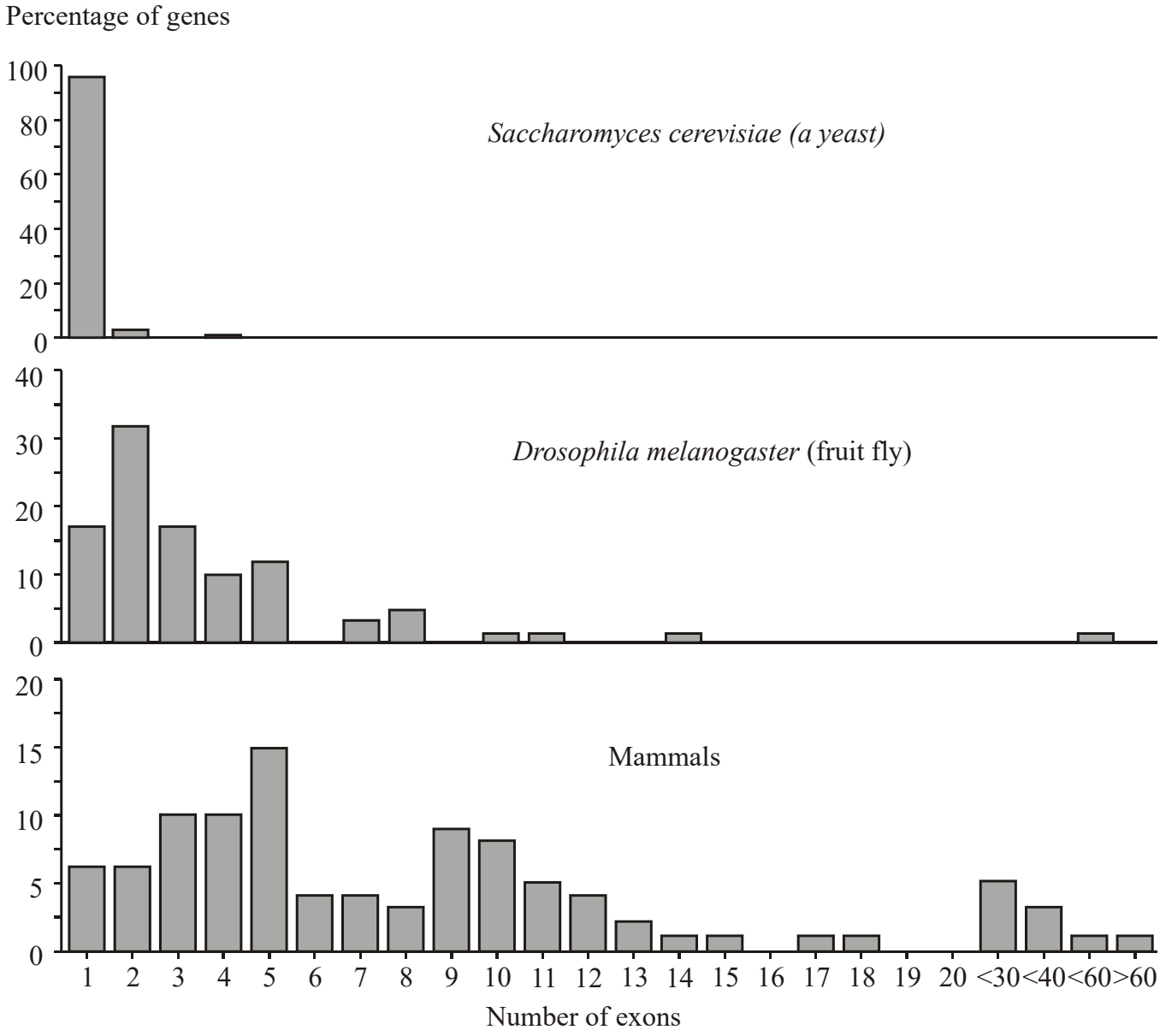
Eukaryotic Genes (17 marks)

