Genetics and inheritance grade 12 notes pdf

and polymerises in a template depended fashion following the rule of complementarity. It somehow also facilitates opening of the helix and continues elongation. Only a short stretch of RNA remains bound to the enzyme. Once the polymerases reaches the terminator region, the nascent RNA falls off, so also the RNA polymerase. This results in **termination** of transcription.

An intriguing question is that how is the RNA polymerases able to catalyse all the three steps, which are initiation, elongation and termination. The RNA polymerase is only capable of catalysing the process of elongation. It associates transiently with **initiation-factor** (σ) and **termination-factor** (ρ) to initiate and terminate the transcription, respectively. Association with these factors alter the specificity of the RNA polymerase to either initiate or terminate (Figure 6.10).

In bacteria, since the mRNA does not require any processing to become active, and also since transcription and translation take place in the same compartment (there is no separation of cytosol and nucleus in bacteria), many times the translation can begin much before the mRNA is fully transcribed. Consequently, the transcription and translation can be coupled in bacteria.

In eukaryotes, there are two additional complexities –

(i) There are at least three RNA polymerases in the nucleus (in addition to the RNA polymerase found in the organelles). There is a clear cut division of labour. The RNA polymerase I transcribes **rRNAs**

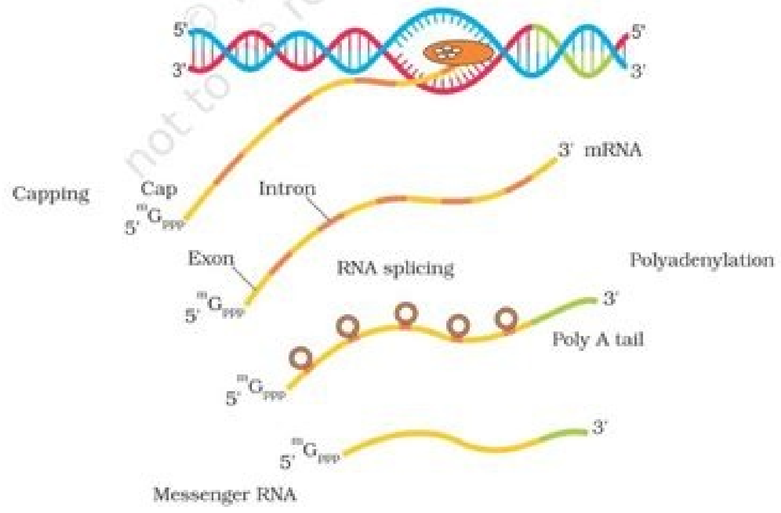


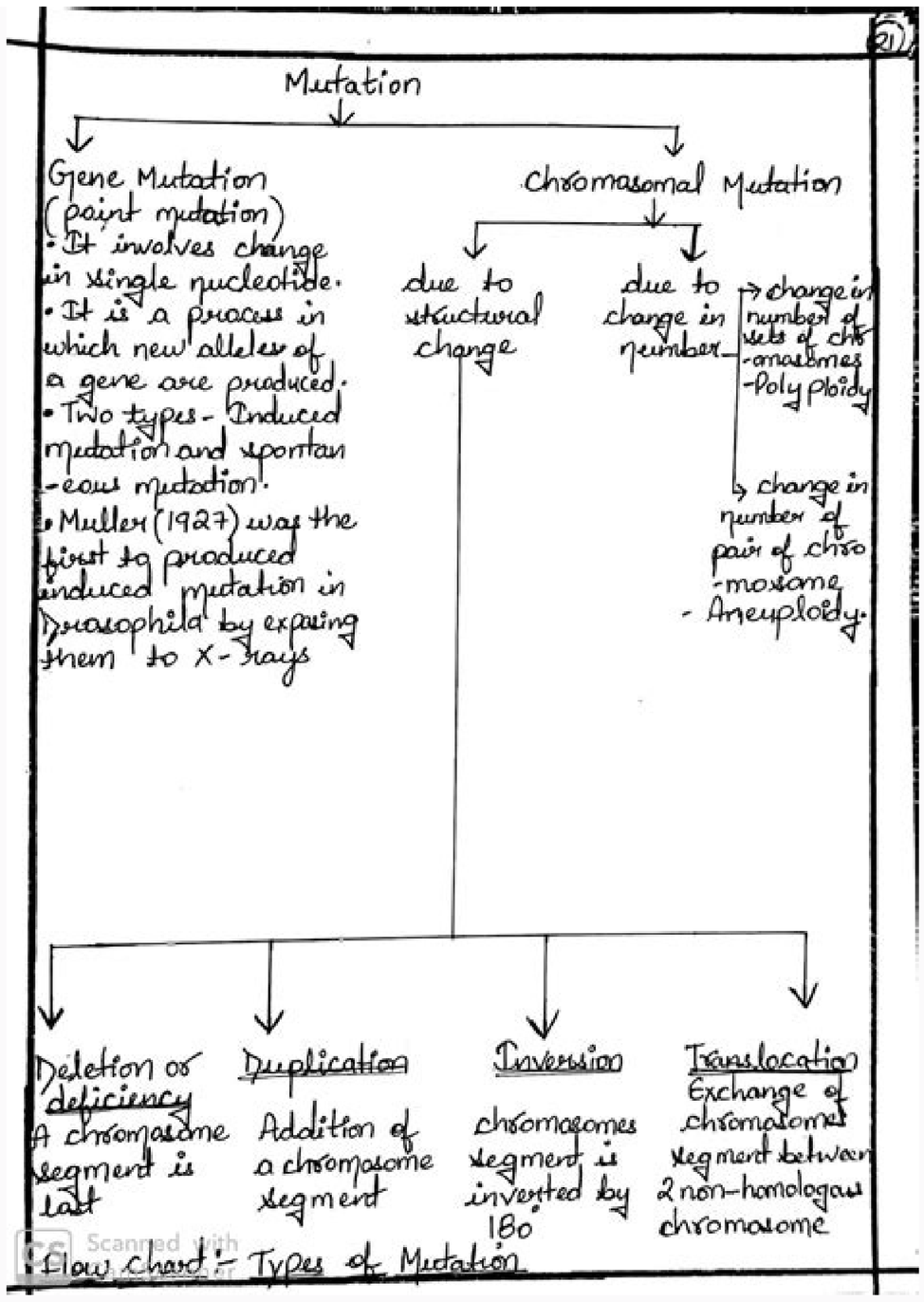
Figure 6.11 Process of Transcription in Eukaryotes

