNA mole sequence that would be produced by transcription.



- b) Haran acids does this sequence code for?
  Filed ino acids. The ribosome reads a codon of three mRNA which amino acid will be placed next in the sequence. The riboseparately, and they do not overlap so 12 bases is equivalent.
- c) An mRNA sequence reads as follows: UGUUCAAAACAUUGC.
  Using the table below, state the amino acid sequence that would sequence. Assume that base 1 is the opening letter of a codon.

Cysteine—Serine—Lysine—Histidine—Cysteine
(Cys-Ser-Lys-His-Cys)

Each three-le\*

'codon', coding

2. Explain why scientists cannot use the amino acid sequence of a protest sequence of the gene it is made from.

The genetic code is degenerate; this means more than one different can stand for the same amino acid. Because of so, it means that acid glutamic acid appears in a seauches, which are transcribed into means include sections called to so, which are transcribed into means which the sequences will not be visited. These sequences will not be visited.

		2000			
UUU	Dh Tan sir v (Dh a)	UCU		UAU	Tyrosino (Tyr
UUC	Ph. 7.9 pine (Phe)	UCC	Carina (Car)	UAC	Tyrosine (Tyr
UUA		UCA	Serine (Ser)	UAA	STOP
UUG		UCG		UAG	3104
CUU	Leucine (Leu)	CCU		CAU	Uictidina (Ui¢
CUC	Leucine (Leu)	CCC	Proline (Pro)	CAC	Histidine (His
CUA		CCA	Profine (Pro)	CAA	Clutamina (Cl
CUG		CCG		CAG	Glutamine (Gl
AUU		ACU		AAU	Asparagine (As
AUC	Isoleucine (Ile)	ACC	Throoping (Thr)	AAC	Asparagine (As
AUA		ACA	Threonine (Thr)		Lysine (Lys)
AUG	Methionine (start) (Met)	ACG		AAG	Lysine (Lys)
GUU		GCU		<u> </u>	Aspartis asid (A
GUC	\(alina (\(al\)	GCC	Alasia (S.	GAĆ	Aspartic acid (A
GUA	Valine (Val)	GCA		GAA	Clutamic acid (
GUG		G		GAG	Glutamic acid (🤇

### Activity

Transcribe NA sequence below to produce an mRNA sequence, and then codons below to identify the amino acid sequence.

1) GAATTATGCCACTAGGCC

3) GTGTCCTATACCTAG

2) TACCGCGACTAA

4) TACGCTTTTTGGCTA

Take the first letter of each amino acid. Use these letters to spell out the name of

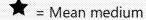
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### **QUESTIONS**

### **KEY**

= Budding beginner





- The DNA triplets GTC, CT, Truend GTG all code for the same are
- The DNA sequer was ASSACAG is read as three, rather than seve
- A AGC codes for the same amino acid in kangaroos
- A triple of DNA reads AAT. From the options below, choose:
  - the mRNA codon that would be made by transcribing this triplet.
    - TTA
    - UUC
    - **AAT**
    - TTU
    - UUA
    - CCG
  - **b)** the tRNA anticodon that would bind to the mRNA codon during
    - UUA
    - AAT
    - UUT
    - GGC
    - AAG
    - **AAU**



- e mRNA sequence that would be produced by transcribin
- Using the table on the previous page, write the amino acid seque using this code. Assume that base 1 is the opening letter of a cod
- A DNA sequence reads as follows: TATGATCAGCCCAGT. During trans single error happens, and one of the bases is substituted for a different sequence made from the 'mutated' mRNA is as follows: isoleucine, leu

Using the sequence and the table on the previous page:

- a) Which amino acid has changed due to the transcription error?
- **b)** Suggest which base in the sequence was incorrectly transcribed.
- Scientists sequence a long section of DNA from Junian chromosome cannot directly convert this seque to mino acid sequences of the



### **Cell Division**

### **Background - key points**

During **mitosis**, cells divide into two (in theory) identical **daughter cells**. Each cell has an exact copy of the and cell's DNA. Mitosis is used so that organisms care produce new cells and repair damaged tissues. Bactures is related, but simpler, process to **reproduce** as a producing two identical daughter cells.



Mitosis has war main stages:

- Prophase: the chromatin condenses so that individual chromosomes
   envelope which surrounds the DNA breaks up into pieces, and the ce
   fibres which stretch across the cell.
- **Metaphase**: chromosomes line up across the equator of the cell, attack
- Anaphase: chromatids are pulled to either pole of the cell by spindle compacted again.
- **Telophase:** the chromatids reach the poles of the cell, and are now kranuclear envelope reforms around each separated set of chromatids.

Telophase is followed by **cytokinesis**, in which the cell actually divides in to membranes to separate the daughter cells.

However, before a cell can divide by mitosic, in macreplicate its DNA, so the with a full complement of genetical series. This stage is called interphase

### DNA re ti 🕠

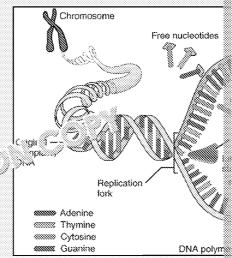
DNA replication involves several enzymes acting in combination, with each specific role within the process.

- DNA helicase breaks the hydrogen bonds between strands, causing
- Free **nucleotides** come into contact with the exposed DNA strands, join with the exposed bases to form a new strand.

An enzyme (called ligase) helps complementary bases to join, forming new hydrogen bonds.

- DNA polymerase creates the 'backbone' of the new strand, joining the sugar of one nucleotide to the phosphate group of the next.
- Ligase joins smaller sear வக்கள்
   DNA together in வெள்ளின்.

The pattername pairing is the same for all organisms: A pairs with T, and C pairs with G.

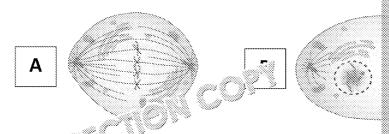


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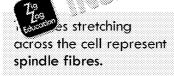
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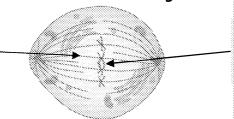
### **Worked examples**

1.



a) Name the stale മാരങ്ങ shown in each of the diagrams abo

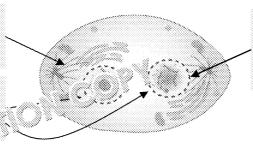




A- Metaphase. The chromosomes have lined up across the equiver yet separated into chromatids. The spindle fibres are visible centromeres of each chromosome.

Spindle fibres are no longer attached to chromosomes.

There are clearly two distinct 'groups' of chromosomes.



B p.s.se. The nuclear envelopes are reforming around the work clearly separated into two bundles.

b) Explain what will happen directly after stage B.

The cell will divide by cytokinesis - the cytoplasm will be share each of which will have a single nucleus.

2. Cancers are diseases in which cells divide incorrectly, in a way that body. Some recently developed chemotherapy (cancer treatment inhibitors of the helicase enzyme. Suggest why drugs would targe

Cancerous tissues divide rapidly in an uncontrolled manner. A drug which inhibits helicase could slow down, or stop, DIA replication in cancerous tissues the double helix wouldn't sep which new ones. If cells can't carry itosis, they also can't divide by cytokinesis to make more cells. This drug could prevent cancerous tissues from expanding, and slow or stop the progression of the disease.

This is the you have biologist about whelicase during Domean if the

Once you'vedoing, you question what drug with the

# 



### **QUESTIONS**

**KEY** 

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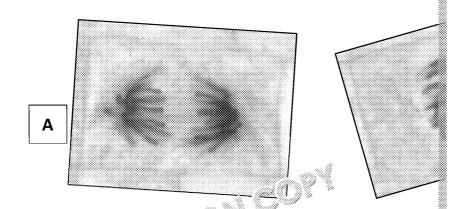
🔻 = Mean medium

■ 1. Complete the table of the stages of mitosis

r.ption در ک

The chromatic's a funed to either end of the cell by spindle fibres.

The condenses so that distinct chromosomes are visible.



- Identify the stage of some nown in each of the plant cell micrograp your arms.
- Describe the stage of DNA replication which occurs after each of the
   a) Helicase breaks hydrogen bonds, leaving two exposed strands of
  - b) DNA polymerase begins to move down the DNA molecule in the
- **4.** DNA replication is regulated by a series of checkpoints the DNA is c whether it is damaged or contains errors. If the DNA contains errors, t undergo a process called apoptosis, or programmed cell death. With mitosis, explain why this process is so important.
- **5.** The periwinkle plant (*Catharanthus roseus*) produces vinca alkaloids, which are toxins that prevent the correct formation of spindle fibres.
  - a) Suggest why vinca alkaloids are used in Incer drugs.
  - **b)** Suggest one limitation of using machod to treat cancer.
  - c) Suggest why vinca all and like not function as antibiotics.







### **Meiosis and Variation**

### **Background - key points**

Mitosis and meiosis are processes with similar names, and the stages of each process resemble each other in many way were witosis is used for processes including growth and the pair, while meiosis is used to produce the representation to fee the production to fee

In addition significant roduces **genetic variation** (i.e. the daughter cells are generally different from each other) while mitosis does not. This is an extremely important distinction – meiosis makes brothers and sisters genetically different from each other, since each sibling inherits a unique set of alleles from each of their parents.

Genetic diversity is introduced in several ways: by the **crossing over** of ho start of meiosis, by the **independent assortment** of chromosomes and chand by random fusion of sex cells when **fertilisation** occurs. For your A Leidentify cells in different stages of meiosis, and you will need to understan

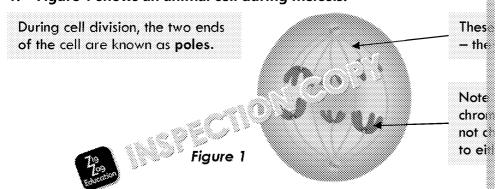
### **STAGES OF MEIOSIS**

Meiosis can be divided into two halves, and then subdivided into eight sta

	Stages	Desci		
	Proch A Janhase I	Homologous chromosome p		
Meiosis 🏣	Prombase I, na, sase I, Telophase I	genetic material, and then ar		
ala se I, Telophase I		of the cell contains one of ea		
"	Duanhasa II Matanhasa II	The sister chromatids in each		
Meiosis II Prophase II, Metaphase II, Anaphase II, Telophase II	so that each gamete ends up			
	Anaphase II, Telophase II	genetic material.		

### Worked example

1. Figure 1 shows an animal cell during meiosis.



a) Which stage of meiosis does Figure 1 show?

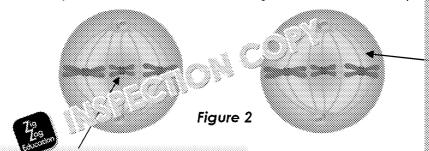
Anaphase I – the homologous chromosome pairs which had previous have been separated, and are being pulled towards either pole of a

# 



### b) Explain what is happening during this stage of meiosis.

Homologous chromosomes (chromosomes with the same genes) the cell by spindle fibres, which have been produced by the cer randomly assorted, so that either daughter cell could end up

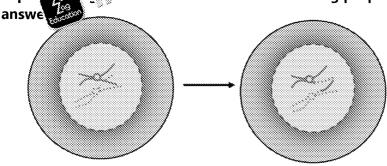


Here, the chromosomes have lined up individually, rather than in pairs.

- c) Identify the stage of meiosis which is happening in Figure 2.

  Metaphase II individual chromosomes, rather than pairs of a across the equator of the cell, attached to spindle fibres.
- d) Give a summary of what has happened between Figure 1 and The chromosomes reached each end of the cell and the spindle as a nuclear envelope formed around each set of chromosomes split into two and the cell membranes reformed around the to (cytokinesis). The chromatin in each cell is a fully condense envelopes began to dissolve called a spindle fibres were for chromosomes migrated and equator of the cell.

2. Explain v Land variation is increased during prophase I. You



During prophase I, homologous chromosomes cross over and swap part or all of an arm. This means that the two chromatids that make up each chromosome combw different, with one chromatid (in each chromosomes chromatid genetic material from the other hand and chromosome.

This means the the Caring metaphase/anaphase II), when chron are independently assorted into the two daughter cells, the chromatids in each daughter cell are non-identical. In addition, the location at which the arms meet and exchange genetic material varies between meiotic events.

# 



### **Activity: Mnatural Mnemonics**

Mnemonics are memory aids, where each of the first letters of the words for a word you are trying to remember. For example, this mnemonic help ranked levels used to classify organisms.

King Philip Can Only Find Green Socks = Kingd Jum, Class, Order

Write a mnemonic to help you remain the stages of meiosis: prophase telophase I, cytokinesis, promase II, anaphase II, telophase



### **KEY**

= Budding beginner

= Mean medium

N

Name the stages of meiosis I shown below, and put them in the order numbers 1–4.







● 2. Fill in the gaps to complete and the ball by either naming a stage of meios

Description

# Hom 750 s

is chromosomes line up in pairs along the equator

The chromatin condenses, and homologous chromosomes cross over, exchanging genetic material.

- **3.** Give three differences between the products of meiosis and the produ
- 🖈 4. Sketch the following stages of meiosis (You only need to draw two to
  - a) metaphase II
  - **b)** anaphase II
- - a) Discuss the ways in which meiosis creates genetic variation.
  - **b)** Explain why this would be important for a rodent living in an area as the Sahel region (between the Sahara Desert and the forests of



### **Polymerase Chain Reaction**

The polymerase chain reaction (commonly known as PCR) was invented by Kary Mullis, a biochemist who worked at the Cetus Corporation, California in the 1980s. Mullis won a joint Nobel Prize in 1993 for his work

Mullis won a joint Nobel Prize in 1993 for his work

PCR is a method of producing large can be of DNA from tiny samples. This is known a samples large enough for gel electron sais, so that individual 'bands' can

be seen on each el. This in turn is required for processes such as **DNA hybridisation**, where labelled DNA probes are used to spot alleles and diagnose genetic conditions.

The polymerase chain reaction is a multistage process, but once the initial sample has been extracted and prepared, all the stages of the process are carried out by an automated machine known as a **thermal cycler**. After each cycle, the amount of DNA in the sample doubles.





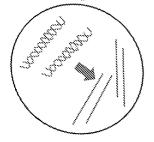
A scientist adding DI

# in excess: Adding more of a reactant than is actually used up in a p

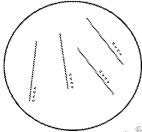
**Preparation:** the sample is extracted and along with:

- nucleany an excess
   a y Jaion of DNA polymerase
- specially designed primers which attach to sequences

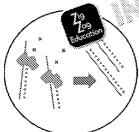
The PCR tube is then placed into the cycler.