It had always been assumed that eukaryotic genes were similar in organization to prokaryotic genes. However, modern techniques of molecular analysis indicated that there are additional DNA sequences that lie within the coding region of genes. Exons are the DNA sequences that code for proteins while introns are the intervening sequences that have to be removed. The graph shows the number of exons found in genes for three different groups of eukaryotes.



a. Calculate the percentage of genes that have five or less exons in mammals. *(1 mark)*

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b. Describe the distribution of the number of exons and percentage of genes in *D. melanogaster*. *(2 marks)*

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c. Compare distributions of the number of exons found in genes of *S. cerevisiae* and mammals. *(2 marks)*

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d. Suggest one reason for the differences in the numbers of exons found in genes of *S. cerevisiae* and mammals. *(1 mark)*

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Human DNA was analysed and details of certain genes are shown in the table below. (kb = kilobase pairs)

Gene	**Gene size / kb***	**mRNA size / kb**	**Number of introns**
Insulin	1.7	0.4	2
Collagen	38.0	5.0	50
Albumin	25.0	2.1	14
Phenylalanine hydroxylase	90.0	2.4	12
Dystrophin	2 000.0	17.0	50

e. Calculate the average size of the introns for the albumin gene. *(2 marks)*

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f. Analyse the relationship between gene size and the number of introns. *(2 marks)*

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g. Determine the maximum number of amino acids that could be produced by translating the phenylalanine hydroxylase mRNA. *(1 mark)*

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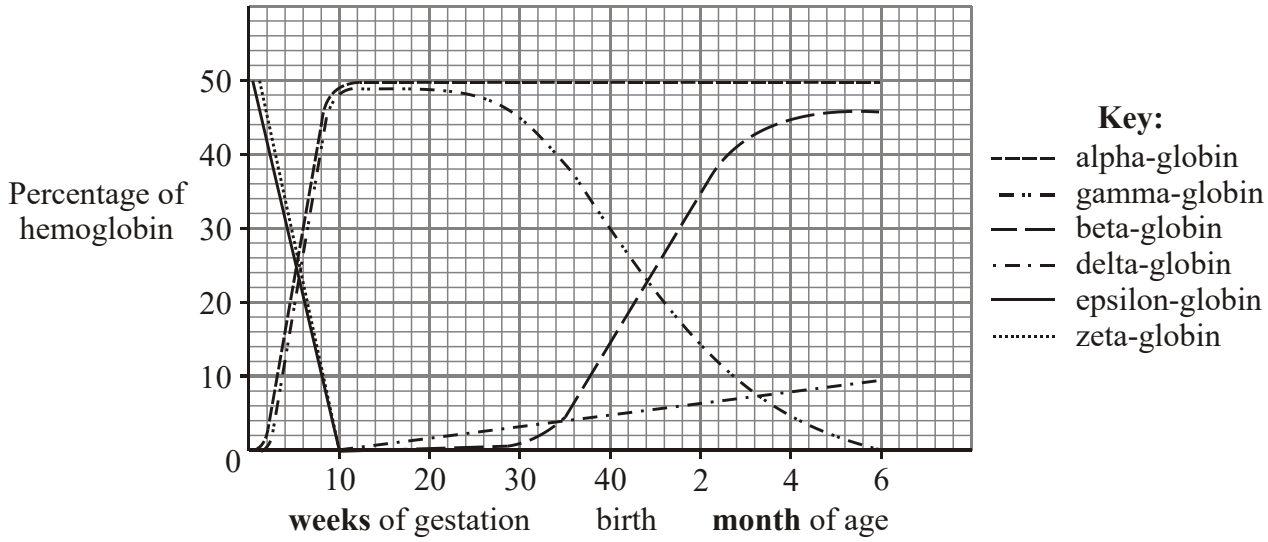
Hemoglobin is a protein composed of two pairs of globin molecules. During the process of development from conception to adulthood, human hemoglobin changes in composition. Adult hemoglobin consists of two alpha- and two beta-globin molecules. Two globin genes occur on chromosome 16: alpha- and zeta-globin. Four other globin genes are found on chromosome 11: beta, delta, epsilon and gamma. The graph below illustrates the changes in expression of the globin genes over time.