CHAPTER 6

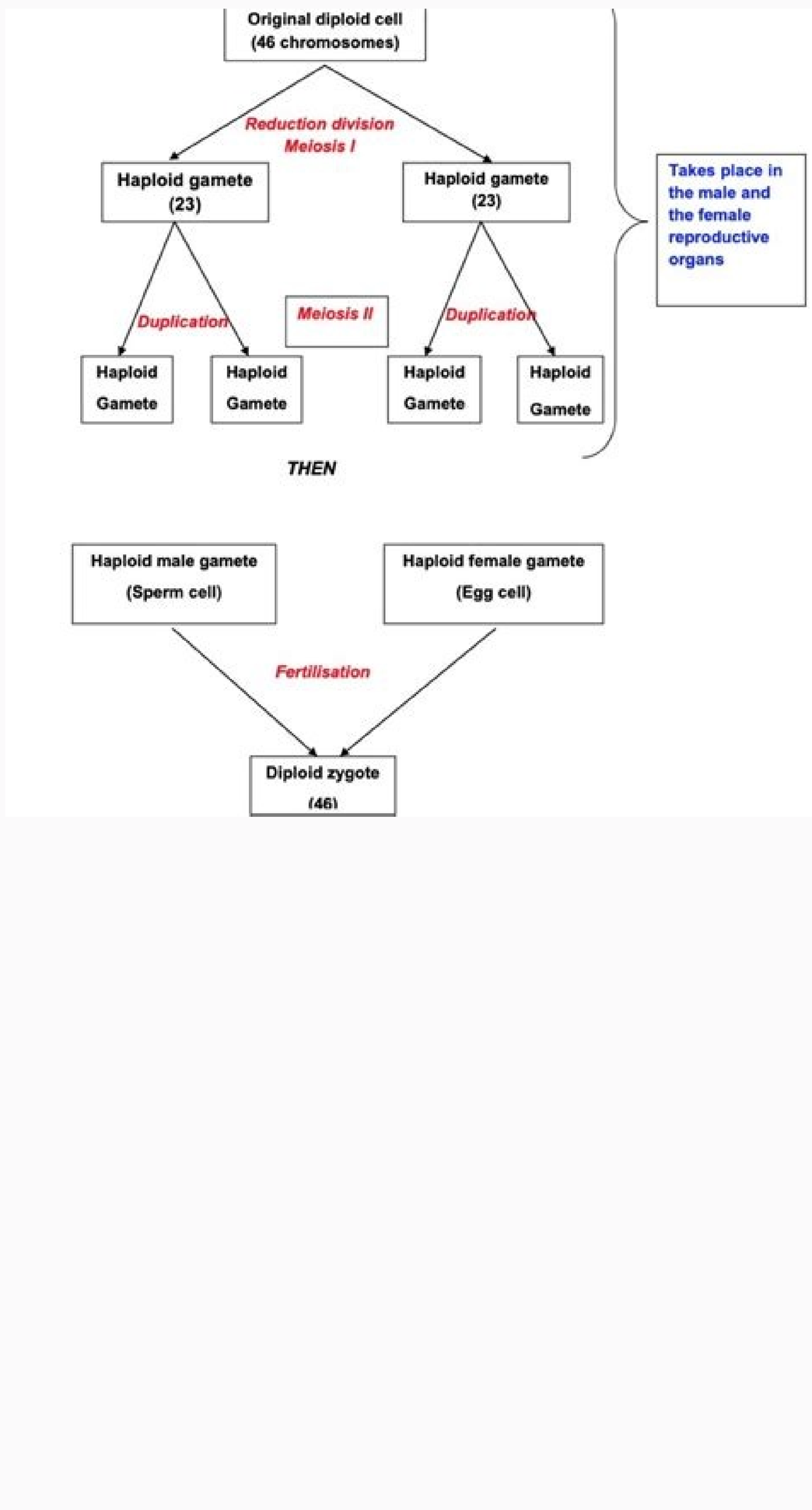
MOLECULAR BASIS OF INHERITANCE

In the previous chapter, you have learnt the inheritance patterns and the genetic basis of such patterns. At the time of Mendel, the nature of those 'factors' regulating the pattern of inheritance was not clear. Over the next hundred years, the nature of the putative genetic material was investigated culminating in the realisation that DNA – deoxyribonucleic acid – is the genetic material, at least for the majority of organisms. In class XI you have learnt that nucleic acids are polymers of nucleotides.

Deoxyribonucleic acid (DNA) and ribonucleic acid (RNA) are the two types of nucleic acids found in living systems. DNA acts as the genetic material in most of the organisms. RNA though it also acts as a genetic material in some viruses, mostly functions as a messenger. RNA has additional roles as well. It functions as adapter, structural, and in some cases as a catalytic molecule. In Class XI you have already learnt the structures of nucleotides and the way these monomer units are linked to form nucleic acid polymers. In this chapter we are going to discuss the structure of DNA, its replication, the process of making RNA from DNA (transcription), the genetic code that determines the sequences of amino acids in proteins, the process of protein synthesis (translation) and elementary basis of their regulation. The determination



Flow chart :- Types of Mutation



of complete nucleotide sequence of human genome during last decade has set in a new era of genomics. In the last section, the essentials of human genome sequencing and its consequences will also be discussed.

Let us begin our discussion by first understanding the structure of the most interesting molecule in the living system, that is, the DNA. In subsequent sections, we will understand that why it is the most abundant genetic material, and what its relationship is with RNA.

6.1 The DNA

DNA is a long polymer of deoxyribonucleotides. The length of DNA is usually defined as number of nucleotides (or a pair of nucleotide referred to as base pairs) present in it. This also is the characteristic of an organism. For example, a bacteriophage known as $\phi \times 174$ has 5386 nucleotides, Bacteriophage lambda has 48502 base pairs (bp), *Escherichia coli* has 4.6×10^6 bp, and haploid content of human DNA is 3.3×10^9 bp. Let us discuss the structure of such a long polymer.

