



Rewarding Learning
ADVANCED
 General Certificate of Education

Centre Number

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Candidate Number

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Life and Health Sciences

Assessment Unit A2 5

assessing

Genetics, Stem Cell Research and Cloning

MV18

[AZ051]

Assessment

Time

1 hour 45 minutes, plus your additional time allowance.

Assessment Level of Control **Tick the relevant box (✓)**

Controlled Conditions	
Other	

Instructions to Candidates

Write your Centre Number and Candidate Number in the spaces provided at the top of this page.
 Write your answers in the spaces provided in this question paper.
 Answer **all nine** questions.

Information for Candidates

The total mark for this paper is **100**.
 Figures in brackets printed at the end of each question indicate the marks awarded to each question or part question.
 You may use an electronic calculator.
 Quality of written communication will be assessed in question **7(a)**.

1 The table below contains statements about DNA, mRNA and tRNA.

(a) Complete the table by writing a tick (✓) if the statement is correct, or a cross (X) if the statement is incorrect.
[3 marks]

Statement	DNA	mRNA	tRNA
Nucleotides contain ribose			
Long single strand			
Nucleotides contain the base thymine			

DNA carries information and its sequence of bases determines the structure of proteins.

(b) Describe and explain how the sequence of bases results in the formation of a protein.
[3 marks]

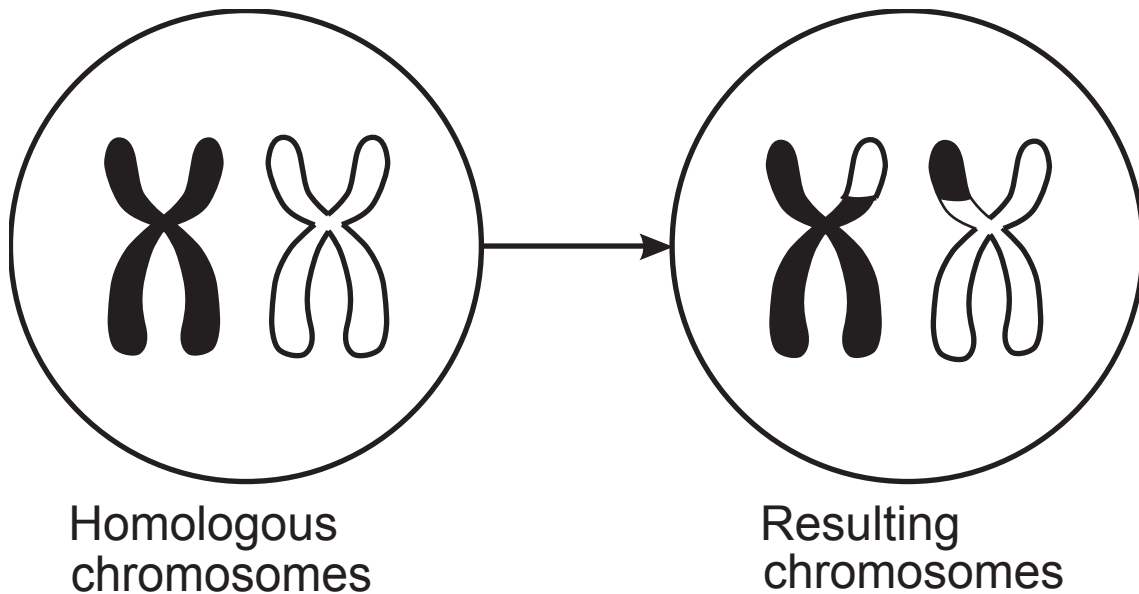
(c) During DNA replication a change in base sequence can occur.

(i) Give the term used to describe this change in base sequence. [1 mark]

(ii) Describe and explain the consequence of a change in base sequence during DNA replication.
[2 marks]

2 Meiosis creates cells that are genetically different.

The diagram below shows homologous chromosomes in a cell. It also shows the resulting chromosomes in a cell when one stage of the process of meiosis has occurred.



- (a) (i) Describe what has occurred to produce the resulting chromosomes. [1 mark]

- (ii) Explain the result of this process shown in the diagram on each of the chromosomes. [1 mark]

(b) Describe and explain **one other** process in meiosis that results in genetically different cells. [2 marks]

- 3 The technique of genetic fingerprinting is based on the uniqueness of each person's genetic make-up.

