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Chapter-23	Chromosomes and DNA	
Q1 what is chromosome. write its discovery and numbers.		
Answers: chromosomes:		
<u>Meaning</u> :- chromo mean coloured and soma mean body.		
<u>Definition</u> :- chromosomes are thread like coloured structures produced from chromatin material, appears inside the nucleus at the time of cell-division.		
<u>Discovery of chromosomes</u> :-		
1: <u>W. Fleming</u> :- chromosomes were first observed by the German embryologist Walter-Fleming in 1882. He discovered the chromosome by examining the rapidly dividing cells of Salamander Larvae. Since their discovery, chromosomes have been found in the cells of all eukaryotes, But in prokaryotes single DNA is also referred as chromosome.		
2: <u>Waldyse</u> :- The term chromosome was proposed by - Waldyse which literally means coloured bodies.		
3: <u>Sutton</u> :- In 1908 Sutton identified the function of chromosomes that they take part in the transportation of hereditary characters.		
<u>Number of chromosome</u> :-		
chromosomes are found in the cells of all eukaryotes. Their number however varies from species to species. While in case of prokaryotes there is single chromosome in a cell.		
<u>For example</u> :- (ETEA-2009, 2011)		

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Species	chromosome No	Species	chromosome No	Species	chromosome No
penicillium	2 (1 pair)	Frog	26 (13) 32 (16)	Dog	78 (39)
mosquito	6 (3 n)	Honey bee	38 (19)	Sugar cane	80 (40)
Fruit fly	8 (4 n)	cat	40 (20)	protozoa	300+ (500)
pea	14 (7)	mouse	46 (23)	Fern	1000 (500)
<u>Terminology related with numbers of chromosomes:-</u>					
on the basis of chromosome number cell or organism may be -					
<u>1: Haploid : - (n) -</u> When a cell or species have single (one) set or half Number of chromosomes is called haploid (n).					
<u>Example :-</u> Gametes and spores are usually haploid cells because they contain half numbers of chromosomes.					
<u>2: Diploid : - (2n) -</u> When a cell or species have two set of chromosomes is called diploid (2n).					
<u>Example :-</u> Human somatic cells contains two set of chromosomes (46-chromosomes) and each set containing 23-chromosomes.					
<u>3: Polyploidy : - (3n, 4n, 5n, 6n, ...)</u> When a cell or species have more than two set of chromosomes is called polyploidy (xn). Polyploidy is found in plants and not in animals because it cause sterility in animals.					
<u>Example :-</u> Triplid - having 3-sets of chromosomes and so no.					
<u>4: Monoploidy : - (x) -</u> Number of chromosomes in which distinction of haploid, diploid and polyploidy is not present is known as monoploidy.					
Q2 Describe the structure of chromosome.					
Answer: <u>structure of chromosome :-</u> The structure of -					

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chromosome is explain with the help of the following heading.

1: components of chromosome :-

structurally the chromosome consists of following parts.

i-chromatids :- A visible and prominent chromosome consists of two arms or thread like structures called chromatids. each chromatid is made up of a long DNA molecule which is highly coiled along with histone proteins.

ii-centromere or primary constriction :- Both the chromatids are connected with each other by a small rounded body -- known as centromere or primary constriction or Kinetochore.

iii-secondary constriction or Nuclear organizer :-

Some chromosomes also contain an additional part or another point of union along the length of chromatids called Secondary constriction. It is associated with the nucleolus i.e- it gives rise to nucleolus during interphase so it is also called nuclear organizers. At least one pair of homologous chromosomes possess nuclear organizer region.

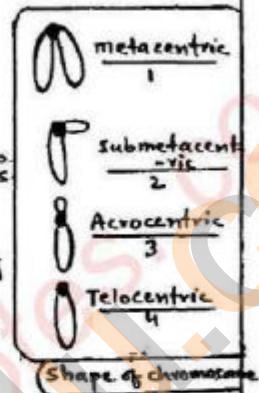
iv-satellite body :- Beside the secondary constriction the remaining part of chromosome is very short and knob like called satellite. satellite is connected to chromatid through secondary constriction. The satellite has a useless sequence of DNA called junk-DNA, which provides support.

parts of chromosome:

The diagram shows a single chromosome consisting of two sister chromatids joined at a central point labeled 'centromere'. A small, distinct loop or knob-like structure extends from one side of the chromosome, labeled 'satellite'. The region near the centromere where the two chromatids meet is labeled 'secondary constriction'.

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<p><u>V-Telomeres</u> :- It is the polar part or end portion of chromosome. It does not allow the attachment of another chromosome.</p>		
<p><u>2: Shape of chromosome</u> :-</p>		
<p>The chromosomes acquire different shapes at the time of anaphase during cell division. The usual shapes are - i, j, L, V.</p>		
<p>Shapes of chromosomes depend upon the location of centromere.</p>		
<p><u>Types of Shape of chromosome</u> :- Based on location of the centromere the chromosomes are of following types.</p>		
<p><u>i-Metacentric</u> :- Those chromosomes having centromere at the centre of two chromatids are called metacentric. These chromosomes acquire V-shape during cell division.</p>		
<p><u>Example</u> :- Metacentric chromosomes in human is - 1, 2, 3, 19, 20.</p>		
<p><u>ii- Sub metacentric</u> :- Those chromosomes having centromere little above or below the centre of two chromatids are called sub-metacentric. These chromosomes acquire L-shape during cell division.</p>		
<p><u>Example</u> :- sub-metacentric chromosomes in human is - 4, 5, 6, 7, 8, 9, 10, 11, 12, 16, 17, 18 and X-chromosome.</p>		
<p><u>iii-Accentric</u> :- Those chromosomes having centromere near the end of chromatid with one large and one short chromatid are called accentric chromosome. These chromosomes acquire J-shape during cell division.</p>		
<p><u>Example</u> :- Accentric chromosomes in human is - 13, 14, 15, 21, 22, Y.</p>		
<p><u>iv-Telocentric</u> :- Those chromosomes having centromere at the tip of chromosome (chromatid) are called Telocentric.</p>		

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<p>These chromosomes acquire I-shape during cell division.</p>		
<p><u>Example:-</u> Humans have no Telocentric chromosomes.</p>		
<p><u>3: Chemical composition of chromosome :-</u></p>		
<p>Chromosomes are mainly composed of DNA and protein called nucleoprotein. A Typical chromosome has the following composition.</p>		
<p>1- DNA — 40% 2- protein — 60%.</p>		
<p>3- RNA — a small amount of RNA is also associated with chromosomes.</p>		
<p><u>i: DNA component of chromosome :-</u> DNA of a chromosome is one very long, double stranded fiber. It extends unbroken through the entire length of the chromosome.</p>		
<p><u>Number of DNA :-</u> Each chromosome has two equal DNA molecules i.e. one in each chromatid and both are connected through DNA of centromere.</p>		
<p><u>Average length :-</u> Average sized human chromosome has approximately 5 cm long DNA which consist of about 140 million nucleotides or base pairs.</p>		
<p><u>charge on DNA :-</u> It is negatively charged due to phosphate group of nucleotides.</p>		
<p><u>Function of DNA :-</u> DNA is the most important component of chromosome because it controls heredity and act a data bank of life.</p>		
<p><u>ii: Histones component of chromosome :-</u> (ETEA-2009)</p>		
<p>Histones are basic proteins due to presence of some basic amino-acids like arginine and lysine which gives positive -</p>		



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<p>charge to histones. Histones and DNA makes the nucleosome, because the histones being positive charged and DNA being negative charged have strong affinity between them to remain intact with each other.</p>		
<p><u>Types of Histones</u> :- These are five (5) types of histones - which have been differently designated according to method of isolation. These are - H₁, H_{2A}, H_{2B}, H₃, H₄.</p>		
<p>1: Histone - H₁ is most easily removed. These are concerned with holding together a chromosome fibre.</p>		
<p>2: Histone - H₃ and H₄ can not be easily removed, they have structural role in chromosome like H_{2A} and H_{2B} role.</p>		
<p><u>Function of Histones</u> :- Histone - proteins play a structural role rather than regulatory role. However, according to some experiments conducted in recent years, some variants of histones are associated with the regulation of gene expression.</p>		
<p>iii: <u>Non-Histone</u> :- a small amount of non-Histones proteins are also found in chromosome which regulate the activity of specific genes i.e. They switch on specific genes.</p>		
<p>4: <u>Organization of chromosome</u> : (ETEA-2012, 2020)</p> <p>During S-phase of cell-cycle DNA and histones are completely disorganized from each other. after DNA-replication both DNA and histones begin to organize again and such process of condensation remains continue till the cell undergoes division and the chromosomes are appeared.</p>		

Levels of chromosomal organization : (ETEA-2005)

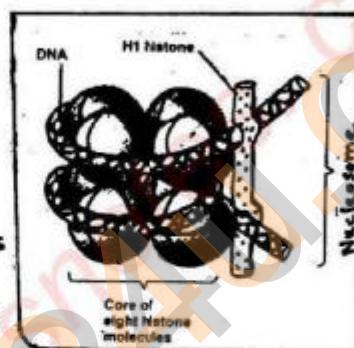
The organization of chromosomes occurs in 4-levels. i.e.—

i: Nucleosome :— chromosome is —

composed of repeating units called — nucleosome which consists of —

Duplex DNA and Histone :— Each —

Nucleosome is composed of 8-histones proteins i.e.— TWO of each H₂A, H₂B, H₃, and H₄. Octameric protein is surrounded



by two turns of about 200-nucleotides of double helical — DNA. Two turns of DNA are stabilized on nucleosome by H₁- Histone protein act as seal or stapler. ← (ETEA-2019)

Core DNA and linker DNA :— The DNA which surround the — Octameric histone proteins is known as core DNA (200 nucleotides) while the DNA in between two nucleosomes is called linker DNA.

ii: Nucleosome string :— The nucleosomes are linked together like beads on a string and such string is called nucleosome string.

Diameter :— Nucleosome string is about 10 nm thick and turn of DNA around the histones is 2nm thick.

iii: Chromatin fiber :— (solenoid) immediately the — nucleosome string begins to coil again about its axis to form further thick fiber called chromatin fiber. During G₁ and G₂-phases of interphase, chromosomes are found in this level of organization.