6.1.1 Structure of Polynucleotide Chain

Let us recapitulate the chemical structure of a polynucleotide chain (DNA or RNA). A nucleotide has three components – a nitrogenous base, a pentose sugar (ribose in case of RNA, and deoxyribose for DNA), and a phosphate group. There are two types of nitrogenous bases – Purines (Adenine and Guanine), and Pyrimidines (Cytosine, Uracil and Thymine). Cytosine is common for both DNA and RNA and Thymine is present in DNA. Uracil is present in RNA at the place of Thymine. A nitrogenous base is linked to the OH of 1′C pentose sugar through a N-glycosidic linkage to form a nucleoside, such as adenosine or deoxyadenosine, guanosine or deoxyguanosine, cytidine or deoxycytidine and uridine or deoxythymidine. When a phosphate group is linked to OH of 5′C of a nucleoside through phosphoester linkage, a corresponding nucleotide (or deoxynucleotide depending upon the type of sugar present) is formed. Two nucleotides are linked through 3′-5′ phosphodiester linkage to form a dinucleotide. More nucleotides can be joined in such a manner to form a polynucleotide chain. A polymer thus formed has at one end a free

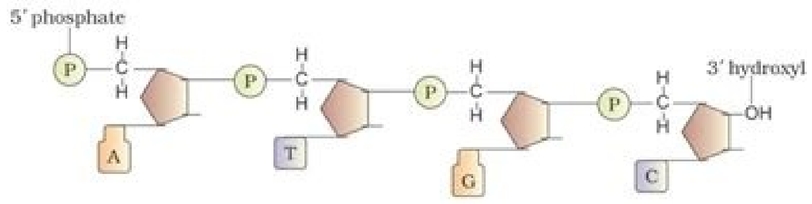


Figure 6.1 A Polynucleotide chain

Non genetic inheritance examples. What are examples of genetic inheritance. Genetics and inheritance grade 12 notes ppt. Genetics and inheritance grade 12 notes pdf. What is genetics and inheritance. Genetics and inheritance grade 12 summary.

