IB · DP · Biology

2 hours | 15 questions

Structured Questions: Paper 2

3.1 Genes & Chromosomes

3.1.1 Genes / 3.1.2 Alleles / 3.1.3 Mutation / 3.1.4 Genome / 3.1.5 Prokaryotic Chromosomes / 3.1.6 Eukaryotic Chromosomes / 3.1.7 Chromosome Number / 3.1.8 Sex Determination / 3.1.9 Karyograms / 3.1.10 Skills: Using Databases

Easy (5 questions)	/40
Medium (5 questions)	/47
Hard (5 questions)	/45
Total Marks	**/132**

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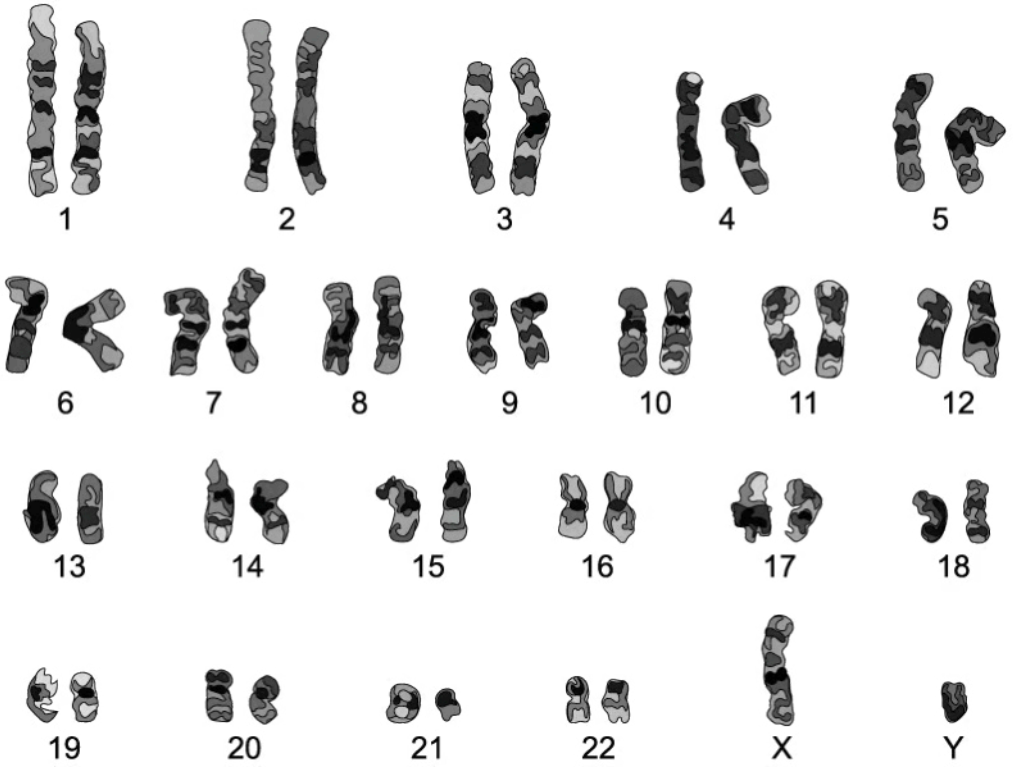
Easy Questions

1 (a) The following diagram shows the karyogram of an individual.



(i) Identify the sex of this individual.

[1]

(ii) State a reason for your answer in part i).

[1]

.....

.....

(2 marks)

(b) Cells in metaphase of mitosis were used to construct the karyogram from part a).

Explain the reason for this.

.....

(1 mark)

(c) List **two** characteristics of the chromosomes that are used to arrange them in a karyogram.

.....

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(2 marks)

(d) Apart from sex determination, state **one other** use of studying the karyotype of an individual.