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<p><u>Diameter</u> :- chromatin fiber is about 30 nm thick.</p>		
<p><u>Parts of chromatin fiber</u> :- The chromatin fiber show 2-regions</p>		
<p>1- <u>Heterochromatin</u> :- Highly condensed or dark portions of chromatin fiber are called heterochromatin. some of these portions remain permanently condensed so that their DNA never expressed or mean it contains inactive DNA (introns).</p>		
<p>2- <u>Euchromatin</u> :- Less condensed or light portions of chromatin fiber are called Euchromatin. It is temporarily condensed during cell division so that a uniform chromatin fiber is established. It contains active DNA (exons).</p>		
<p><u>iv: Chromatid</u> :-</p>		
<p><u>Supercell</u> :- when cell division begins the higher order coiling of chromatin fiber gives to supercoil which has a diameter of 200 nm.</p>		
<p><u>chromatid</u> :- super coiling continues and at last much coiled and condensed structure of 700nm is formed called — chromatid.</p>		
<p><u>Q3</u> what is gene. write its Historical background and structure</p>		
<p><u>Answer</u> : Gene :</p>		
<p><u>Definition</u> :- A part or segment of DNA which can control a single specific character during development of an individual is called gene. —OR— "A short segment of DNA which is responsible for the formation of a particular polypeptide chain or protein is known as gene".</p>		
<p><u>Historical background</u> :- Different views of different —</p>		

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Biologist about gene are given below -

1: Charles Darwin :- In 1868, Charles Darwin proposed the theory of pangenesis. According to this theory there are specific substance circulating in blood called as Germmules. These Germmules contribute to sex (germ) cells and then transmit characters to next generation. Hence, he declared that Germmules are the units of heredity.

2: Francis Galton :- In 1870, Pangenesis was disapproved by F. Galton. He used blood transfusion in rabbits to show that transfused Germmules molecules does not affect the heredity of recipient rabbit. He came to the conclusion - that Germmules are not units of heredity and hence he rejected the theory of pangenesis of Charles Darwin.

3: Hugo De Vries :- In 1890, Hugo De Vries reduces the term pangenesis to pangen as unit of inheritance. He said that pangenes do not circulate in the blood and not migrate from one cell to another as said by Charles Darwin. He argued that pangenes remain inside the cell. In this way he finds that Mendel had discovered 30-years earlier with contrasting traits in garden pea plants that these are units of inheritance called factors which transmitted from parents to offspring during reproduction.

4: W. Johansson :- Later on, in 1909 Johansson introduce the term gene as a unit of inheritance or heredity.

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S: W. Bateson :- Three years prior than gene term (i.e-1906) the term genetics was proposed by W. Bateson and opened a new field of biology in the form of heredity, variation and evolution.
Modern concept of gene:

This concept was put forward by Mendel when he was — performing experiment on pea plant. According to this concept each trait in the pea plant is controlled by separate units called as factors or elements. These factors are now — considered as gene which is a segment of DNA having sequence of nucleotides for the sequence of amino-acids of particular polypeptide.

Where do gene reside :— (Gene location) — Genes are present on chromosome in linear fashion. The location of gene on — the chromosome is called locus. most genes exist in — alternate forms called alleles. alleles responsible for the same character are always present on the same locus.

For example :— The gene of ABO-blood group is I. it is polymorphic gene because it has 3-different alleles - I^A , I^B , i. These 3-alleles lie on the same locus at chromosome No-9.

Structure of gene :—

Gene is a segment of DNA which consists of 2-regions i.e-

1: Regulatory region :— This region again consists of 2-parts.

I-promoter :— promoter is the site where RNA-polymerase enzyme bind during transcription. promoter is located to the

5'-end of coding strand of a gene.

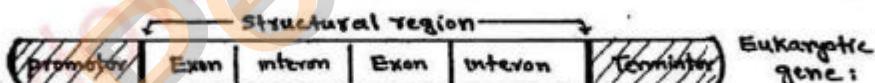
ii-Terminator :- Terminator is the region where RNA-polymerase enzyme stop transcription. Terminator is located on 3'-end of coding strand of the gene.

2: Structural Region :- Structural region is present between the promoter and terminator region of a gene. It is second part of gene which is also called open reading frame. This region posses genetic information in the form of Triplet code or genetic code. Structural region contains exon <functional region or sequences> or intron <non-functional sequence> or both.

In Eukaryotes :- Eukaryotic genes are monocistronic as - they posses exon and intron in structural region of the gene.

1-Exon :- The functional sequence of nucleotides on DNA is called exon. It is the active part of DNA. It forms only 2% of DNA.

2-Intron :- The non-functional sequence of nucleotides on DNA is called intron. It is inactive part of DNA. It forms about 98% of DNA.

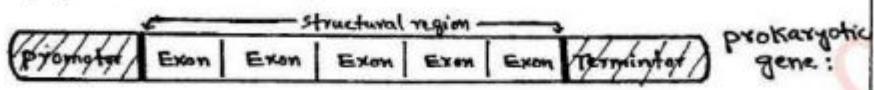


In prokaryotes :- The genes of prokaryotes are present in the form of clusture called as operon. prokaryotic gene are - polycistronic as they possess continuous exon in structural region of the gene.

Operon :- many adjacent structural genes or regions that can synthesize different polypeptides by the single promoter

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and single terminator region in a coordinated way and such - group of genes is called operon.


Diagram illustrating a prokaryotic gene structure. It shows a promoter region followed by a structural region composed of multiple exons. The structural region is labeled "Structural region" with arrows above and below it. The entire unit is enclosed in brackets and labeled "Prokaryotic gene".

Q4 Explain the chromosomal theory of inheritance.

Answer: chromosomal theory of inheritance:

Introduction :- The chromosomal theory of inheritance is the idea that genes (units of inheritance) are found in the chromosomes so chromosomes act as carriers of heredity.

History of chromosomal theory of inheritance:

1: Karl Correns :- The chromosomal theory of inheritance idea was put forward in 1900 for the first time by German geneticist - Karl Correns in one of his papers announcing the rediscovery of Mendel's work, but he had no supportive evidence for this at that time.

2: W. Sutton and T. Boveri :- The actual credit of chromosomal theory of inheritance goes to both Walter Sutton (an American Biologist) and Theodor Boveri (a German biologist). In 1902, these scientists recognized independently that the behaviour of Mendel's factors (gene) is parallel to the behaviour of chromosome at meiosis i.e -

Parallel behaviour of gene and chromosomes:

During meiosis the behaviour of genes are same or parallel to the behaviour of chromosomes as following -

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<u>Beh; of chromosomes:</u>		<u>Beh; of genes:</u>
1: Before meiosis diploid cell ($2n$) possess two copies of each - chromosome (Homologous Chrom)		1: Before meiosis, diploid cell possess two copies of each gene. For example - In pea plant - diploid cells have a pair of - each gene like - Rr, Yy, Tt, etc before meiosis.
For example - in pea plant there are 7-pairs of chromosomes - before meiosis.		2: Gametes after meiosis have only copy of chromosome. For example - after meiosis gametes of pea plant have only 7-chromosomes instead of 7-pairs
2: Gametes after meiosis have only copy of chromosome. For example - after meiosis gametes of pea plant have only 7-chromosomes instead of 7-pairs		2: Gametes after meiosis have only copy of gene. For example - after meiosis the gametes of pea plant have only one allele - R or r, Y or y, etc.
3: Homologous chromosome pair separate during meiosis.		3: Pairs of gene for each trait also segregate during meiosis.
4: Each homologous member of pairs independently assort during meiosis & inheritance. e.g - $XX \rightarrow \otimes \otimes$		4: Each gene pair or alleles also independently assort from other members during meiosis/inheritance e.g - $RrYy \rightarrow \textcircled{R} \textcircled{Y} \textcircled{R} \textcircled{Y} \textcircled{r} \textcircled{Y} \textcircled{r} \textcircled{Y}$
5: In fertilization, the gametes unite and restore the original number of chromosomes.		5: In Fertilization, gametes unite and restore the original - number of genes or alleles.
6: The individual chromosomes remains constant from one generation to the next.		6: The individual genes also remains unchanged from one generation to the next.

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<p><u>Mechanism of Sexual Reproduction :-</u> In addition to the above parallel behaviours of genes to chromosomes we can also analyze that mechanism of sexual reproduction which involves egg and sperm. i.e—</p>		
<p>1: If Mendel's work is correct then each gamete must make equal heredity contribution. Although Sperm had much less cytoplasm but its contribution of nucleus is same as that of egg.</p>		
<p>2: So, it means that chromosomes are present in nucleus which contains genes which indicates that genes are located at chromosomes.</p>		
<p><u>Objection on Sutton's work :-</u> Many investigators of that time pointed out a serious objection on Sutton's theory or work. i.e “The numbers of genes (characters) assort independently in a given kind of organism often greatly exceed than the number of chromosomes of that organism.” ← (ETEA-2014)</p>		
<p><u>Solution of the objection :-</u> Later on, in 1910, it was cleared after the experimental work of T.H. Morgan on Drosophila, that many genes are carry by a single chromosome. e.g – sex-linkage and inheritance of eye-colours of drosophila.</p>		
<p>Q5 prove an evidence of DNA as heredity material.</p>		
<p>Answer: DNA as a Heredity material:</p>		
<p><u>Heredity material :-</u> The material which transfers the characters from parents to offspring is known as heredity</p>		

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<p>material. So, DNA act as heredity material because it transfers the characters from parent to offsprings.</p>		
<p><u>Explanation :-</u> Chromosomes are composed of DNA and proteins. To know whether DNA or protein is the heredity material, biologists performed a series of experiments. It was confirmed through the following experiments that DNA is the heredity material. i.e.—</p>		
<p><u>1: Griffith's experiment :-</u> The evidence of heredity nature of DNA was provided by a British microbiologist known as — Frederick Griffith's in 1928. ↙ (ETER-2007)</p>		
<p><u>Work of F. Griffith :-</u> In 1928, Griffith worked on two different stains (types) of bacterium called pneumococcus i.e—</p>		
<p>i: <u>Rough type bacterium : - (R-type)</u> — It is having rough colonies, non-capsulated, non-pathogenic and do not cause pneumonia.</p>		
<p>ii: <u>Smooth type bacterium : - (S-type)</u> — It is having smooth colonies, capsulated, pathogenic and cause pneumonia disease.</p>		
<p><u>Griffith experiments :-</u></p>		
<p>Griffith performed the following experiments on mice—</p>		
<p><u>1-Experiment No-1 :-</u> He injected R-type bacteria to mouse, as result they did not show any symptoms of pneumonia and survived.</p>		
<p><u>2-Experiment No-2 :-</u> He injected S-type bacteria to mouse, they were infected with pneumonia and died.</p>		
<p><u>3-Experiment No-3 :-</u> He injected heat killed S-type bacteria to mouse, but the mice survived. He made hypothesis that</p>		

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capsule in S-type bacteria is responsible for pathogenesis.

4-Experiment No-4 :- He injected the mixture of R-type - bacteria and heat killed S-type bacteria (as both were non-pathogenic) to mouse, as a result the mice died. He was much surprised with this result having dead mice.

Conclusion of Exp; -4 :-

- 1: From the blood of the dead mouse in experiment No-4 , Griffith recovered smooth type living bacteria (S-type).
- 2: From this experiment he concluded that Living-R-type - bacterium had transformed into Live-S-type bacterium by absorbing some material which control characters (capsule) from the heat killed S-type bacteria. Such process is - therefore called as Transformation.

