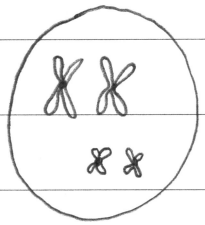
Start here.

a)

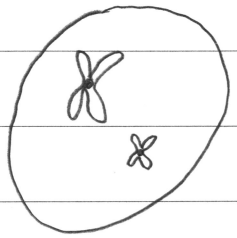
| Mutation | no. of Chromosomes |
|---|---|
| Trisomy | (n) + 1 |
| Polyploidy | (n) x 2 |
| Base Substitution | (n) |

note: where (n) stands for the normal number of chromosomes

b) Diploid cell 2(N)



Haploid cell (N)



c) (i) Vision defect – recessive

Limb defect – reeessive

(ii) For the vision defect (V = gene for normal v = defect)

ind. ~~8 x 6~~ 9 = Vv (heterozygous as has affected father of v

ind. 10 = VV or Vv (because ind. 7 is unknow whether Vv or VV)

∴ either

| | Option 1 | |
|---|---|---|
| | V | v |
| V | VV | Vv |
| v | VV | Vv |

or

| | Option 2 | |
|---|---|---|
| | V | V |
| V | VV | VV |
| v | VV | VV |

= 50%

∴ either 1:1 chance of vision defect or 100% chance if gene not linked

For the Limb defect (where L = normal gene and l = defect gene)

ind. 9 = ll (recessive)

ind. 10 = Ll (gene not expressed however mother ll)

∴

| | l | l |
|---|---|---|
| L | Ll | Ll |
| l | ll | ll |

∴ 1:1 chance of limb defect if not linked

If genes were linked

option 1 VL/vl x vl/vl

| | VL | vl |
|---|---|---|
| vl | vvLl | vvll |
| vl | vvLl | vvll |

∴ phenotypes = ~~normal vision no limb defect : normal vision limb defect : vision defect limb defect~~

1 normal vision no limb defect : 1 normal vision limb defect

VL/vl x vl/vl

option 2

| | VL | vl |
|---|---|---|
| vl | vvLl | vvll |
| vl | vvLl | vvll |

∴ Phenotypes =

1 normal vision no limb defect : 1 normal vision limb defect

∴ If the genes are linked the phenotypic ratio is 1 normal vision no limb defect : 1 normal vision limb defect

or 50% : 50%

∴ If the genes are not linked there is a 1:1 chance of offspring having a limb defect and either a 100% chance or a 1:1 chance of a vision defect.

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d) (i) The relative position of linked genes can be identified through finding out the number of recombinant offspring produced when ~~organisms~~ a dihybrid organism is crossed with a double recessive. The ~~number of~~ percentage of recombinant offspring ( $100 \times \frac{\text{no. of recombinant offspring}}{\text{total no. of offspring}}$ ) is equal to a map unit on a linkage map thus enabling the relative position of linked genes to one another.

(ii) 1. linkage maps can only be used to find already known genes which account for ~~only~~ about 50% of the human Genome.

2. Linkage maps can only provide the relative position of genes, not the actual position.

3. Linkage maps would be unpractical and time consuming ~~for~~ studying the human genome as it would be necessary to pedigree traits in humans and the recombination of these traits over many generations as experiments on the breeding and crossing over of humans are highly unethical

e) As advancements in technologies grow, ~~and~~ these are able to be applied ~~to produce~~ to Scientific developments. Over the years, our understanding of gene cloning (the production of a genetically identical gene) and gene cascades (a series of chemical signals which operate the ~~construction of~~ turning 'on' and 'off' of genes leading to the development of an organism) has led to new applications for these discoveries. The recent production of an artificial chromosome into a surviving, reproducing bacteria is an example of this. The creation of an artificial chromosome would have been due to the understanding that gene cascades produce the formation of an organism and that if an artificial chromosome was inserted, ~~gene clon~~ ~~from~~ DNA transcription, replication and translation would enable the chromosome to influence all parts of the organism. Our understanding of gene cascades has grown in recent years due to ~~the~~ experiments done with the differentiation of cells. For example, it was found that if, in the early stages of development, an arm cell was placed in the 'leg' area, it would still produce a leg as it was not yet differentiated, it was found however that after a time, an arm would grow where it should have been a leg. These experiments showed how genes in cells were switched 'on' and 'off' to specialise and differentiate them through the process of gene cascades. This led to the ~~development~~ research into stem cells in order to attempt to cure genetic diseases such as cystic fibrosis as it was now known that if stem cells could be manufactured then they could produce more

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correct genes and hence provide a more effective ~~[illegible]~~ cure for genetic diseases. Similarly, the ability to clone genes led to humans being able to create transgenic species. For example, the creation of a 'Super pig' with 10 growth hormones, which benefits the human meat industry but possibly als has ethical issues due to the arthritus caused in the pig. It was the understanding of gene cloning and gene cascades ~~[illegible]~~ that thus led to their application in technologies such as transgenic species and insertion of genes or cells.

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