

Name	Date started	Target end date
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GCE A LEVEL – BIOLOGY UNIT 4 QUESTION PACK

1075 (Legacy BY5) · New spec Unit 4 Topic 5 · A2 unit, first sat 2017, 90 marks, 2h paper

REVISE

.wales

BIOLOGY – UNIT 4 · MEIOSIS, MUTATION & SOURCES OF VARIATION

4.4 Variation and evolution – meiosis, sources of genetic variation, gene and chromosome mutations

Meiosis as the source of genetic variation through independent assortment of homologous chromosomes and crossing-over, together with random gamete fusion at fertilisation, and the role of point mutations (substitution, insertion, deletion) illustrated by the β -globin variants of haemoglobin including sickle-cell.

LEGACY 2008 SPECIFICATION

Estimated time for entire question pack: ~26 min

Derived from the legacy BY5 papers' pace of ~1.6 min/mark, padded for long-prose answers (16 marks over 2 questions).

*You are advised to **not** attempt to complete all of this in one sitting.*

ABOUT THIS QUESTION PACK

This is a **comprehensive practice question pack**, not a single mock paper. It contains every question from the legacy WJEC BY5 papers (2008 modular spec, 2011–2017) that maps onto new-spec A2 Unit 4 Topic 5 (4.4).

Questions are ordered by source paper date.

INSTRUCTIONS

Use black ink or black ball-point pen. Show all working – quality of written communication will affect marks. A calculator is allowed. Diagrams included in answers must be fully annotated.

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Q	Source	Max	Mark	Q	Source	Max	Mark
1	BY5 Jun 14 Q3	8		2	BY5 Jun 17 Q1	8	
Total						16	

Meiosis, Mutation & Sources of Variation – what the new spec asks

WJEC GCE A Level Biology (from 2015) · Unit 4: Variation, Inheritance & Options · Topic 4.4.

Meiosis I – reduction

- Homologous chromosomes pair (synapsis) at prophase I.
- Crossing-over at chiasmata exchanges DNA between non-sister chromatids.
- Independent assortment of homologue pairs at metaphase I.

Meiosis II – equational

- Sister chromatids separate as in mitosis.
- Four haploid daughter cells produced from one diploid parent.
- Each gamete is genetically unique.

Sources of variation

- Independent assortment ($2^{23} \approx 8.4$ million combinations in humans).
- Crossing-over – recombines maternal and paternal alleles.
- Random fertilisation – any sperm with any egg.

Gene mutations

- Substitution – one base replaced; can be silent, missense, or nonsense.
- Insertion / deletion – shifts reading frame; usually severe.
- Sickle-cell – single base change (GAG → GTG) in β -globin.

Chromosome mutations

- Non-disjunction – failure to separate; aneuploidy (e.g. trisomy 21).
- Inversion, translocation, duplication, deletion at chromosome level.
- Detected by karyotyping.

Continuous vs discontinuous

- Continuous (e.g. height, mass) – polygenic + environmental.
- Discontinuous (e.g. blood group) – monogenic; distinct categories.
- Normal distribution often emerges from polygenic inheritance.

Meiosis, Mutation & Sources of Variation in one page

Quick-reference notes – revisit before each question.

Meiosis I

Homologous chromosomes pair (bivalents).
Crossing-over at chiasmata.
Independent assortment of bivalents.

Meiosis II

Sister chromatids separate.
Like mitosis but no DNA replication beforehand.
Four haploid cells per parent cell.

Variation sources

Independent assortment.
Crossing-over.
Random fertilisation.
Mutation (rarer but creates new alleles).

Substitution

One base for another.
Silent (synonymous), missense, or nonsense.
Sickle-cell: GAG → GTG, Glu → Val in β -globin.

Frameshift

Insertion or deletion of bases not multiples of 3.
Shifts reading frame downstream.
Usually catastrophic.

Chromosome mutation

Non-disjunction → aneuploidy (e.g. Down's trisomy 21).
Inversion, translocation, deletion, duplication.