**Separation:** the thermal cycler is heated to 94 streak the hydrogen bonds between DNA base template and complementary strands.



Annealing: the thermal cycler is cooled to arou the oligonucleotide (sr., "I and le-stranded DNA of the separator DNA swands. This only happer complements to the DNA.



**Synthesis:** the thermal cycler is heated to aroun optimum temperature for a special Archaean verpolymerase. The enzyme starts at the primers at to each exposed DNA strand, making two new molecules. The process then starts again.



### **Activity: Complete the cycle**

The phrases below are not in order. Add the phrases below to the boxes to complete the diagram.

Draw pictures to represent what is happening to DNA at each stage of the

Synthesis of complementary strands

Separation of strands.

Primers join to the DNA strands, at locations where they are complementary to the sequence

DNA polymerase joins free nucleotides to each molecule, starting at the primers

Annealing of oligonucleot

Stage: .....

Temperature:

What's happen

Temper

What's happening: .....

Stage: ......

Temperatur

What's happ

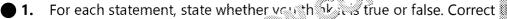
### QUESTIONS

### **KEY**

= Budding beginner







- A) Separation must occur have smalled ing of primers in each PCR cy
- B) Separation and mappen at the same temperature, but use
- C) Duanth Sesis stage of the PCR cycle, the cycler is heated to
- D) D ynthesis, new nucleotide bases are added to each DNA st
- **2.** Explain why the polymerase chain reaction is repeated multiple times
- ★ 3. A forensics team wants to identify a DNA sample recovered from a cride do so, the DNA sample must be amplified using the polymerase chain PCR produces many copies of a DNA sequence. You can use bullet polymerase that the polymerase chain produces many copies of a DNA sequence.
- $\bigstar$  4. Write a paragraph to explain how the polymerase chain reaction could
  - a) testing for the presence of a genetic condition in an embryo
  - b) detecting the presence of an infectious disease in the blood
- DNA replication in humans is regulated by a series of checkpoints which DNA. Artificial DNA replication through PCR is gulated in this was issue for some people using PCR.





### **DNA Profiling**

### **Background - key points**

Just like coding DNA, non-coding DNA varies from individual to individual. In particular, there are sequences of DNA – TACCGATT — xample – repeated over and over consecutive processor of the chromosor particular person to person in the number of repeats – one person might have 60 repeats of an STR while another has 130 in the same location, for example. STRs can be further divided into minisatellites (longer) and microsatellites (very short; 2–5 base pairs repeated 10–15 times).

These kinds of variation between individuals make DNA profiling possible. This means that if a **restriction endonuclease** enzyme cuts up a large enough sample of DNA, only cutting at specific sequences called **recognition sites**, the fragments it produces will be slightly different from person to person.



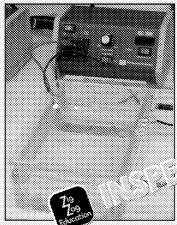


Agarose

Some DNA fragments produced by a restriction of some will be bigger in of the length of STRs. As a result, when any produced DNA samples are people (except for identical possible sets of factors approduced is greater than the number of people

This technic mass many real-world applications. It can be used to connect and (just as importantly) to rule people out as suspects. It can also be use relationships, since individuals inherit their DNA equally from each parent.

### **Electrophoresis**



Gel electrophoresis apparatus

In order to produce a DNA profile, a tecle electrophoresis is used. The DNA fragmer restriction endonuclease are placed in an attached to two **electrodes**, and a current

Because the DNA for joinents contain man groups, the major a slightly **negative ch** D\*13 or joinents are attracted to the **posi** 

However, larger fragments are less able spaces in the gel matrix, so they move matrix fragments move quickly, and fragments distinct 'bands' on the gel. The DNA bases using a dye that specifically binds to DNA Southern blotting technique.

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### **Worked example**

1. A couple, Paul and Kate, have one biological daughter, Jo, and a Bradley. In addition, Paul has an adult daughter, Paige, from his f therefore, Kate's stepdaughter.



The number of recognition sit the size of a 🛭

a) Look at the DNA profile above. Briefly, describe how DNA from to produce this sample, and explain what the white bands co

The DNA fragments for each individual are produced by cutting a sample of DNA with a restriction endonuclease enzyme, which cuts the DNA at a specific sequence called a recognition site.

The fragments are then 'run' on a 🕮 📉 process called elect move towards the position in the due to their slightly negative

The size of the lagments determines how far across they will move. Therefore, each band consists of ragments of DNA of a specific length, which are collectively visible as a band.

b) Use the DNA profile above to identify the members of the far

The child who is the biological daughter, Jo, will have W inherited half her DNA from her father and half from stre her mother. As a result, all of her DNA bands will be rel by: shared by one or other of her parents. This is only wo: true for Child 3, so Jo is Child 3. stu

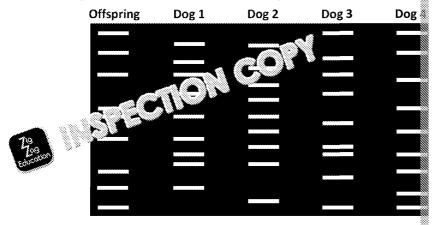
Most of Child 2's DNA bands are not say a ky either parent, which suggests Child 2 is not tell to either parent – this must be Bradley in chive's adopted son.

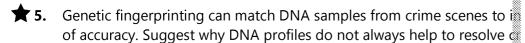
Call has several DNA bands in common with 🌌 , but not with Parent 1. This suggests that the two 🕯 are related – so Child 1 is Paige, and Parent 2 is Paul. Finally, Parent 1 must be Kate.



### **QUESTIONS**

- 1. Using one sentence for each definition, define:
  - non-coding DNA
  - recognition site
- **2.** For each statement about electrophoresis, state the her you think it is true or false. Correct the false sate and.
  - A) DNA fragments have a slightly gative charge due to the carbox backbone of each and help and a slightly gative charge due to the carbox
  - B) During el resis, the smaller fragments of DNA move throu
  - C) En the heresis often uses a thick agarose gel, which is solid but lall molecules to move.
  - **D)** DNA fragments move towards the negative electrode (or cathode)
- During gel electrophoresis, DNA fragments are 'run' on an agarose gel, p of DNA at different distances from the positive electrode. Explain why ma
- **4.** A puppy has its DNA sequenced and profiled by a dog breeder, to che breeder knows that Dog 1 is its mother, but isn't sure which of dogs 2
  - a) Using the DNA profile output below, explain which dog is the pur
  - **b)** Why does each individual's DNA produce a unique pattern of bar
  - c) Give an exception to the rule described in question ii).







- a) forensic criminologists trying to identify DNA from a crime scene
- **b)** an adopted child who is searching for his biological parents
- c) a medical professional who is helping a family understand a famil
- 7. The set of markers to the right shows the regime of a DNA profiling for a foal (young horse). Pare to the results, draw two sets of plausible markers, a present the mother and father of the foal
- **8.** A received by has suggested that the repeat number of an STR on character be an individual will develop Character Masyndrome, a neurodegenerative disorder which is normally first observadulthood. Explain how this could be detected using a DNA profile, are would be useful for medical professionals and patients.



### **Mutation and Expression**

### **Background - key points**

Mutations are changes in the base pair sequence of a DNA molecule, which occur during DNA replication. They can be caused by **ionising radiation**, poisons or accidental daing the process of DNA replication.



Nai To	ar Too		
Insertion	A new base (or bases) is inserted into the		
insertion	DNA sequence.		
Deletion	One or more bases is completely lost from	Т	
Deletion	the DNA sequence.		
Cubetitution	One or more bases is replaced by a different		
Substitution	base (or bases) in the DNA sequence.		

However, gene products and functions can be altered in other ways, beside DNA. Any given cell only needs to produce some of the proteins coded for a cell in the skin doesn't need to produce the hormone insulin. In addition the same proteins all of the time. Regulation of gene expression is essential differentiated cells.

Transcription factors – this is a general term of fire sins which affect the genes. They may bind to promoters (a see extilation or methylation)

**Promoters** – genes have the sequence before the start of the codin binds to the promoter to transcribe the gene. Sometimes, several g

**Acetylation** — cells can add **methyl** (CH<sub>3</sub>) groups to cytcharder for RNA polymerase to bind to the section of DNA. Alternatively, **ac** added to the **histone proteins** which chromosomes are wrapped around. complex to become less densely packed, and makes it easier for RNA polymerase.

### Post-transcriptional modification

Cells can also modify mRNA to affect how a gene is expressed. For example, **introns** are spliced out of pre-RNA to form **mature RNA**, but transcription factors can also affect which **exons** are included. This can create two or more similar but distinct proteins the same original DNA sequence.

Small interfering RMA (miRNA) and micro RNA (miRNA) (miRNA) sabotage (miRNA), pinding to it in an imperfect way so that it can be translated by a ribosome.

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MENA

### **Worked examples**

- 1) Identify and describe the type of mutation in each case:
  - a) CCAACTGAGGAC
- CCAACTGCAGGAC
- b) TACGAAAGTTCATAA
- → TACGACAGTTCATAA
- c) TTTCGT GGTACCT
- → TT;:\\GGTACCT
- d) ACAGCGCTGTAAA
- ACAGCGCTGTATTGAA
- a) Insertion of a Cina mases 7 and 8
- b) S: 70

tic vac for an A at base 6

Addition and mutations con base pair, and

c) De sales of bases 7-8

d) Insertion of TTG between bases 11 and 12

2) A gene known as p53 triggers programmed cell death in tumour coding DNA sequence from bases 1171–1200 reads as follows:

GTCGCCTGCA ACCTAGGCGG ACGCTACCAT...

However, a rare, mutated allele of the gene has a different DNA second GTCGCCTGAA CCTAGGCGGA CGCTACCAT...

Explain why this mutation could affect the function of a protein I

The mutation could drastically change the shape of the protein. Be just one base, the ribosome will start reading and mRNA codon wrong place. This frame shift will complete manage the amino action of the protein for the probably lose its form of the protein of the protein of the protein. Be the protein of the protein of the protein of the protein of the protein. Be the protein of the protei

- 3) Which the examples below describes post-translational modifies
  - a) Al ative splicing, so that different exons are included or ex
  - b) Addition of a phosphate group, to 'activate' a kinase protein
  - c) Methylation of cytosine residues in DNA
  - a) This is not an example of post-translational modification. Exorpart of a translated protein. This happens after transcription an example of post-transcriptional modification.
  - b) Yes, this is an example of post-translational modification a 'completed' by being activated with a phosphate group (phosp
  - c) No, this method is used to down-regulate or prevent transcriptranslation of a gene. It occurs before the large on happens.

### **Activity: Storyboard**

Draw a series of discussion, in the style of an instruction manual, to show of modification a gene can go through to produce a protein. You could in

- facting how the gene is transcribed
- splicing out of introns and alternative patterns of splicing
- post-translational modifications, such as addition of prosthetic gradules

# 



### **QUESTIONS**

	KEY	
= Budding	= Mean m	

● 1. a) Identify the type of mutation in each cas

Sequence Number	Original	Ne
	ACT AAG CCC AAT	ACT AGC
EEE	GGT TAA CGA CTC	GGT TAA
3	TTT TGC ACA CAG	TTT TGC AG
4	GAT TTG AAC CCT	GAC TTT G

- **b)** Suggest, giving a reason, whether mutation 3 or mutation 4 is months the protein produced.
- 2. Some gene mutations do not cause changes in the protein produced. properties of DNA, explain why.
- **3.** Explain how methyl groups can be used to regulate gene expression.
- **4.** People with two defective alleles of the F-X spene develop Tay–Sachs gradually deteriorate and die and repeats with Tay–Sachs have a short common mutation which are safety as Tay–Sachs is the deletion of a single of the general safety.

Explair the deletion of a single DNA base can have a major impactor protein produced.

- **5.** siRNA molecules can be designed in laboratories to be complementar Explain how this technology can be used to study protein function.
- A stretch of genome in a nematode worm contains a promoter and are as two genes which code for subunits of an acetylcholine receptor. According to the postsynaptic membrane of neurons, where they recognise the and transmit a nervous impulse.
  - a) A substance is discovered which binds permanently to the promocould be fatal for the nematode worm.
  - b) Suggest why both genes are controlled with same promoter.



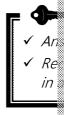




### **Gene Technology**

### **Background - key points**

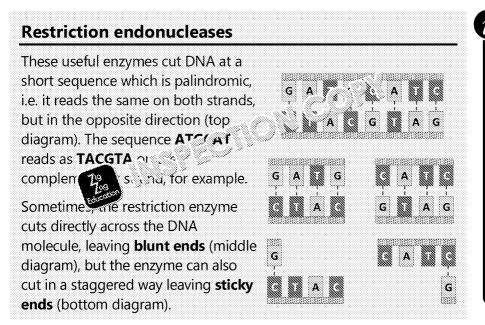
**Genetic modification**, the ability to produce or isolate genes and insert them into the genome of target organisms undoubtedly a great scientific revolution of or a a. be process makes use of several different technological archimethods, but it can be broken down into five hash persecution or production of a gene, is a population of the gene into a larger



DNA mole (transportation) (causing the gene to be taken up by a taken which have up the gene, and **growth/cloning** of the transgenic organization to form natural clones).

There are three ways in which a gene can be produced or isolated:

- Convert an mRNA sequence to complementary DNA (cDNA) using a re
- Type a known amino acid sequence into a computer attached to a general assemble an appropriate DNA sequence.
- Use a restriction endonuclease an enzyme which 'cuts' DNA contains fragments, specifically cutting it at a set of bases called a recognition



### Insertion and transformation

Once the fragment containing the gene has been recognised using a **DNA probe** (a sequence complementary to part of the cane), the DNA fragment is prepared by having a **promoter** sequence added to it. This means once is significantly cell, RNA polymerase will be able to bind to it and turned an allow a plasmid. The fragment can then be inserted into a **vect** promolecule used to transfer DNA into another or any assually a **plasmid**, but other vectors can be used – such uses, which insert their DNA into the host cell.

If the vector molecule is cut using the same restriction endonuclease enzyme used to cut the DNA into fragments, the two molecules will have complementary sticky ends. This means that a **DNA ligase** enzyme can join the fragment to the vector, creating **recombinant DNA**.