A gene is a short length of DNA found on a chromosome that codes for a particular characteristic (expressed by the formation of different proteins) Alleles are variations of the same gene As we have two copies of each chromosome, we have two copies of each gene and therefore two alleles for each gene One of the alleles is inherited from the mother and the other from the father This means that the alleles do not have to 'say' the same thing For example, an individual has two copies of the gene for eye colour but one allele could code for brown eyes and one allele could code for blue eyes The observable characteristics of an organism (seen just by looking - like eye colour, or found – like blood type) is called the phenotype The combination of alleles that control each characteristic is called the genotype Alleles can be dominant or recessive A dominant allele only needs to be inherited from one parent in order for the characteristic to show up in the phenotype A recessive allele needs to be inherited from both parents in order for the characteristic to show up in the phenotype. If there is only one recessive allele, it will remain hidden and the dominant characteristic will show If the two alleles of a gene are the same, we describe the individual as being homozygous (homo = same) An individual could be homozygous dominant (having two copies of the dominant allele), or homozygous recessive (having two copies of the recessive allele) If the two alleles of a gene are different, we describe the individual as being heterozygous (hetero = different) When completing genetic diagrams, alleles are abbreviated to single letters The dominant allele is given a capital letter and the recessive allele is given the same letter, but lower caseAlleles are different forms of the same gene. You can only inherit two alleles for each gene, and they can be the same (homozygous) or different (heterozygous).We cannot always tell the genotype of an individual for a particular characteristic just by looking at the phenotype – a phenotype associated with a dominant allele will be seen in both a dominant homozygous and a dominant heterozygous genotype If two individuals who are both identically homozygous for a particular characteristic are bred together, they will produce offspring with exactly the same genotype and phenotype as the parents - we describe them as being 'pure breeding' as they will always produce offspring with the same characteristics A heterozygous individual can pass on different alleles for the same characteristic each time it is bred with any other individual and can therefore produce offspring with a different genotype and phenotype than the parents - as such, heterozygous individuals are not pure breedingKey Terms & Definitions for Genetic Inheritance Table Sex is determined by an entire chromosome pair (as opposed to most other characteristics that are just determined by one or a number of genes) Females have the sex chromosomes XX Males have the sex chromosomes XY As only a father can pass on a Y chromosome, he is responsible for determining the sex of the child Sperm cells determine the sex of offspring The inheritance of sex can be shown using a genetic diagram (known as a Punnett square), with the X and Y chromosomes taking the place of the alleles usually written in the boxesPunnett square showing the inheritance of sex On occasion, both alleles within a genotype are expressed in the phenotype of an individual - this is known as codominance Inheritance of blood group is an example of codominance There are three alleles of the gene governing this instead of the usual two I represents the gene and the superscript A, B and O represent the alleles Alleles IA and IB are codominant, but both are dominant to IO IA results in the production of antigen A in the blood IB results in the production of antigen B in the blood IO results in no antigens being produced in the blood These three possible alleles can give us the following genotypes and phenotypesBlood Phenotypes Table We can use genetic diagrams to predict the outcome of crosses that involve codominant alleles:'Show how a parent with blood group A and a parent with blood group B can produce offspring with blood group O' Punnett square showing the inheritance of Blood Group The parent with blood group A has the genotype IAIO The parent with the blood group B has the genotype IBIO We know these are their genotypes (as opposed to both being homozygous) as they are able to produce a child with blood group O and so the child must have inherited an allele for group O from each parent Parents with these blood types have a 25% chance of producing a child with blood type O Page 2Remember that the number of chromosomes found in each species differs, for example, humans have 23 pairs