Continuous vs discontinuous

Continuous: polygenic + environment; normal distribution.
Discontinuous: distinct categories (e.g. ABO).

Causes of mutation

Replication errors (rare with proofreading).
Mutagens: UV, X-rays, chemicals (mustard gas).

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3. The photograph below shows the pairs of chromosomes found in a body cell of a mouse.

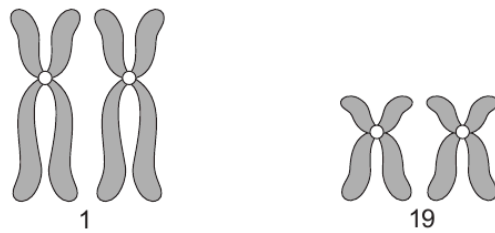
Examiner only



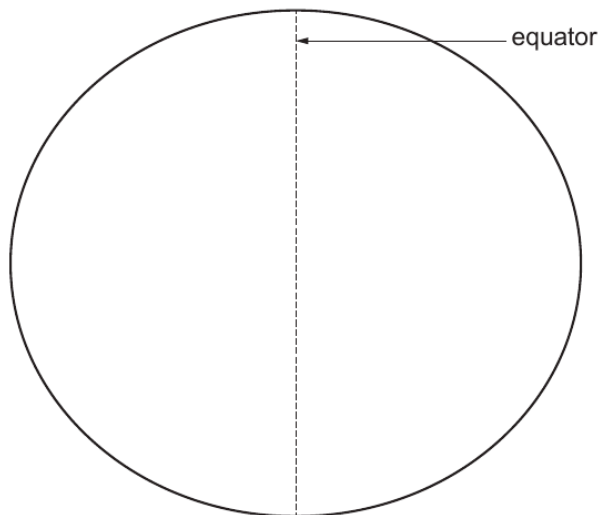
(a) What is the diploid number of the mouse? [1]

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(b) The chromosomes in pairs 1 and 19 are commonly represented diagrammatically as:



(i) Using the cell outline below draw diagrams to show how these pairs of chromosomes are arranged in **metaphase I** of meiosis. [1]



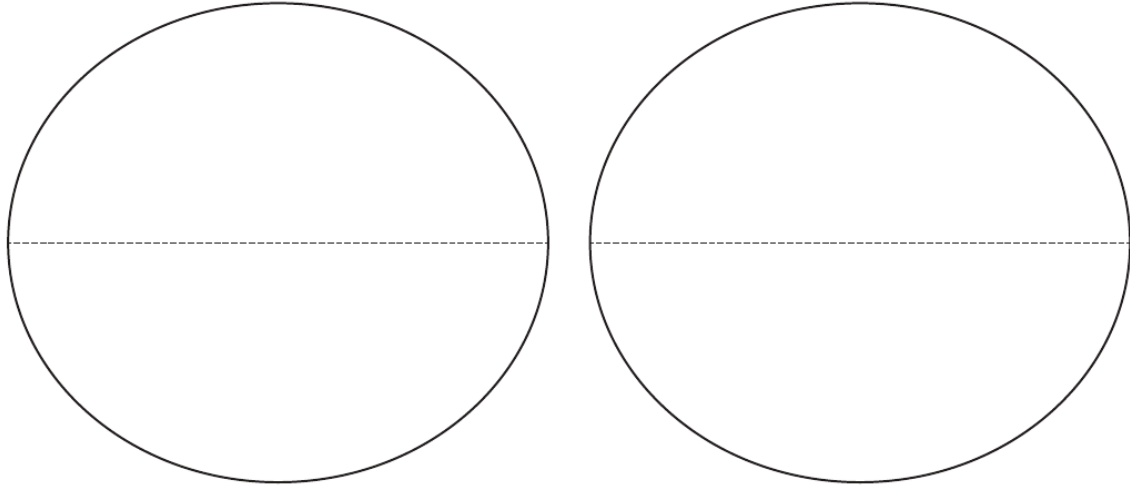
(ii) On your drawing label; chromatid, centromere, centriole, spindle fibres. [2]