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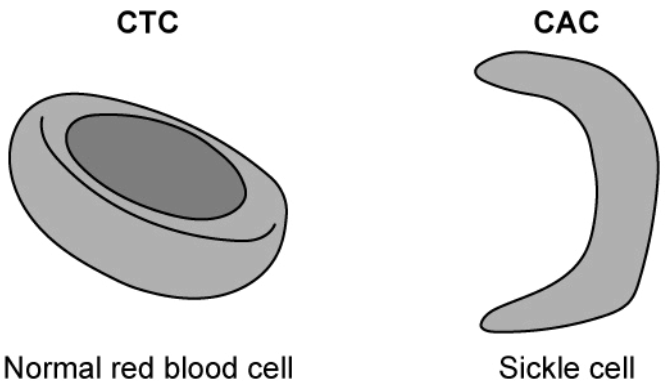
(1 mark)

2 (a) Sickle cell anaemia is a genetic disease where a DNA triplet base in the gene coding for alpha-globin in haemoglobin changes from CTC to CAC. This mutation results in sickle shaped red blood cells that can cause a range of different symptoms.The diagram below shows the DNA base triplet change and the resulting change in the structure of red blood cells.



(i) Identify the type of mutation that is illustrated in the diagram.

[1]

(ii) State a reason for your answer in part i).

[1]

.....

.....

(2 marks)

(b) List **two** consequences of the change in shape of the red blood cells in a person suffering from sickle cell anaemia.

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(2 marks)

(c) Some mutations, such as those causing sickle cell anaemia, may be inherited by offspring.

Describe how mutations can be inherited by offspring.

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(2 marks)

(d) Gene mutations lead to the formation of new alleles in a population.

Define the term 'allele'.

.....

(1 mark)

3 (a) During the process of fertilisation, haploid sperm and egg cells fuse together to form a diploid zygote.

Define the term 'haploid'.

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(1 mark)

(b) There are certain advantages to cells being diploid.

List **two** advantages of cells containing two sets of chromosomes.

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(2 marks)

(c) A couple is expecting a child and wondering which sex the baby will be.

The following genetic diagram shows the sex chromosomes present in the gametes of both parents.

Gametes	X	X
X		
Y		

Calculate the percentage chance of the baby being a girl by completing the genetic diagram.

Show your working.

.....

.....

.....

(3 marks)

(d) One of the genes carried on the Y chromosome is the SRY gene.

State **one** role of this gene in the development of male embryos.

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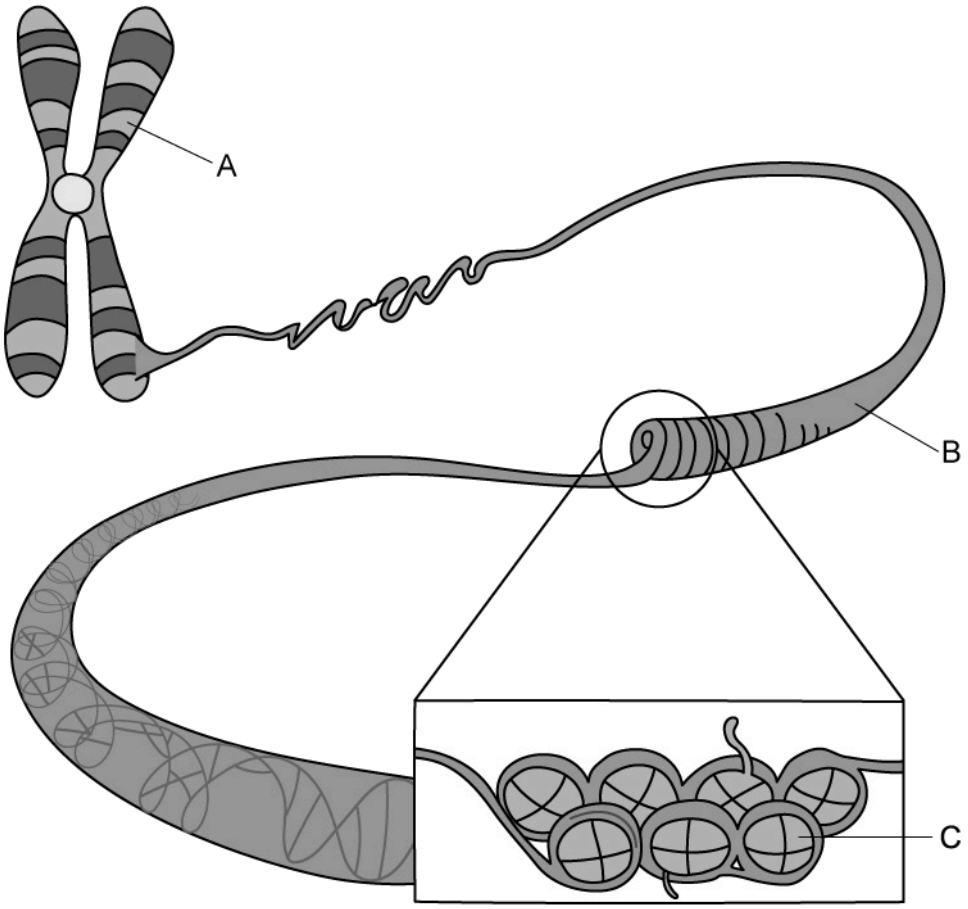
(1 mark)