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The plasmid can then be inserted into a bacterial cell using a range of met bacteria can be placed in a dilute **calcium chloride** (CaCl<sub>2</sub>) solution, and the warmed to around 42 °C. This causes the membrane to become highly per to cross it and be taken up by the cell. The cell can now be described as a

An alternative method of getting recombinant DNA into a cell is to use a control DNA is coated with a very thin heavy metal such a sind, tungsten, silver of microscopic particles are fired into a ties of cultural under high pressure. The mostly used for plant genetic engineering.

### Identific 🔑 vach ma

### งมเท marker genes

Not all cells will actually take up the plasmid – in fact, the majority won't. In order to check whether a cell has taken up a plasmid, the DNA fragment may contain both the target gene (the one which is being used to make a protein) and a **marker gene**, which has some easily observed effect. The marker gene might be something such as GFP, the gene that makes **green fluorescent protein** – if the bacteria has taken up the plasmid, it can be caused to produce this glowing protein, which is easily observed under UV light.

### Worked examples

1. Researchers are studying the effects of oxidate ficiency in humpeptide hormone which is important in social reproduction and a want to produce large amount to produce the periods.

Sugget the scientists might use oxytocin produced by gene the hormone from blood samples.

This method is likely to be cheaper than taking oxytocin from hum molecules are only produced in small quantities in the body. This molecules are only produced in small quantities in the body. This molecules are only produced in small quantities in the body.

2. A plasmid may be coated in a small lipid droplet known as a lipos Using your knowledge of the properties of membranes, suggest to

Cell surface membranes are made of phospholinid, and are highly Liposomes are able to fuse with the cell, and are, mimicking the releasing the contents of the "ros", into the interior of the cell. plasmids are macro, and are not able to cross the cell mornor distribution.

# 



### 2. These statements describe alternative ways of carrying out stages which stage is being described in each statement, and explain the

a) A bacterial cell culture is given a mild electric shock, which m temporarily very permeable. This is known as electroporation This is an alternative way to carry out transformation – inserting a plasmid vector into a (hadira cell. The plasmid contains the tarring and, therefore, the cell can be convince and produce the 📭t , 🔭 📆 , without needing to achieve the difficult plicing the gene into a bacterial genome.

b) The herbicide Bromoxynil is added to a plant tissue culture. T incorporated the bxn gene, as well as target gene A into their the herbicide and survive.

This method uses a marker gene to identify which cells have taken up the gene. The DNA fragment has the bxn gene added to it, and it is inserted into the plant's genome. If a plant cell has successfully taken up the target gene, it will also be able to survive exposure to the herbicide.

Lots of different methods a gene has been taken up via and re taken up the to colle methods destroy cells

### he glossary Activity

A lot of new words appear in this topic, and you will gain valuable marks accurately and in the correct context. To help you practice, unscramble t a definition for each scientific term. All of the terms relate to gene technology

CRANE BADMINTON	INFANTS ROOM ART	ONCI
AID ANGLES	NICER ANGST	



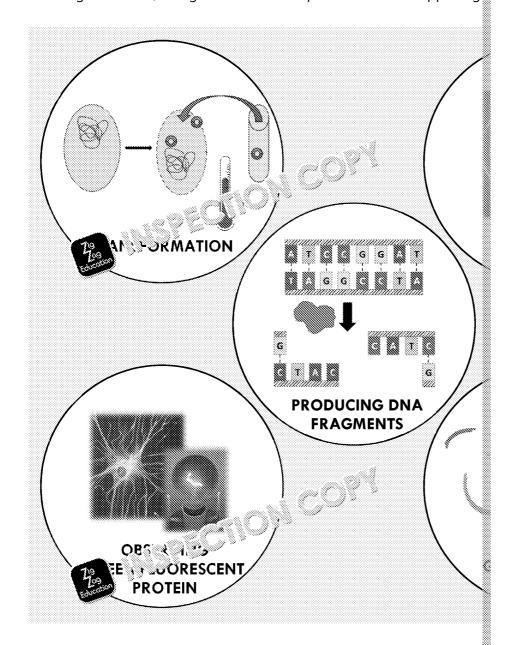


### **QUESTIONS**

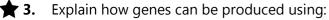
● 1. Fill in the gaps to complete the paragrap.

Sometimes scientists we not produce large quantities of a protein, su this is to splice the second a \_\_\_\_\_\_, such as a plasmid, using endor the second cut the plasmid at a specific \_\_\_\_\_\_ s been in the day of the second cut the plasmid at a specific \_\_\_\_\_ s been in the day of the second cut the plasmid at a specific \_\_\_\_\_ enzyre from different sources, and so it can be described as \_\_\_\_\_ DI

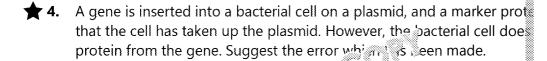
**2.** The bubbles below show stages of a process in which a useful gene is the stages in order, and give a short description of what is happening







- a) reverse transcriptase
- **b)** a gene machine



**5.** Organisms may be genetically a data for many different reasons. For silkworm proteins (1) advection in their milk, while GM tomato a long and of sever, genetic modification is considered a continuous people apposed to the production of genetically modified organisms.

Discuss the ethical arguments for and against genetic modification. To four of each.

- **✗ 6.** Explain how genetic modification could be used:
  - a) to treat somebody with a genetic disorder caused by a non-funct the liver
  - b) to reduce the need for pesticides in common crop plants
- **7.** 'Restriction enzymes will cut a DNA molecule wherever they encounte sequence.'

From the observation above, as well as your own in wledge, suggest are used to insert genes into bacterial sells, it stand of inserting the ge





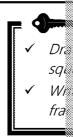
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### **Dihybrid Inheritance**

### **Background - key points**

At GCSE, you used genetic cross diagrams to represent the inheritance of alleles for a single gene. However, and always want to study the inheritance of changes of the geneticists want to know how (or the genes interact with each other. One way the is to carry out **genetic crosses** using diagrams to represent the inheritance of changes of the genetic crosses using diagrams to represent the inheritance of alleles for a single gene. However, the inheritance of changes of the genetic crosses using the genetic crosses are genetic crosses and genetic crosses are genetic crosses are genetic crosses and genetic crosses are genetic crosses ar



With two parents and two genes, there are as many as 16 possible combined passed on to offspring. Probability calculations get a bit more complicated individuals are bred, the offspring may have several different phenotypes.

Genetic cross diagrams aren't as useful for **dihybrid inheritance** as they a — lots of arrows crossing over one another can become difficult to interpre and **branch diagrams** are used.

### Worked example

- 1. Two rice plants are crossed by agronomists to didy the inheritance of two genes:
  - Gene D controls stem leng ாத் a cominant D allele makes rice plants grow t இருந்த while the recessive d allele causes show ( ''asa') stems.
  - G Lat at sets grain yield plants with the L allele produce lowgrain, while the recessive /allele causes less grain production.

The rice plants crossed are believed to be heterozygous for both genes.

a) Draw a Punnett square to show the predicted offspring.

		UL	υı	dL.	d)
The sidebars show all the possible genotypes of each parent's gametes.	ದ	DDLL	DULI	DdLL	Do
Each parent is heterozygous (DdLI) and they will all	سُ	שטטIL	DDII	DdlL	Do
pass on the lele of Decay one allele of L. Any combination	<b>★</b> 평	dDLL	dDLI	ddLL	dd
is possible.	T	dDIL	dDII	ddlL	da



# b) The agronomists are trying to breed a short-stemmed rice planamount of grain. Which genotypes would produce this result Plants with the genotypes ddLL and ddLl will have short-stem and will produce a large amount of grain (at least one domination).

c) What proportion of the offspring would the gronomists expenses Give your answer as a fraction.

Punnett square shows 16 1000 genotypes.

Three of the 1600 his outcomes have the

'control of the offspring outcomes short stems and produce excess grain.

Unless other other outcomes have the outcomes have short stems and produce excess grain.

d) Give your answer to the previous question as a percentage.

3/16 possible genotypes (ddLL, ddlL, ddLl) will create short-stemmed plants with excess grain.

3/16 = 0.1875

0.1875 x 100 = 18.75 % (or 19 % to nearest 100 whole percentage)

- 2. Two flies are crossed multiple times to study the inheritance of two genes.
  - Gene E affects eye colour flies with the designant allele, E, have deep red eyes, and flies with two recessive e alleles have orage.
  - Gene W controls wir ງ ເຄັງ !... flies with the dominant a!' ເພັ້ນ ເຄັດພາຍ normal wings, while flies wife ເພັງ ຂອງເຂດ allele, w, have short wings.

An orange-eyed, normal-winged fly (P1) is crossed with a red-eye
The ratio of phenotypes in the offspring is:

6 red eyes, normal wings : 6 orange eyes, normal wings : 2 red ey 2 orange eyes, short wings

- a) Suggest the genotypes of the two parents.
  - For eye colour, we can see that there are eight red-eyed f
    flies (1:1)
  - We know that fly P1 has orange eyes, so it must have the alleles are needed for orange eyes). It will pass the same a
  - However, fly P2 has red eyes. I have two of the dominal these allele on to all of the pring and they would all have
  - In fact, half suggest spring have orange eyes. This suggest le La eyes and the alleles for orange eyes equally the stype Ee.





- For wing length, we can see that there are 12 normal-wi winged flies (3:1).
- P1 must have at least one dominant Wallele, because it W
- However, if it had two dominant Walleles, it would alway offspring; therefore, all the offspring would have normal v
- This is not the case; therefore, it is not the genotype
- The same logic applies second fly, P2. So both flies



### b) Draw a Punnett square to illustrate the possible results of the

	eW	ew	eW	ew 💌
Š	EeWW	EeWw	EeWW	EeWw
Š	EewW	Eeww	EewW	Eeww
3	eeWW	eeWw	eeWW	eeWw
3	eewW	eeww	eewW	een w
40				

Fly P1 has two

Here, you can Punnett square whether it mata question.



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### **QUESTIONS**

**● 1.** For each pair, say which of these proport ໂຄງ ເລື່ອreater

A) 3/5	P) 0.55
A) 0	B) 75 %
A) 50 Education	B) 9/16
A) 0.000046	B) 0.046 %
A) 8/20	B) 3/10



- 2. Two coreopsis plants which are doubly heterozygous for genes Y and R are crossed.
  - Gene Y has two alleles the dominant allele, Y, causes yellow outer petals, and the recessive allele, y, causes white outer petals.
  - Gene R has two alleles the dominant allele, *R*, causes red inner petals, while the recessive allele *r* causes yellow inner petals.
  - a) Explain what 'doubly heterative's means.
  - b) Using the Punnett ware come right, suggest the ratio of phenotypes with conspring.
- 3. Match redicted phenotypic ratios to the dihybrid crosses.

All the same (16:0)

9:3:3:1

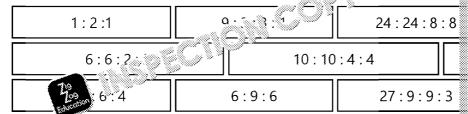
1:1:1:1

Two parents who are each heterozygrecessive genes.

One parent has only recessive alleles heterozygous.

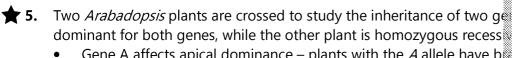
One parent is homozygous dominar parent is homozygous recessive for

🛊 4. Identify three pairs of ratios which are equivalent 🚳 you can simplify



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Zig Zag Education



- Gene A affects apical dominance plants with the A allele have be (lateral growth), while plants with two a alleles have a rapidly growbranches (apical dominance).
- Gene B affects flower colour plants with the rece B allele have yet two b alleles have white flowers.
- a) Suggest the ratio of of  $f_{1}$  is the  $F_{1}$  generation.
- b) Two plants from peneration are cross-bred. Draw a Punnet the ss.
- c) St ratio of phenotypes you would expect to see in the F<sub>2</sub> g
- **d)** What proportion of branched, yellow-flowered plants would you generation? Give your answer as: i) a percentage; ii) a fraction.



- A research team is studying the inheritance of two genes in mice:
  - Gene M controls fur colour; one allele causes chestnut (red-brown white fur.
  - Gene T controls tail length; the dominant allele causes long tails, causes short tails.

The researchers think both genes have a dominant-recessive relations mice with brown fur and long tails, expecting to produce offspring with

9 chestnut, long tail: 3 white, long tail: 3 chestall; short tail: 1 white,

Instead, the scientists' resulting as a liows:



Phenotype	Number c
Chestnut, long tail	
Light red, long tail	1
White, long tail	

- a) Write this as a ratio in its simplest form.
- **b)** Suggest **two** reasons why the researchers' results differed from the







### **Codominance and Multiple Alleles**

### **Background - key points**

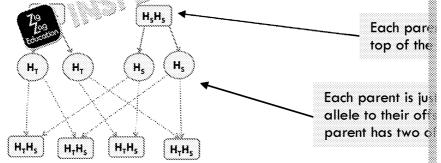
The simplest cases of inheritance involve single genes with two alleles and simple dominant–recessive relationships have been alleles. However, genetics often isn't that simple; sometime, there will be multiple alleles for the same gene. For the pie, common rabbits have at least four competing time of the pie, controlled (C<sub>c</sub>), agouti (Himalayan (C<sub>h</sub>) and allowed C<sub>s</sub>, each rabbit still only has two copies of the but it can approximation of these alleles.

In other cases, there may only be two important alleles, but both of them can affect an organism's phenotype. For example, a plant with one allele for flowers and one allele for red flowers may produce pink flowers, or red flowith white edges. This pattern of inheritance is known as codominance.

### **Worked example**

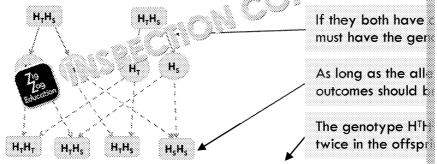
- 1. In a certain species of antelope, a gene for antler shape is codominal alleles have twisted, spiral horns, while antelopes with two H<sup>s</sup> alleles with one of each allele have 'curved' horns, which twist slightly but
  - a) Why is this an example of codominance?

    Both alleles contribute to the phenotype in lividual.
  - b) Draw a genetic cross diagram is show the result of breeding and a parent with a significant horns.



- c) Describe the phenotypes present in the offspring (from 1b).

  All the offspring have curved horns, since they all have the same
- d) Draw another genetic diagram to show the effect of crossing horns. Give the ratio of phenotypes in an of spring.