h. State which globin genes are the first to be expressed after fertilization. *(1 mark)*

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i. Compare the expression of the gamma-globin gene with the beta-globin gene. *(3 marks)*

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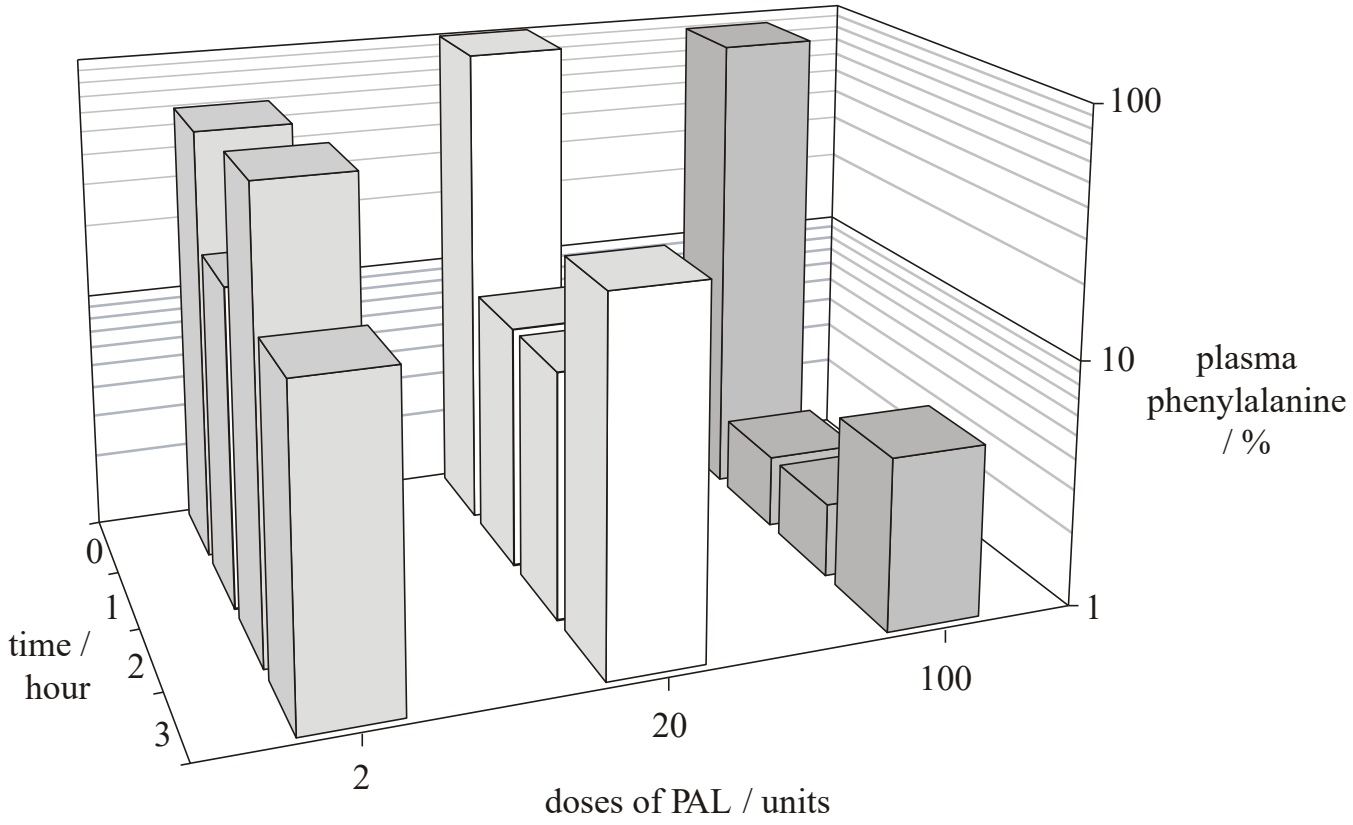
j. Deduce the composition of the hemoglobin molecules at different time periods. *(2 marks)*

10 weeks of gestation: .....