whereas dogs have 39 pairs and rice has 12 pairs.Page 3 GENETICS LIFE SCIENCES STUDY GUIDES AND NOTES GRADE 12 CHAPTER 5: GENETICS 5.1 Key concepts Make mobile notes (see instructions on page x) to learn these key concepts. Term Explanation Diagram/Additional notes Gene A small portion of DNA coding for a particular characteristic. Alleles Different forms of a gene which occur at the same locus (position) on homologous chromosomes. Genotype Genetic composition (make-up) of an organism. Phenotype The physical appearance of an organism determined by the genotype, e.g. tall, short. Dominant allele An allele that is expressed (shown) in the phenotype when found in the heterozygous (Tt) and homozygous (TT) condition. Recessive allele An allele that is masked (not shown) in the phenotype when found in the heterozygous (Tt) condition.It is only expressed in the homozygous (tt) condition. Heterozygous Two different alleles for a particular characteristic, e.g. Tt. Homozygous Two identical alleles for a particular characteristic, e.g. TT or tt. Term Explanation Diagram/Additional notes Monohybrid cross Only one characteristic or trait is being shown in the genetic cross. Example: Flower colour only, e.g. yellow flower or white flower or shape of seeds only, e.g. round seeds or wrinkled seeds. Complete dominance A genetic cross where the dominant allele masks (blocks) the expression of a recessive allele in the heterozygous condition. In this type of cross the allele for tall (T) is dominant over the allele for short (t). The offspring will therefore be tall because the dominant allele (T) masks the expression of the recessive allele (t). Incomplete dominance A genetic cross between two phenotypically different parents produces offspring different from both parents but with an intermediate phenotype. Example: If a red-flowered plant is crossed with a white-flowered plant and there is incomplete dominance - the offspring will have pink flowers (intermediate colour). Co-dominance A genetic cross in which both alleles are expressed equally in the phenotype. Example: If a red-flowered plant is crossed with a white-flowered plant and there is co-dominance the offspring has flowers with red and white patches. Multiple alleles More than two alternative forms of a gene at the same locus. Example: Blood groups are controlled by three alleles, namely IA, IB and i. Sex-linked characteristics Characteristics or traits that are carried on the sex chromosomes. Examples: Haemophilia and colour-blindness The alleles for haemophilia (or colour-blindness) are indicated as superscripts on the sex chromosomes, e.g. XHXH (normal female), XHXh (normal female), XhXh (female with haemophilia), XHY (normal male), XhY (male with haemophilia). Karyotype The number, shape and arrangement of all the chromosomes in the nucleus of a somatic cell. Cloning Process by which genetically identical organisms are formed using biotechnology. Example: Dolly the sheep was cloned using a diploid cell from one parent; therefore it had the identical genetic material of that parent. Genetic modification The manipulation of the genetic material of an organism to get desired changes. Example: The insertion of human insulin gene in plasmid of bacteria so that the bacteria produce human insulin. Human genome The mapping of the exact position of all the genes in all the chromosomes of a human. Example: Gene number 3 on chromosome number 4 is responsible for a particular characteristic. Activity 1 Choose an item from COLUMN 2 that matches a description in COLUMN 1.Write only the letter (A to I) next to the question number (1-5), for example 6. J. COLUMN 1 COLUMN 2 The allele that is not expressed in the phenotype when found in the heterozygous condition Different forms of a gene which occur at the same locus on homologous chromosomes A sex-linked condition where blood fails to clot properly The pair of chromosomes in a diploid organism that have the same size and shape and control the same set of characteristics The physical and functional expression of a gene A. GeneB. RecessiveC. HaemophiliaD. DominantE. HomologousF. GenotypeG. PhenotypeH. AllelesI. Karyotype [5] Answers to activity 1 B✔ H✔ C✔ E✔ G✔ (5 × 1) [5] 5.2 Genetic crosses Use the following genetic problem format or template to solve all monohybrid genetic problems: The problem on the next page shows that a cross between a heterozygous parent (Tt) and a homozygous recessive (tt) parent produces F1 offspring that are 50% heterozygous (Tt) and 50% homozygous recessive (tt). A cross between a homozygous dominant (TT) parent and a homozygous recessive (tt) parent produces F1 offspring that are 100% heterozygous (Tt). A cross between a homozygous dominant (TT) and a heterozygous (Tt) parent produces F1 offspring that are 50% homozygous dominant (TT) and 50% heterozygous (Tt). A cross between two heterozygous (Tt) parents produces F1 offspring that are 25% homozygous dominant (TT), 50% heterozygous (Tt) and 25% homozygous recessive (tt). 