Transformation :- The transfer of genetic material of one organism (donor) to another (recipient) which could alter the genetic make-up of recipient is called transformation.

The diagram illustrates the four groups of mice used in Griffith's experiment:

- Group 1: Injected with R-type Bacteria → mouse lives
- Group 2: Injected with S-type Bacteria → mouse dies
- Group 3: Injected with Heat killed S-type Bacteria → mouse lives
- Group 4: Injected with a mixture of R-type and Heat killed S-type Bacteria → mouse dies

A vertical column on the right is labeled "Griffith's Experiment".

2: Avery's Experiment :- Avery, MacLeod and Mac Carty -

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<p>repeated the Griffith experiment and proved that DNA is heredity-material in 1944. They demonstrated that transforming activity is lost if DNA of heat killed S-type bacteria is destroyed. But transforming activity is shown if DNA of heat killed S-type bacteria present. ←(ETEA-2013)</p>		
<p><u>Experimental work:</u></p>		
<p>1- <u>Experiment No-1: <Removing of protein></u> Avery and his co-workers prepared mixture of dead-S-type and live R-type bacteria that Griffith had used in his last experiment. Then they removed as much of the protein by treating it with protease enzyme. After removal of nearly all protein (99.9%) then they injected the mixture of R-type and S-type into mice, again transformation occurred and mice died. So, they said that proteins are not transforming agent.</p>		
<p>2- <u>Experiment No-2: <Removing of RNA></u></p> <p>In second experiment all the RNA contents were removed with the help of RNase enzyme and then the mixture is injected to mice, but still transformation occurred and mice died. so, they said that RNA is also not transforming agent.</p>		
<p>3- <u>Experiment No-3: <Removing of DNA></u></p> <p>In this final experiment the mixture was treated with DNase enzyme in order to remove DNA contents of S-type. Such mixture then injected to mouse, result no pneumonia disease symptoms develop on mouse because no transformation occur</p>		

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<p><u>conclusion :-</u> In this way it was confirmed that transforming agent in the Griffith's experiment was DNA.</p> <p><u>3: Hershey and chase experiment:</u> (ETEA-2017)</p> <p>Hershey and chase in 1952 demonstrated that DNA is genetic material. They used bacteriophages for their experiments.</p> <p><u>Bacteriophage :-</u> Bacteriophages are viruses which attack on bacteria. Bacteriophage body consists of protein and DNA. just after infection, many copies of bacteriophages are produced in the host within 20-25 minutes.</p> <p><u>Heredity material of bacteriophage :-</u> It was not known till 1952 that whether protein or DNA is responsible for the heredity information of bacteriophage and even it was also not known that whether protein or DNA or whole virus-body enters to bacteria. In 1952, Hershey and chase carried out experiment to identify the viral part responsible for infection.</p> <p><u>Experimental work :</u></p> <p>1: They labeled the DNA of some viruses with radioactive phosphorus (^{32}P) and protein coat of others with radioactive sulphur (^{35}S).</p> <p>2: Two bacterial cultures of E. coli were used, one-bacterial-culture was -</p>		
<p style="text-align: center;">(HARSHY AND CHASE EXPERIMENT)</p>		

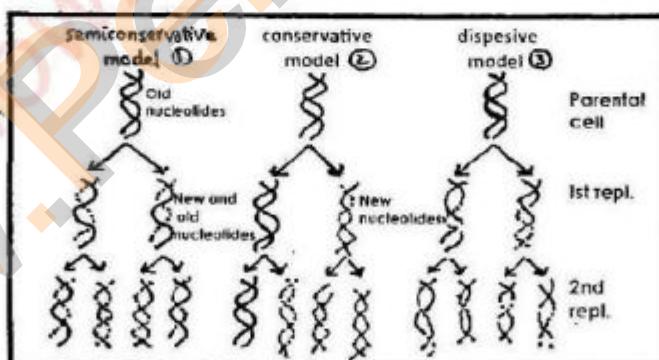
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<p>exposed to phages with radioactive DNA and the other bacterial culture was exposed to phages with radioactive protein.</p>		
<p>3: After a few generation, bacterial cells from both cultures were tested for radioactivity.</p>		
<p>4: Those bacteria which were infected with radioactive-DNA phage showed radioactivity while the other did not show radioactivity.</p>		
<p><u>Conclusion</u> :- This proved that only phage-DNA enters the bacterial-cell while the protein-coat remains outside. Thus through a series of experiments it was proved that DNA is the heredity material not proteins. DNA was discovered in 1869 by the German biochemist Friedrick Miescher (who called it "nuclein").</p>		
<p><u>Q6</u> What is DNA replication. write its different model.</p>		
<p><u>Answer:</u> DNA Replication:</p>		
<p><u>Meaning</u> :- Replica mean to copy.</p>		
<p><u>Definition</u> :- The process by which a parental DNA duplicates to produce two daughter DNA molecules is called DNA replication.</p>		
<p>—OR— “The process of self-synthesis of DNA during life cycle of cell is called DNA replication.”</p>		
<p><u>parent DNA</u> :- The DNA molecule which is to be replicated is called parent DNA.</p>		
<p><u>Daughter DNA</u> :- The DNA molecules which is produced as a result of replication is called daughter DNA. (ETEA-2014)</p>		
<p><u>Occurrence</u> :- DNA-replication occur in S-phase of a cell cycle.</p>		

Model of DNA replication:- There are various model for DNA replication, However the following 3-models are well accepted.

1: semi conservative Model:-

Semi conservative model is the most accepted model - presented by Watson and Crick. According to this model each strand of parental DNA molecule separate or unzipped and each act as template strand for the formation of new strand. In this way two daughter DNA-molecules is formed with one old and one new strand i.e. 50% conservation of old or parental strand occurs.

2: conservative Model:- According to this model the two - daughter DNA molecules always get new synthetic strand. whereas parental-DNA molecule remains in double helical structure as previous was, so parental DNA is fully - conserved in the next generation.



3: Dispersive Model:- According to this model parental - DNA completely divides into fragments. New fragments

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<p>or nucleotide segments are also synthesize. After which new and old fragments of nucleotides are mixed and rejoin to make daughter DNA strands or molecules. so this type of replication in which new and ^{old} fragments of nucleotide - segments are mixed to form daughter DNA is called - dispersive model.</p>		
<p>Q7 Elaborate the work of Meselson and Stahl to justify the semi conservative replication as a correct model of replication</p>		
<p>Answer : Meselson - Stahl experiment:</p>		
<p><u>Introduction :-</u> In order to prove that DNA-replication occurs according to which model, Meselson and Stahl of California institute carry out experiment in 1958. They evaluated all the 3-models and it was finally concluded that DNA replication occurs according to semi-conservative model.</p>		
<p><u>Experimental work :-</u></p>		
<p>Meselson - Stahl experiment consists of the following steps.</p>		
<p>1: culturing of bacteria :- Normally bacteria (e.g E.coli) - contain ¹⁴N in the nitrogenous bases of DNA. But they grow such bacteria in a medium containing heavy isotope of - nitrogen ¹⁵N which is radioactive. This isotope was then - incorporated into the bases of the bacterial DNA. after - many generations the DNA of these bacteria was denser than that of bacteria grown in a medium containing the - Lighter isotope of nitrogen ¹⁴N.</p>		

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<p>Meselson and Stahl then transferred the bacteria from - ^{15}N-medium to 3-separate plates which containing ^{14}N-medium.</p>		
<p><u>2: Sampling of DNA</u> :- when bacteria with ^{15}N DNA were - transferred to ^{14}N-medium then 3-samples were taken - from it at different times i.e -</p>		
<p>i- <u>Sample at 0-minute</u> :- First sample was obtained from 1st plate just after the transfer of culture called sample at 0-minute. as there has no replication occurred so both strands of DNA will have heavy isotopes ^{15}N.</p>		
<p>ii- <u>Sample at 20-minute</u> :- The second sample was taken from second plate after 20-minutes called sample at 20 minute. as one replication has occurred, so one strand will have - ^{15}N and other will have ^{14}N because new strand is formed in ^{14}N-medium.</p>		
<p>iii- <u>Sample at 40-minute</u> :- The 3rd-sample was taken from 3rd plate after 40-minutes called sample at 40-minute. In 3rd sample two replications has occurred. In this sample 4-cells were produced with 2-types of DNA.</p>		
<p><u>control sample</u> :- A control sample was taken from the - bacteria culture, which were grown separately in ^{14}N-medium which both strands of DNA will have ^{14}N only.</p>		
<p><u>3: centrifugation of DNA sample</u> :- Meselson and stahl - prepared cesium-chlorid solution and dissolve the DNA samples in that solution. The Samples was centrifuge in centrifugation</p>		

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<p>mechanism for several hours. Result the DNA-strands - fragments of different densities got separated. Due to centrifugation the cesium-chlorid ions are sedimented at bottom.</p>		
<p>4: Result of centrifugation :-(observation)</p>		
<p>1: DNA of control-sample was appeared lightest as formed sediment at the top of the test tube, because it contains only light isotope i.e. ^{14}N in both DNA strands.</p>		
<p>2: The DNA of sample at 0-minute was appeared heaviest as it formed sediment at the bottom of the test tube.</p>		
<p>3: The DNA of Sample at 20-minute formed sediment intermediate level to that of control sample and sample at 0-minute.</p>		
<p>4: The sample at 40-minute had two sediments, one at the top and others at intermediate level.</p>		
<p>5: Interpretation of result :-</p>		
<p>Meselson and Stahl interpretation their result as follow.</p>		
<p>1: The DNA of control-sample appeared lightest because it had both strands of ^{14}N, grown in ^{14}N-medium.</p>		
<p>2: The DNA of sample at 0-minute appeared heaviest because it had both strands of ^{15}N, grown in ^{15}N-medium.</p>		
<p>3: DNA of sample at 20-minute formed sediments at - intermediate level because after first replication of parental ^{15}N-strand in ^{14}N-medium, daughter DNA has one parental strand of ^{15}N & one new strand of ^{14}N. (hybrid DNA).</p>		
<p>4: DNA of sample at 40-minute formed two sediments, one at</p>		

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intermediate level and one at top. This is because in first replication two hybrid DNA (^{15}N and ^{14}N) were produced which on further replication produced two hybrid DNA (^{15}N and ^{14}N) and two newly synthesized DNA containing only ^{14}N . Hybrid DNA formed sediment at intermediate level while new DNA formed sediment at top.

Conclusion :- The above experiment clearly confirmed the prediction of the Watson and Crick model that DNA replicates in a semi-conservative manner.