4 (a) The following diagram shows the arrangement of DNA in a eukaryotic chromosome.



(i) Label structure **A** in the diagram.

[1]

(ii) State what is represented by the banding pattern on structure **A**.

[1]

.....

.....

(2 marks)

(b) Use the information in the diagram in part a) to:

(i) Identify structures **B** and **C**.

[1]

(ii) Describe the relationship between structures **B** and **C**.

[1]

.....

.....

(2 marks)

(c) Chromosomes in diploid cells will occur in homologous pairs.

Define the term 'homologous chromosomes'.

.....

(1 mark)

5 (a) *One mark is available for clarity of communication throughout this question.*

The Human Genome Project was an international, collaborative research programme to sequence the entire human genome.

List **four** of the main applications of the Human Genome Project.

(4 marks)

(b) Plasmids are small, circular DNA molecules commonly found in prokaryotic cells.

Describe the role of a plasmid.

(3 marks)

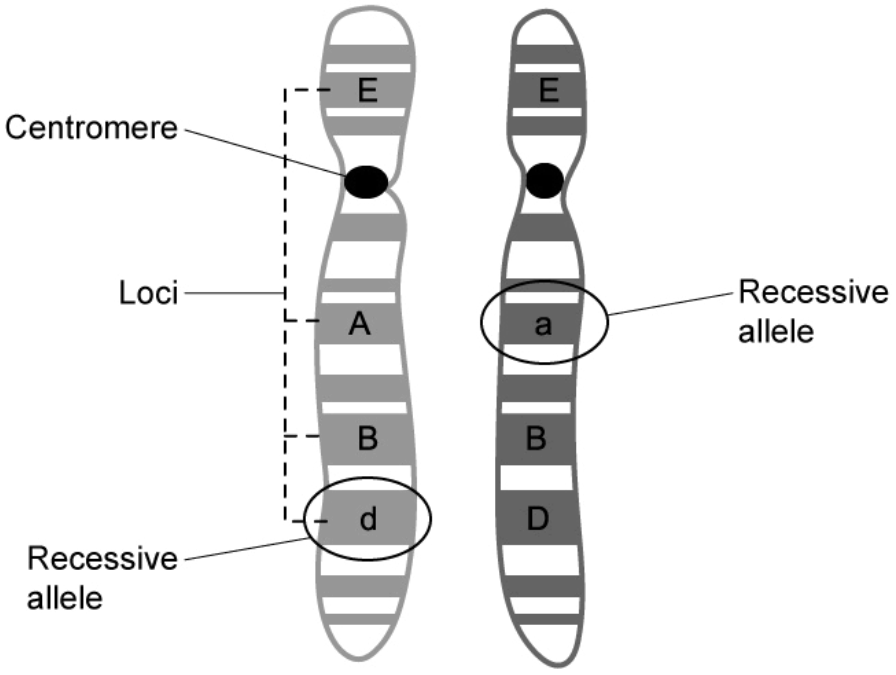
(c) Advancements in genome sequencing has led to developments in scientific research.

Outline the technique used to sequence a genome.

(8 marks)

Medium Questions

1 (a) The diagram shows one pair of homologous chromosomes



(i) Label the diagram to identify the following:

- Locus
- Centromere

(ii) Draw a circle around a section of chromosome that contains a recessive allele.