1 spiral horned: 2 curved horned: 1 straight horned

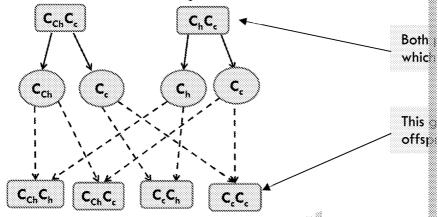
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## 2. Rabbits have four alleles for coat colour. They have a straightforwagouti (C<sub>a</sub>) is dominant to all other coat colours, chinchilla (C<sub>ch</sub>), is and albino (C<sub>c</sub>), and Himalayan is dominant to albino.

- a) A rabbit has the genotype  $C_hC_c$ . What phenotype does it have The rabbit has a Himalayan coat  $-C_h$  is the cominant allele of
- b) A chinchilla rabbit is crosse in a imalayan rabbit, and so albino coat colour l'an anetic diagram, show how this is.

  The albino a'll a seessive to all other alleles, so only rabbits he iver coat colour. This means both the other rabbits must be minant allele which gives them their coat colour.



c) Using percentages, give the predicer, oportions of phenoty question 1b).

d) Why would the offspring of a Himalayan rabbit and a chinching the agouti allele is dominant to all other alleles; therefore, if each the allele, they would be agouti (rather than Himalayan or ching)

### **Activity:**

Spot and correct the errors in the paragraph below.

Co-dominance means that an allele can be recessive dominant, depersonly occurs in plant species, due to their couples an appear which were a seent in either parent. When a are not written using capit and were case letters; instead, they are writinumbers. An example comminance is found in human blood type – to O antige comminant, while type B is recessive.

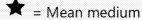
# 



## QUESTIONS

## KEY

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- 1. State whether these sentences describe conditional multiple alleles
  - A) An allele is not expressing the law of copies are present in the general
  - **B)** A gene has through  $B_a$ ,  $B_b$  and  $B_c$ , all of which are quite compared to  $B_c$ , all of  $B_c$ , all of which are quite compared to  $B_c$ , all of  $B_c$ .
  - າເພື່ອໄດ້ເຂົ້າເຂົ້າເຂົ້າ another gene from being expressed at all.
  - erent alleles of the same gene both contribute to the phe
  - Human blood type has multiple alleles I<sup>A</sup>, I<sup>B</sup> and I<sup>O</sup> which control the cells in the immune system.
    - People with genotype I<sup>A</sup>I<sup>A</sup> or I<sup>A</sup>I<sup>O</sup> have type A blood.
    - People with genotype I<sup>B</sup>I<sup>B</sup> or I<sup>B</sup>I<sup>O</sup> have type B blood.
    - People with genotype I<sup>A</sup>I<sup>B</sup> have type AB blood.
    - People with genotype I<sup>O</sup>I<sup>O</sup> have type O blood.
- a) Describe the relationship between the three genes.
  - **b)** Draw a genetic cross diagram to show the possible offspring of tw IAIO and IBIO.
- Uniquely, people with type O blood can donate blood to people parents have the genotypes IAIO and IBIO, who is the chance that a able to donate blood to either pagen?
- A gene, M, is involved is god and flower colour in an orchid. Flowers petals, and flowers. Flowers wi yellow 4 vian blue streaks.
  - Drugenetic cross diagram to show the possible offspring you plants which are heterozygous for M.
  - **b)** Suggest the genotypes of the parents which would produce offsp
    - 2 blue petals : 2 streaked petals
    - ii) All streaked petals
    - iii) All yellow petals
- Researchers identify two genes in a spiny mouse population from East alleles. They think both genes have a simple dominant-recessive relat
  - Gene P controls fur pattern the allele thought to be dominant (...) other produces straight (flattened) fur.
  - Gene C controls fur colour the allele that is be dominant (A brown) fur, while the other produces who will ur.

The researchers cross and read, mocha mouse with a flat-furred, several primary as Fi generation.

a) Si the phenotypic ratio whi

- the phenotypic ratio which the researchers would expect
- Instau, the researchers find that all of the mice in the F<sub>1</sub> generati brown) fur. The researchers now think that both genes may be co Describe how the researchers could test this theory by cross-bree give the expected results.

# 



## Sex Linkage

## **Background - key points**

Humans have 22 pairs of **autosomal chromosomes**; in each pair, the chromosomes have the same generation most vertebrates, we also have one pair of the chromosomes – females have the chromosomes, while males have one X chromosome. The Y chromosome and so there are yield one Y chromosome, which females have two copies of in their genome, but males only have one copy of these genes. We call these **X-linked** genes. There are also some y-linked genes, found only on the Y chromosome.

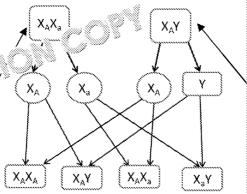
Since males only have one copy of X-linked genes, if they have an allele that causes a genetic trait, it will be expressed wherever it is present. However, recessive X-linked traits will only be expressed in females if they have two copies of the recessive allele. As a result, many X-linked recessive traits are significantly more common in males than in females.

The other implication of x-linkage is that not ergody pass on X-linked gewithout necessarily expressing supplies to a genetic disorder. By contrast linked trait to a daughter is a second a son, who will always inherit a Y challinkage can be a second patterns of inheritance, and you should study deeper united admig.

## Worked example

- 1. X-linked agammaglobulinemia is a recessive X-linked condition when 200,000 children. It causes B-immune cells to form incorrectly, and condition has to have regular injections of antibodies so that their
  - a) Draw a genetic diagram to show the possible genotypes of the and Tania. Neither parent has X-linked agammaglobulinemia carrier for the condition.

Tania's genotype is XAXa, because she carries the allele causes X-lir and police of the causes x-lir and police of the causes the recessive condition.



 $X_nX_n$ 



If the test reveals that the child is male, he will have a 50 % c linked condition (genotypes XAY and XAY are agually likely), bu chance of inheriting the condition (has all show will always inh allele from her father). Eith, wowing the gender of the prediction.

- T re s, Mike (M) and Saanvi (F) are having genetic c) columning. Saanvi expresses the genetic condition X-linked agammaglobulinemia, but Mike does not.
  - State the probability that Mike and Saanvi will have a child with X-linked agammaglobulinemia.
  - ii) State the probability that a son born to Mike and Saanvi will have X-linked agammaglobulinemia.
  - 50 % Saanvi will always pass on an Xa allele, while Mike will either pass on a 'normal' XA allele or a Y chromosome which is silent for this gene.
  - ii) 100 % Saanvi will always pass on an Xa allele, June while Mike will pass on a Y chromosome to a

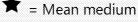


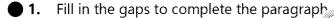


## **QUESTIONS**



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Humans have \_\_\_\_ pairs of \_ chromosomes, and a single which vary depending the individual is male or female. Fe chrom the same males have one \_\_\_ chromosome and one \_\_\_ c

Colour blindness is an X-linked recessive condition. A man is colour-b partner is homozygous for the 'normal' allele (XBXB). Draw a genetic cr possible genotypes of the couple.

Match the sentence to the type of genetic trait it best describes: X-link dominant or Y-linked.

## Description

A woman can be a carrier for the trait, but a male will always express trait if the correct allele is present.

Only men can inherit the trait.

If one copy of the correct allele is present in eit male or a femal will express the trait.

Explain why X-linked row sive that's are more common in males than a

isan X-linked condition. Thierry (M) has haemophilia, wh ney are expecting a child.

a) Is the condition recessive or dominant? Explain your answer.

- **b)** Using appropriate symbols, suggest the genotype of:
  - i) Adila
  - ii) Thierry
- Evaluate whether these statements are true or false: c)
  - Thierry and Adila have a 75 % chance of having a child with h
  - ii) A boy born to Thierry and Adila cannot express the condition
  - iii) A girl born to Thierry and Adila could express the condition h



## **Chi-squared Tests**

## **Background - key points**

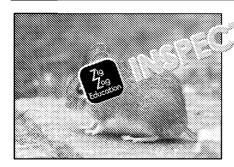
When two organisms are bred to study the inheritance of a specific gene or genes, the **phenotypic ratio** (rational particular pattern of inheritance of the genes. However, and he is rarely as tidy as the diagrams in your textbook and a simple, in blood type pattern of



inheritance I alleles are codominant, while the O allele is receboth blood Ab (IAIB) have four children, on average you would expect two children to be type AB (IAIB), and one child to be type B (IBIB). However might inherit two IA alleles, or each child could inherit an IA allele and an IB

The results we actually see (**observed results**) from a genetic cross can so from the results we are predicting (**expected results**). The **chi-squared** tes whether this difference between the observed and expected results can be chi-squared value is small, it's likely that the difference between observed due to random fertilisation, and other chance factors. However, if the chi-seans our original prediction is likely to be wrong. This means that we may explanation of the results.

## Worked example



^ .e ્રા ) f scientists is studying the t Nood mice.

- Gene A has an effect on face lenged
   allele have regular faces, while notes
   unusually long faces and large notes
- Gene B has an effect on fur color allele have brown fur, while mice grey fur.

The scientists believe that the two genes may be autosomally linked. The heterozygous mice (*AaBb*) several times, producing 48 offspring, and respectively.

Phenotype	Observed Results	Expected Results
Long face; brown fur	19	27
Long face; grey fur	13	٠, ١
Short face; brown fur	_/5	9
Short face: gree for	1	3

The expecte 9:3:3:1, you crossed unlinked do add up to 4

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## **NULL HYPOTHESIS**

A null hypothesis predicts that there is no significant difference between two observed and expected results. Most of the time of the sure inherited in predifferences between observed and expected sources between by

The opposite of the null becomes the alternative hypothesis – that the difference. Scientic Control hope to prove the null hypothesis wrong

b) Caut a chi-squared test on the results, using the formula

First, you need to take the observed number of individuals for each phenotype away from the expected number, and then square the answer.

Then, divide each volume Make sure you divide results for each phese number of results!

It will probably help you to draw a table like this, so you can keep track of your sums and don't miss out anything important.

Observed	Expected	• (0
19	27	(-8)
13	9	(4) <sup>2</sup>
15	9	(6) <sup>2</sup>
	3	(-2

The chi-squared for this set of results is: 2.37 + 1.78 + 4

(7° ) (10°	Chi-so	quared Distrib	ution	
Legrees of		Critica	l Value	
Freedom	p = 0.1	p = 0.05	p = 0.025	p = 0.0
1	2.706	3.841	5.024	6.635
2	4.605	5.991	7.378	9.210
3	6.251	7.815	9.348	11.34
4	7.779	9.488	11.143	13.27

c) Look at the table of critical values above. Based on the availant the genes are linked? Justify your answer.

Yes, the evidence suggests that the genes are (autosomally) linked. The chi-squared value with three decays of freedom is 9.48 (3 sf), which is greater than an object value at p = 0.05. This means that  $t^{1/2} = 0.05$  than a 5% chance that the observed respected by the genes assorted independently.

The area suggests that, in this  $F_1$  generation, A is more likely to be inherited with b, and a is more likely to be inherited with B, creating a large number of Axbb and aaBx offspring.

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## **QUESTIONS**

■ 1. Use ticks and crosses to show appropriate inappropriate uses of the chick are crest.

Chi-s

Usage

Composition of the composition o

Compare the actual results of a cross test with the predicted results.

Discover whether two genes are likely to be linked.

Work out how closely correlated different alleles are with the development disease, such as breast cancer.

- 2. What is the general null hypothesis in a chi-squared test?
  - **3.** A team of geneticists breeds snapdragon plants which are heterozygo
    - T controls stem length, and has two alleles: T (tall stem) and t (short
    - Q controls flower colour, and has two alleles: Q hellow flowers) and

The team gets the following results:



ype	Expected Results	Obse
Tall, yellow	36	
Tall, white	12	
Short, yellow	12	
Short, white	4	

- a) Why do the geneticists expect to see more tall plants with yellow white flowers?
  - **b)** Using a table like the one below, carry out a chi-squared test on t

Observed	Extracted $(O-E)^2$	

c) Use table of critical values (overleaf), explain whether you the results the geneticists were expecting.

## 



	Chi	-squared Distributi	on
Degrees of		Critica	al Value
Freedom	p = 0.1	p = 0.05	p = (
1	2.706	ا الحراب	5.0
2	4 / <sub>O</sub> .	5.991	7.3
3	6.251	7.815	9.3
Los 4	7.779	9.488	11.

- **4.** Genetic researchers are studying the production of flower colour in enzyme-producing genes (B and R), both of which are involved in p think that the genes work as follows:
  - Plants with at least one dominant B allele and two recessive ra
  - Plants with at least one dominant R allele and two recessive b
  - Plants with at least one dominant B allele and at least one dom flowers, as the two pigments will mix together.
  - Plants with two recessive b alleles and two recessive r alleles w won't produce any pigments.

The researchers cross two doubly heterozygous plants (BbRi) with @ to see offspring in the ratio:

9 purple: 3 red: 3 blue: 1 whice

The actual ros ់្នៅ the researchers observe are shown bel



Phenotype	Numbe
Purple flowers	46
Red flowers	0
Blue flowers	0
White flowers	34
Total	80



**b)** The researchers conclude that their original theory was incorre would have reached this conclusion.



Explain an alternative explanation or howesuits. 







## **Population Genetics**

## **Background - key points**

Population genetics is the study of the frequency of alleles in populations, how this changes over time, and he can be predicted.

**Using** ₹

All of the alleles of all of the genes present in a collectively to a recollectively to a recollective to a recollec

## Hardy-Weinberg equation

Most of the time, we don't know how common an allele is within a populable to see the phenotype caused by a specific allele. If the gene in quest dominant and one recessive, then we can use this information to work ou **Hardy-Weinberg equation**:

 $p^2 + 2pq + q^2 = 1$ , where p is the frequency of the dominant allele and q is the recessive allele. In the equation,  $p^2$  is the number of payons dominant the number of heterozygous (e.g. Tt or TT in TV). As, with one copy of each of homozygous recessive (e.g. Tt) as. Remember, since there are of

The Hardy–Weinberg േയ് എന്ന് a magic bullet; it works perfectly whe

- Ther proposed affeles being studied.
- The appeare equally likely to be passed on to the next generation.
- The rate of mutation is **negligible**, so the effects can be ignored.
- The population is genetically isolated from others.
- The population is large.
- The characteristic doesn't have any impact on which organisms chock

In practice, these conditions are never met. But as long as a population is conditions, the Hardy–Weinberg equation can still be a powerful tool.