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- (iii) Using the cell outlines below draw diagrams to show how the chromosomes would subsequently be arranged in **metaphase II** of meiosis. [1]

Examiner only



- (iv) State **three** ways in which meiosis contributes to variation in mouse offspring. [3]

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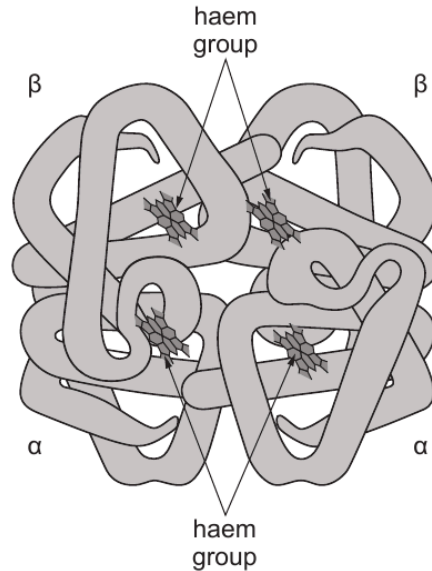
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Answer **all** questions.

Examiner
only

1. A haemoglobin molecule contains four polypeptide chains, two α chains and two β chains, as shown:



- (a) (i) What is the highest level of protein structure shown by this molecule? [1]

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- (ii) According to the one gene one polypeptide hypothesis, how many genes are required to code for this haemoglobin molecule? [1]

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The diagram below shows the first nine amino acids of a β chain, together with the nucleotide base sequence of the gene which codes for its synthesis:



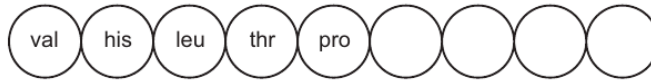
base sequence GTGCATCTGACTCCTGAGGAGAAGTCT

More than 300 variations in the amino acid sequence of the β chain have been recorded. The table below shows some of them.

Name of variation	Amino acid number	Amino acid substitution	Mutation
Raleigh	1	val to ala	GTG to GCG
Deer Lodge	2	his to arg	CAC to CGC
Warwickshire	5	pro to arg	CCT to TCT
S	6	?	GAG to GTG
Rio Grande	8	lys to thr	AAG to ?

Examiner only

- (b) Complete the diagram below to show the amino acid sequence in the Rio Grande variation. [1]



The table below shows the amino acids coded for by different DNA base triplets.

		Second Letter				
		T	C	A	G	
First Letter	T	TTT } phe TTC } TTA } leu TTG }	TCT } TCC } ser TCA } TCG }	TAT } tyr TAC } TAA stop TAG stop	TGT } cys TGC } TGA stop TGG trp	T C A G
	C	CTT } CTC } leu CTA } CTG }	CCT } CCC } pro CCA } CCG }	CAT } his CAC } CAA } gln CAG }	CGT } CGC } arg CGA } CGG }	T C A G
	A	ATT } ATC } ile ATA } ATG met	ACT } ACC } thr ACA } ACG }	AAT } asn AAC } AAA } lys AAG }	AGT } ser AGC } AGA } arg AGG }	T C A G
	G	GTT } GTC } val GTA } GTG }	GCT } GCC } ala GCA } GCG }	GAT } asp GAC } GAA } glu GAG }	GGT } GGC } gly GGA } GGG }	T C A G

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- (c) Use the table to find:
- (i) One mutation which causes the Rio Grande variant. [1]
AAG to
 - (ii) The amino acid substitution that occurs in the S variant. [1]
..... to

People with the S variation suffer from a condition called sickle cell anaemia. In this condition, their red blood cells change shape when blood oxygen concentration is low.

- (d) Explain how the substitution of an amino acid in the β chains can bring about this change. [3]

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END OF QUESTION PACK

2 questions · 16 marks · ~26 min

Source: WJEC BY5 (2008 modular spec, 2011–2017)

Curated for WJEC Biology 2015 spec A2 Unit 4 – Topic 5 (4.4)

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