.....

.....

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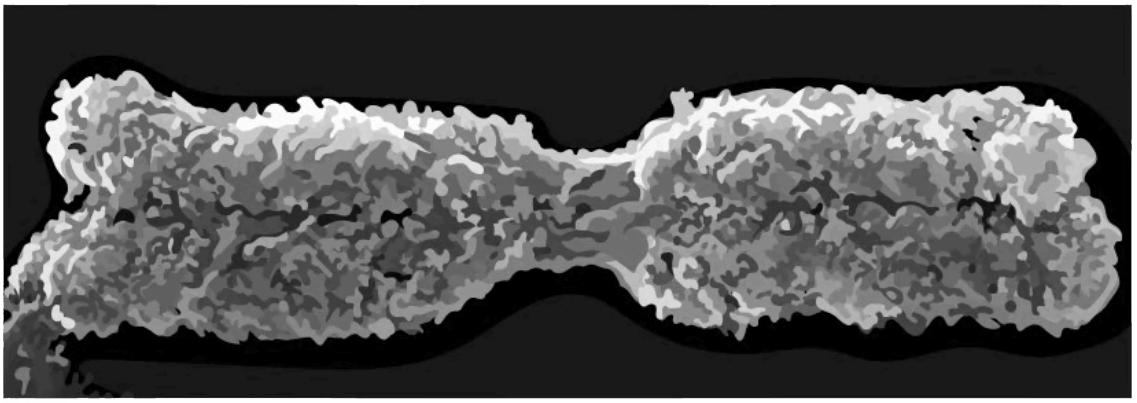
(3 marks)

(b) Outline the process which leads to the production of a chromosome with the appearance as shown in the image below, when viewed with an electron microscope.



(2 marks)

(c) In meiosis, homologous chromosomes pair up during metaphase 1.

Explain why these homologous chromosomes are similar but not identical.

(3 marks)

2 (a) The following base sequences represent sections of two different alleles of the gene which determines an individual's ability to roll their tongue.

Allele A (tongue roller): GCCGTAAC
Allele B (non-tongue roller): GCGCTTAC

Outline why two different alleles result in different expressions of a gene.

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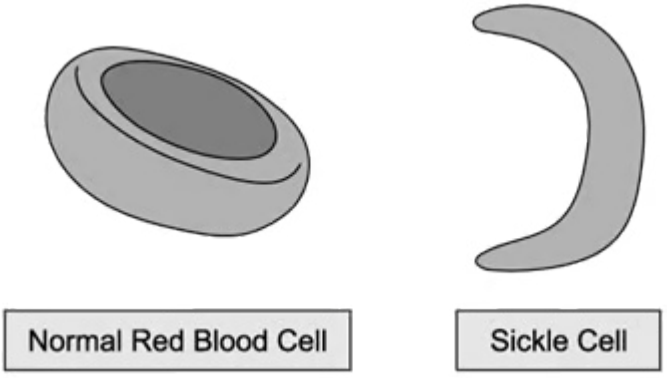
.....

(3 marks)

(b) Sickle cell anemia is a genetic disorder with symptoms such as dizziness, a rapid heart rate and fatigue. It is caused by an allele that leads to altered haemoglobin proteins. These altered proteins undergo aggregation (sticking together), an event which changes the shape of red blood cells. This can be seen in the image below.



Suggest how sickled red blood cells may result in the symptoms noted above.

.....

.....

.....

(3 marks)

(c) Explain why the shape of white blood cells is not affected by sickle cell anaemia.

.....

(1 mark)

(d) Mutations such as the one seen in sickle cell patients are usually caused by an error during DNA replication.

Identify the enzyme that is responsible for catalysing the process of DNA replication.

.....

(1 mark)

3 (a) Outline the developments in technology which have enabled the successful sequencing of the genome in the Human Genome Project.

.....

.....

(2 marks)

(b) Suggest **two** medical applications of genome projects like the Human Genomet Project.

.....

.....

(2 marks)

(c) It was decided that all data generated for the Human Genome Project would be made publicly available so that access would be free for everyone.