2 months after birth: .....

Phenylketonuria (7 marks)

Phenylketonuria (PKU) is a disease caused by a gene mutation that makes too much phenylalanine which may cause brain damage. The enzyme phenylalanine ammonia lyase (PAL), converts phenylalanine into harmless products. Mice with PKU were injected with PAL. The levels of phenylalanine in blood plasma were measured immediately after the injection (0 hour) and every hour for the next three hours. Different groups of mice with PKU were injected with three different doses of PAL. The results are shown below as a percentage of the levels of phenylalanine before the PAL injection.



a. Calculate the approximate percentage reduction in phenylalanine at 0 hour when the mice were injected with a dose of two units of PAL. *(1 mark)*

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b. Outline the effect of a dose of twenty units of PAL on phenylalanine levels. *(2 marks)*

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c. Discuss the effectiveness of the different doses of PAL to treat PKU mice. *(3 marks)*

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d. Outline how the type of mutation that causes PKU differs from Klinefelter's syndrome (XXY). *(1 mark)*

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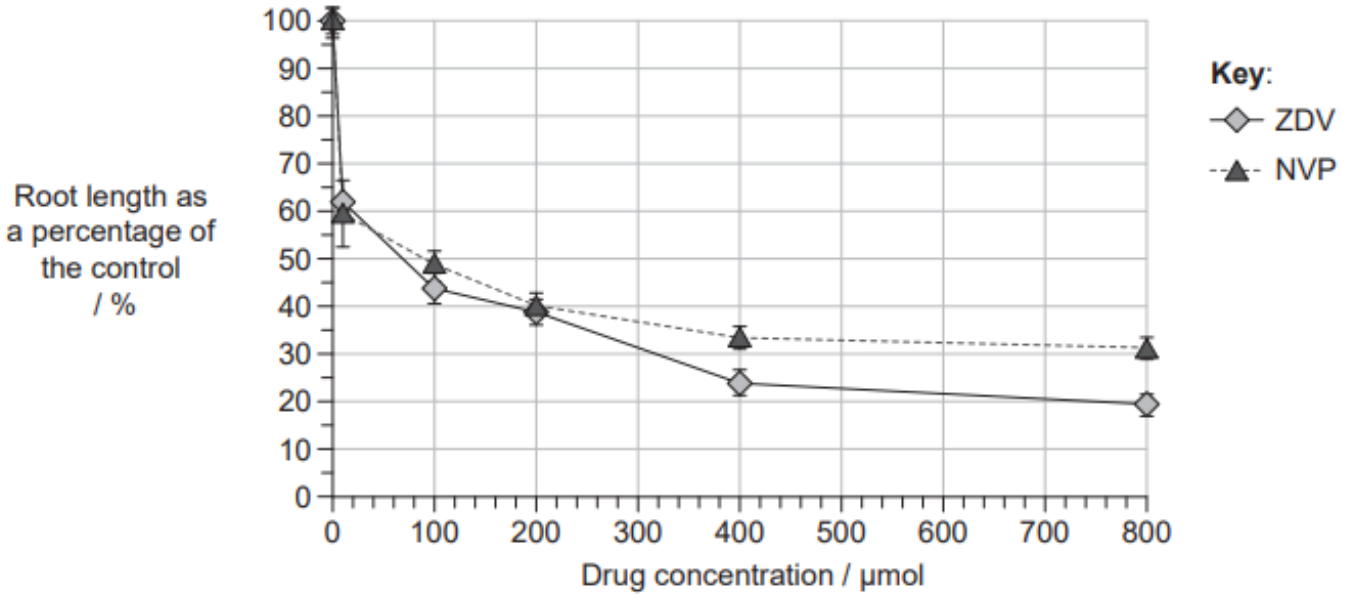
Mitosis (16 marks)