5.2.1 Complete dominance This refers to a genetic cross where the dominant allele masks (blocks) the expression of a recessive allele in the heterozygous condition.The following problem represents a genetic cross which shows complete dominance:e.g. Genetic problem 1In humans the ability to roll the tongue is due to a dominant allele. A man who is heterozygous for tongue-rolling and a woman who cannot roll her tongue have children. Use the symbols T and t for the alleles of the tongue-rolling characteristic and represent a genetic cross to determine the possible genotypes and phenotypes of the children. (6)Read the problem carefully and note the following steps: Identify the phenotypes of the man and the woman (parents/P1), i.e. the man is a tongue-roller and the woman is a non-tongue-roller Step 1 Identify the genotypes of the two parents, i.e. the man is heterozygous (Tt) and the woman can only be a non-tongue-roller if she is homozygous recessive for this characteristic, i.e. she must have the genotype (tt) Step 2 The next step is to show how the alleles are separated through the process of meiosis into separate gametes, i.e. in the man the gametes (sperm) will contain either the 'T' allele or the 't' allele.In the woman the egg can only contain the 't' allele Step 3 The next step shows that fertilisation takes place. Indicate all possible combinations of how sperm cells fuse with a possible egg cell to show the possible genotypes of the F1 generation that could arise Step 4 Interpret the phenotypes of all the possible genotypes from the cross Step 5 Solution to genetic problem 1 5.2.2 Incomplete dominance This refers to a genetic cross between two phenotypically different parents producing an offspring different from both parents but with an intermediate phenotype. The following problem represents a genetic cross that shows incomplete dominance. e.g. Genetic problem 2A homozygous snapdragon plant with red flowers (R) was cross-pollinated with a homozygous snapdragon plant with white (W) flowers. All the plants that grew from the cross had pink flowers. Represent a genetic cross to show the possible genotypes and phenotypes of the F1 generation of plants. Solution to genetic problem 2 5.2.3 Co-dominance This refers to a genetic cross in which both alleles are equally expressed in the phenotype.The following problem represents a genetic cross which shows co- dominance. e.g. Genetic problem 3A plant with white flowers was cross-pollinated with a plant with red flowers. All the plants that grew from the cross had flowers with equal distribution of red and white colour. Represent a genetic cross to show the possible genotypes and phenotypes of the F1 generation of plants. Solution to genetic problem 3 5.2.4 Inheritance of sex The following problem represents a genetic cross which shows inheritance of sex. e.g. Genetic problem 4A couple has three sons and the woman is pregnant again. Show diagrammatically by means of a genetic cross what the percentage chance is of the couple having a baby girl. Solution to genetic problem 4 5.2.5 Inheritance of sex-linked characteristics Sex-linked characteristics are characteristics (traits) that are carried on the sex chromosomes.The following problem represents a genetic cross which shows the inheritance of sex-linked characteristics. e.g. Genetic problem 5Haemophilia is a sex-linked hereditary disease that occurs as a result of a recessive allele on the X-chromosome (Xh). A normal father and heterozygous normal mother have children. Represent a genetic cross to determine the possible genotypes and phenotypes of their children.The alleles for haemophilia are indicated as superscripts on the sex chromosomes,e.g. XHXH (normal female), XHXh (carrier/heterozygous normal female), XhXh (female with haemophilia), XHY (normal male), XhY (male with haemophilia). Solution to genetic problem 5 Activity 2 Question 1Try solving this problem on your own before you look at the solution.Fur colour in mice is controlled by a gene with two alleles. A homozygous mouse with black fur was crossed with a homozygous mouse with brown fur. All offspring had black fur. Using the symbols B and b to represent the two alleles for fur colour, show diagrammatically a genetic cross between a mouse that is heterozygous for fur colour and a mouse with brown fur. Show the possible genotypes and phenotypes of the offspring. (6) Question 2In rabbits the dominant allele (B) produces black fur and the recessive allele (b) produces white fur. Use a genetic cross to show the possible phenotypes and genotypes