Suggest **one** advantage and **one** disadvantage of making scientific data publicly available.

.....

.....

(2 marks)

(d) When carrying out the genome project the DNA from individuals was obtained via blood samples.

Identify, with a reason, which cell type the DNA is obtained from when using a blood sample.

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.....

(2 marks)

4 (a) The human genome is approximately 3 billion, or 3 000 000 000, base pairs long. A DNA sequencing machine allows for 5.5×10^8 base pairs to be sequenced per hour.

Using this information calculate the number of days it would take to sequence 1500 genomes of hospital patients using this machine. Give your answer to the nearest day.

.....

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(2 marks)

(b) The table below shows part of the DNA base sequence coding for β-haemoglobin and two mutations of this sequence detected in a sickle cell sufferer.

DNA base sequence coding for β-haemoglobin												
mRNA sequence for β-haemoglobin	A	C	U	C	C	U	G	A	G	G	A	G
DNA base sequence with mutation 1												
mRNA base sequence with mutation 1	A	C	U	C	C	U	G	U	G	G	A	G
DNA base sequence with mutation 2												
mRNA base sequence with mutation 2	A	C	U	C	C	U	G	A	A	G	A	G

Complete the table with the DNA sequences that will undergo transcription to produce β-haemoglobin, mutated protein **1,** and mutated protein **2**.

.....

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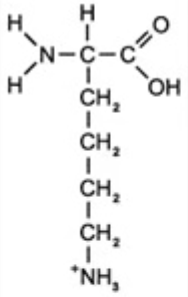
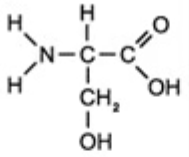
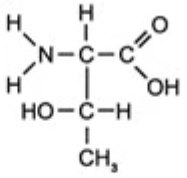
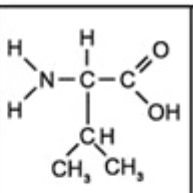
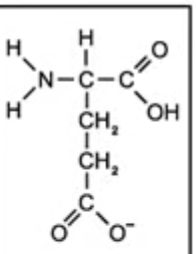
(3 marks)

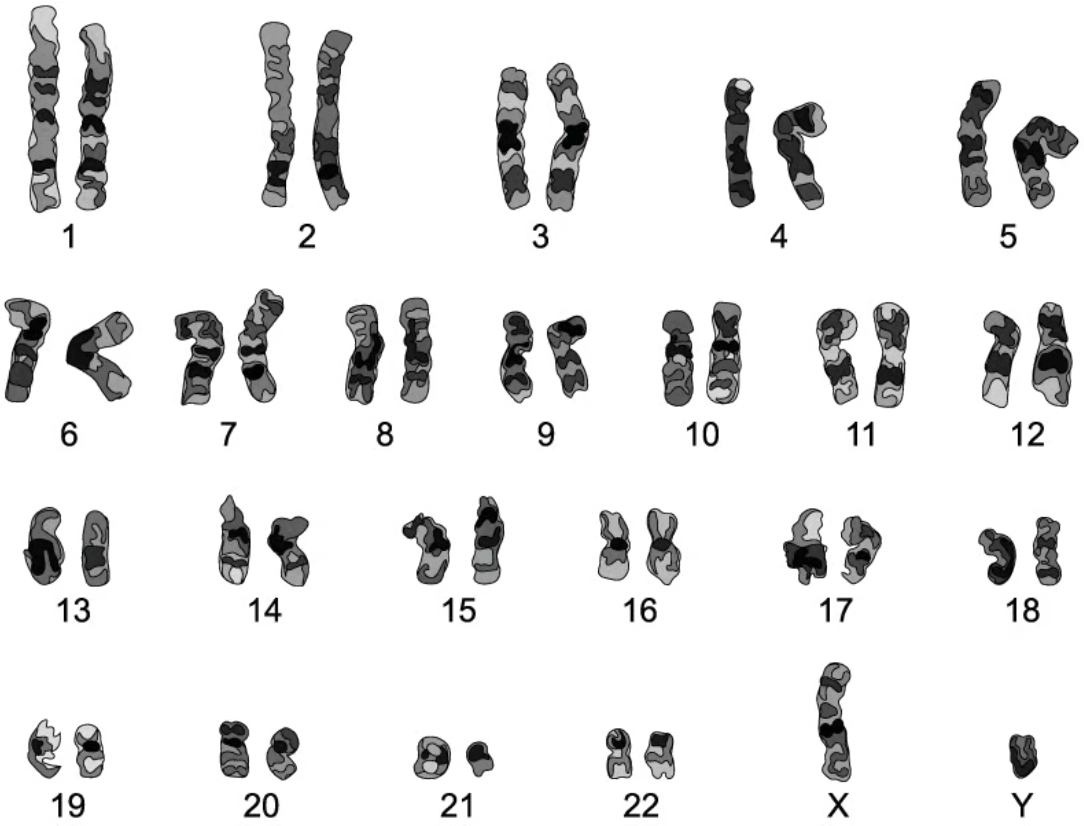
(c) The table below shows some examples of amino acids, their structures, and the mRNA codons that code for them.