Antiretroviral drugs are used to treat Human Immunodeficiency Virus (HIV) infections. Zidovudine (ZDV) and nevirapine (NVP) are examples of antiretroviral drugs. There are concerns that these drugs may be toxic to body cells in mitosis. In a study using *Allium cepa*, root tips were exposed to the drugs for 96 hours at a range of concentrations. The control treatment was a drug concentration of 0µmol. In the graph, root lengths after the 96-hour treatment period are expressed as a percentage of the length of the control.



a. Deduce the concentration of ZDV that would cause a 50 % reduction in root growth compared to the control. *(1 mark)*

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b. Identify root length, as percentage of the control, resulting from ZDV concentration of 400 µmol. *(1 mark)*

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c. Compare and contrast the effect of ZDV and NVP on the growth of *Allium* roots. *(2 marks)*

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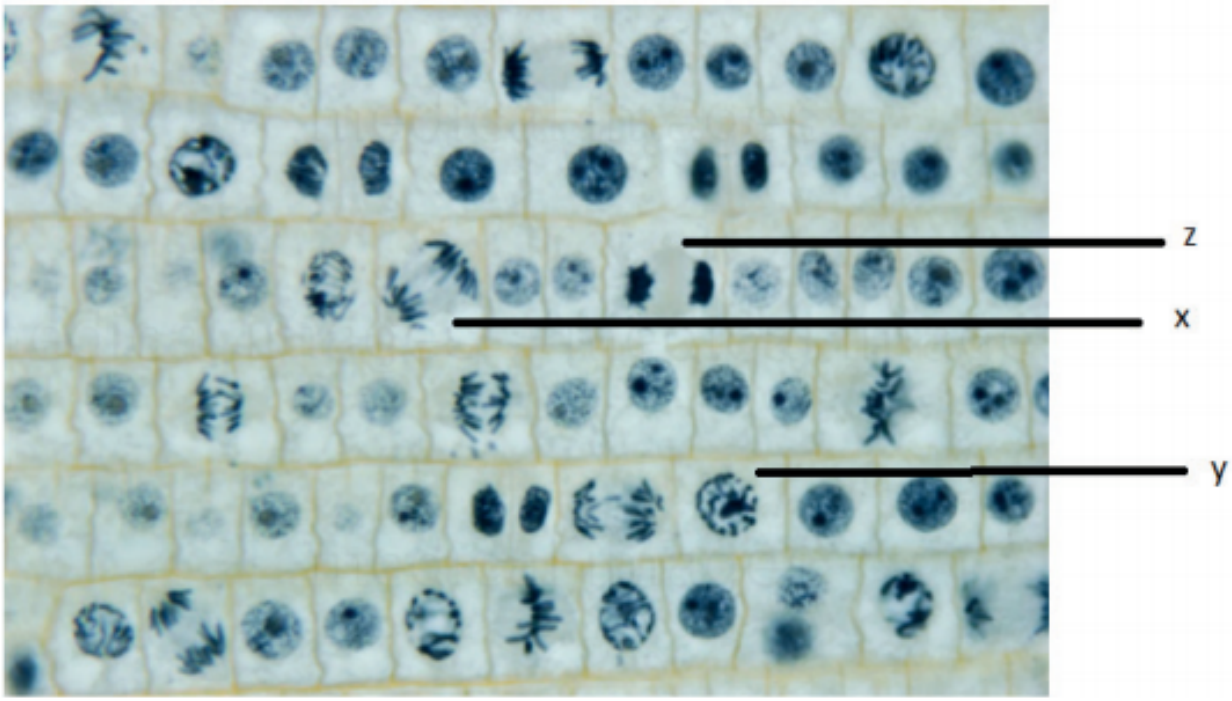
Both ZDV and NVP are believed to have a damaging effect on the process of mitosis but ZDV in particular is believed to block the formation of the spindle.



d. Based on the information, suggest with a reason which of the labelled cell types will become more common in *Allium* root tips treated with ZDV. *(2 marks)*

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e. *Allium* root tips continue to show some growth even at high concentrations of NVP. Suggest a possible reason for the growth seen in root tips with 800µmol NVP. *(1 mark)*