The diagram illustrates the Meselson-Stahl experiment. It starts with a bacterial cell containing DNA labeled with ^{15}N . This cell undergoes three rounds of division in a medium containing ^{14}N , resulting in three daughter cells. At 0 minutes, a sample is taken from the first division (labeled 1). At 20 minutes, a sample is taken from the second division (labeled 2). At 40 minutes, a sample is taken from the third division (labeled 3). The DNA is then extracted and suspended in cesium chloride solution, followed by centrifugation. The resulting bands are labeled 1 through 4. Band 1 (control) contains unlabeled DNA. Band 2 (labeled parent) contains DNA with both strands labeled ^{15}N . Band 3 (F1 generation) contains DNA with one strand labeled ^{15}N and one strand labeled ^{14}N . Band 4 (F2 generation) contains DNA with both strands labeled ^{14}N .

Q8 Describe the events of the process of DNA replication.

Answer: DNA replication process :-

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Definition :- The process of formation of two daughter DNA molecules from single parent DNA is called DNA replication.

Steps of replication :- The process of DNA-replication -
Consists of the following 3-steps or phases-

1: Initiation phase :- initiation phase is characterized by the formation of replication-bubble and replication-fork which are formed at a particular site called origin of replication.

Origin of replication :- A sequence of particular nucleotides or a very specific point along the length of DNA from where DNA-replication begins is known as origin of replication. In Eukaryotic DNA, there are more than one origin of replication sites but in prokaryotic-DNA there is only one origin of replication site.

Role of enzymes in initiation phase :- The enzymes used in the initiation phase are - DNA Gyrase and Helicase i.e -

i: DNA Gyrase :- Topoisomerase - DNA gyrase opens the turns of DNA duplex molecule results in the conversion of - Spiral ladder like form to straight ladder like form. This process is also called unwinding.

ii: Helicase :- Helicase enzyme moves along the length of DNA very fastly and reaches the origin of replication and starts breaking base pairs of DNA by broken hydrogen bonding between nucleotides. Thus, the two strands separate from each other and form a bubble-like structure called -

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<p><u>replication bubble</u>. This process is also called unzipping.</p>		
<p><u>Replication Fork</u> :- The two ends of replication bubble on either side of bubble is like tuning fork hence called ^{fork} replication fork.</p>		
<p><u>single stranded binding proteins (SSB)</u> :- The two separated strands are prevented from rejoining by single stranded binding proteins which attached to the exposed nucleotides.</p>		
<p><u>Template</u> :- Both single strand of unzipping-DNA act as template. (supportive) strands for the formation of new strands or complementary strands.</p>		
<p><u>2: Polymerization phase</u> :- (Extension phase) (ETEA-2014)</p>		
<p><u>Definition</u> :- The formation of new or daughter strand over template is called polymerization or extension.</p>		
<p><u>Enzymes in polymerization</u> :- During polymerization the daughter strands are synthesized by DNA-polymerase enzyme.</p>		
<p><u>Polymer</u> :- primer is a short oligonucleotides strand of RNA which is responsible for the initiation of DNA-polymerase activity. DNA polymerase cannot work until primer is attached to the template strand. For this purpose primase enzyme is involved to arrange some nucleotides called primer on template strands as a result the synthesis of daughter strands begins by the DNA-polymerase enzyme. ←(ETEA-2019)</p>		
<p><u>Types of DNA-polymerase</u> :- DNA-polymerase has 3-types.i.e.</p>		
<p>i: <u>DNA polymerase-I</u> :- It is dual function enzyme. (i.e.-act as polymerase as well as exonuclease). It plays an important</p>		

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sole in termination phase so it provides support to DNA — polymerase-III in the main replication process.		
<u>ii: DNA polymerase-II</u> :- It is involved in the repairing process of damaged DNA during life time of a cell.		
<u>iii: DNA polymerase-III</u> :- It is the main enzyme which synthesizes daughter strands along the length of template strands.		
Mechanism of DNA polymerase-III activity :-		
1: DNA-polymerase-III is dimeric molecule i.e.—it consists of two units—one act as catalyst site and other act as — proofread site. both the units are joined by small polypeptide chain. one unit will act on one strand and the other on another strand or template-strand. (ETEP-2017, 2018)		
2: DNA-polymerase-III can add nucleotides on 3-OH group so new bases or nucleotides are added on 5-end of — primer and the direction of replication become 5' to 3' end.		
3: as a result the DNA-polymerase-III synthesizes both daughter strands along the template during replication process.		
4: During the process of replication if any nucleotide is — added mistakenly, then they are removed by DNA-polymerase-II enzyme and new correct nucleotides is inserted by DNA-polymerase-III. This process is called proof-reading.		
<u>Leading strand</u> :- One unit of DNA-polymerase-III — synthesizes one daughter strand along the parental — template strand continuously towards replication fork or		

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<p>in the direction of movement of helicase enzyme. This — continuously growing daughter strand is called leading strand.</p>		
<p><u>Lagging strand</u> :- another unit of DNA-polymerase-III synthesizes daughter strand along the other parental — template strand discontinuously away from the replication — fork or opposite to the movement of Helicase enzyme. This discontinuously growing daughter strand is called lagging strand.</p>		
<p><u>Okazaki fragments</u> :- The lagging strand is synthesized in the form of short fragments called Okazaki fragments. The second unit of DNA-polymerase-III synthesizes the new — daughter strand upto certain length (called Okazaki — Fragment) on parental template strand and then jump back (100–200 nucleotides in Eukaryotes and 1000–2000 — nucleotides in prokaryotes) to new primers and start — synthesizing new Okazaki fragment. When this Okazaki is synthesized upto already synthesized Okazaki fragment then jump back again. In this way lagging strand is formed in the form of fragment with small gaps among them. These gaps are later sealed by ligase enzymes in termination phase.</p>		
<p><u>3: Termination phase</u> :-</p>		
<p><u>Introduction</u> :- Termination phase occurs in the presence of enzyme called DNA-polymerase-I which is characterized by the replacement of primers by DNA-nucleotides and joining of Okazaki's fragment to form continuous strand.</p>		

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DNA polymerase acts as exonuclease :- DNA-polymerase-I also act as exonuclease which replace the primer by DNA-nucleotides. Exonuclease attached to 3'-end of okazaki - fragments and 5'-end of primers. Exonuclease removes the RNA nucleotides from 5'-end of primer and adds DNA - nucleotides to the 3'-end of okazaki fragment until primer is completely replaced by DNA-nucleotides. Finally the DNA ligase enzyme joins and seals the two okazaki fragments and form continues strand.

One DNA

Two DNAs

DNA-Polymerase (Pol α)

DNA-Polymerase (Pol β)

DNA ligase

RNA primer

Lagging strand

Leading strand

Okazaki fragment

Helicase

Topoisomerase

Single strand Binding proteins

DNA-Replication

Q9 What is central dogma of gene expression -OR- Explain the function of a gene -OR- protein synthesis.

Answers: Central dogma of gene expression :

Introduction :- All living organism follow the same basic mechanism of gene expression and protein synthesis. This is called central dogma of biology or central dogma for life.

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Steps of central dogma :- There are 2-steps of central-dogma of gene expression i.e.- Transcription and Translation.

1: Transcription : (ETEA-2014, 2020)

Definition :- The process in which the formation of mRNA from DNA occurs during protein synthesis is called transcription.

Occurrence :- In eukaryotes, transcription occurs in the nucleus of the cell while in prokaryotes transcription occurs in the cytoplasm of the cell due to lack of true nucleus.

Steps of Transcription :-

Transcription process consist of the following steps.

1: Initiation phase : (ETEA-2020)

RNA-polymerase :- The main enzyme for transcription process is RNA-polymerase which binds on specific site called promoter which is located on 5'-end of gene.

Promoter :- A specific sequence of nucleotides to which RNA polymerase attaches with the help of initiation factor and begin the process is called promoter. Promoter is a regulatory region because it controls the process of transcription. RNA-polymerase cannot attach to the promoter by itself but initiation factor moves along the length of gene (DNA) in order to identify promoter site, so initiation factor directs RNA-polymerase to attach with promoter.

Promoter region in prokaryotes :- In prokaryotes there are two binding sites or promoters. i.e.-

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1-TATAAT :- It is also called -10 sequence. The negative sign shows that it is located 10 nucleotides before the initiation site of the structural gene.

2-TTGACCA :- It is also called -35 sequence.

promoter region in Eukaryotes :- In Eukaryotes, there are also two binding sites or promoters. i.e -

1-TATA box :- <TATTAAA> It is also called -25 sequence.

2- CAAT box :- < CAAT > It is also called -75 sequence.

RNA polymerase structure and role :- RNA-polymerase

consists of 4-subunits i.e- Alpha, Beta, Beta prime, sigma. The first 3-parts (i.e -alpha, beta, B') are considered to be the core enzymes of the process. while the 4th-part (i.e-sigma) is required for attachment of RNA-polymerase to the - promoter site.

Role :- RNA polymerase can read the DNA strand (gene) only from 3'-end to 5'-end and the mRNA will formed from 5'-end to 3'-end. It also adds nucleotides to 3'-end of the growing polypeptide chain but unlike DNA-polymerase it does not require primers to perform polymerase activity.

Types of RNA polymerase :-

i. iii. Case of Eukaryotic cell RNA-polymerase enzyme has 3-types as following.

i: RNA-polymerase-I which synthesize rRNA.

ii: RNA-polymerase-II which synthesize mRNA.

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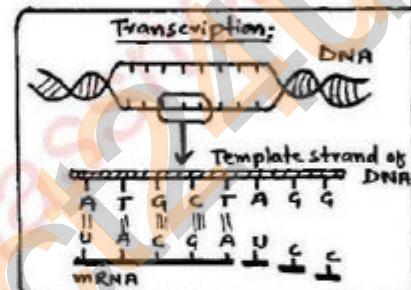
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iii: RNA-polymerase III which synthesize tRNA.

2: while in case of prokaryotes, they have only one type of RNA-polymerase which synthesis all types of RNAs.

Transcription bubble :- After binding of RNA-polymerase subunits with promoter region of gene (DNA) the double-stranded DNA become unzipped and open, result transcription bubble is appeared.

2: Elongation phase :- As the RNA-polymerase attaches to promoter, Sigma part (unit) is released and the remaining parts (core enzyme) extend the polymerization of RNA strand. One of the two strand of the gene acts as template for transcription.



Antisense strand :- The template strand to which complementary mRNA is synthesized is called antisense strand, because mRNA is complementary to it.

Coding strand :- The other strand whose sequence of nucleotides is exactly same to mRNA nucleotides sequences except 'T' which is replaced by 'U' is called coding strand.

