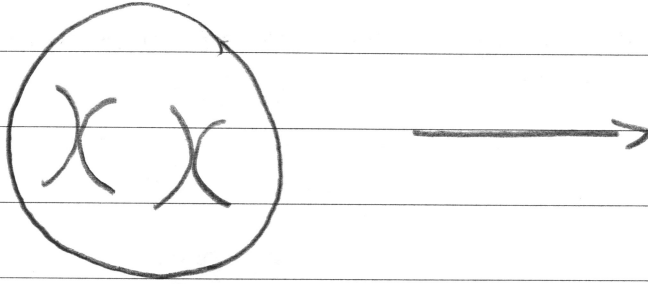


Start here.

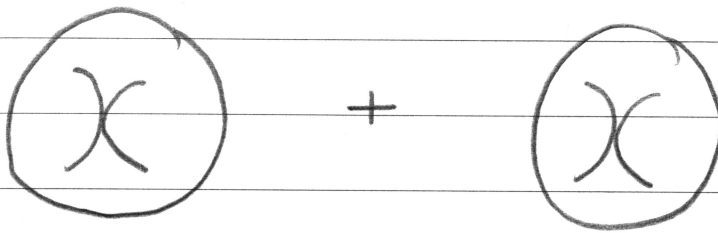
(a)

Mutation Type	Features of mutation	Affect on chromosome
Base substitution	Three amino acids which code for a polypeptide may be changed due to bases swapping chromosomes. or being substituted	This doesn't affect the number of chromosomes in an organism, however it does change the production of amino acids
Trisomy		There is a gene defect as there are 3 chromosomes for a certain cell functioning.
Polyploid.		There is double the information on the chromosome
		& this leads to polypeptide production & gene expression being effected.

(b) Diploid cell is a somatic cell. It has 2 sets of chromosomes and 2 pairs of genes.



A haploid cell is a gametic cell. It has one set of chromosomes and one pair of genes.



(c) (i) { Vision Defect: Recessive
 Limb Defect: Recessive

(ii)

For vision defect Individual 9.

	$X^R X^r$	$X^r Y$	\therefore individual 9 must be heterozygous for the vision defect
X^R	$X^R X^r$	$X^R Y$	
X^r	$X^r X^r$	$X^r Y$	

For vision defect Individual 10

It would be assumed that individual 10 would be homozygous dominant for the gene and not have the vision defect

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∴ Their child would have a 100% chance of **not** having the vision defect if they were **not** linked

	R	r
R	RR	Rr
r	Rr	rr

Limb defect for individual 9

It would be assumed that individual 9 would be homozygous recessive for the gene and have a limb defect.

Limb defect for individual 10

Individual 10 could be homozygous dominant for the trait or heterozygous dominant for the trait, meaning that they would not have a limb defect.

therefore ...

	r	r		r	r
R	Rr	Rr	R	Rr	Rr
r	Rr	rr	r	rr	rr

their child has a 25% chance of **having** a limb defect if the traits were **not** linked.

If the genes were linked:

You may ask for an extra Writing Booklet if you need more space.

Start here.

(d)

(i) ~~DNA hybridisation~~ Use a restriction enzyme to cut out a piece of the gene, from two individuals who both have both traits. Use DNA hybridisation to compare & line up, showing relative position of linked genes.

(ii) The Human Genome Project could not be studied during linkage maps because:

- * we do not fully understand how genes are linked and why they are linked.

- * we have not discovered all of the genes in the human body

- * Through assumption we believe that certain genes are linked and linked to specific diseases, but we are unsure.

- * we don't necessarily know the distance of genes between each other on the chromosomes.

(e) Gene cloning has led to the development of technologies that allow for the replacement of defective genes in people suffering from genetic problems such as cystic fibrosis. Through research, it has been discovered that these genes can easily be reproduced and inserted into a gene. Gene cascades are when genes are 'switched' on and begin to form a limb. Hox genes initiate and suspend the production. The morning sickness drug that caused children to have deformed limbs was in relation to the chemical switching off the hox genes for limb production and mutating them

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