Amino Acid	 Lys	 Ser	 Thr	 Val	 Glu
mRNA codons	AAA AAG	AGU AGC	ACU ACG ACA ACC	GUU GUG GUC GUA	GAA GAG

Suggest why mutation **2** from part (b) is of no concern to the scientists studying this patient's DNA

(2 marks)

(d) A karyogram, such as the one shown in the image below, can be used to detect some Mutations



State why this karyogram could not be used to detect sickle cell anaemia.

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(1 mark)

5 (a) *One mark is available for clarity of communication throughout this question.*

Outline how a mutation leads to the development of Down syndrome.

(3 marks)

(b) Distinguish between prokaryotic and eukaryotic DNA.

(5 marks)

(c) Outline the technique developed by John Cairns to measure the length of DNA, and how his methods contributed to further discoveries about DNA.

(7 marks)

Hard Questions

1 (a) Part of the gene coding for a specific polypeptide contains the following base sequence:

CATAGTTGGCCA

The following table contains some of the codons on messenger RNA and the amino acids that they code for:

Codons	Amino acid coded for by codons
AUU / AUC / AUA	Isoleucine (Ile)
UUA / UUG / CUU / CUC / CUA / CUG	Leucine (Leu)
GGU / GGC / GGA / GGG	Glycine (Gly)
ACU / ACC / ACA / ACG	Threonine (Thr)
UCU / UCC / UCA / UCG	Serine (Ser)
GUU / GUC / GUA / GUG	Valine (Val)
UAA / UAG / UGA	STOP

Using the information provided, identify the amino acid sequence for this part of the polypeptide.

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.....

.....

(3 marks)

(b) The cells that synthesise this polypeptide was exposed to a mutagen that caused a substitution mutation that changed the DNA base sequence in the following way:

CATACTTGGCCA

Using the table from part a), explain the effect this mutation will have on the polypeptide produced.

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.....

.....

(3 marks)

(c) This polypeptide forms part of the active site of an enzyme that catalyses a metabolic reaction.

Suggest the effect that the mutation mentioned in part b) would have on the enzyme.

.....

.....

(2 marks)