## Activity: What went wrong?

A study is conducted on the frequency of a recessive allelo which causes a secondition. The disease causes individuals to have your one tissue, organ temperature regulation, and individuals have with the disease have a reduce

The population consists of 200 per desiring in a village in the crowded Notice as is identified in the population which is the population of the population and people as the property of the recessive allele.

A genetic tudying genetic diversity in the region, and sequences the people in the community. She discovers that the number of carriers is suprediction made by the Hardy–Weinberg equation.

Suggest reasons why this might be the case.



## **Worked example**

1. The chemical phenylthiocarbamide (PTC) tastes bitter and unpala other people cannot taste it at all. This odd characteristic is determined people with at least one A allele can taste the chemical, and people taste it. These two alleles are the only significant alleles of the general contents.

The frequency of the A allele in a remo u alian outback com

a) What proportion of passible two copies of the A allele?

When p is the  $f_{n,n}$  is of the dominant allele,  $p^2$  is

the people who have two copies of the all the homozygous dominant).

C N

D.

tv.

$$P^2 = (0.45)^2 = 0.2025$$

Expressed as a percentage, this is 20.25 %.

b) What proportion of people cannot taste PTC?

To find out the proportion of people who cannot taste PTC, we frequency of the a allele. Since there are only two alleles, p + a

P = 0.45. Therefore, q = 1 - 0.45 = 0.55

Since A is dominant, only people with two copies of the a allele cannot taste PTC. To calculate the number of homozygous rec

 $q^2 = (0.55)^2 = 0.3025$  or 30.25%

c) In a small town in Namilian Sequency of the A and a alleled group of 200 per starts asked to try and taste paper infused they can a second people from the group are able to taste

In this case, we don't know the frequency of the alleles to star with. However, we do know that the number of people who castaste PTC, and therefore have either one or two copies of the

We the frequency of allele A in the population.

allele, is 164 out of 200.

So 
$$p^2 + 2pq = 164/200$$
, or 0.82.

Since  $p^2 + 2pq + q^2 = 1$ , we can say that  $q^2 = 1 - 0.82 = 0.18$ .

 $q^2$  is the number of people who are homozygous recessive, and have two a alleles. To find out q, the frequency of the recessive allele, we need to square-root our  $\frac{1}{2} \frac{1}{2} \frac{1}{2} \frac{1}{2} \frac{1}{2} \frac{1}{2}$ .

$$\sqrt{q^2} = \sqrt{0.18} = 0.424$$

First in 3, the frequency of the A allele must be 1-q. 1 24 = 0.576 or 57.6 %.

## 



## **QUESTIONS**

KEY

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• 1. Fill in the gaps to complete the para ு p. நில்ல

The Hardy–Weinberg end thou a useful tool	which uses a known qu
individuals exກາງ ເຂົ້າສໍ particular	, to calculate the
gene It can also be used in reve this toate what proportion of a	erse; if you know how co
this to the what proportion of a	is homozygous do
homozygous	

● 2. In each of these examples, there are only two alleles – one is dominangene, work out the percentage of homozygous dominant and homozy the population. (You can use your calculator.)

Allele frequency	Homozygous dominant	Homozygous recessive
T = 0.9, t = 0.1		
Q = 0.6, q = 0.4		
R = 0.008, r = 0.992		
b = 0.27		

**3.** Roughly 25 % of a pondion he immune to the toxin urushiol, which immunity is called A, a dominant allele; individuals with two recess painful the last if they touch poison ivy.

Use the equation to calculate:

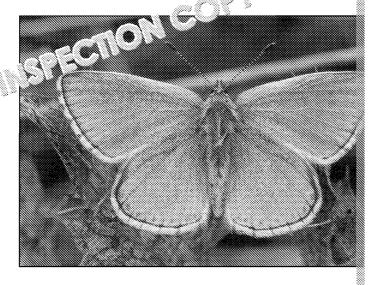
- **a)** the frequency of the *r* allele
- **b)** the proportion of the population who are heterozygotes, and, the they will pass on immunity to their children
- **4.** A horticulturist fills an enormous greenhouse with 10,000 Californian have brilliant orange flowers (caused by the dominant allele, *R*). He no have a trait caused by a recessive allele (*r*) which causes them to prod
  - The plants with yellow flowers have shorter stems than other plan
  - The characteristic is not linked to a sex chromosome.
  - There are 127 plants with pale yellow flower
  - The plants with pale yellow flowers a செரிக்க உள்ளை
  - a) What proportion of the fire heterozygotes (Rn)?
  - b) The horticultural around the greenhouse, and picks two corange-flowered offspring?



## **Exam-style Questions**

Use these exam-style questions to test what you have learnt throughout you questions will require knowledge from both years of your A Level, and are challenges you will encounter in your final exam.





a) Adonis blue butterflies (*Polyommatus bellargus*) have a bright blu This trait is controlled by the Wsc gene. A few butterflies have a d which causes the wings to be white instead of bine.

A section of the Wsc gene seguing e am a 'normal' Adonis blue

## **TATATAGATGGTATA**

e butterfly has a nearly identical gene sequence, oves a single base from the sequence.

Explain, using the sequence above as illustration, how small r	
widespread effects.	







**b)** Another individual of the same species undergoes a mutation, ch

## **TATATAGATGGTATA**

to:

## **TATATCGATGGTATA**

i)	Identify the type of mutation which is signal above.
11	The restauron does not cause any change in the protein prodes any change in the protein prodes are suggest why no change is observed. Explain your answer in the protein prodes are suggested.
) Ot	<u>.</u>
	her factors besides mutations can affect how and where a gene these.
of	her factors besides mutations can affect how and where a gene these.
of	these.
of	these.
of Fa- 	these.
of Fa- 	these.





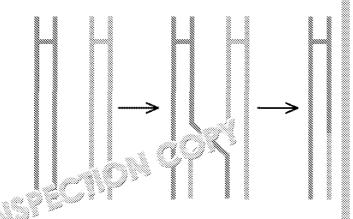


Figure 2: Crossing-over of homologous chromo

- 2. A cell divides during meiosis to form gametes. At one stage of this processir up and 'cross over', exchanging part, or all, of one of their arms (see a) i) Give the stage of meiosis during which this process happens.

  ii) This process contributes to genetic diversity in gametes. Described in the process contributes to genetic diversity in gametes.
- 3. Researchers have discovered a new trait in the *Arabidopsis* (rock cress 'over florescent' (they have far more flowers than 'normal' plants), but researchers want to study what causes this trait; they know that the traillele of gene F, but they don't know where gene F is located.
  - a) A heterozygous plant is crossed with a homozygous recessive plane Explain what the term homozygous means.



b) Using appropriate symbols, draw a genetic cross diagram to show crossing a heterozygous plant with a homozygous recessive plant.

State the expected proportions of phenotypes in the offspring, us



**c)** The researchers think that the gene which causes over florescence which controls stem height. *Arabidopsis* plants with the dominant whereas plants with two recessive *t* alleles have short stems.

Two doubly heterozygous Arabidopsis plants are crossed, and the

Phenotyc >	Observed	Expe
ดิเทลาสเการะเลกซุ tall	74	6
\	10	2
Over florescent, short	13	2
Normal flowers, short	15	



Use the chi-squared equation to calculate an X<sup>2</sup> value for these re

$$\chi^2 = \Sigma \frac{(O - E)^2}{E}$$





## d) The researchers conclude that the two genes are linked.

i)	Suggest how the researchers would have reached this conclu	J.
		•

ii) Based on the information have read, suggest why this corresearchem.



- **4.** a) The wild tulip (*Tulipa agenesis*) has a gene which controls the bre
  - Plants with at least one copy of the dominant Nallele break late, in May.
  - Plants with two recessive n alleles do not break down the hor April.

In a wild population, 7 % of tulips flower in April. Use the Hardy—Nethe proportion of heterozygotes in the population. Assume that / gene N.

Hardy–Weinberg equation:  $p^2 + 2 pq + \sqrt{2}$ 



b) The tulip also has a gene, F, which control with r colour. Plants we flowers, and plants with two Figure 1016 Fave white flowers. Plants with flowers.



## **ii)** Draw a Punnett square to show the possible offspring, when heterozygous for both the F gene and the N gene are crossed write the expected ratio of phenotypes in the offspring.



5.	The bacterium Pantoea agglomerans can be genetically engineered to
	Plasmodium falciparum, the parasite that produces malaria.

A gene sequence which will produce the toxin is a parated from another endonuclease enzyme. The enzyme cuts is a very that leaves the gene exposed DNA sequences known as a sequences.

Explain how the cross as a low inserted into a plasmid, and how this presented into a plasmid, and how the presented into a plasmid into a	·c
sample ac which generate the useful toxin.	

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## **Answers**

Note: answers to activities are given where appropriate.

### **DNA: the Code of Life**

Activity: the countries are: 1) Latvia; 2) Mali; 3) Haiting a

### Questions:

1.

The DManiple మాగ, GTA and GTG all code for the same amino acid.

The D pence ATAGGACAG is read as three, rather than seven, amino aci

The DNA triplet AGC codes for the same amino acid in kangaroos, ferns and ye

- 2. a) UUA
  - b) AAU
- 3. a) UCAUGUCCGAUUACG
  - Serine-Cysteine-Proline-Isoleucine-Threonine (Ser-Cys-Pro-Ile-Thr)
- **4.** a) The valine (Val) which 'should' be at position three in the amino acid sequences (Gly).
  - **b)** Base 8 (incorrectly transcribed as guanine, rather than uracil).
- 5. Many reasons, including:
  - Some of the DNA may code for RNA molecules which the never translated
  - Eukaryotic gene sequences contain intron , wic ire spliced out before tree
  - Transcription factors can affect vice and of a gene are used to produ
  - There are long sections contact and DNA outside of genes, which do not



1.

## Description

The chromatids are pulled to either end of the cell by spindle fibres.

The chromatin condenses so that distinct chromosomes are visible.

The chromosomes at each pole of the cell are surrounded by a new nuclear en Spindle fibres, attached to each chromosome at the centromere, cause the chromosomes to line up across the equator of the cell.

- 2. A Anaphase. The separated chromatids are being pulled to either end of the
  - B Prophase. The chromatin has condensed, because individual chromosomes chromosomes have not yet started to line up across the equator of the cell
- 3. a) Free nucleotides with bases which are cor pathers ary to the bases on the Adenine always pairs with thymically specific always pairs with guanine.
  - b) DNA polymerase joins the solution to be solved by the body strand phosphate group (1) to make the 'backbone' of a new DNA strand
- 4. The air to to so produce daughter cells with identical genetic material, so different and repair themselves. If DNA contains errors, then tissues will consets of instructions. This will reduce the effectiveness of the tissue, since cells no structures, or react in different ways to the same stimulus.

Alternatively, damage to DNA could cause the development of a tumour. By sto from dividing, the body reduces the risk of cancer.



Cancerous tissues divide rapidly and in a disorganised way. Vinca alkaloids ca process, since they will be particularly effective in cells that divide quickly, what

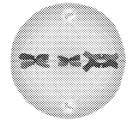
- b) Other cells carry out mitosis too; it's difficult to ta live larges so that they of toxin is likely to affect the function of other a sussicausing damage.
- Bacterial cells don't have centriole no makes or spindle fibres they div than eukaryotic mitosis The Common be interrupted by a toxin which p

## Meiosis









2: Metaphase I



1: Prophase I

2.

### Description

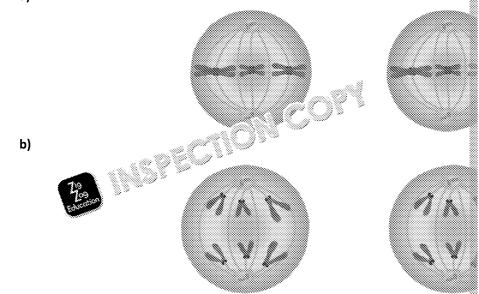
Homologous chromosomes line up in pain a angline equator of the cell.

Separated chromatids are prived in the end of the cell by spindle fibres.

າະ ົອform around the separated chromatids, and four gam

n condenses, and homologous chromosomes cross over, exchan

- 3. Mitosis produces genetically identical daughter cells, while meiosis produc other and from the original parent cell.
  - Mitosis produces diploid cells (with two full sets of genetic material), while
  - Mitosis produces two daughter cells, while meiosis produces four daughte
- 4. a)

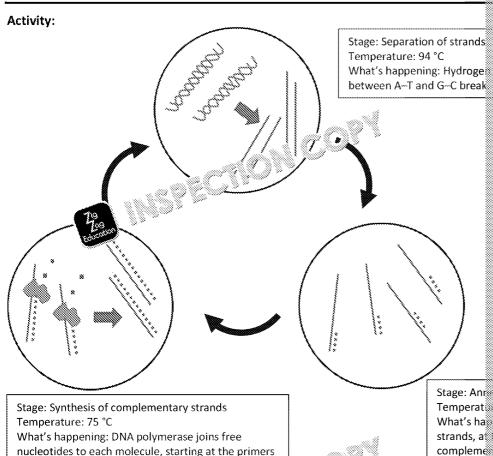




## During metaphase I, homologous pairs of chromosomes line up across the equa Down's syndrome has three copies of chromosome 21, there is one chromoson any other, and this disrupts the process of meiosis.

- 6. Meiosis produces genetic diversity in several stages. Firstly, during propha join up and cross over, exchanging genetic material by swapping part of or chromatids on each homologous chromosome are no longer identical. The independently assorted into the two daughter during metaphase/ana of 2<sup>23</sup> (over 8 million) ways. Finally ുറി വ്യവത്തെ can be oriented in e II, so that each chromatid control is neither daughter cell during anapha
  - Genetic diversion for sitant because it means that the rodent's offs b) differ ়ে ১
    - າ ເພື່ອເດັກment like the Sahel, which is getting drier, having geneti probability that at least one individual will have alleles that allow
    - Individuals which thrive will have higher reproductive success, reachi own offspring.
    - Also, the rodent's offspring can potentially spread out into a broader ecosystems).

## The Polymerase Chain Reaction



### Questions:

- A) True; B) False anne-" an salar a temperature of 55–60 °C, which is mu temperature for a new separation; C) True; D) False – they are added by a polyme
- Each cycle can only double the amount of DNA present, so several cycles are ne quantities of DNA from tiny samples.