Growing of mRNA :- In elongation phase RNA polymerase (II) move from 3' end to 5' end of antisense strand towards terminator, besides transcription bubble also moves along the DNA, leaving the growing mRNA protruding out from the

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<p>transcription bubble. This event continues till the RNA - polymerase reaches the terminator region of the gene.</p>		
<p>3: Termination phase :- This is the last phase of transcription after which transcription is complete.</p>		
<p>Terminator :- The region or sequence of nucleotides which end the process of transcription is called terminator.</p>		
<p>Sequence of terminator :- Terminator region consists of 'GC'-base pairs followed by 'AT'-base pairs. i.e.-</p>		
<p>i: GC Hairpin :- The part of mRNA which is transcribed in this region is called GC-hairpin and it is the form of a loop like structure. GC-sequence is found on coded strand.</p>		
<p>ii: AU Tail :- GC-hairpin is followed by small tail of AU - nucleotides. The formation of GC-hairpin and AU-tail is indication of complication of transcription.</p>		
<p>2: Translation : <for detail plz see Question No-12> The formation of polypeptide or protein with the help of RNAs after transcription is called Translation.</p>		
<p>Q10 Describe the post-transcriptional modification of mRNA.</p>		
<p>Answer: Post transcriptional modification of mRNA :-</p>		
<p>Definition :- The process through which primary or immature mRNA is converted into mature or functional RNA is called post transcriptional modification of mRNA.</p>		
<p>Occurrence :- This process only occurs in eukaryotes, -</p>		

However in prokaryotes the mRNA formed is ready for translation due to absence of nucleus and lack of introns.

Need of post transcription modification:- Due to the following two reason the post-transcriptional modification occur.

1: Hydrolytic enzymes:- In eukaryotes the mRNA formed as a result of transcription has to travel long distances to reach ribosomes in the cytoplasm. In this journey it faces some enzyme like phosphatases and nucleases which can degrade this mRNA. To protect this mRNA from the degradation it is modified and then it is allowed to go to cytoplasm. This process occurs inside nucleoplasm.

2: Non-functional intron:- Another problem with newly formed mRNA is the non-coding protein sequence. As mRNA contains two regions - exon (protein coding sequence) and intron (non-protein coding sequence). These introns are to be removed to convert mRNA into functional one.

Steps of post transcription modification:-

post-transcriptional modification of mRNA involves two steps.

1-Addition of cap and tail:- phosphatases and nucleases enzymes attack only on ends of mRNA. So therefore, one end of mRNA is protected by cap and other end by tail and hence it is protected from the action of enzymes. i.e-

cap:- The cap is in the form of 7-methyl GTP. It is attached to 5'-end of mRNA.

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Tail :- It is a small chain of 300-500 adenine nucleotides called poly-A-tail. It is attached to 3'-end of mRNA.

2: RNA splicing:

Definition:- The removal of introns (non-coded part) from primary mRNA to form a secondary and well mature RNA is called RNA splicing.

Explanation:- The newly formed mRNA is very long because it contains both introns and exons. The introns are non-coding sequences or non-functional sequences so they are to be removed. This removal of introns is catalyzed by small nuclear spliceosome (an RNA-protein complex). now this - mature mRNA will now enter to translation.

The diagram illustrates the process of RNA splicing. At the top, 'Primary-mRNA' is shown as a horizontal bar with segments labeled 5' m⁷G-cap, Exon, intron, Exon, intron, Exon, and 3' poly-A-tail. An arrow points down to 'mature mRNA', which is shown as a bar with segments labeled 5' m⁷G-cap, Exon (spliced from the original intron position), and poly-A-tail. A bracket between the two bars indicates that the intron was excised and the exons were spliced together. The word 'RNA Splicing' is written diagonally across the bottom right of the diagram area.

Q11 what is genetic code. write its characteristics

Answer: Genetic code:

Meaning :- Code mean data or information on gene or DNA

Definition :- The sequence of nucleotides in DNA or RNA - which determines the specific amino-acids sequence of the proteins is called genetic code.

Explanation :- DNA in the nucleus is considered as a store

house of genetic information. This information is in the form of a code called genetic code. Thus the sequence of nucleotides in the DNA for the specific sequence of amino-acids for the synthesis of particular protein is called genetic code. It is the basis of heredity and nearly universal in all organisms. —

Genetic code is stored on coding or sense strand of a DNA molecules as a linear, non-overlapping sequence of the 2 nucleotides.

Codons :— The genetic code is a coded language which is based on 4-types of nucleotides or nitrogenous bases i.e.— Adenine (A), Guanine (G), cytosine (C) and thymine (T).

These bases are variously arranged to form code words — called codons. Each codon consists of 3-nitrogenous bases or 3-Latters called Triplet form that will eventually be interpreted as a single amino-acid in a polypeptide chain.

Types of codons :— There are 64-possible codons which are arranged into different types i.e.—

1: Start codons :— one codon act as start codon. e.g.— AUG is a start codon for amino-acids chain. It code for — methionine amino-acid. (ETEA-2016, 2017)

2: Stop codons :— 3-codons such as — UAG, UGA, UAA act as stop codons that indicate that the message is over. These 3-codons do not encode for any amino-acid, hence they are also called non-sense codons.

3: Sense codons :— Beside the above 3-stop codons, all the

the other 61 codons encode specific amino-acids are called sense codons. (ETEA-2018)

Characteristics of genetic code :- some characters are -

- 1: Code will be Triplet i.e. all codons consists of 3-nucleotides.
- 2: The code is non-overlapping i.e. end to end sequence - AUA.
- 3: There are no internal punctuation marks. e.g. - AUA.
- 4: Code will be degenerative, which refers that an amino-acid can be encoded by more than one codon, but a particular codon does not specify more than one amino-acid.
So the genetic code has redundancy but no ambiguity. For example, codons - GAA and GAG both specify glutamic acid (called redundancy), but neither of them specifies any other amino-acid (called no ambiguity).
- 5: Total codons are 64, in which 61 codon-code for specific A.Acid.

- 6: Genetic code is universal – except for mitochondrial DNA.
i.e. Genetic code is identical in all organism e.g. - AGA codon specifies Arginine amino acid in bacteria, in human, and all other organisms whose genetic code has been studied.
- 7: The genetic code has polarity, that is, the code is always

SECOND BASE				THIRD BASE	
FIRST BASE	U	C	A		
U	UUU Phe UUC UUA UUG	UCU UCC UCA UCG	UAU UAC UAA UAG	UAU UAC UAA Stop UAG Stop	CYS C A G
C	CUU CUC CUA CUG	CCU CCC CCA CCG	CAU CAC CAA CAG	CGU CAC GAA GAG	U C A G
A	AUU AUU AAA AUG Met or stop	ACU ACC ACA ACG	AAC Tyr AAA AAG	AAT Asn Lys	AGU Ser AGC Arg AGA Arg AGG
G	GUU GUC GUA GUG	GCU GCC GCA GCG	GAU GAC GAA GAG	GGU GGC GGA GGG	U C A G

Dictionary of Genetic Code

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<p>read in a fixed direction, i.e. in the $5' \rightarrow 3'$ direction.</p>		
<p><u>Mitochondrial genetic code</u> :- The study of genetic code of mitochondrial-DNA shows that genetic code is not that much universal. \leftarrow (ETEA-2015)</p>		
<p><u>For example</u> :- 1:- UGA codon is normally a stop codon but in mitochondria it reads as Tryptophan amino-acid.</p>		
<p>2: Like wise AUA was read as methionine in mitochondria instead of Isoleucin amino-acid.</p>		
<p>3: AGA and AGG code for termination of protein synthesis in mitochondria instead of Arginine amino-acid.</p>		
<p>Q12 Explain the process of translation of mRNA into polypeptide</p>		
<p><u>Answer</u>: Translation:</p>		
<p><u>Definition</u> :- The process of the formation of protein with the help of RNAs is called translation.</p>		
<p><u>Explanation</u> :- In translation the mRNA which produced by transcription is decoded by the ribosome to produce a specific amino-acid chain or polypeptide that will later fold into an active protein. \leftarrow (ETEA-2007)</p>		
<p><u>Second phase of gene expression</u> :- Translation is the second phase in gene expression. The information stored in mRNA is translated into protein with the help of RNAs and ribosomes.</p>		
<p><u>Occurrence</u> :- In prokaryotes, translation occurs in the cell cytoplasm where the large and small-subunits of ribosomes</p>		

are located. while in case of eukaryotes, translation - occurs across the membrane of endoplasmic reticulum where Ribosomes are located.

Steps of Translation :- The process of translation completed in 4-phases as following. ↴ (ETEA-2019)

1: Activation of amino-acids :- Amino acids are present in inactive form in cytoplasm. Amino-acids are first activated by ATP inside the cytoplasm in order to make bonds with - tRNA. Various amino-acids that are to be take part in - polypeptide formation have been continuously activated - throughout the process of translation. ↴ (ETEA-2019)

Aminoacyl-tRNA-complex :- For every amino-acid there is a specific tRNA. with the help of activation enzyme, i.e - aminoacyl-tRNA-synthase, a specific amino-acid binds to it tRNA . formed aminoacyl-tRNA-complex. each amino acid attaches to the 3'-end of particular tRNA molecule.

2: Formation of initiation complex :-

Definition :- Aminoacyl-tRNA-complex , ribosomal-subunits , and mRNA combine to form a complex called initiation complex.

Initiation complex :- The process of translation actually - begins with the formation of initiation complex. First a tRNA molecule which carrying methionine (starting amino-acid) binds to the smaller ribosomal subunit. This binding is controlled by an enzyme called initiation factor. at the same time -

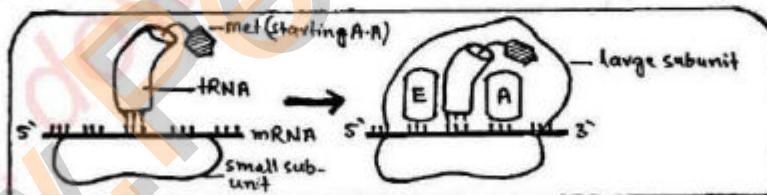
5'-end of mRNA also attaches to the smaller subunit of - ribosome with the help of another initiation factor or enzyme. Initiation complex is completed when larger subunit of - ribosome is also placed upon smaller subunit. The complex then move along the mRNA in a 5' → 3' direction until it locates the AUG-initiation or start codon.

Sites on Ribosome :- There are 3-different sites on ribosome

i: peptidyl site :-(p-site) - The region on ribosome where first amino-acid-tRNA attaches is called p-site. In this - region peptide bond is formed between successive amino-acids during elongation phase. ←(ETEA-2018)

ii: Aminoacyl site :-(A-site) - The region on ribosome where tRNA coming and bearing amino-acids, will be attached - during elongation phase.

iii: Exit site :-(E-Site) - The region on ribosome where - empty tRNA will leave the ribosome during elongation phase.



3: polypeptide elongation :- During this phase, ribosomal units move along mRNA and amino-acids are brought by tRNAs toward ribosome continuously, which joined together to form a polypeptide chain.