Enzymes are used to cut DNA at specific recognition sites to create different sized fragments.

- (a) Name the type of enzyme used to cut DNA into fragments. [1 mark]
-

- (b) The enzyme **Hae**111 cuts DNA **only** at the recognition site GGCC with the cut then taking place between adjacent G and C bases or C and G bases.

The cut takes place right across the DNA strand. A DNA strand is cut with enzyme **Hae**111.

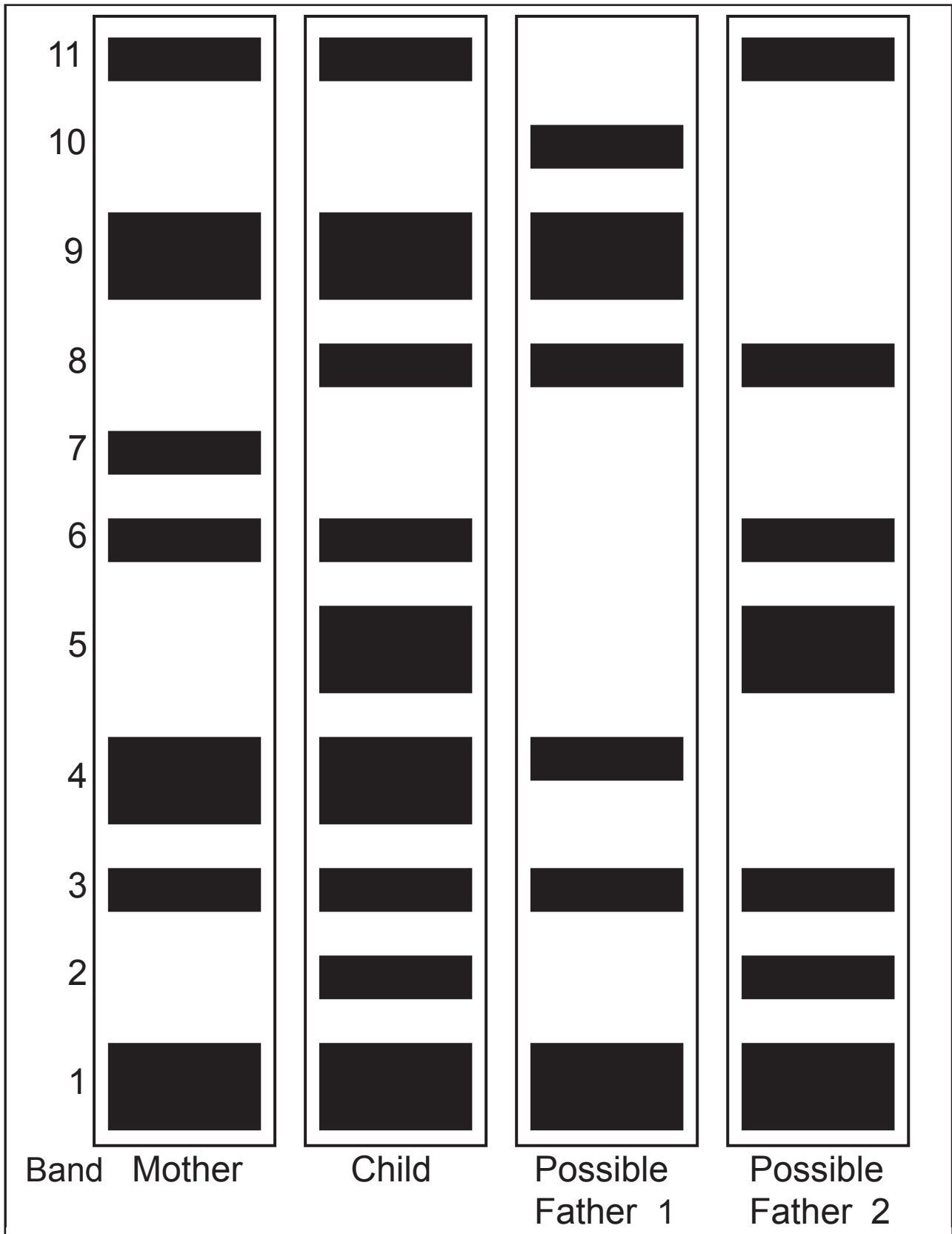
- (i) Show the positions of the cuts in the DNA strand with thick black lines on the diagram below. [1 mark]



- (ii) Give the total number of DNA fragments produced. [1 mark]
-

(c) The process of gel electrophoresis produces a unique set of DNA bands for each individual.