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nucleotides to each molecule, starting at the primers

## **3.** • First, the DNA is placed into the thermal cycler, along with a high-tempera polymerase enzyme.

- Specially designed short primer sequences are also added; these will be compared sequence.
- An excess quantity of free nucleotides are also added.
- The cycler heats to roughly 94 °C, breaking the hydrogen bonds between t
- The cycler then cools to 55–60 °C. This allows the Shaw DNA primers to an
- The cycler then heats to 75 °C. This is the continued emperature for the spentides to each strand of DN.
- The process is repeated നുത്തി സ്ഥാനി the sample of DNA is large enough
- 4. a) Parent mixing found out whether their child has a familial genetic discount of the second of t

However, getting a large enough DNA sample from an early embryo is diffiembryo. Because of this, it's better to take a small sample of embryonic District that surround the child, or from the amniotic fluid). This can then be amplified to the control of the child, or from the amniotic fluid.

- b) Pathogens often actively avoid detection by the immune system by hiding evasive tactics. However, small amounts of pathogenic DNA may still end be amplified using PCR.
- 5. Because DNA is not 'proofread' during replication, there is a risk that the DNA sereplicated. If this happens often, or early in the cycling process, there is a risk the DNA produced will not match the original sample. Since the issues as a series to amplify sereening/testing, this could possibly lead to interpret agreement of genetic conditions.

## **DNA Profiling**

1. Non-cu NA DNA which is found within the genome, but which doesn't comprotein. A molecules.

Recognition site – a short sequence of DNA bases at which a restriction endonuc

- 2. A) False DNA molecules are negatively charged due to their phosphate groups fragments move towards the positive electrode (or anode).
- 3. All DNA molecules are attracted to the positive electrode; each molecule has a phosphate groups. However, the smaller molecules are able to move more rapis polymer molecules in the gel, and so they move further. The process of cutting endonuclease creates fragments of various sizes, which move at various speeds same length form distinct bands on the gel.
- 4. a) Dog 4 is the puppy's father. We can tell this because a of the puppy's DNA or Dog 4, and the puppy gets all of its genome in the instance and father, DNA repeats which affect the length a frequence.
  - b) The VNTR lengths and generic field in present in each individual both af recognition sites. The field are that, generally, no two individuals produce D when their FNL and it using a restriction endonuclease enzyme.
  - c) Ic two produce the same pattern of DNA bands in a DNA profile.

## 



### As stated in the question above, identical twins produce the same pattern of DN cannot be distinguished by genetic fingerprinting. However, there are also othe

- A DNA sample may prove that a suspect was at a crime scene, but it doesr role in a crime.
- The sample obtained may be too small to be useful, or it may be damaged.
- Small samples are more prone to being contaminated and incorrectly amp is incorporated into the DNA sequence.
- Criminal juries are not experts in genetic finge and may misinterp
- Forensic criminologists can use the maiduality of DNA to identify crimina 6. unique set of DNA fraggemes the work variations in their DNA (particularly n DNA profile of an individual, al the scene.
  - prtion of DNA bands which are shared between individuals can be b) Ti rel ships. An individual's DNA can be compared with that of a potential particularly used for paternity testing, but in rare cases records of materni for adopted children.
  - Medical professionals can use DNA profiles to identify signs of genetic disc risk for conditions with a genetic component. For example, a mutation ma band of DNA to appear.
- 7. Any pattern meets the criteria, if:
  - All of the DNA bands seen in the foal's profile are also seen in either of the between the two parents).
  - The parents also have other DNA bands, which are not seen in the foal.
- 8. The length of an STR sequence will determine how far apart two recognitions In turn, this will affect the size of a specific DNA fragment which is produc
  - For individuals with more STR repeats, the DNA fragility will be longer and so, during electrophoresis, it will move a share of a face across the gel.
  - Therefore, a specific band produced ുടിെക്കാന്oresis should appear in a on how many STR repeats and income
  - This means that in the least be assessed to find out whether they have
  - Charge No. She syndrome.

    The professionals can help a patient to sefore they actually develop the condition. This may in turn impr them to prepare for any changes they will have to make to address the co

## Mutation and Expression

### **Activity:**

1. a)

Sequence Number	Original	New	
1	ACT AAG CCC AAT	ACT AGC CCA AT	D
2	GGT TAA CGA CTC	GGT TAA CGA TTC	
3	TTT TGC ACA CAG	TTT T , SC ACA CAG	
4	GAT TTG AAC CCT	ارگر TTT GAA CCC T	

The mutation in sequence 4 is love likely to have a major effect on the pr mutation involved the form of a single base, so the ribosome will start ກe າ ເປັນ ເກe left. This is a frame-shift mutation; every triplet aft cely different amino acid sequence could be produced, and the p mt 3D shape and function.

The mutation in sequence 3 is an insertion of three bases, so an additional protein sequence produced. However, the triplets after this are not affect sequence remains identical.

# 



Alternatively, the mutation could occur in an intron – a sequence of DNA bases mRNA, but then is spliced out and not translated into protein.

- 4. This change will be a frame shift in the overall protein.
  - Significant start reading every codon after the mutation one place to the start reading every codon after the mutation one place to the start reading every codon.
  - The sequence of amino acids produced 'downstream' from bp 1510 will be
  - The protein may also be cut short due to the creation of a stop codon whe
- **5.** If siRNA joins with an RNA-induced silencing complex (RISC) protein and bi will prevent that sequence of mRNA from being translated at a ribosome.
  - In the longer term, this will cause the mRNA to be broken down.
  - If siRNA is inserted into a living cell/organism, it can be used to temporaril
    protein from being produced.
  - The change in the organism can be observed, and this can be used to make the protein.
  - This is known as a gene knockout method.
- 6. a) If the substance gets into the nervous system, it may hind to the promoter polymerase from being transcribed. The affect of using will no longer prosonervous impulses will no longer be faited. This could prevent the control movement or the function of the organs. Alternatively, the substant permanently 'switches of ', no slucing excessive and unnecessary copies of could disturb the factor of the nervous system, causing acetylcholine resplant of the system of communication.
  - b) Single genes code for subunits of the same structure, the nematode will the same time.

## **Gene Technology**

### **Activity:**

CRANE BADMINTON = RECOMBINANT DNA – genetic material from one organism we to contain genes from another source.

AID ANGLES = DNA LIGASE – an enzyme which can be used to join sections of DNA, between nucleotides.

KEENER GRAM = MARKER GENE – a gene which is inserted along with another gene of the genes can be easily verified.

INFANTS ROOM ART = TRANSFORMATION — the modification of an organism by humanother organism.

NICER ANGST = TRANSGENIC – an ( all in inch has been modified by humans to a organism.

ONCE UNSE ON SOLUTION ENDONUCLEASE — an enzyme which short sequence.

# 



### **Questions:**

- 1. vector; restriction; recognition/base pair; ligase; recombinant
- Number 1 Producing DNA fragments. A section of DNA containing the target restriction endonuclease enzyme, leaving 'sticky ends'.

Number 2 – Gene insertion. A plasmid vector is cut using the same restriction endends which match those of the DNA fragment. The fragment can then be joined to Number 3 – Transformation. Calcium chloride set and mild heat shock as membrane highly permeable, so that the piesr, it is able to cross it.

Number 4 – Observing green flux are protein. This protein is produced by a inserted along with the transport of the marker gene is visible due to its fluores whether the plantage and the genes and if it has been successfully taken up Number to the hadden to the bacteria are cultured on an agar plate, where the from the gene. The bacteria could alternatively be grown in a liquid grown resistance marker gene has been used, the bacteria may be grown in an environcheck they have taken up the plasmid.

- 3. i) mRNA is easier to extract from a cell than a specific gene, and so mRNA catreated with a reverse transcriptase enzyme. Reverse transcriptases product (complementary DNA) from mRNA, using free nucleotides. The cDNA mole polymerase and more free nucleotides, to produce double-stranded DNA, genome of another organism.
  - ii) a known amino acid sequence is entered into a specific computer program appropriate DNA sequence, and controls a machine which makes this DNA
- **4.** The gene may have been inserted without a promoter and terminator sequence cannot bind to the gene and use it to produce mRNA.
- **5.** Arguments for:
  - Generates valuable products which is product in hunger, help human socie
  - May reduce waste (e.g. t alonger shelf life) and create a more
  - Possibility to sav land ware medical advances, e.g. production of cheap cure at a land ware formers.
  - In the genome.
  - Pulse of science is to understand the world better; scientific curiosity of

### Arguments against:

- Unethical to allow animals to suffer for human benefits.
- Companies carrying out GM may have unethical business models (e.g. mak seedless crops yearly).
- Research tends to be expensive, and budgets could be spent elsewhere.
- Risk of transgenic organisms 'escaping' and affecting wild populations, esp plasmids to other bacteria.
- Many aspects of genetics are not fully understood, so we may be unaware
- May lead to people demanding 'designer babies' individuals wanting their c traits.
- Religious objections to altering genetic material.
- from the liver. These cells care the planted into the body, where the Alternatively, a functional livers could be inserted into the genome of liver stem into the body. These cells care the planted into the genome of liver stem into the body. The could be inserted into the genome of liver and will be considered into the liver, and will be considered into the body.
  - b) In the reinfino plant tissue cultures, which produces a protein that is to specific from the pest species. This would deter pests, reducing the need for pesticides.

# 



### 7. A restriction enzyme could cut the bacterial genome in the middle of an essay. bacteria would not be able to survive and produce the desired protein. Ho without any of the genes on plasmids, since plasmids carry non-essential

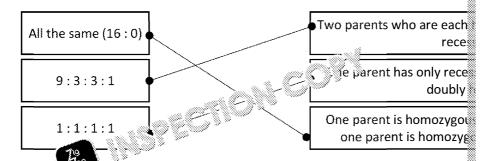
- Inserting a whole genome into a cell is difficult, since the membrane is not plasmids are small, and can cross the cell membrane more easily.
- Plasmids are small and easy to sequence, so they can easily be assessed for recognition site.

## Dihybrid Inheritance

### Questions:

- 1. a)
- b)
- $75(\%) \div 100 = 0.75$
- c)
- B is bigger:  $\frac{9}{16} = 9 \div 16 = 0.5625$ ;  $50 \ (\%) \div 100 = 0.5$ B is bigger:  $0.046 \ (\%) \div 100 = 0.00046$ A is bigger:  $-\frac{8}{20} = 8 \div 20 = 0.4$ ;  $\frac{3}{10} = 3 \div 10 = 0.3$
- 2. If an organism is doubly heterozygous, it means that it has two different a studied – in this case, it means that both plants have the genotype YyRr.
  - 9 yellow outer petal, red inner petal: 3 white outer, red inner: 3 yellow o yellow inner

3.



- 4.
  - 1:2:1=4:8:4
  - 24:24:8:8=6:6:2:2
- All (100%) of the plants in the F<sub>1</sub> generation will have yellow flowers and a 5.

h	1
v	,

	A8	Ab	88	ab	
8	AABB	AABb	AaBB	AaBb	
An	AAbB	AAbb	AabB	Aabb	
80	aABB	aABb	aaBB	29 <u>(</u> )	•
Q.	18 B	r	aabB	aabb	

- now, lateral growth: 3 yellow, apically dominant: 3 white, lateral grow c)
- 56.25 %;

## 



In addition, it's probable that the gene for fur colour is codominant, since colour which is intermediate between the two parents.

## **Codominance and Multiple Alleles**

### Activity:

Codominance means that when the recessive or dominant<sup>1</sup>, depending on the plant species to remplex genetic expression, and it means that, sometime were not presented expression. When alleles are codominant, they are not written letters; instead of the parent. When alleles are codominant, they are not written letters; instead of the parent. When alleles are codominant, while type B is recessive.

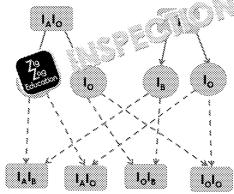
### **Corrections:**

- 1 Codominant alleles are neither recessive nor dominant; they are partly expresse
- 2 Codominance occurs in animals and plants, and in other organisms such as fungi
- **3** Codominant alleles are written using the same first letter, and then variable lettersubscript); for example, C<sup>w</sup> or D<sub>S</sub>.
- **4** The alleles for type A and type B antigens (I<sup>A</sup> and I<sup>B</sup>) are codominant alleles, while recessive.

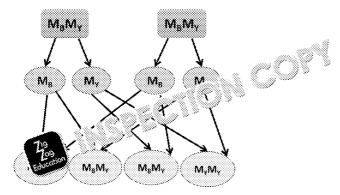
### **Questions:**

- 1. A) Neither describes a recessive allele; B) Multiple alleles; C) Neither describ
- 2. a) I<sup>A</sup> and I<sup>B</sup> are codominant, while allele I<sup>O</sup> is received to both.

b)



- c)  $\frac{1}{4}$ , or 25 % this is the proportion of the offspring which are expected to h
- 3. a)



- b) i)  $M^BM^B$  and  $M^BM^Y$ 
  - ii) M<sup>B</sup>M<sup>B</sup> and M<sup>Y</sup>M<sup>Y</sup>
  - iii) M<sup>Y</sup>M<sup>Y</sup> and M<sup>Y</sup>M<sup>Y</sup>



F<sub>1</sub> generation mice are all doubly heterozygous, because they have been producing dominant and homozygous recessive mice. Therefore, they will all have the P<sub>S</sub> causes spiny fur, P<sub>F</sub> causes flattened fur, C<sub>M</sub> causes mocha fur and C<sub>W</sub> causer correct, mice with P<sub>S</sub>P<sub>F</sub> will always have curly fur, and mice with C<sub>M</sub>C<sub>W</sub> v

The Punnett square shows all the possible genery of the offspring:

P.C., P.C.

	r.sew	2.25M	Prom.	100
S. C.	P <sub>S</sub> P <sub>S</sub> C <sub>A</sub> ,C <sub>h</sub>	S. S. C. S. W	P <sub>S</sub> P <sub>F</sub> C <sub>M</sub> C <sub>M</sub>	P <sub>S</sub> P <sub>F</sub> C <sub>M</sub> C <sub>W</sub>
a.	P <sub>S</sub> P <sub>S</sub> C <sub>W</sub> C <sub>M</sub>	P <sub>s</sub> P <sub>s</sub> C <sub>W</sub> C <sub>W</sub>	$P_sP_FC_WC_M$	P <sub>s</sub> P <sub>F</sub> C <sub>W</sub> C <sub>W</sub>
Ž a	P <sub>F</sub> P <sub>S</sub> C <sub>M</sub> C <sub>M</sub>	P <sub>F</sub> P <sub>S</sub> C <sub>M</sub> C <sub>W</sub>	P <sub>F</sub> P <sub>F</sub> C <sub>M</sub> C <sub>M</sub>	P <sub>F</sub> P <sub>F</sub> C <sub>M</sub> C <sub>W</sub>
Ç.	P <sub>E</sub> P <sub>S</sub> C <sub>W</sub> C <sub>M</sub>	P <sub>F</sub> P <sub>S</sub> C <sub>W</sub> C <sub>W</sub>	P <sub>F</sub> P <sub>F</sub> C <sub>W</sub> C <sub>M</sub>	P <sub>F</sub> P <sub>F</sub> C <sub>W</sub> C <sub>W</sub>

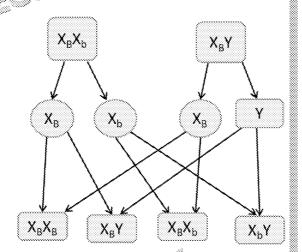
If the researchers' predictions are correct, based on the Punnett square the following ratio of phenotypes in the offspring:

1 spiny, mocha fur : 2 curly, mocha fur : 1 straight, mocha fur : 2 spiny, san straight, sandy fur : 1 spiny, white fur : 2 curly, white fur : 1 straight, white

## Sex Linkage

- 22; autosomal; X; X; Y
- 2.