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<p><u>Steps of polypeptide elongation</u> :- The process of polypeptide elongation occurs by 3-steps which are repeated again and again.</p>		
<p><u>1-1st step</u> :- One of the codons of mRNA is always exposed at site-A. anticodon bearing amino-acyl tRNA complex binds to site-A with the help of elongation factor.</p>		
<p><u>2-2nd step</u> :- Then an enzyme (i.e-peptidyl transferase) - emerges from p-site. It removes amino-acid (may be a chain) from p-site and binds it to the new coming amino-acid with the help of peptide bond.</p>		
<p><u>3-3rd step</u> :- In this step translocation occurs. The ribosomal subunit moves slightly along the mRNA from 5'-end to 3' end direction so that new codon is exposed at A-site. This movement is called translocation. As a result, empty tRNA at p-site is shifted to E-site to leave ribosome. The tRNA at A-site bearing chain is shifted to p-site and in this way new codon is exposed at A-site.</p>		
<p><u>Completion of polypeptide</u> :- In this way the whole mRNA is translated i.e- these 3-steps are repeated again and again according to the coding message on mRNA and the required protein is synthesized. The elongation process terminates when stop-codon reaches at A-site.</p>		
<p><u>4: Termination</u> :</p>		
<p><u>Stop codon</u> :- polypeptide elongation is continues until a chain terminating non-sense (stop) codon is exposed at -</p>		

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A-site: non-sense (stop) codons do not bind to any tRNA but all 3-termination (stop) codons (i.e.- UAG, UGA, UAA) are recognized by release factor and terminates the translation process.

Releasing of polypeptide chain :- upon termination, the completed polypeptide chain is released from the tRNA. The tRNA is also released from the ribosome and the two - two Ribosomal-subunits separate from the mRNA. and this way polypeptide chain or protein synthesis completed.

Q13 Discuss the regulation of gene expression with the help of lac operon Model.

Answer :- Regulation of gene expression:

Definition :- The process of switch ON and switch OFF of a gene is called regulation of gene expression.

Explanation :- Regulation of function of a gene or protein synthesis is necessary in all cells. i.e -

- 1: In the prokaryotic cell, the regulation of protein synthesis

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is achieved by means of genetic units called operons.

2: In multicellular organisms (eukaryotes) different cells perform different function (different protein synthesis) which is due to different genes expression during embryonic conditions or during metamorphosis. Though all cells of an individual have same genome (genetic materials) but having different functions due to expression of gene's set according to functions or needs.

Methods of gene regulation :-

There are two methods for gene regulation as following—

1: positive Gene Regulation :- The expression of gene in the presence of special regulatory protein or activator is — called positive gene regulation.

2- Negative gene regulation :- The suppression of a gene in the presence of special repressor protein is called -ve regulation.

Example of gene regulation :- (Lac operon)— In prokaryotes, Lac operon having dual—positive and negative control of gene expression. Bacteria obtain energy from a carbohydrate called lactose which is chemically known as β -galactosides. But this only occurs when β -galactosidase enzyme is present because this enzyme break-down the β -galactosides into Galactose and glucose which then take up by bacteria for getting energy. For this purpose bacterial DNA possess Lac-operon region in their DNA which encode for —

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<p>protein (enzymes) necessary to perform coordination – function such as – “ catabolism of substrate obtained from outside (called lac operon) or biosynthesis of a given amino-acid – Tryptophan (called trp-operon)”.</p>		
<p><u>Structure of lac operon</u> :- The lac operon consists of – genes</p>		
<p>1: <u>Structural genes</u> :- There are 3-structural genes in lac operon i.e. cistron-β, cistron-γ and cistron-α.</p>		
<p>2: <u>Regulatory genes</u> :- There are 3-regulatory genes in lac operon i.e. Regulator gene (i), promoter gene (p) and operator gene (o).</p>		
<p><u>polycistronic mRNA</u> :- mRNA that is transcribed (form) from prokaryotic operon is called polycistronic which mean that many proteins are encoded in a single mRNA.</p>		
<p><u>Mechanism of operon function</u> :- (Dual function)</p>		
<p>1: <u>Negative gene regulation</u> :- (if inducer is absent) In the absence of inducer (e.g. Lactose) in a medium, regulator gene (i) will synthesize a repressor protein. The repressor protein will stop operator gene (o) from its function. as a result operator gene (o) will not be able to activate structural genes to form mRNA. When no mRNA is formed it means enzymes for the catabolism of β-galactosides are not formed.</p>		
<p>2: <u>positive gene regulation</u> :- (if inducer is present) in the presence of inducer (e.g. Lactose) in a medium, The –</p>		

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repressor protein will not formed or inactivated. In such case operator gene (O) will activates structural genes to form their respective mRNA. These mRNAs of 3-structural genes will form 3-different enzymes (proteins) for metabolism of β -galactosides. i.e -

- 1: Structural Gene- Z codes for β -galactosidase enzyme which breakdown β -galactosides into glucose and galactose.
- 2: Structural Gene- y codes for permease enzyme which increases permeability of bacterial cell wall to glucose.
- 3: Structural Gene- a codes for transacetylase enzyme which assists the activity of an enzyme β -galactosidase.

The lac Operon

The diagram illustrates the lac operon structure and its regulation. It shows two horizontal gene sequences:

- Absence of inducer:** The sequence is $P - I - p - o - z - y - a$. A wavy arrow labeled "repressor mRNA" points to a "repressor" protein, which is shown binding to the "operator" region (the I and p genes). A note states: "repressor binds to the operator region and prevents RNA polymerase from transcribing the operon".
- Presence of Inducer:** The sequence is $P - I - p - o - z - y - a$. An "Inducer" molecule (represented by a circle with a plus sign) binds to the repressor protein, forming an "Inactive repressor" complex (represented by a single black dot). This inactive repressor no longer binds to the operator, allowing RNA polymerase to bind and transcribe the structural genes (z, y, a). The resulting mRNA is labeled "lac mRNA", which encodes three proteins: β -galactosidase, permease, and transacetylase.

Q14 what is mutation. write its different types.

Answer: Mutation :-

Definition :- any change in the structure of genetic -

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<p><u>material (DNA) is called mutation.</u></p>		
<p><u>Explanation :-</u> mutation is a permanent and non-reversible changes in the structure of chromosome or DNA or genes. A chromosome is an assembly of genes. the number of genes, structure of genes and position of genes on the chromosomes is fixed. occasionally changes occur in the sequence of nucleotides in the DNA molecule. This permanent change in the nucleotides of DNA is called Mutation.</p>		
<p><u>Mutagens :-</u> The agents that cause mutations are called mutagens. For example- mutations are caused by - radiation viruses, Transposons and mutagenic-chemicals as well as errors occur during DNA-replication.</p>		
<p><u>Mutant :-</u> The organisms carrying mutation are called mutant.</p>		
<p><u>Concept of mutation:-</u> The concept of mutation as the cause of the sudden appearance of a new characteristic was first proposed by the Dutch botanist Hugo de Vries in 1901, following his work on inheritance in the evening primrose. Nine years later T.H. Morgan began a series of investigation into mutations in drosophila and, with the assistance of geneticists throughout the world, identified over 500-mutations.</p>		
<p><u>origin of mutation :-(Based on Type of Cells)</u></p>		
<p>There are the following 3-origin of mutation. ←(ETEA-2016,2017)</p>		
<p><u>1: Hereditary mutation :-</u> This type of mutation always passed from parents to offspring through gametes. This is</p>		

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also called germ line mutation because it occurs in the germ cells - (germ tissues) of body. This type of mutation is present throughout a person's life in every cell, ^{in the body} .		
<u>2: De-novo-mutation</u> :- This type of mutation occurs in gametes (sperm or egg). This may also occur even after fertilization. It is also called new (de novo) mutation. This type of mutation leads to congenital abnormalities (birth defect).		
<u>3: Somatic mutation</u> :- This type of mutation occurs in DNA of individual cells at some time during a person's life. They do not pass to next generation. They can be caused by environmental factors such as UV-radiation or can occur during DNA-replication.		
<u>Types of mutation</u> :-(Based on mode of mutation)		
<u>1: Spontaneous mutation</u> :- This type of mutation occurs automatically or naturally because of some internal factors.		
<u>2: Induced mutation</u> :- This type of mutation is induced artificially in the living organism by exposing them to abnormal environment (such as - radiation, certain chemicals).		
<u>Types of mutation</u> :-(Based on size and Quality)		
on the basis of quality mutation has the following 2-types.		
<u>1: point mutation</u> :		
<u>Definition</u> :- When mutation occurs in a small part of DNA molecule, either in single nucleotide or a pair of nucleotide it is called point mutation.		

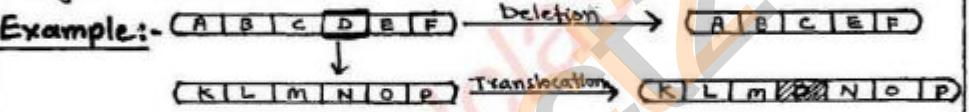
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<p><u>Types of point mutation</u> :- point mutation has 3-subtypes i.e-</p>		
<p>i: <u>Deletion point mutation</u> :- The removal of one or few nucleotide from a particular segment of DNA is called deletion.</p>		
<p>Example:- <u>(ATTAGCCTTAGAACT)</u> → <u>(ATTAGCCT<u>A</u>GAAT)</u></p>		
<p>ii: <u>Insertion point mutation</u> - the addition of one or few nucleotide in a particular segment of DNA is called insertion.</p>		
<p>Example:- <u>(ATTAGCCTTAGAACT)</u> → <u>(ATTAGCC<u>A</u>TAGAACT)</u></p>		
<p>iii: <u>Substitutional point mutation</u> :- The replacement of one or few nucleotide in a particular segment of DNA is called substitution.</p>		
<p>Example:- <u>(ATTAGCCTTAGAACT)</u> → <u>(ATTAGCC<u>C</u>ATAGAACT)</u></p>		
<p><u>2: chromosomal mutation</u> :- <u>(gross mutation or aberration)</u></p>		
<p><u>Definition</u> :- The mutation which causes changes in the structure or number of chromosome is called chromosomal.m.</p>		
<p><u>Types</u> :- There are two types of chromosomal mutation i.e-</p>		
<p>i: <u>structural changes in chromosome</u> :- structural changes in chromosomes take place during meiosis when due to certain mutagen chromosome is split down into several fragments but later on when these fragments reunite , its new pattern become changed from original one.</p>		
<p><u>Types of structural changes</u> :- structural changes may be-</p>		
<p>a: <u>Deletion</u> :- The removal of a segment of chromosome - comprising single or few genes.</p>		
<p>Example:- <u>(A B C D E F)</u> → <u>(D) (A B C E F)</u></p>		
<p>b: <u>Inversion</u> :- In this mutation, a portion of chromosome -</p>		

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break off, turns around and joins again in such a way that the sequence of gene gets reversed.

Example:- 

c: Translocation :- It involves shifting of a segment of one chromosome to another non-homologous chromosome. Thus both the chromosomes are affected. The donor chromosome suffers a deletion while the recipient chromosome become longer than normal.