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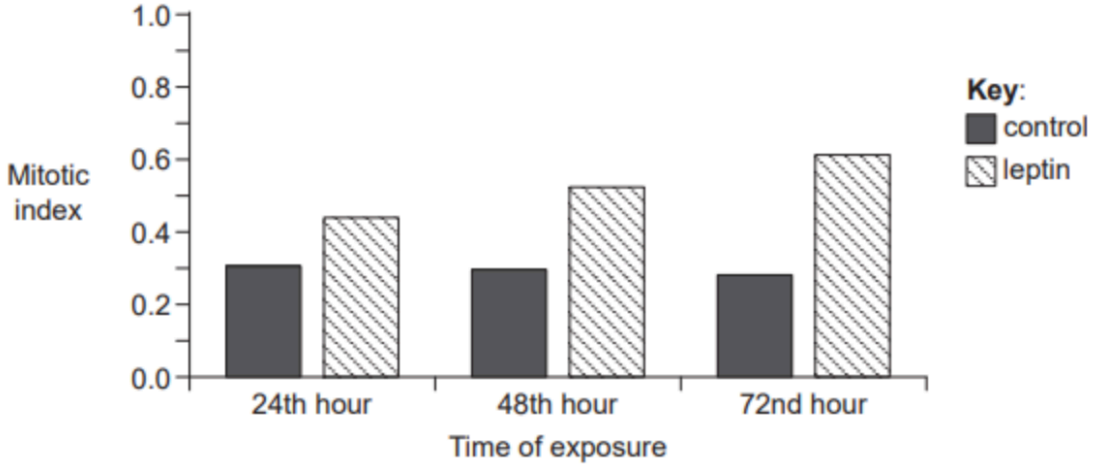
Mitosis plays an important role in tissue regeneration and can be an important factor in recovery from surgery. The hormone leptin has been shown to promote mitosis in certain circumstances. The bar chart shows the mitotic indices of liver tissue exposed to leptin and control tissue during 72 hours after surgery.



f. Deduce the change in mitotic index after 72 hours compared to the control. *(1 mark)*

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g. Based on the data, evaluate the evidence for leptin promoting regeneration of liver tissue. *(2 marks)*

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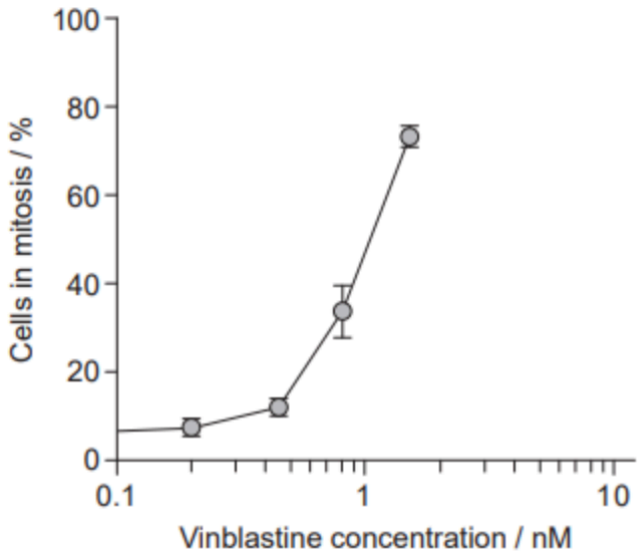
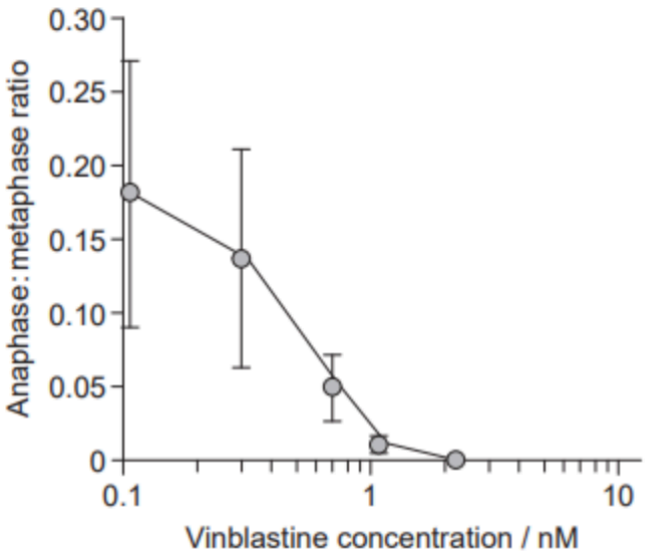
Anticancer drugs are used to prevent uncontrolled cell division. The mechanism of action of vinblastine, an anticancer drug, was investigated over a range of concentrations. Vinblastine is an alkaloid isolated from the periwinkle plant (*Catharansus roseus*). The percentage of cells in mitosis and ratio of anaphase to metaphase in cells exposed to this drug *in vitro* for a fixed time were recorded. Data was displayed in graphs.