The genetic fingerprints below show the bands obtained from a child, the child's mother and two men, one of whom may be the child's father.



(i) Describe and explain how the process of gel electrophoresis separates the different fragments of a person's DNA. [2 marks]

(ii) Give the number of DNA bands the child and mother have in common. [1 mark]

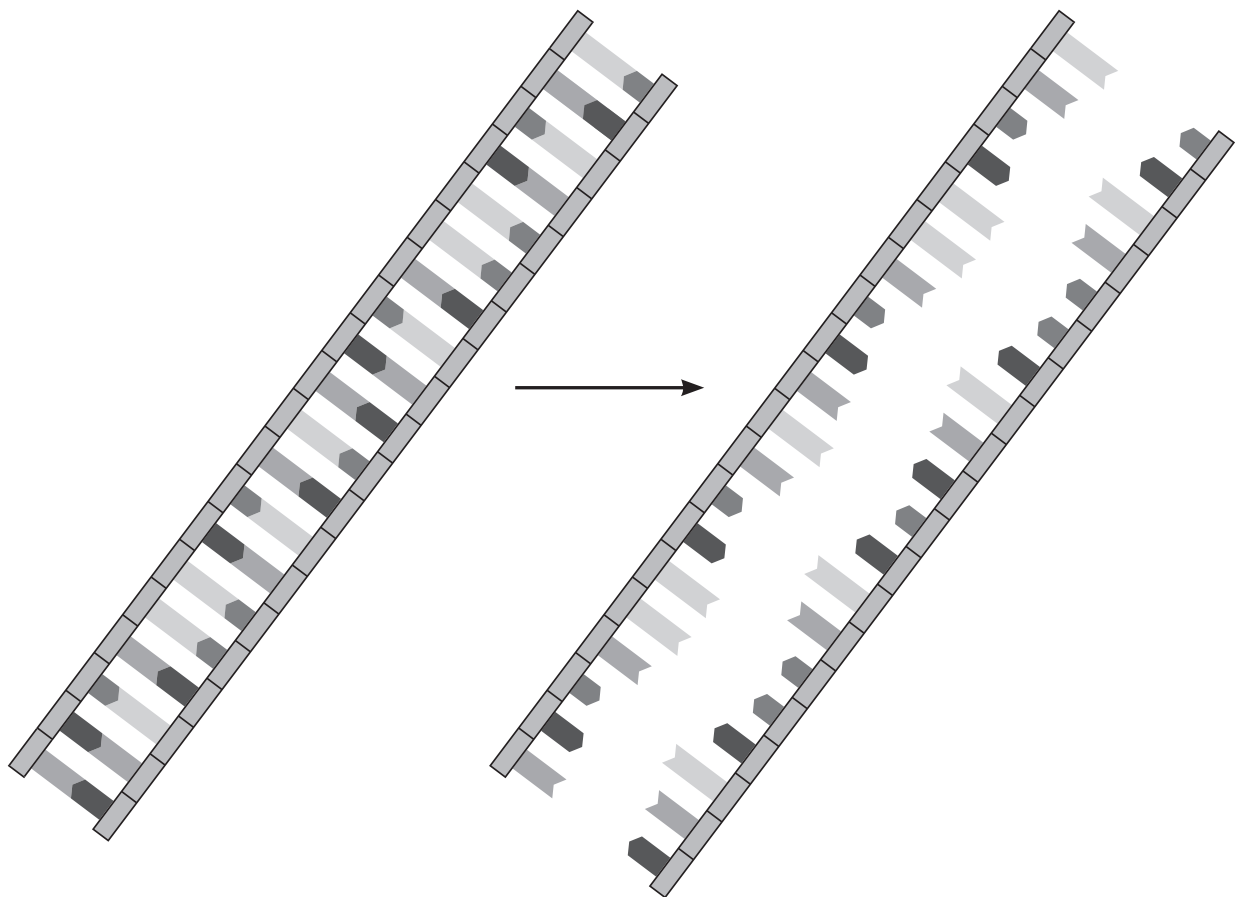
(iii) Use the genetic fingerprints to identify who is the father of the child. [1 mark]

(iv) Explain your answer using the DNA band numbers shown. In your answer refer to the genetic fingerprints of the mother, child and both possible fathers. [3 marks]

- 4 The polymerase chain reaction (PCR) enables many copies of DNA to be made from a small sample.