Example:- 

d: Duplication :- The repetition of one or few genes in the same chromosome (homologous chromosome) is called Duplication

Example: 

ii: change in number of chromosome :- Due to non-disjunction during meiosis change occurs in the numbers of chromosomes.

Types :- Change in number of chromosome has two types i.e-

1: Aneuploidy :- The change in the number due to addition or loss of one or more chromosome is called aneuploidy. It occurs due to non-disjunction of chromosomes during meiosis. The number of chromosomes in aneuploidy can be greater or smaller than the number of chromosomes in the wild type.

Types :- Aneuploid has the following subtypes.

i: Monosomy :- $(2n-1)$ It is the result of loss single -

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<p>chromosome from diploid set ($2n$). i.e. $46 - 1 = 45$.</p>		
<p>ii: <u>Nullisomy</u> :- ($2n-2$) - It is the result of loss of a pair of homologous chromosome from the diploid set i.e. $46 - 2 = 44$</p>		
<p>iii: <u>Trisomy</u> : ($2n+1$) - It is the result of addition of single chromosome in the diploid set i.e. $46 + 1 = 47$.</p>		
<p>iv: <u>Tetrasomy</u> : ($2n+2$) - It is the result of addition of a pair of homologous chromosome in the diploid set i.e. $46 + 2 = 48$.</p>		
<p>2: <u>Euploidy</u> :- Change in the number of chromosome due to addition or loss of one or more complete set of chromosome is called euploidy. Euploid is also called polyploidy. Polyploidy can be possible in plants but rarely found in animals because in animals sex-balance is important. Variation in diploid numbers or sex-balance results in sterility. ← (ETEA-2018)</p>		
<p><u>Types</u> :- Euploidy or polyploidy has the following subtypes.</p>		
<p>i: <u>Monoploid</u> :- These organisms contain one set of chromosomes in the nuclei of their body cells. They are haploid. Such organisms are usually weak and sterile.</p>		
<p>ii: <u>Triplid</u> :- These organism have 3-sets of chromosomes ($3n$).</p>		
<p>iii: <u>Tetraploid</u> :- Having four sets of chromosomes ($4n$).</p>		
<p>iv: <u>Pentaploid</u> :- Having five sets of chromosomes ($5n$).</p>		
<p>v: <u>Hexaploid</u> :- Having six sets of chromosomes ($6n$).</p>		
<p>vi: <u>Heptaploid</u> :- Having seven sets of chromosomes ($7n$).</p>		
<p>vii: <u>Octaploid</u> :- Having eight sets of chromosomes ($8n$).</p>		
<p>viii: <u>Nonaploid</u> - 9-sets. ix: <u>Decaploid</u> :- 10-sets.</p>		

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<p>Q15 what is mutagen. write the types of mutagen.</p> <p>Answer: Mutagen:</p> <p>Definition :- The agents which cause mutations are called mutagen.</p> <p>Example :- certain radiation, mutagenic chemicals.</p> <p>Types of mutagen :- Mutagens has the following types.</p> <p>1: Physical mutagen :- These are physical agents which brings mutation. These are external as present in environment. These can breaks the chromosomes and thus bring mutation. H.j. Muller was the first who induces mutation using x-rays in Drosophila. other than x-rays, gamma-rays and ultra-violet radiation can be used to induce mutation. Spontaneous mutation is caused by cosmic rays coming from sun.</p> <p>Effects of physical mutagens :</p> <ol style="list-style-type: none">1: Radiation cause breaks in the chromosomes.2: Mutant-cells then show abnormal cell division (mitosis).3: Different types of cancers are the result of radiation.4: UV-rays affect the structure of DNA-helix.5: UV-rays also affect the replication process. <p>2: chemical mutagens :- These are chemical-agents which brings mutation, some time they produce polyploidy. First chemical mutagen discovered was mustard gas which was used as a weapon by German army in 1917 world war 1st.</p> <p>Examples :- Nitrous-acid, formaldehyde, mustard gas, 5-bromouracil, acridines, caffeine, nicotine, pesticides, colchicine.</p>		

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<p><u>Effects of chemical mutagens :-</u></p> <p>1: certain chemicals are like DNA-nucleotides and may be wrongly attached to DNA and cause mutation.</p> <p>2: certain chemical mutagens remove amino-group (NH_2) from adenine or cytosine and cause changes in DNA.</p> <p>3: certain chemicals attach hydrocarbon with nitrogen bases and damage the DNA structure.</p> <p><u>3: Biological mutagens :-</u> sometime microbes also causes changes in the sequence of nucleotides of DNA.</p> <p><u>Example :-</u> Transposons, viruses and certain bacteria, etc</p>		
<p><u>Q16</u> Describe the different diseases induced by chromosomal mutation.</p> <p><u>Answer:</u> Diseases induced by chromosomal mutation:</p> <p>The following diseases are caused due to chromosomal mutation.</p> <p><u>1: Klinefelter's syndrome : (xxY syndrome)</u></p> <p><u>Condition :-</u> It is sex chromosomal non-disjunction or Trisomy (2n+1)</p> <p><u>Introduction :-</u> Klinefelter's syndrome is a genetic disorder which results when a boy is born with an extra copy of the X-chromosome i.e. xxY. It is a common genetic condition - affecting males. - (ETEA-2010)</p> <p><u>Dr. H. Klinefelter :</u> Klinefelter's syndrome is named after Dr. Henry Klinefelter who in 1942 first described a group of symptoms found in some males. In 1959, these males were described to have an extra X-chromosome (xxY) than usual</p>		

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<p>males sex complement (XY).</p>		
<p><u>Causes and risk factors:</u></p>		
<p>1: This is due to non-disjunction of sex chromosome during oogenesis in mother. as a result one egg receive two X-chromosomes with autosomes. (XXA). </p>		
<p>2: if a male inherits two X-chromosomes from mother egg (XXA) and Y-chromosome from father sperm (YA) so such individual have sex-chromosome Trisomy i.e. -XXX.</p>		
<p>3: For old mother the risk of Klinefelter syndrome is higher.</p>		
<p>4: Klinefelter syndrome does not occur in females.</p>		
<p><u>Signs and symptoms:</u></p>		
<p>1: person of Klinefelter syndrome will be sexually sterile (95-99%)</p>		
<p>2: They do not make Testosterone hormone as other boys do.</p>		
<p>3: less muscular-body, less facial and body hairs.</p>		
<p>4: They may sit up, crawl, and walk-later than other infants.</p>		
<p>5: Broader hips than other boys.</p>		
<p>6: Breast are larger, Bones are weaker.</p>		
<p>7: over all energy level is lower and reduced strength.</p>		
<p>8: Trouble in fitting in with other kid because they tend to be quiet and shy as compare to other boys.</p>		
<p>9: other problems like - Trouble in Language, problem in reading and hearing, etc.</p>		
<p>10: more FSH secretion.</p>		
<p>11: Taller than average stature</p>		
<p>12: - Decreased sex drive.</p>		
<p>13: Small testicles and penis.</p>		
<p>14: - Difficulting expressing feeling</p>		

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<p><u>Treatment of Klinefelter syndrome:</u>—There is no treatment to change pattern of xxy in each cell of the body ,but there are variety of ways to treat the syndrome of the xxy-condition</p>		
<p>1: Testosterone replacement therapy can bring their — testosterone level to normal which lead to male characters. so that the voice become deep ,muscles are stronger ,grow facial and body hairs and person can enjoy normal social life.</p>		
<p>2: Breast tissue removal. 3: speech and physical therapy.</p>		
<p>4: Educational support. 5: psychological counseling.</p>		
<p><u>2: Turner's syndrome :</u></p>		
<p><u>Condition</u> :—It is sex-chromosomal non-disjunction or Monosomy 2 (2n-1).</p>		
<p><u>Introduction</u> :— This is the disorder of female in which she possess one x-chromosome instead of normal xx-chromosome. so that the number of chromosomes in such individual will be 45-chromosomes instead of 46-chromosomes. As these persons do not have y-chromosome so they are always female.</p>		
<p><u>Dr. H. Turner</u> :— In 1938 , H.Turner first described Turner Syndrome.</p>		
<p><u>Causes and Risk factors:</u></p>		
<p>1: This is non-disjunction disorder of sex-chromosome during oogenesis in mothers. so such mother produce egg (gamete) having no x-chromosome called Nullo-gamete (0A). when such egg is fertilize by sperm having x-chromosome the resulting baby will be of turner syndrome (45-chromosomes).</p>		
<p>2: The complete absence of x-chromosome generally occurs</p>		

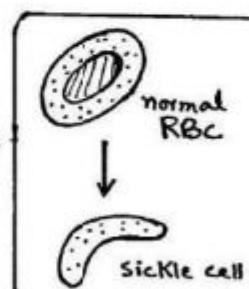
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because of an error in the father sperm or in the mother egg.		
3: In some cases, an error occurs in cell division during early stages of fetal development.		
<u>Signs and symptoms :-</u> Some common symptoms are below-		
1: More than 95% women have short stature.		
2: Such female will be sterile, ovaries will be non-functional, sex-hormones like estrogen and progesterone are not produced as required so they don't start menstruation at puberty.		
3: Breast are poorly developed. However other genitalia are - normal so pregnancy with donor embryo may be possible.		
4: Middle ear infection at childhood leading to hearing loss, <small>some time.</small>		
5: Normal intelligence with good verbal skill and reading. — However problems may occur in memory skills, fine finger movement, statistics and mathematics like subjects.		
6: Difficulty in social situations and Delayed growth.		
7: wide or web like neck and drooping eyelid.		
8: Swelling of the hands and feet especially at birth.		
9: Small lower jaw and low set ears at birth.		
10: Broad chest and short fingers and toes at birth.		
<u>Treatment of Turner Syndrome :-</u>		
Having appropriate medical treatment and family support — allows a woman with Turner syndrome to lead a normal, healthy and happy life. Treatments include —		
1: Estrogen and progesterone replacement therapy can help		

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<p>in breast development, menstruation and prevents osteoporosis.</p>		
<p>2: Some women with Turner syndrome can become pregnant with the donation of an egg or embryo.</p>		
<p>3: Growth Hormones are injected to increase body height and growth.</p>		
<p><u>3: Down Syndrome :—< Mongolism > (ETEA-2018)</u></p>		
<p><u>condition :-</u> It is autosomal non-disjunction or Trisomy ($2n+1$)</p>		
<p><u>Introduction :-</u> The autosomal non-disjunction of chromosome pair No-21 is called Down syndrome. Such individual will have one extra copy of 21st-chromosome i.e. They have 47-chromosomes ($2n+1$) instead of 46-chromosomes.</p>		
<p><u>Doctor Langdon Down :-</u> Down syndrome is named after Doctor Langdon Down who in 1866 first described the syndrome as a disorder. In 1959, Dr. Jerome Lejeune first time discovered the genetic origin of Down syndrome.</p>		
<p><u>Causes and risk factors :</u></p>		
<p>1: It is due to non-disjunction of chromosome No-21, in which 21st-chromosome fails to separate and thus one-gamete receiving 24-chromosomes. If this gamete is fertilized by normal gamete (n) than individual will have 47-chromosomes instead of 46-chromosomes.</p>		
<p>2: Advancing maternal age increase the incidence of Down syndrome.</p>		
<p>3: Both men and women can pass the genetic translocation for Down-Syndrome onto their offsprings.</p>		

