HSC 2010 - Biology
Band 4/5 - Sample 1
Question 33

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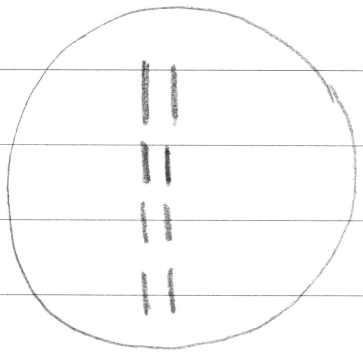
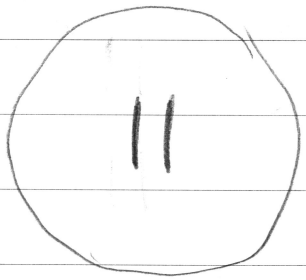
HOW MUTATIONS AFFECT CHROMOSOME NUMBERS.

Q33 (a)

TYPE OF MUTATION	HOW IT EFFECTS THE CHROMOSOME:
Trisomy	A trisomy mutation basically when, a pair of chromsomes don't segregate into each cell during the division of meiosis. The pair together goes into one cell. this means the gamete is lacking a whole chromosome.
Polyploidy	A polyploidy mutation is when, all the pairs of chromosomes during meiosis don't segregate into different cells during meiosis. this results in one gamete having no chromosomes. And the other gamete cell having a diploid number, when it should be haploid.
Base Substitution	Base substitution means during the replication of the strands of DNA, one base is substituted for another. For example (A instead of T) This results in the incorrect combination of bases, which can lead to incorrect protein produced.

* Note: all three types of mutations can lead to things such as death or genetic disease if the DNA does not repair itself, one mutation taken place.

(b) The simularities and differences between:

Diploid cell (4) (somatic cell)	Haploid cell (2) (gamatic cell)
	

Simularity: Both have (as shown) chromosomes. Both are cells, and a product of cell devision

Differences: The Diploid cell has full number of chromosemes (4) and Haploid has half the number of chromosomes.

There are two different types of cells, the diploid cell is a somatic cell,

Key
II = pair of chromosomes

but the other cell in order for it to be Haploid has to be a gamatic cell or (germ cell) therefore two different types of cells.

(c) (i) Both are recessive genes.

(ii) Phenotypes & Ratios of vision Defect & Limb Normal vision is dominant (N) over vision defect (n) Normal Limb dominant (L) over limb defect (l) NOT LINKED, Individual both 9 and 10 don't have the vision disease, but could have carrier. Individual 9 has limb defect so carries the gene, but Individual 10, could have the defective, gene this is unknown. Dvl to Mendals laws and Dihybrid crosses we know if the genes are not linked, the ratio would be 9:3:3:1. But if they are linked they could be any possible combination, but they aren't predictable due the the possible variation there is. In mendals law of segregation, he recognises that there are circumstances were genes are linked and if these genes are linked. Then we know that the recombinant DNA will be less than parental, in the offspring so they will be more

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parental DNA showing. ~~RATI~~ So therefore we can conclude that the ratio is for linked will be less then 50% for the RECOMBINANT DNA, but stronger for the parental DNA. The ratio could be anything ~~otem~~ depending on how far away or close the linkage is. But if linked the recombinant DNA would be smaller ratio.

- For example:

  Parental DNA (7) : Recombinant DNA (3)

  ∴ (7:3)

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(d)

(i) To determine linked genes, we can use Recombinant DNA technology, We can die a section of DNA by extracting a section of a gene (via restriction enzymes). Put it back into a cell and watch under a microscope, the transcription and translation. to help us identify how close the genes are actually together. We can do this many times and compare it with many secondary sources and other results. To try to hypothesise, the position of the linked gene and

(ii) how close they are together.

(1) Firstly the gene linkage maps, do show the relative position and how close the genes are together on the chromosome but they do not reveal them what each gene, actually does and what the gene's trait is. (which defeats their aim in the project of identifying the 30,000 genes.

(2) Secondly gene linkage does not actually reveal the base sequence of DNA which is what the Humane genome project was aiming to do. So therefore can't be used.

(3) Thirdly gene linkage maps don't deal with the ethnical side of the human genome project. They donated 3-5% of their budget in tacting their issue. The gene linkage simply shows how close the genes are it does not deal with the wider issue.

(e) Gene cloning, is the cloning of one particular gene, many times to form an organism of life. Gene cascade on the other hand, is the role of the HOX genes in producing an organism and how they develop the genes and limbs in proportion and in a specific linear order. The discoveries of these new techniques have resulted in the development of New Artifical life. with the knowledge of cascades and HOX genes, we have been able to study the way in which the organism grows and transforms. this knowledge has allowed us to monitor our new life that we have created to make sure that what we are creating is producing properly and efficently. What this does effectively is that it allows us to produce new life. new technologies to replicate this process. What we can also do now is take the HOX genes from other organisms and place them into the bacterium. We are able to do this, because with the knowledge that in some organisms. (ie mouse and fruit fly) These HOX genes are infact 100% identical.

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With this knowledge and these new technologies as a result we are artificially able to replicate the process's that are needed to start life. For example 'Gene cloning' has been used for many different things such as to produce Bt cotton and insulin for the human body. We are also able to produce sheep such as Dolly, through a different form of cloning rather whole organism cloning. The most important thing is that gene cloning and the understanding of gene cascades go hand in hand together. It is because of one that we are able to create the other.

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