The technique of PCR is a modified version of the process of DNA replication.

- (a) The diagram below illustrates the initial stage of PCR. A section of DNA is heated to 95°C. This breaks the hydrogen bonds holding the two strands together.



- (i) Give the name of the enzyme that breaks the hydrogen bonds holding the two strands together in DNA replication. [1 mark]

(ii) Suggest why it is important that hydrogen bonds are broken between the two strands. [1 mark]

(iii) Using your knowledge of DNA replication, suggest **two** substances that should be added to complete the copying of DNA in PCR. [2 marks]

1. _____

2. _____

Scientists carried out PCR on a molecule of DNA.

At the end of the first cycle, there are **two** molecules of DNA.

(iv) How many molecules of DNA will there be at the end of **six** cycles? [1 mark]

(b) PCR can be used to produce DNA probes that can screen patients for clinically important genes.

(i) Outline how a DNA probe is used to locate a specific DNA fragment. [2 marks]

(ii) Give **one** way that the result of screening with a DNA probe can be used in genetic counselling. [1 mark]

- 5 (a) There are four blood groups in the ABO system. Individuals can be blood group A, B, AB or O. Blood group is an example of the inheritance of multiple alleles.

The ABO blood group system is controlled by three alleles I^A , I^B and I^O .

I^A allele is co-dominant with allele I^B .

I^A and I^B alleles are dominant to I^O allele.

- (i) Complete the table below to show the genotypes of blood groups in humans.

Some blood groups may have more than one genotype. [3 marks]

Blood group	Possible genotypes
A	$I^A I^A$ and $I^A I^O$
B	
AB	$I^A I^B$
O	

(ii) Explain the terms dominant and co-dominant in terms of a person's blood group. [2 marks]

(b) A woman who is heterozygous for blood group A has a child with a man whose blood group is AB.

(i) Determine the probability of the woman and man having a child with blood group B.

Show your working in a Punnett square in the space below. [4 marks]

Probability of having a child with blood group B _____

(ii) This woman and this man could not have a child with type O blood group.

Explain why. [2 marks]

6 A farmer keeps two different herds of cattle.

The cattle in herd 1 produce a lower quality meat than the cattle in herd 2.

(a) The farmer wants to improve the quality of his meat produced from herd 1.

(i) Describe how the farmer can use traditional breeding to obtain a higher quality of meat from his cattle.
[3 marks]

(ii) Suggest **one** disadvantage of traditional breeding in cattle. [1 mark]

(b) Human genes have been introduced into farm animals, such as sheep, to breed animals that can produce useful human proteins.

(i) Give **one** advantage of this process. [1 mark]

(ii) Name **one** therapeutic chemical used to treat burns that is produced in this way. [1 mark]

(b) Gene technology is used to manufacture substances of medical importance such as insulin.

(i) Name the group of organisms used to produce insulin by genetic engineering. [1 mark]

(ii) Give **two** sources of insulin used to treat diabetes before the advent of genetic engineering. [2 marks]

1. _____

2. _____

(iii) Give **two** disadvantages of using insulin from these sources. [2 marks]

1. _____

2. _____

- 8 (a) In guinea pigs, dark fur colour is dominant (**D**) and light fur colour (**d**) is recessive.

Rough coat texture (**R**) is dominant, while smooth coat texture (**r**) is recessive.

The genes for these characteristics are located on separate chromosomes and are independently inherited.

They are not sex-linked.

- (i) State the **genotype** of a guinea pig, homozygous for dark fur and heterozygous for coat texture.

[1 mark]

- (ii) Give the **phenotype** of a guinea pig with the genotype **Ddrr**. [1 mark]

- (iii) Define the term 'phenotype'. [1 mark]

(b) Two guinea pigs of the following genotypes were crossed.

DdRr x ddRr

(i) Complete the Punnett square to show the parental gametes and the genotypes of the offspring.
[4 marks]

(ii) Give the numbers of each of the phenotypes produced in this Punnett square. [2 marks]

dark fur, rough coat _____

dark fur, smooth coat _____

light fur, rough coat _____

light fur, smooth coat _____

(c) The number of offspring produced from several **other** breeding pairs of guinea pigs were recorded.

- dark fur, rough coat 21
- dark fur, smooth coat 27
- light fur, rough coat 24
- light fur, smooth coat 28

A total of 100 guinea pigs were recorded.