3.

A woman can be a carrier or the trait, but a male will always express the trait if the and the is present.

Only 12 an sherif the trait.

If one of the correct allele is present in either a male or a female, they will express the trait.

4. X-linked recessive traits are more common in males, because they only have on will express the trait when only one allele is present. By contrast, females will o copies of the recessive allele are present – with rare alleles, this is highly unlike



b) i) X<sup>H</sup>X<sup>h</sup>; ii) X<sup>h</sup>Y

c) i) False – drawing a genetic diagram will show that two out of four offst cause haemophilia, so there is a 50 % chance.

iii) True – although girls with haem of his operare in the wider population male with haemophilical in machild has a 50 % chance of inheriting



1.

Usage	
Compare two populations to see whether they have different average body sizes.	(X <sup>2</sup> does not compar student's t-test for t
Compare the actual results of a cross test with the predicted results.	✓
Discover whether two genes are likely to be linked.	✓
Work out how closely correlated different alleles are with the development of a disease, such as breast cancer.	(X <sup>2</sup> does not measure This would involve us such as regression as

2. The null hypothesis of a chi-squared test in the state of the same of the s

3. a) The vellow fine of deal stem traits are controlled by dominant alleles are of the watever one copy of each allele is present.

b)

KCO.		
Observed	Expected	$(\mathbf{O} - \mathbf{E})^2$
33	36	$(33 - 36)^2 = (-3)$
15	12	$(15 - 12)^2 = (3)^2$
10	12	$(10 - 12)^2 = (-2)$
6	4	$(6-4)^2 = (2)^2$

 $\chi^2 = 0.25 + 0.75 + 0.333 + 1 = 2.33 \, I_{\odot}$ 

Yes, the results are consistent to the geneticists were expect freedom, and the year auch less than 7.815, which is the critical value high stable to the difference between the observed results and the boxes.

## 



	Observed	Expected	$(0-E)^2$
	46	9 x 5 = 45	$(46 - 45)^2 = (-1)^2 =$
	0	3 x 5 = 8	$(0-15)^2 = (-15)^2 =$
	•	» 3 x 5 = 15	$(0-15)^2 = (-15)^2 =$
7-3	34	1 x 5 = 5	$(34-5)^2 = (29)^2 = 8$
Total	80	$16 \times 5 = 80$	

 $\chi^2 = 198 (3 sf)$ 

- the chi-squared value is greater than the critical value for p<0.05. This means 5 % probability of achieving results skewed by chance alone, if the null hypothesis (In this case, the chi-squared value is so enormous that checking the critical but you should always take the time to confirm your suspicions.)
- c) This looks like an example of epistasis (an inheritance pattern where an all another gene is expressed, or stops another gene from being expressed).

  In this case, at least one dominant allele is needed from both genes (BxRx) two recessive alleles of either gene are present, then no pigment is produce purple flowers: 7 white flowers, and the actual (observed) ratio is 9.2 purple two genes might produce enzymes which we in sequence to produce (recessive alleles), the pigment is not read to the control of the

## **Population Genetics**

### **Activity:**

Although the econditions of Hardy–Weinberg are never perfectly met by any close enough meeting the conditions that you can use the equation to predict allet true for our case study, for several reasons:

- A village in a densely populated region like the Niger Delta is not likely to have \( \biglie{\pi} \)
- Immigration and emigration from the community could be substantial.
- The population size is quite small (200 people), which means the effects of chardistribution of alleles.
- People with two copies of the recessive allele have a reduced life expectancy, a
  under-represented in the actual population. In addition, this factor could be mo
  country such as Nigeria, where access to healthcare is not universal.

### **Questions:**

1. characteristic/trait; allele; pool; population; heterozygous; recessive

2.

Allele frequency	Homozygous dom'nant	)nozygous recessive	
T = 0.9, t = 0.1	T <sup>2</sup> (.84	$t^2 = 0.1^2 = 0.01$ 1 %	
Q = 19 = 0	$Q^2 = 0.6^2 = 0.36$ $36\%$	$q^2 = 0.4^2 = 0.16$ $16 \%$	
R = 0.008, r = 0.992	$R^2 = 0.008^2$ = 0.00064	$r^2 = 0.992^2$ = 0.984064	
b = 0.27	$B^{2} = (1 - 0.27)^{2}$ $= 0.5329$	$b^2 = 0.27^2 = 0.0729$ $7.29 \%$	
	53.3 %	1.25 70	

## 



a) Since the allele for urushiol immunity is dominant, both dominant homozy immune to urushiol, so:

$$p^2 + 2pq = 0.25$$
.

We know that  $p^2 + 2pq + q^2 = 1$ , so  $q^2 = 1 - 0.25 = 0.75$ .

If  $q^2 = 0.75$ , then the frequency of  $q = \sqrt{0.75} = 0.866$ . This is equal to the

- b) p+q=1, so  $p=(1-q)=(1-0.866)=0.13^{4}$ Finally, to work out the number of heteror  $pq=2(0.866\times0.13)$
- 4. a) 127 out of 10,000 plants have a least own flowers this is a recessive trait. Hardy–Weinberg 6

$$q^2 \mathcal{I}_{log}^{\circ} \stackrel{27}{\underset{000}{\sim}}$$

be used to work out q, the frequency of the r allele:

$$q = \sqrt{0.0127} = 0.113$$
 (3 sf), or 11.3 %.

Since we can only see evidence of two alleles, we can assume that these a the population, which means that p + q = 1. So, p = (1-q) = (1-0.11). To calculate the number of homozygous dominant plants:  $p^2 = (0.887)^2$ . Finally, the number of heterozygotes:  $2pq = 2 \times 0.887 \times 0.113 = 0.200$ . so 20 % of the population are heterozygotes (Rr).

**b)** Of all the plants in the greenhouse, 7,870 out of 10,000 (78.7 %) are homozhorticulturalist can exclude the 127 pale-yellow plants by sight. So he has a homozygote on his first try

But we need to know the probability that the horticular ralist will pick two a row. To find this out, we multiply the change of cking the first poppy ( $\frac{7}{9}$  second poppy (since one has already en picked, this is actually  $\frac{7869}{9872}$ ). This rule of probability. 7870 7869



\_ (63.5 %)

## Questions:

- 1. vector; restriction; recognition/base pair; ligase; recombinant
- 2. Number 1 Producing DNA fragments. A section of DNA containing the target restriction endonuclease enzyme, leaving 'sticky ends'.

Number 2 – Gene insertion. A plasmid vector is cut using the same restriction enends which match those of the DNA fragment. The fragment can then be joined to Number 3 – Transformation. Calcium chloride solution and a mild heat shock a membrane highly permeable, so that the plasmid is able to cross it.

Number 4 – Observing green fluorescent protein. This protein is produced by a inserted along with the target gene. The marker gene is visible due to its fluores whether the plasmid contains the genes and if it has been successfully taken up Number 5 – Growth. Here, the bacteria are cultured on a gar plate, where the from the target gene. The bacteria could alternative my grown in a liquid grow

- mRNA is easier to extract from a ce in a specific gene, and so mRNA can treated with a reverse at nsc chase enzyme. Reverse transcriptases production of the complementation of the com
  - **b)** A known amino acid sequence is entered into a specific computer program appropriate DNA sequence, and controls a machine which makes this DNA

## 



## 4. The gene may have been inserted without a promoter and terminator sequence cannot bind to the gene and use it to produce mRNA.

### **5.** Arguments for:

- Generates valuable products which may prevent hunger, help human socie
- May reduce waste (e.g. tomatoes with a longer shelf life) and create a mo
- Possibility to save lives and make medical advances, e.g. production of chear cure/treat genetic disorders.
- Increases understanding of the genome.
- Purpose of science is to under think would better; scientific curiosity out

### Arguments against:

- Unetbical \*: "Windle to suffer for human benefits."
- C 1 es scriying out GM may have unethical business models (e.g. makes section section).
- Research tends to be expensive, and budgets could be spent elsewhere.
- Risk of transgenic organisms 'escaping' and affecting wild populations, esp plasmids to other bacteria.
- Many aspects of genetics are not fully understood, so we may be unaware
- May lead to people demanding 'designer babies' individuals wanting their traits.
- Religious objections to altering genetic material.
- 6. a) Insert a functional version of the gene which produces the enzyme directly from the liver. These cells can then be implanted into the body, where the Alternatively, a functional gene could be inserted into the genome of liver stem into the body. These cells will divide to produce tissues within the liver, and will
  - b) Insert a gene into plant tissue cultures, which produces a protein that is to species. Alternatively, the protein could be an enable which creates a protein the pest species. This would deter pests, roug in the need for pesticides.
- 7. A restriction enzyme could at a saterial genome in the middle of an established would not a saterial genome in the middle of an established with a restriction and produce the desired protein. However, and are saterial genome in the middle of an established are saterial genome in the middle of an established with a restriction enzyme could be a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the middle of an established with a saterial genome in the s
  - with arm of plasmids, since plasmids carry non-essential governments on plasmids, since the membrane is not play are small, and can cross the cell membrane more easily.
  - Plasmids are small and easy to sequence, so they can easily be assessed for recognition site.



# 



## **Exam-style Answers**

	Question	ū	Answer
1.	a)		Any restrict for the first of t
			A completely different sequence of amino acids is produc, D, therefore, the protein has a different function / no function. <
	(q	i)	Substitution mutation ✓
		( <del>)</del>	The DNA code is deger of my retrian one triplet can code for the same amino acid. V  The rip may have been substituted for the same aminacle.
	(C)		Many possible answers:  Transcription f  Affect ::  Affects whi
			Means the RNA is <b>not translated</b> and is broken down instead. ✓

INSPECTION COF

Genetics Skills Pack for A Level Edexcel B Biology

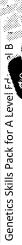
Page 66 of 78



## INSPECTION COPY

	Question	_ c	Answer	Marks
2.	a)	i)	Prophase I	1
		(ii)	During meiosis II / metaphase II / anaphase II, the <b>chromatids</b> that separate into each daughter cell are <b>not identical</b> / no longer identical / different from one another.	1
		(E)	(Offspring produced will ideally have a highliched and a five sity.) Important because environmental control and are not a five sity.) Important because environmental control and a range of different environmental control and size and exceptions of at least some offspring surviving to adulthood and six and offspring of their own.	к
3.	a)		Means that an organism has two of the same allele for a specific gene.	1
	(q		F f f f f f f f f f f f f f f f f f f f	ε





## INSPECTION COPY

Question	_ _			Answer	er		Marks
		Phenotype	Observed	Expected	$(O-E)^2$	$\frac{(O-E)^2}{E}$	4
		Tall, yellow	74	63	$(11)^2 = 121$	$\frac{121}{63} = 1.921$	
		Tall, white	10	21	= (-,-)	$\frac{121}{21} = 5.762$	
		Short, yellow	13		$(-8)^2 = 64$	$\frac{64}{21} = 3.048$	
		Short while	\\ \\ \\ \\ \\ \\ \\ \\ \\ \\ \\ \\ \\	7	$(8)^2 = 64$	$\frac{64}{7} = 9.143$	
		×	(2 = 1.921 + 1.	5.762 + 3.04	$X^2 = 1.921 + 5.762 + 3.048 + 9.143 = 19.9 (3 sf)$	3 sf)	
	<u>:-</u>	They would have looked at a critical values table.	looked at a cr	ritical values t	able. 🗸		2
		If chi-squared value is greater than the critical of freedom), it is likely the genes are linked. •	ue is greater t likely the gen	than the critic es are linked.	If chi-squared value is greater than the critical value for p<0.05 (with three degrees of freedom), it is likely the genes are linked. $\checkmark$	with three degrees	
	(ii	(The researchers	want to find 1	the gene caus	(The researchers want to find the gene causing the trait so that they can study it.)	they can study it.)	2
		The chi-squared t	test suggests i	the gene is or	The chi-squared test suggests the gene is on the same chromogome as the stem-	ome as the stem-	
		Means the resear	rchers can naı	rrow down th	Means the researchers can narrow down their starciests, greene a smaller amount of DNA. ✓	e a smaller amount	
		(Hardy\Moinherg	T GOTTO T		(Hardy—Weinhard activation of the dominant allela's	minant allala's	_
		frequency and q is the second allele's frequency.	اع دو حراراً الا	allele's freq	uency. ✓		-
		Al Tog ec. 3311	ve, so it is rep	resented by q	ecasive, so it is represented by q in the equation.		
			1				
		$q = \sqrt{0.07} = 0.2$	= 0.265 ×				
		(With only two alleles, $p+q=1$ )	leles, $p+d=$	= 1)			
		p = 1 - q = 0.735	35				
		$\begin{vmatrix} p^2 = 0.541  \checkmark \\ 2pq = 1 - 0.07  - \end{aligned}$	-0.541 = 0.	= 0.39 (2 sf)  or  39 %	/ %6		
	<u>:-</u>	Codomínance					1
				]			

## INSPECTION COPY

Mark	4	9
Answer	FRIN FRENN FWENN FWEN FWE	A clear, thorough explanation of the processes of insertion, transformation and bacterial growth, which considers multiple aspects of the process.  A well-constructed and detailed step-by-step explanation in minology is used consistently and correctly.  Level 2 (3-4 marks)  A considerable effort the process is generally bacterial growther with any part of the process may be lacking in detail bacterial growther with any but one part of the process may be lacking in detail and the process in mostly consistent and correct.  Level 1 (1-2 marks)  Some effort to explain the processes involved in genetic engineering.  The explanation is incomplete and includes some incorrect information and terminology. Only some parts of the process are explained. The process may not be explained in a methodical way.  O marks  No response or no relevant interval interval.
_ ء	(::	
Question		
		5.