h. By referring to both graphs, evaluate the hypothesis that vinblastine targets cells in mitosis and prevents them from completing the process. *(3 marks)*

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i. Some anticancer drugs inhibit mitosis by blocking the formation of the spindle. Suggest **one** other way in which vinblastine could block mitosis. *(1 mark)*

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j. Discuss **one** advantage and **one** disadvantage of using plant tissue to investigate drugs intended to treat cancer in humans. *(2 marks)*

Advantage:

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Disadvantage:

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Answers

1a *(1 mark)*
47–49% (*units are not needed*)

1b *(2 marks)*
a. *D. melanogaster* / *Drosophila* has few genes with one exon
b. highest percentage has 2 exons
c. most genes have 5 or fewer exons
d. a few genes have 10 or more exons / more than 8
e. maximum number of exons does not exceed 60

1c *(2 marks)*
a. *S. cerevisiae* / yeast has most genes with only 1 exon while mammals 5 exons is most frequent
b. no yeast genes have more than 5 exons while some mammal genes have greater than 60 exons
c. mammal genes contain more exons on average
d. with a wider distribution than yeast

1d *(1 mark)*
a. *S. cerevisiae* / yeast is a unicellular organism / mammals are multicellular / complex
b. mammals have more transcriptional regulation
c. *S. cerevisiae* smaller in size / more compact genome

1e *(2 marks)*
a. gene size – mRNA size = intron size / 25.0 – 2.1 = 22.9 kb
b. average size of intron = 1.6 (± 0.1) kb
(*unit required*)

1f *(2 marks)*
a. smaller genes usually have less introns / larger genes have more introns / relationship not clear
b. dystrophin and collagen have same number of introns but the dystrophin gene is larger
c. albumin has more introns but is smaller than the gene for phenylalanine hydroxylase

1g *(1 mark)*
2.4 kb (1 amino acid / 3 bases) = 800 amino acids ***or*** 799 amino acids

1h *(1 mark)*
epsilon and zeta (globin)

1i *(3 marks)*
a. gamma genes (mostly) expressed before birth and beta genes expressed after birth
b. beta-globin levels rise at 28(±2) weeks of gestation while gamma levels decrease / as one rises, the other falls
c. gamma-globin expression starts at 0–2 weeks whereas beta-globin starts at 26 (±2) weeks / gamma expression starts earlier
d. one month after birth hemoglobin has equal mixture of beta-globin and gamma-globin
e. gamma levels go to zero while beta becomes a regular part of hemoglobin

1j *(2 marks)*
a. *10 weeks after gestation:* two alpha-globins with two gamma-globins / 49 (50)% alpha and 48 (49)% gamma
b. *2 months after birth:* variety of molecules all containing alpha and two chains from the other three types / 6% delta, 14% gamma, 35% beta, 50% alpha

2a *(1 mark)*
(100% − 60% =) 40 (±4) *(units not required)*

2b *(2 marks)*

a. no reaction immediately after injection
b. decreases then remains steady / constant / plateaus / between 1 hour and 2 hours
c. rises again at 3 hours

