Year 11 Human Biology – Inheritance, variation and mutations test Mark: /58

Name: Answer key Teacher: _____

PART A: MULTIPLE CHOICE

1. To display recessive characteristics you must have:
   a. Have both alleles recessive.
   b. Have both alleles dominant.
   c. One recessive allele and one dominant allele.
   d. Physical characteristics due to the genotype.

2. Choose the **incorrect** statement regarding the chromosomes.
   a. Human beings have 46 chromosomes in a normal body cell. ✓
   b. One pair of chromosomes control the formation of asexual characteristics.
   c. In humans there are two types of sex chromosomes.
   d. The 23rd pair of chromosomes are the sex chromosomes.

3. Choose the **incorrect** statement regarding mitochondrial DNA:
   a. It is found only in the ova.
   b. It is inherited from the mother.
   c. It is inherited from the father.
   d. It is located in the mitochondria.

4. Choose the **incorrect** statement regarding sex chromosomes.
   a. The X chromosome is much bigger than the Y chromosome. ✓
   b. Some genes found on the X chromosome are not found on the Y chromosome. ✓
   c. A female has only one copy of all the genes on the X chromosome.
   d. A male has only one copy of certain genes on the X chromosome.

5. A person with Rh antibodies:
   a. Is said to be Rh positive.
   b. Can produce anti-Rh antibody.
   c. Is said to be Rh negative.
   d. Can produce anti-Rh antigens.

PART B: SHORT ANSWER

1) Fill in the table below. (4 marks)

Blood group	Antigens on red blood cells	Antibodies in plasma
A	Antigen A	Anti B
B	Antigen B	Anti A
AB	Antigen A & B	no antibodies
O	no antigens	both anti A & B

-1 for every incorrect answer

2) A homozygous male tall pea plant is crossed with a homozygous female short pea plant. What are the possible offspring? Show full working out and include genotypes, phenotypes and percentages.

T=tall, t=short. (5 marks)

Parent genotypes Male = TT (1) female = tt (1)

	T	T
t	Tt	Tt
t	Tt	Tt

(1)

Genotype Tt 100%. (1)

Phenotype Tall 100%. (1)

3) Fill in the table below. (2 marks)

	Germline mutation	Somatic mutations
Cells it affects	gametes (sex cells)	Body cells
Inherited or not inherited	Inherited	not inherited

4) **Explain** the main difference between gene mutations and chromosomal mutations. (2 marks)

Gene mutation - single gene is mutated / altered

Chromosomal mutation - all or part of chromosome is affected

5) **List** two examples of conditions caused by gene mutations. ANY TWO (2 marks)

Albinism Duchenne form of muscular dystrophy

Cystic Fibrosis Tay-Sachs disease

6) **List** two examples of conditions caused by chromosomal mutations. ANY TWO (2 marks)

Down Syndrome Klinefelter's syndrome

Patau Syndrome Turner's syndrome

Cri du chat sydrome

7) **Fill** in the missing words. (3 marks)

Point mutation – if one base is changed, the protein could be altered, no effect may occur or the protein for which it codes may be missing or abnormal.

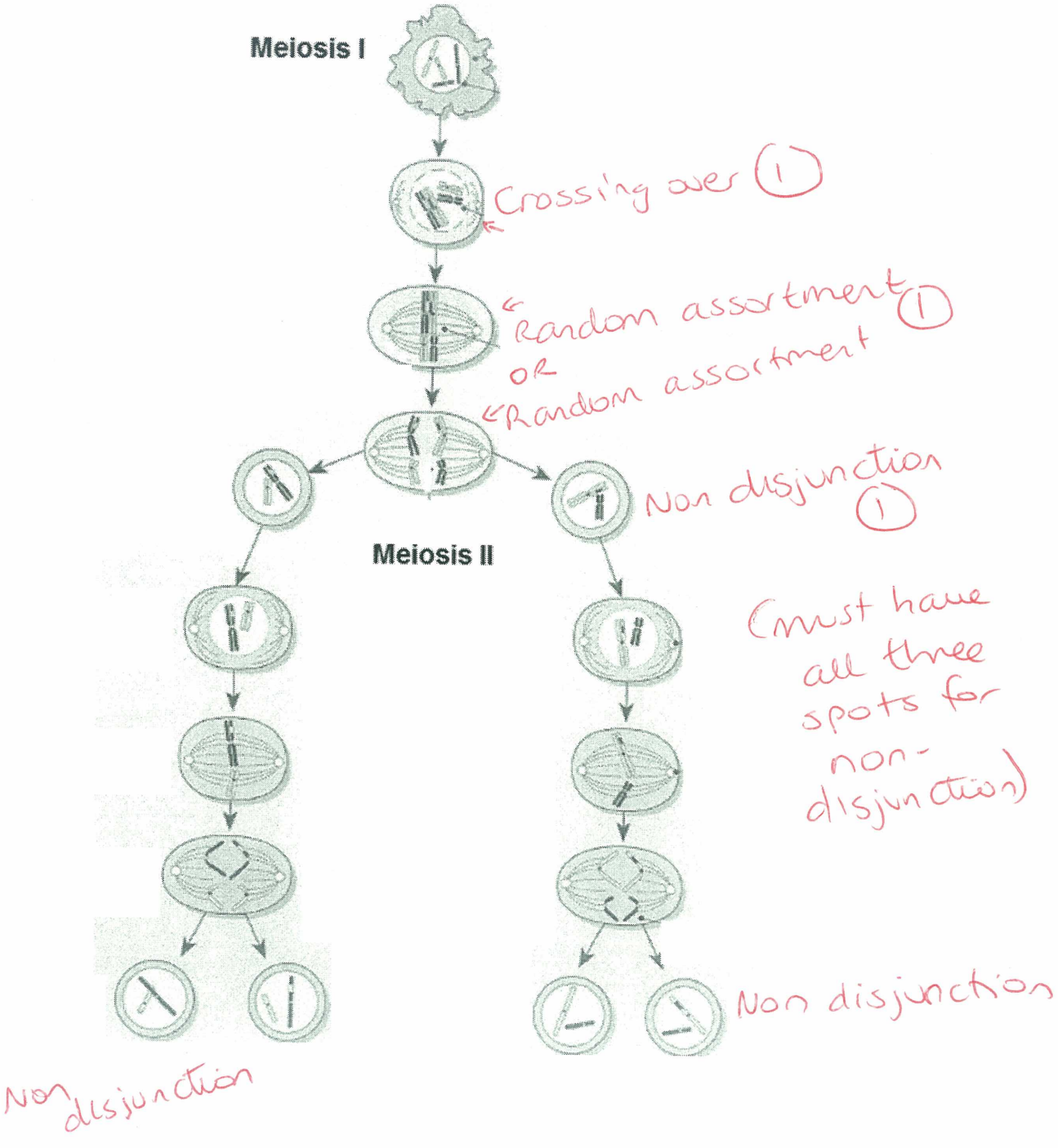
One missing or abnormal protein can have an enormous effect on the whole body.