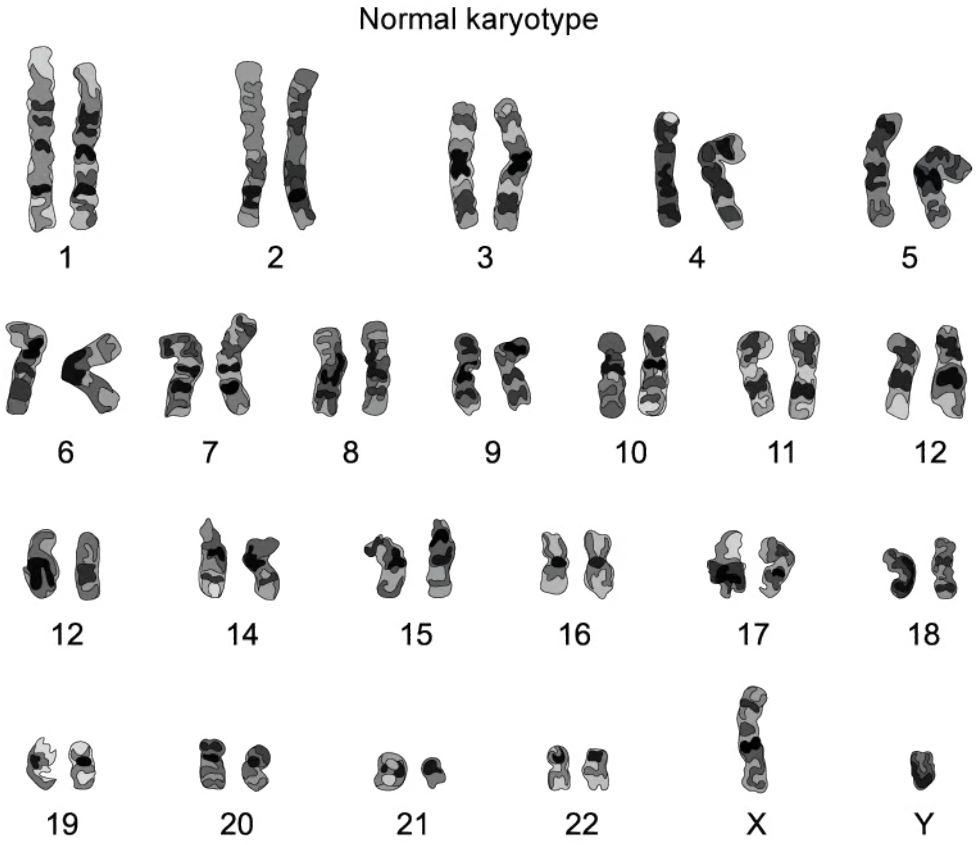
(d) Based on the information provided in part a), explain how it could be possible for a mutation to have no effect on the polypeptide.

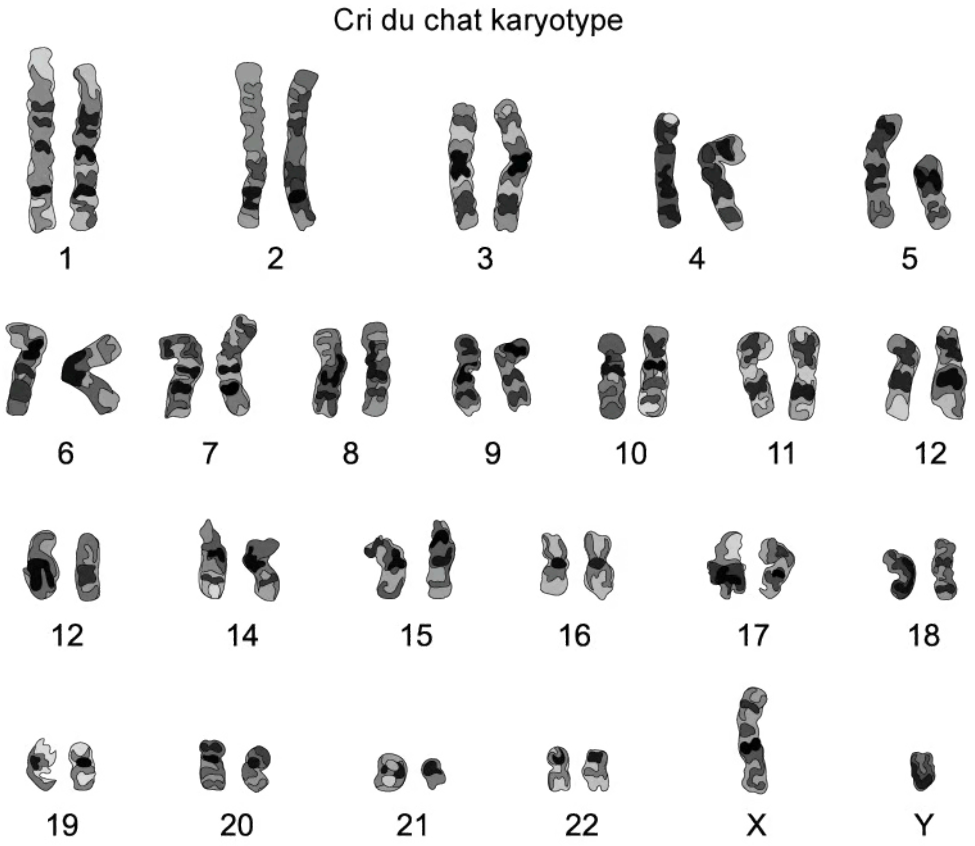
.....