of the F1 generation for fur colour if two heterozygous rabbits are crossed. (6) 5.2.6 Dihybrid cross A dihybrid cross involves the inheritance of two characteristics. Mendel explained the results obtained from dihybrid crosses according to his Law of Independent Assortment. According to the Law of Independent Assortment, alleles of a gene for one characteristic segregate independently of the alleles of a gene for another characteristic. The alleles for the two genes will therefore come together randomly during gamete formation. This means that the two characteristics are transmitted to the offspring independently of one another. The above law only applies if the genes for the two characteristics are not on the same chromosome. Steps you should follow in working out a dihybrid cross:e.g. ExampleIn pea plants, the allele for tallness (T) is dominant and the allele for shortness (t) is recessive. The allele for purple flowers is dominant (P) and the allele for white flowers is recessive (p). Two plants, heterozygous for both tallness and purple flowers, were crossed. STEP What to do generally What to do in this problem Step 1 Identify the phenotypes of the two plants for each of the two characteristics. According to the statement of the problem, both parents are tall and have purple flowers. Step 2 Choose letters to represent the alleles for the gene responsible for each characteristic. Use the letters, e.g. T for tall, t for short, P for purple, and p for white as provided in the question. Step 3 Write the genotypes of each parent. According to the statement of the problem, both parents are heterozygous for each characteristic. Their genotype will therefore be TtPp. Step 4 Determine the possible gametes that each parent can produce. Remember that each parent will have two alleles for each gene. The gametes of each parent will have only one allele for each gene because of segregation during meiosis Remember that because of the principle of independent assortment an allele for one gene could appear in the same gamete with any of the alleles for the other gene. Each parent has the genotype TtPp. If we represent the alleles for each gene in the following format, then we can see how these alleles could come together randomly (principle of independent assortment) to form the four types of gametes: TP; Tp; tP and tp as shown below. Alleles T t P TP tP p Tp tp Step 5 Enter the possible gametes at the top and side of a Punnett square. Please refer to the solution that follows. Step 6 Because of random fertilisation, gametes from both parents could fuse in different combinations to form the offspring In the punnet square, write down the genotypes of the offspring that will result from each possible combination of gametes. Please refer to the solution that follows. Step 7 Determine the phenotypes of the offspring from the genotypes obtained in the punnet square. Please refer to the solution that follows. Solution to the problemP1 Phenotype Tall, Purple × Tall, Purple..... Step 1 Genotype TtPp × TtPp..... Step 2,3 F1 Genotype 9 different genotypes, as in the table above Phenotype 9 tall, purple flowered plants (T-P-); 3 short, purple flowered plants (ttP-); 3 tall, white flowered plants (T-pp), and 1 short, white flowered plant (ttpp) . Step 7 Activity 3 QuestionIn hamsters, the allele for black coat colour (B) is dominant over the allele for white coat colour (b). The allele for rough coat (R) is dominant over the allele for smooth coat (r). If you cross a hamster that is heterozygous black and homozygous rough, with one that is heterozygous black and heterozygous rough, what will be the phenotypes and genotypes of the offspring? (Use the steps 1-7 to arrive at an answer). 5.3 Mutations A mutation is any sudden unexpected change in the genetic structure of a cell. Mutations occur suddenly and randomly and may be caused by many environmental agents such as X-rays and certain chemicals.Mutations may be harmful or harmless to the organism in which they occur. Harmful mutations cause changes in DNA that can cause errors in protein sequencing, that can result in partially or completely non-functional proteins. Harmless mutations have no effect on the structure or functioning of the organism. Useful mutations can be advantageous to the organism and they are passed on from parent to offspring.Gene mutations are mutations that affect a single or a few base pairs in just a single gene, while Chromosomal aberrations refer to changes in the normal structure or number of chromosomes.Mutations result in new genotypes as we move from one generation to the next. This leads to variation within a species.Gene mutations can cause genetic disorders: Haemophilia: Absence of the protein needed for the formation of blood clots due to a mutant gene. Colour blindness: Absence of the proteins that make up either the red or green cones/photoreceptors in the eye. Albinism: Absence of the protein that forms the pigment melanin. Chromosomal aberrations e.g. Down syndrome is where there is an extra chromosome (47 instead of 46) in the zygote. 