The chi-squared test can be used to check if these results statistically fit an expected ratio of **1:1:1:1**.

The formula for the chi-squared test is:

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

Where O = observed frequency
 E = expected frequency
 Σ = sum of

- (i) Complete the table below and then calculate the χ^2 value for these results.

Category	Observed (O)	Expected (E)	(O-E)	(O-E) ²	$\frac{(O-E)^2}{E}$
dark fur, rough coat	21				
dark fur, smooth coat	27				
light fur, rough coat	24				
light fur, smooth coat	28				

Calculated χ^2 value _____ [5 marks]

The table below gives probability values for use in χ^2 test with $n-1$ degrees of freedom (d.f.) where n is the number of categories.

χ^2 values

d.f.	probability = 0.900	0.500	0.100	0.050	0.010	0.001
1	0.016	0.455	2.71	3.84	6.63	10.83
2	0.211	1.39	4.61	5.99	9.21	13.82
3	0.584	2.37	6.25	7.81	11.34	16.27
4	1.06	3.36	7.78	9.49	13.28	18.47
5	1.61	4.35	9.24	11.07	15.09	20.52
6	2.20	5.35	10.64	12.59	16.81	22.46
7	2.83	6.35	12.02	14.07	18.48	24.32
8	3.49	7.34	13.36	15.51	20.09	26.13
9	4.17	8.34	14.68	16.92	21.67	27.88

(ii) Between what **range of probabilities** does your χ^2 value fit? [1 mark]

Use the table **above** to assist you.

between _____ and _____

(iii) Do the results statistically fit the expected ratio of **1:1:1:1**?

Explain your answer. [2 marks]

(iv) Give the genotypes of the guinea pigs that would produce a ratio of **1:1:1:1**. [2 marks]

_____ and _____

- 9 (a) Cystic fibrosis is caused by a mutation of the gene producing the protein CFTR.

This protein consists of 1480 amino acids.

- (i) Calculate the number of bases required to code for the amino acids in the CFTR protein. [2 marks]

You are advised to show your working.

_____ bases

The digestive system is one of the body systems affected by cystic fibrosis.

Cystic fibrosis also affects the lungs.

- (ii) Give **two** digestive system symptoms caused by cystic fibrosis. [2 marks]

1. _____

2. _____

One method for treating patients with cystic fibrosis involves the transfer of healthy CFTR genes into epithelial cells in their lungs.

(iii) Identify **two** vectors for introducing healthy CFTR genes into patients. [2 marks]

Vector 1 _____

Vector 2 _____ [2]

(iv) Describe how these vectors might be delivered into the lungs of the patient. [1 mark]

Patients who have been treated by this method do not pass on the healthy CFTR gene to their children.

(v) Explain why. [2 marks]

(b) Scientists investigated the effect of two drugs, Lumacaftor and Ivacaftor, in patients with cystic fibrosis.

The most common CFTR mutation is Phe508del.

45% of patients with cystic fibrosis are homozygous for this allele.

(i) Using the example of Phe508del, explain the term homozygous. [1 mark]

The drug Ivacaftor increases the chances of opening CFTR channels in the lung epithelial cells.

When it is used in combination with Lumacaftor it increases chloride transport in these cells.

(ii) Using the information provided and your knowledge, suggest how the use of both drugs reduces the symptoms of cystic fibrosis in the lungs caused by the presence of a thick sticky mucous. [3 marks]