(1 mark)

2 (a) Cri du chat syndrome is a rare genetic disorder caused by a chromosomal abnormality that occurs very early in embryonic development. Babies born with cri du chat syndrome suffer from a variety of symptoms and have a characteristic cry which sounds like the meowing of a cat.

The karyograms below compare the karyotype of a normal child with one that suffers from cri du chat syndrome.





Contrast the karyotype of a normal child with that of a child suffering from cri du chat syndrome.

.....

(1 mark)

(b) Two genes, SEMA5A and CTNND2, are believed to be involved with brain development in a foetus. These genes are missing from children suffering from cri du chat syndrome.

Suggest **two** possible symptoms of children with cri du chat syndrome as a result of this.

.....

.....

(2 marks)

(c) There are some individuals with cri du chat syndrome that do not differ developmentally from their peers in a significant way.

Based on the information provided in the karyograms at part a), explain this occurrence.

.....

.....

(2 marks)

(d) Most of the people affected by cri du chat syndrome do not have a family history of the condition.

Suggest what this means in terms of the heritability of the syndrome.

.....

(1 mark)

3 (a) The table below shows the genome size and haploid chromosome number of different organisms.

Organism	Genome size / base pairs	Chromosome number / n
Polychaos dubium (single celled eukaryote)	6.7×10^{11}	> 100
Trumpet lily (plant)	9.0×10^{10}	12
Mouse	3.5×10^{9}	20
Human	3.2×10^{9}	23
Carp (fish)	1.7×10^{9}	49
Chicken	1.2×10^{9}	39
Housefly	9.0×10^{8}	6
Tomato plant	6.6×10^{8}	12

Calculate the percentage difference in the chromosome number found in the zygotes of chickens compared to those of humans.

Show your working and give your answer to three significant figures.

.....

.....

(2 marks)

(b) The diploid number in an organism is always an even number.

Using your knowledge on the behaviour of chromosomes during meiosis, explain the importance of the diploid number in an organism.

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(2 marks)

(c) Scientists hypothesised that a high chromosome number leads to the development of a more complex organism.

Discuss this hypothesis using the data provided in part a).

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(3 marks)

(d) Based on your knowledge of chromosomes, suggest a reason why the genome size of a species does not always seem to correlate with the chromosome number.

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(1 mark)

4 (a) Wilson's disease is a condition caused by a mutation of gene ATP7B located on chromosome 13, which codes for an ion transport enzyme. This enzyme is responsible for transporting copper ions (Cu^{2+}) into bile so that it can be removed from the body through the digestive tract. There are several mutations which may lead to Wilson's disease; one of these mutations involves the replacement of the amino acid histidine by glutamine.

Describe the type of mutation that could have led to this disease.

.....

.....

(2 marks)

(b) Based on the information provided in part a), explain the effect this mutation would have on the transport of copper ions.

.....

.....

(2 marks)

(c) In most cases of Wilson's disease, a sufferer must have two copies of the mutated ATP7B allele before the disease is present.

Suggest what this may indicate about the dominance of the ATP7B allele.

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(1 mark)

(d) People suffering from Wilson's disease have high levels of free copper in their bloodstream which have been shown to cause damage to the cell membranes of red blood cells.

Explain the consequences of this to the sufferer.

.....

.....

(2 marks)

5 (a) *One mark is available for clarity of communication throughout this question.*

Compare and contrast the X and Y chromosome.

(5 marks)

(b) The sex of a foetus is determined by the father.

Explain this statement.

(3 marks)

(c) Outline how a substitution mutation can alter the amino acid sequence of a polypeptide by using sickle cell anaemia as an example.

(7 marks)