2c *(3 marks)*

a. all doses reduce amount of phenylalanine / positive correlation
b. 100 units reduce it the most / 2 units reduce it the least
c. at 2 hours level remains lower except for the 2 units of PAL / rise at 2 hours for 2 units of PAL
d. 2 units of PAL will be the most appropriate as less enzyme needed / 2 units of PAL reacts the fastest (0 hour)
e. effect of 100 units lasts longer
f. no knowledge of possible side effects / more data on time / units / dosage required

2d *(1 mark)*

(PKU is a gene mutation while) Klinefelter's is a chromosome disorder

3a *(1 mark)*

75 µmol. *(Allow answers in the range of 70 µmol–80 µmol)*

3b (1 mark)

24%. *(Allow answers in the range of 23–25%)*

3c *(2 marks)*

Similarities:

a. reduce root length (compared to the control)
b. (ZDV and NVP) have greatest change (in growth) at lowest concentrations (of ZDV and NVP)
c. same effect at 200 µmol
d. above/at about 400 µmol effect levels off

Differences:

e. the change/differences become significant after 200 µmol
f. ZDV has a (slightly) larger effect / NVP has a (slightly) smaller effect / WTTE ***OR*** above 400 µmol effect of ZDV remains higher / NVP remains lower

Answer must contain one similarity and one difference. Credit may be given for numeric differences when accurately stated.

3d *(2 marks)*

a. Y (will become more common)
b. spindle not formed yet ***OR*** cells x and z have spindles
c. cells in Y cannot progress (into Z/into metaphase)

3e *(1 mark)*

a. still some mitosis
b. individual cells grow/elongate (expand by absorbing water)
c. NVP is not 100 % effective / does not enter all of the cells / not all cells have come in contact with NVP
d. roots have reached maximum saturation of NVP and are no longer functionally affected
e. some cells are resistant to the drug

Accept reasonable suggestions.

3f *(1 mark)*

0.3 / 100 % increase / doubling. *(Do not penalize errors in significant figures. For example, 0.34 would be acceptable)*

3g *(2 marks)*

a. mitotic index in treatment greater than in control/leptin appears to promote mitosis
b. mitotic index increases with time suggests ongoing regeneration/growth ***OR*** positive correlation between exposure to leptin and increased mitotic index
c. but experiment limited to 72 hours/regeneration/recovery may take longer than 72 hours
d. no error bars shown/no information on significance/sample size

Acknowledge WTTE

3h *(3 marks)*

Evaluation of evidence in graph on left

a. increase in (percentage of) cells in mitosis (as vinblastine concentration rises)
b. supports hypothesis that cells get stuck in/cannot complete mitosis

Evaluation of evidence in graph on right

c. drop in anaphase-metaphase ratio due to fewer cells in anaphase/more cells in metaphase
d. cells not progressing from metaphase to anaphase/get stuck in metaphase

Do not allow mpa if the candidate is arguing that the hypothesis is not supported.

3i *(1 mark)*

a. causes microtubules/spindle fibres to break up / tubulin molecules to depolymerize
b. prevents contraction of spindle microtubules/fibres
c. disrupts/damages kinetochores/centromeres/microtubule motors/centrioles/centrosomes
d. prevents separation/pulling apart of (sister) chromatids/chromosomes/centromeres
e. prevents microtubules/spindle binding to chromatids/chromosomes/centromeres/DNA

Mark the first suggestion only in answer. *Do not allow answers about DNA replication or other processes that precede mitosis*.

3j *(2 marks)*

Advantage:

a. avoids risks for humans/harm to humans / more ethical (than with human patients/volunteers)

Disadvantage:

b. differences between plant and human cells so humans may not respond in same way ***OR*** plants have cell wall / no centrioles / other relevant difference between plant and human cells

Not enough for mpa to say 'not using humans'. For mpb there must be either a statement that differences between cells may cause a different response, or a specific example of a cell difference.