5.4 Pedigree diagrams A pedigree diagram is used to study the inheritance of characteristics in a family over a number of generations. A pedigree diagram is also called a family tree. Remember the following steps when interpreting pedigree diagrams:Step 1 Study any key and opening statement/s and look for dominant and recessive characteristics and phenotypes.Step 2 Write in the phenotypes of all the individuals as given in the problem.Step 3 Fill in the genotype of all the individuals with the recessive condition - it must have two recessive alleles (two lower case letters, e.g. ff).Step 4 For every individual in the diagram that has the recessive condition, it means that each allele was obtained from each of the parents. Work backwards and fill in one recessive allele for each parent.Step 5 If the parents showed the dominant characteristic, fill in the second letter which represents the dominant allele (a capital letter, e.g. F).Step 6 Any other individual showing the dominant characteristic will most likely be homozygous dominant (FF) or heterozygous dominant (Ff). Activity 4 The pedigree diagram in Figure 5.1 shows inheritance of eye colour in humans over three generations of a family. Brown eye colour (B) is dominant over blue eye colour (b). Study the diagram and then answer the questions that follow. Figure 5.1 Pedigree diagram showing inheritance of eye colourNote the following in the pedigree diagram on page 38: Squares represent males and circles represent females. The horizontal line between a square (Joshua) and a circle (Ronel) shows that they have mated. The vertical line flowing from the horizontal line represents the offspring (Sarah and Peter) of the two parents (Joshua and Ronel). Brown eye colour (B) is dominant over blue eye colour (b) - stated in problem Step 1 Joshua, Jack and John are males with blue eyes. Veronica and Marlena are females with blue eyes. Peter and Frank are males with brown eyes. Step 2 Ronel, Sarah and Gayle are females with brown eyes. Joshua, Veronica, Marlena, Jack and John will have the genotype 'bb'. The recessive characteristic only shows up in the homozygous condition Step 3 Example: The genotype of Peter is 'Bb' - working backwards from the offspring Marlena or Jack or John who are homozygous recessive.This means that one of the recessive alleles of Marlena, Jack and John, i.e. 'b', must have come from parent Peter and the other one from parent Veronica Steps 4 and 5 Ronel could be homozygous dominant (BB) or heterozygous dominant (Bb) Step 6 Questions1. How many members of the family have blue eyes? (1)2. Is Veronica homozygous or heterozygous for eye colour? (1)3. Write down the genotype of: a) Joshua (2)b) Ronel (2)c) Frank (2) 4. If Frank marries a woman with the same genetic composition as Sarah, what is the percentage probability of them having a child with brown eyes? (1) [9] Answers to activity 41. 5✔ (1)2. Homozygous✔(1)3. a) bb✔✔ (2)b) BB/Bb✔✔ (2)c) Bb✔✔ (2)4. 75%✔ 5.5 Genetic engineering Genetic engineering is the process whereby the genes on the DNA are changed, transferred or manipulated to produce a different organism. Activity 5 QuestionState FOUR disadvantages and FOUR advantages of genetic engineering. [8] Answer to activity 5Four disadvantages of genetic engineering: Expensive✔/research money could be used for other needs Interfering with nature✔/immoral Potential health impacts✔ Unsure of long-term effects✔ (4) Four advantages of genetic engineering: Production of medication/resources cheaply✔ Control pests with specific genes inserted into a crop✔ Using specific genes to increase crop yields✔/food security Selecting genes to increase shelf-life of plant products✔ (4) [8] 5.6 Genetic counselling Couples with a risk of a genetic disease can undergo genetic counselling to enable them to make informed decisions on whether they want to have children or not. Activity 6 QuestionA young couple wants to have a child, but they are aware of a serious genetic disorder in one of their families that could be carried through to their offspring. State THREE benefits of genetic counselling in this case. [3] Answer to activity 6Three benefits of genetic counselling: To be given advice on the risk of transferring the defective gene✔/ to find the probability of passing on the defective gene to the offspring To be given an explanation of the procedure involved in DNA testing✔ To be given an explanation of the results of DNA testing✔ [3]

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