Start here.

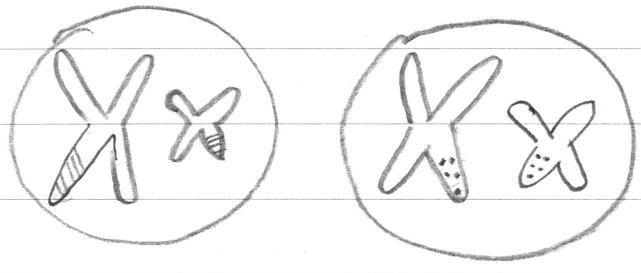
a)

Trisonomy	chronosomal eg: Downs aaitional chromosone added to pair 2n + 1 Syarome (trisonomy in chromosone 21)
Polyploidy	aaaitional ~~[illegible]~~ chromosone pair 2n
Base subshitution (gene mutation)	The alteration of a chromosonal nucleotide ~~specific~~ base sequence which codes for the particular functioning of a protein.

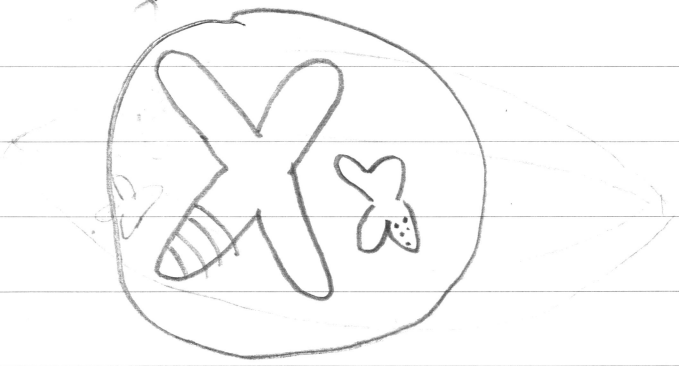
no

b)

Diplad = 2



Haploid = 1



c)

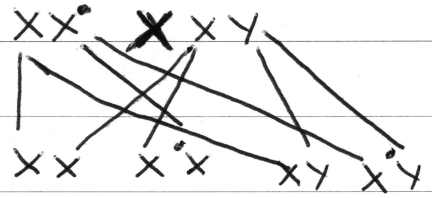
i)

vision defect: recessive

limb defect: ~~Recessive~~ Dominant

ii) Limb:

linked: XX° X XY



$XX = 25\%$

$X^{\circ}X = 25\% = 1:1$ ratio

$XY = 25\%$

$X^{\circ}Y = 25\%$

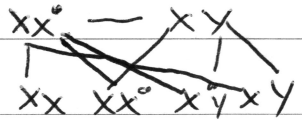
∴ it is a 50% chance that the child will have a ~~vision~~ limb defect

not linked: ~~50~~ % As mother is a ~~ca~~ infected

75 % chance as father could be a carrier and mother is infected.

Vision:

linked: XX X $X^{\circ}Y^{\circ}$



= 50 % chance

1 : 1

not linked: 25 % chance

1 : 2 : 1 → co-dominant / recessive.

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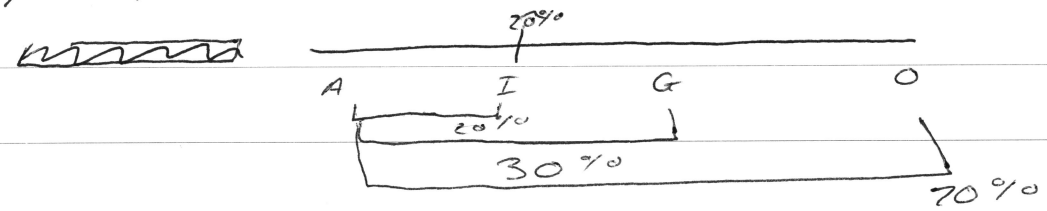
d)

i) Data can be collected through the use of linkage maps to determine the relative position ~~the inheritance patterns of the gene~~ i.e of linked genes ~~linkage maps~~ on the chromosome.

eg: 

etc. This use of linkage maps can be used to identify the position of linked genes when combined with the use of a pedigree to determine the relevant heredity of the gene.

ii)

1. The human genome is determined to map the entire chromosome, linkage maps only show the distance between genes not their position.
2. The human genome also seeks to determine the base pair of each chromosome and in function whereas again linkage only shows the relative distance of the genes not their location or purpose.
3. linkage maps do not identify the genes only their position.

The process of gene cloning is the process by which identical organisms are produced. This process occurs through embryonic splitting and nuclear transfer in which a sample of the original organism's DNA is extracted using restriction enzymes and is then inserted into the empty nucleus of a zygote and fertilised in pure culture creating a genetically identical organism. The development of such an organism has led to significant new applications for technology such as the development of recombinant DNA as referenced in the source and Gene therapy.

Through development such as gene cloning and gene cascades scientists have been able to develop the concept of recombinant DNA, a technology particularly significant to sufferers of diabetes. The procesn of recombinant DNA involves a bacterial plasmid in which a section of foreign DNA is cut using restriction enzymes and paired using annealing techniques and ligation to create for example insulin producing bacteria which with further development could decrease the incidence of diabetes in individuals.

These developments have also

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led to the development of whole artificial chromosones. Such technologies as this are significant to suffers for cystic fibrosis with the development of gene therapy. Gene therapy is the process of replacing defective genes with healthy ones from another healthy organism. Developments such as this have significantly impacted suffers of cystic fibrosis who are now able to receive somatic gene therapy of having the recombinant AVV virus dripped into their lungs gradually replacing defective genes causing the illness. Although such technology is only in its first stages the possibilities of such technology are only just begining as recombinant DNA may be able to be used to cure some of ~~(a book of time)~~ the most prelevant hereditary and somatic diseases.

(c

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