8) **Describe** the difference between trisomy and monosomy. (2 marks)

Trisomy - three copies of a ~~normal~~ chromosome

Monosomy - One copy of a chromosome.

9) On the diagram of meiosis below, label where crossing over, random assortment and non-disjunction occur. (3 marks)



10) **Explain** how random fertilisation contributes to variation. (2 marks)

- the particular egg that is released from the ovary is random. (1)
- the particular sperm that fertilises the egg is random (1)

11) Write a definition for the term 'natural selection'. (2 marks)

Process where species becomes better adapted to its environment. The individuals with favourable characteristics survive & the ones that don't perish (1) (1)

12) Write a definition for the term 'mutation. (2 marks)

A new variation, resembling neither parent, that occurs quite suddenly & purely by chance. (1) (1)

13) For each main group below, give a specific example of a mutagen. (4 marks)

Choose one for each

Ionising radiation: (1) Nuclear, ultraviolet, x-rays

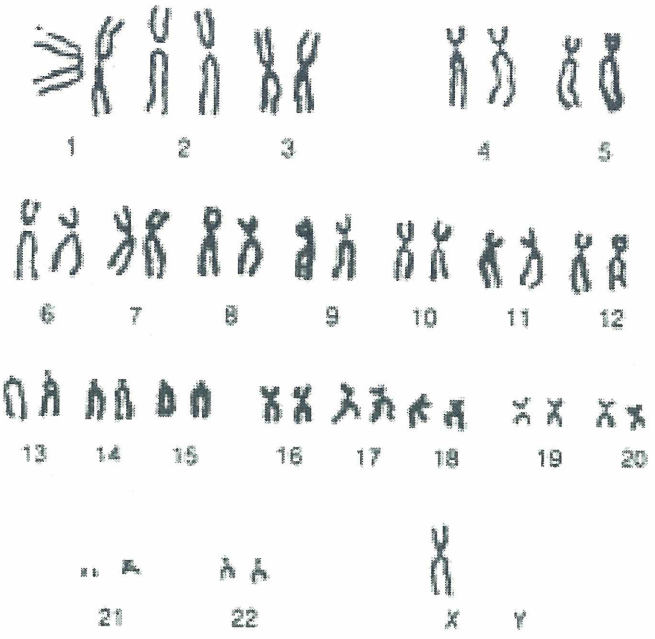
Alcohol and diet: (1) Diet high in fat, high alcohol intake, smoking

Viruses and microorganisms: (1) HIV, Hepatitis

Poisons and irritants: (1) Asbestos, dyes, tobacco tar

14) **State** the sex of the individual whose karyotype is shown below. (1 mark)

Female



b) **State** whether the individual has monosomy or trisomy. **Explain** your answer. (2 marks)

- Monosomy (1)
- A pair of X chromosomes is missing (1)

c) **State** the name of the specific mutation that this individual suffers from. (1 mark)

Turner's syndrome

15) **Fill** in the missing spaces. (3 marks)

A person can have large numbers of mutations in their genes but not be aware of them. If that person reproduced with a partner who had the same recessive mutation, the recessive condition could appear in their offspring.

Some recessive mutations are lethal if they are not masked by a recessive normal allele. These are called lethal recessives.

~~Two~~ one disorders caused by this type of mutation are Tay sachs and _____.

-1 for each missing

PART C: EXTENDED ANSWER

16) Variation in the human species occurs due to chromosome variation. For each type of chromosome variation listed below, **state** when it occurs, **describe** how it occurs and **list** two examples of disorders caused by the type of variation (only if it causes disorders).

Crossing over, random assortment of chromosomes during meiosis and non-disjunction.

Hint: use a table!

(11 marks)

	When it occurs	Describe how it occurs	Disorders
Crossing over	prophase 1 (1)	When chromosomes (1) pair up, chromatids tangle with eachother chromosomes then have new combination of alleles (1)	/
random assortment	metaphase 1 Anaphase 1 (1)	chromosomes line up at equator in random way (1) chromosomes split to either pole in random way (1)	/
Non-disjunction	During meiosis 1 & 2 (1) OR Anaphase 1 & 2	chromosome pairs do not separate properly when cell divides (1) Results in some daughter cells missing or gaining a (1) pair of chromosomes	Down syndrome (1) Turners syndrome (1)