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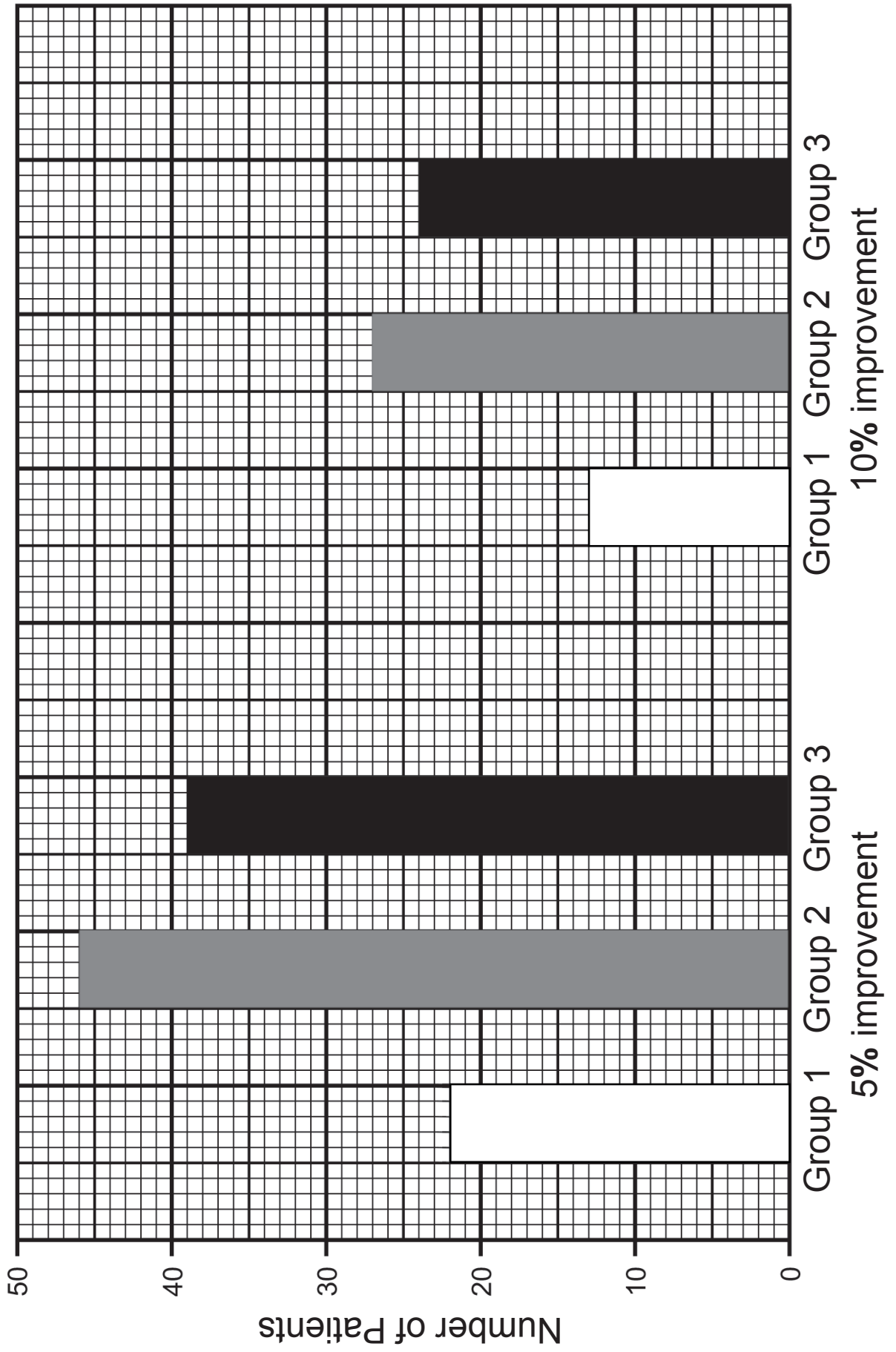
(c) Scientists investigated the effects of Ivacaftor and Lumacaftor on the forced expiratory volume in one second (FEV_1) of cystic fibrosis patients.

They recorded the FEV_1 at the start of the investigation and at 24 weeks when the investigation ended.

Patients were divided into 3 groups:

- Group 1: Placebo group (did **not** receive Lumacaftor or Ivacaftor)
- Group 2: Received 600 mg of Lumacaftor once daily in combination with 250 mg of Ivacaftor every 12 hours.
- Group 3: Received 400 mg of Lumacaftor every 12 hours in combination with 250 mg of Ivacaftor every 12 hours.

The graph opposite page shows the number of patients in each group with a 5% and 10% improvement in FEV_1 after the 24 week study.



(i) Using the information provided and the graph, state which group had the greatest increase in FEV₁ for the investigation. [1 mark]

(ii) Compare and analyse the outcomes for each group in the investigation. [4 marks]

(iii) Explain the purpose of **Group 1** in this study. [2 marks]

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SOURCE

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Q9(c)..... "From Supplement to: Wainwright CE, Elborn JS, Ramsey BW, et al. Lumacaftor–ivacaftor in patients with cystic fibrosis homozygous for Phe508del. Page No.14, Figure S5 Copyright © (2015) Massachusetts Medical Society. Reprinted with permission from Massachusetts Medical Society."

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