## **APPENDIX 1 – Fundamentals of Genetic**

### **Timeline of Discoveries**

1854 – Gregor Mendel begins work on plant hybridisation.

1859 – Charles Darwin publishes *On the Cig 5 c* Species, unaware of Mendel's experied in

1865–1866 – Mendel pulsus Spenetic findings, and speaks New York Society meeting.

1910 – The Hunt Morgan identifies the first known generic trait linked to the X chromosome.

1951 – Rosalind Franklin uses X-ray crystallography to produce images of DNA.

1953 – Francis Crick

1977 – Frederick San DNA sequencing teck

1985 – the Cetus Corpolymerase chain reac

1986–1987 – DNA p

1990–2003 – The Hu

## The Language of Genetics

Genes are sections of DNA which usually code for the production of a single process part (or all) of a protein. They are found on much larger structures can have 46 of these in each body cell. Because we inherit a set of 23 chromosome copies of each gene in our cells. These two copies may be exactly the same, or different versions of the same gene alleles. If an allele is expressed, this means the anatomy, physiology or behaviour of an organism.

## Talking about the

**Dominant alleles** – are as a long as the individual has at least one copy as a low sown using an upper-case letter.

For example gene in rabbits has two alleles, A (causes long ears) and a (causes short ears), rabbits with the alleles A and A, or A and a, will have long ears.

Recessive alleles – are only expressed if the individual has two copies of the allele. Shown using a lowercase letter. To continue the example above, only rabbits with the alleles a and a will have short ears.

**Genotype** – the specific alleles that an organism has for one or more genes.

**Homozygous** – an organism with two of the same allele for a specific, studied gene (e.g. AA or aa).

Heterozygous – an organism with two differer will be or the same gene (e.g. Aa).

Phenotype – an organien in Servable characteristics), caused by a mixture of gradual stransforment. For example, human height is affected gradual stransforment, but reaching full height requires a good diet too.

**Genome** – an organism's full set of genetic material, including all of its genes as well as the non-coding DNA between them.

# 



## **Sex Chromosomes**

Humans usually have 46 chromosomes, in 23 pairs. 22 of these pairs contain the females. However, the sex chromosomes vary between males and females. Females have one X chromosome and one Y chromosome. Although the two changes, there are also some genes which are found only on the X chromosome on the Y chromosome).

## Mendel and Monohybrid ( o . es

Gregor Mendel (1822-1855) are abbot of a small monastery in Brno, which was are are Austrian Empire (but is now in the Czech Republic). We are Austrian Empire (but is now in the Czech Republic). We are Austrian Empire (but is now in the Czech Republic). However, Mendel received a good education, had access to a wide range of books and was supported by the liberal-minded monastery to teach science courses at the city's university. He also had the advantages of space and enough free time to carry out his research.

Mendel experimented using pea plants, which turned out to be a very good idea. Peas have lots of visible characteristics which are controlled by pairs of simple dominant/recessive alleles. This means that cross-breeding (or crossing) two pea plants for these characteristics will produce simple ratios of offspring, and it's easy to compare predictions to actual results. When two plants are crossed to study how a single gene is inherited and its effects on the offspring, we call organ monohybrid cross.

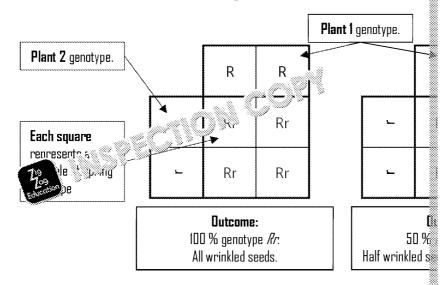
## Example

We can stry and the crosses using one of Mendel's own experiments.

There is a good in pea plants which controls seed shape.

- The dominant allele, R, causes wrinkled seeds.
- The recessive allele, r, causes round seeds.

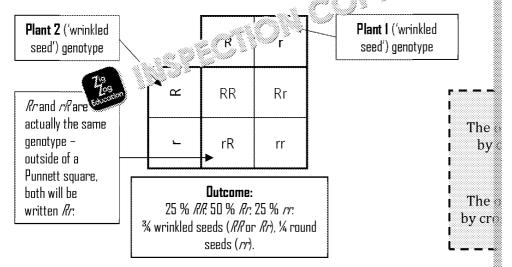
Because the r allele is recessive, only plants with the genotype rr will have round wrinkled seeds could have the genotypes RR or Rr. To find out the genotype (Plant 1), we can cross it with a round-seeded plant (Plant 2).





The **Punnett squares** above show the possible offspring, depending on the gend the outcome of the cross depends on what genotype the wrinkled-seeded plant as the  $F_1$  generation) have wrinkled seeds, we know that Plant 1 must have the

So let's say that all of the  $F_1$  generation have wrinkled seeds. We can see from previous page that all the offspring will have the genotype  $R_r$ . What if we crosproduce a new generation of offspring (the  $F_2$  generation)



Two heterozygous, wrinkled-seeded pea plants, which have the same genotype crossed. This has resulted in three different genotypes, and two different phenomena.

We can also write these numbers as ratios:

- 25 % RR, 50 % Rr, 25 % rr means that on we be for every one RR off there will be two with the genotype rr. The second s
- For every three plants a swankled seeds (RR or m), on average there we (m). The results in R : 1.



# 



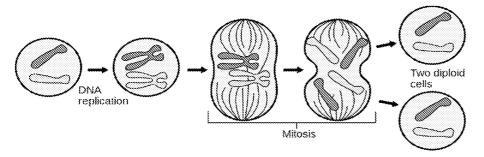
### **Mitosis and Meiosis**

Mitosis and meiosis are both processes of cell division, where a cell copies its genetic material and then splits into two or more daughter cells. Each of the new cells will have some of the original cell's organelles, and some of its cytoplasm.

Mitosis happens in body cells, so that the body calls

- grow
- b) repair itself

The body cells which will go mitosis start out with two we can write this as 2n). copies of e.

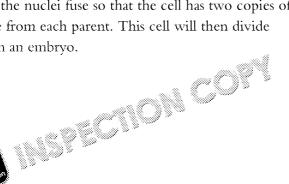


- 1. Before mitosis, the DNA is replicated, so there are few versions of each g
- During mitosis, the chromosomes are senar red r. arawn to each end of 2.
- The cell then divides by pinch is initially middle, this is called cytokinesis. 3.
- The genetic material and evenly between the two daughter cells. This is 4. ve pies of the genetic material (2n), like most of our box

during the production of haploid gametes, such Meiosis hap as sperm and egg cells in mammals. During meiosis, two divisions occur to produce four daughter cells, which are:

- genetically different from each other
- only have one copy of the genetic material. This means they have half the normal number of chromosomes (n).

During sexual reproduction in humans, the sperm cell fertilises the egg cell, and the nuclei fuse so that the cell has two copies of every gene – one from each parent. This cell will then divide and grow to form an embryo.





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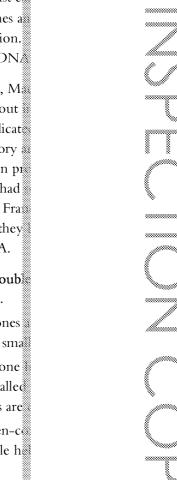
A cell

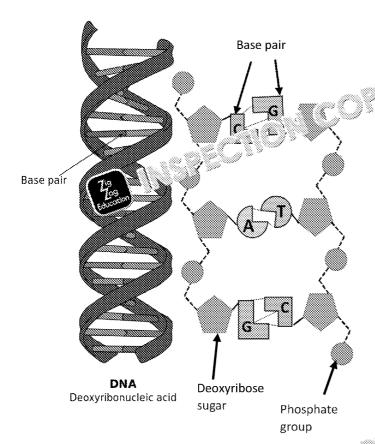
gen

A ce

amo: theref

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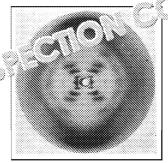




**Top left:** helical structure of a DNA molecule.

Top right: backbones, bases 7 pt pt

**Bottom right:** Rosalind Franklin's 'photo 51'.



In 1944, a scientist can DNA carries genes as genetic information.

Seven years later, Macould be drawn out fibres, which indicate the same laboratory Rosalind Franklin processed DNA had Finally, in 1953, Franch announced that they structure of DNA.

- DNA is a doub 'backbones'.
- The backbones pattern of a small
- Each backbone molecules called and its bases are of the double hapairs.
- However, they adenine (A) always
   guanine (G) always
- This means the always has A wl
- DNA is built free each nucleotide and one of the fee

## **Mutations and Genetic Disorders**

When DNA is copied, it usually produces two molecules fDNA, each with epairs as the original. However, sometimes sometimes sometimes one during the will change. Sometimes bases are added an are sometimes one base is swap mutation. If mutations happen from merosis, they can be inherited by the organizations affect the proteins which are produces two molecules fDNA, each with epairs as the original produces two molecules fDNA, each with epairs as the original. However, sometimes sometimes one base is swap mutation. If mutations happen for a produce fDNA, each with epairs as the original. However, sometimes sometimes one base is swap mutation. If mutations happen for a produce for a produce fDNA, each with epairs as the original.

Many muta ave no effect at all, and some have a positive effect on an orgodynamic of their will be harmful, and may cause diseases. If a disease is caused by having known as a **genetic condition**. Some genetic conditions, such as cystic fibrosis, others, such as Huntington's chorea, are caused by dominant alleles.



## Pop Quiz

You can use this guiz to test how solid your knowledge of GCSE genetics is. I content well, you'll be able to understand the A Level course much better, and

Draw lines to match the terms to their definitions. (Two of the terms have

Monohybrid cross

Heterozygous



Homozygous

Allele

Genetic cross

Chromosomes

3.

Read til

Favi (2) So different versions of

One of two or more versions of

Breeding two individuals to stud single gene.

Large DNA structures which com

A version of a gene which is onl present.

Fill in the gaps in the sentences below.

A human skin cell has 46 \_\_\_\_\_\_, while an agg cell only has \_\_ If an individual which is homozygous dominated fit gene is crossed with homozygous recessive, all their offs will will be scale ared the basic rules of inheritance, using

plants to carry cut a tigations.

A gene for flower colour in pea plants has two alleles, B and b. The domin flowers, while the recessive allele causes white flowers. Plant 1 is heterozy Plant 2 is homozygous recessive. The two plants are bred to produce offsp

ssage below, and answer the questions which follow.

- What is the genotype of Plant 1?
- b) What is the phenotype of Plant 2?
- Will all of the offspring have purple flowers? Explain your answer.
- Explain how the number of copies of the genetic material in a cell change
  - before and during meiosis
  - b) during fertilisation

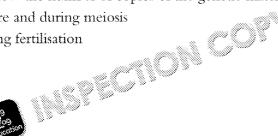


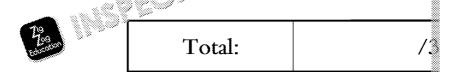




Figure 2: Forke

ਜett square to show the possible offspring genotypes whei

- (A the crossed.
- **b)** State the proportion of phenotypes expected in the F<sub>1</sub> generation. Gi as percentages, and as a ratio.
- 6. What is the difference between a genotype and a phenotype? Give examp
- 7. A DNA strand has the sequence of base pairs: ACCTGA. What sequence strand have in the same location?
- 8. How are the sex chromosomes different from the other 22 pairs of chrom
- 9. Briefly, explain how an accidental change in the genetic material of an organized genetic disorder.
- 10. Describe the structure of DNA

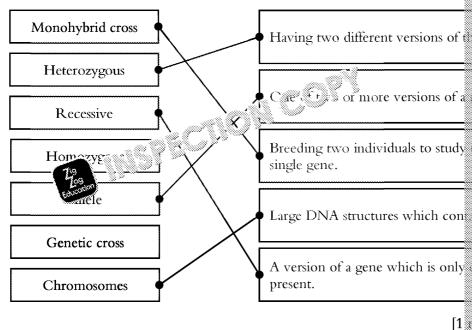






## Pop Quiz - Answers

1.



- 2. chromosomes; 23; heterozygous; Mendel; pea.
- 3. a) Bb [1]
  - b) white flowers [1]
  - c) No; half of the offspring will inherit a flowers [1], a flowers [1].
- 4. a) Before the Aris replicated, so the cell has four copies of each two aring meiosis, producing four haploid (n) cells with only one
  - b) Few ation happens when two haploid (n) gametes meet and fuse. Each gene [1], and so the zygote (fertilised egg) has two copies of each
- 5. a)

	М	m
Σ	ММ	Mm
ш	mM	mm

k for correct axes; 1 mark for co

b) 75 19 un; 25 % forked fin [1]. This is a ratio of 3:1 [1].

# 



## 6. Many examples are possible. The key is that genotypes are the specific ger phenotypes are outward appearances based on how genes interact with the correct definitions of genotype and phenotype; 1 mark each for correct examples are possible.

## Example mark scheme:

A genotype is the specific alleles which an individual has for a gene [1]. For stem height, a plant may have the genotype Tt (in this case, the dominant)

The plant's phenotype is its actual height beginn both its genes and its esthe soil that the plant is grown in the soil that the plant is grown in the plant's grown as tall. [1]

- 7. TGGA Tog
- 8. The sex chromosomes, X and Y, have different genes (although they do a common). [1] Females have two X chromosomes, while males have one X chromosome. [1]
- 9. A mutation may occur in which bases are inserted, deleted or swapped for happens, the DNA code will read differently, and the protein produced fre different in shape (sometimes radically different). [1] The protein may have function altogether, or acquire an unexpected function. [1] This can cause was important, or if the new protein disrupts some other process in the box
- 10. Your answer could include the following points:

11 A fc each correct point

- Two complementary strands, which are intended together by bonds bet
- Each strand has an alternation gar-phosphate backbone
- Nitrogen-car gasses (A, C, G, T) which 'stick out' from each l
- E Go e pairs with one specific other base A joins to T, C joins to commentary base pairing.
- Results in a double helix shape
- 'Sides' of the ladder are sugar phosphate backbones, and 'rungs' of the
- Assembled from individual units which are called nucleotides



# 