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<u>Signs and Symptoms of Down Syndrome:</u>		
1: Small mouth, short neck, flat face and nose.		
2: Small ears, upward slanting eyes with small skin fold at inner corners. 3:- large protruding tongue.		
4: Inner corner of eye may be rounded instead of pointed.		
5: Hands are short and broad with short fingers and with single crease or line in palm. susceptibility to respiratory infection.		
6: Iris of eye will have white spots. 7:- poor muscle tone.		
8: Over all growth poor. 9: Mentally retarded.		
10: Excessive flexibility. 11: Average IQ is low (around 50)		
12: loose ligaments are also common. Heart defects.		
<u>Treatment of Down syndrome</u> :- Down syndrome is a genetic disease and every cell of an individual is diseased but still no specific treatment is known, However it can be managed by taking some measures like—		
1: Corrective surgery for heart defects, stomach, etc.		
2: Regular check up of ear-infection, hearing loss, obesity, hypothyroidism, visual impairments and other medical conditions.		
3: Down syndrome individual should have to supported by family.		
4: Special programs after children with Down syndrome — stimulation at an early age with appropriate sensory, motor and cognitive activities.		
Q17 Discuss different diseases induced by Gene mutation		
<u>Answer: Diseases induced by Gene (point) mutation:</u>		

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The following diseases caused by gene or point mutation.		
<u>1: Sickle cell Anemia :</u>		
<u>Biological Name :-</u> <u>Dyserythropoiesis.</u>		
<u>Definition :-</u> sickle cell anemia is an <u>autosomal recessive</u> genetic disorder characterized by abnormal, rigid, sickle shaped red blood cells which leads to shortage of RBCs (Hb)		
<u>Explanation :-</u> sickle cell anemia is an <u>autosomal recessive</u> genetic blood disease in which the red blood cells become rigid and sticky and are shaped like sickles or crescent moons and loss flexibility. These irregularly shaped -cells can get stuck in small blood vessels which can slow or block blood flow and oxygen to part of the body.		
<u>causes and risk factors :</u> (ETEA-2005)		
1: Sickle cell anemia is caused by point mutation in the gene (HbA) located on chromosome No-11, that tell the body to make hemoglobin, as a result abnormal hemoglobin is produced which cause red blood cells to become rigid, sticky and mis-shaped. Such mutation produce recessive allele.		
2: It is caused by a recessive allele (Hbs) which encode - defective β -globin-chain as a result abnormal hemoglobin is formed called haemoglobin-S.		
3: In sickle cell anemia the Glutamic acid (amino-acid) is replaced by valine amino-acid due to point mutation. as a result abnormal β -globin chain or Hbs is produced.		

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4: Hbs gene is express only in Homozygous recessive form. if an individual has one recessive allele (Heterozygous) then the individual will be normal.		
5: The sickle cell gene is passed from generation to generation in a pattern of inheritance called autosomal recessive — inheritance i.e. the patients inherit two such alleles from both parents.		
<u>Sign and symptoms:</u>		
1: Anemia — low number of RBC. 2: infections and episodic pain. 3: shortness of breath and fatigue 4: Repeated infection. 5: Yellow of eyes and skin called jaundice. 6: painful episodes (called crises) which affect back-bone, long bones and chest. 7: Swollen hands and feet may be the first signs of sickle cell anemia in babies. 8: — Vision problems. 9: Delayed growth and development in children.		
<u>Treatment of Sickle Cell anemia:</u>		
1: Folic-acid supplement. 2: Hydroxyurea (Hydrea) medicine for severe pain. 3: Antibiotics (penicillin) 4: Analgesics and plenty of fluids. 5: Vaccination to prevent infection. 6: Blood transfusion in severe anemia case. 7: Supplemental oxygen 8: — Stem cell or bone marrow transplant. 9: phenylketonuria :—<PKU>		



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<p><u>Definition</u> :- The condition in which phenylalanine appears in urine is called phenylketonuria —OR— “The condition in which baby is born without the ability to break down — phenylalanine into tyrosine is called phenylketonuria.”</p>		
<p><u>Causes and risk factors</u> :- (ETER-2014)</p>		
<p>1: PKU is an autosomal recessive genetic disorder whose gene is located on chromosome No-12. It is express only in homozygous recessive condition.</p>		
<p>2: For a child to inherit PKU, both the mother and father must have and pass on the defective (recessive) gene.</p>		
<p>. The point mutation cause PKU. The PKU gene contains the instructions for making an enzyme called phenylalanin-hydroxylase needed to breakdown the amino-acid called phenylalanine. In a person with PKU, this gene is defective, causing a completely or nearly complete deficiency of the enzyme, as a result the level of phenylalanine build up in the body which damage nerves in central nervous system.</p>		
<p>4: A dangerous build up of this phenylalanine can develop — when a person with PKU eats foods that are high in protein.</p>		
<p><u>Signs and symptoms</u> :</p>		
<p>1: Skin, hair, eye, colour lightens due to less or no melanin production.</p>		
<p>2: Head size of baby abnormal and mental retardation.</p>		
<p>3: Jerky movement of arms and legs.</p>		
<p>4: Tremors and unusual posturing of hands.</p>		

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5:	musty odour from skin , breath and urine.	
6:	Hyperactivity.	7: Stunted growth.
8:	skin rashes.	9: Small head size
10:	Behavioral or social problems.	
Treatment of phenylketonuria :		
1:	Treatment involves a diet which is extremely low in - phenylalanine particular when the child is growing.	
2:	Avoid milk, eggs, and meat due to high protein content.	
3:	Avoid aspartame (Nutra sweet) and artificial sweet.	
4:	Neutral amino-acid therapy powder or tablets.	
5:	Lofenadac formula for babies can be used throughout life because it has low level of phenylalanine.	
 End		
For Your Information		
The average IQ of children with Down syndrome is around 50, compared to normal children with an IQ of 100. A small number have a severe to high degree of intellectual disability.		

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Exercise		
Exercise MCQs		
A . Multiple Choice Questions (MCQs).		
<p>1. which of the following is not exhibited by DNA double helix ?</p> <p>a. antiparallel configuration b. complementary base pairing c. major and minor grooves <input checked="" type="radio"/> d. uracil</p> <p>2. If DNA of a particular species was analyzed and it was found that it contains 27% Adenine, what would be the percentage of Cytosine?</p> <p><input checked="" type="radio"/> a. 27 % b. 30 % c. 23 % d. 54 %</p> <p>3. Hershey and Chase experiment confirmed that DNA was the hereditary material on the basis of the finding that radioactive :</p> <p>a. phage were found in the pellet b. cells were found in the supernatant c. sulfur was found inside the cell <input checked="" type="radio"/> d. phosphorus was found in the cell</p> <p>4. During proofreading, which of the following enzymes reads the DNA?</p> <p>a. primase b. topoisomerase c. DNA ligase <input checked="" type="radio"/> d. DNA polymerase-III</p> <p>5. UGA, UAG, and UAA are regarded as:</p> <p>a. complementarity <input checked="" type="radio"/> b. nonsense codons c. universality d. degeneracy</p> <p>6. Nullisomy can be represented as:</p> <p><input checked="" type="radio"/> a. $2n - 2$ b. $2n + 2$ c. $2n - 1$ d. $2n$</p> <p>7. A chromosome with unequal length of its arms is:</p> <p>(a) Metacentric <input checked="" type="radio"/> (b) Sub metacentric (c) Acrocentric (d) Telocentric</p> <p>8. In Meselson & Stahl experiment, the DNA from sample at 20 minutes, after centrifugation it made sediments at the:</p> <p>(a) Top (b) Bottom <input checked="" type="radio"/> (c) Intermediate (d) Top & intermediate</p> <p>9. Which of the following act as a stop codon?</p> <p>(a) UGG (b) UGC <input checked="" type="radio"/> (c) UAG (d) UGU</p> <p>10. In mitochondria UGA codon act to specify ----- instead stop codon:</p> <p>(a) Arginine (b) Valine (c) Glutamic acid <input checked="" type="radio"/> (d) Tryptophan</p> <p>11. If an mRNA is synthesized with all the different codons, what is the minimum number of amino acids in the protein that is formed by mRNA:</p> <p>(a) 64 <input checked="" type="radio"/> (b) 62 (c) 60 (d) 66</p> <p>12. In eukaryotic mRNA molecule there are 90 nucleotide involved in translation process. What is the number of amino acid in the protein formed by this mRNA molecule? <input checked="" type="radio"/> (a) 29 (b) 30 (c) (d) 90</p> <p>13. In Griffith experiment mice developed pneumonia when they were injected with</p> <p>(a) R-type bacteria (b) heat killed S-type bacteria (c) heat killed R-type bases <input checked="" type="radio"/> (d) heat killed S-type bacteria along with live R-type bacteria.</p> <p>14. If the codon consisted of only two nucleotides, there would be how many possible codons? (a) 4 (b) 8 <input checked="" type="radio"/> (c) 16 (d) 20</p>		

Short Questions

Q1: — Differentiate the concept of monoploid and haploid.

Answer: Monoploid vs Haploid:— $(x:n)$

1: Monoploid :— The term monoploid refers to a cell or an organism that has a single set of chromosome (x).

Example :— $2n = x = 7$ in barley, or $2n = x = 10$ in corn.

2: Haploid :— Haploid describe a cell that contain a single set of chromosomes which are not paired (n).

Example :— In humans, gametes are haploid cells that — contain 23-chromosomes.

Difference b/w monoploid and haploid :— monoploids have a single basic set of chromosomes while haploids, on the other hand represent individuals having half the somatic chromosome number found in normal individual.

Q2: — List are the types and role of histone protein in chromosome

Answer :— See Question No-2 (write only Histone topic in DNA chemical composition of Question No-2).

Q3: — Give any two evidences provided by Sutton in favour of chromosome theory of inheritance.

Answer :— See Question No-4 for following explanation →

1: 1st Evidence :— plz write only parallel behaviour of gene + chm.

2: 2nd Evidence : write only mechanism of sexual reproduction.

Q4: — what was the conclusion of Avery's experiment.

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<p><u>Answer</u> :- See Question No-5. (write only - Avery's exp;).</p>		
<p><u>Q5</u>:- Differentiate b/w conservative and semi conservative models of DNA-replication.</p>		
<p><u>Answer</u> :- See Q-6 (write only conservative and semiconservative)</p>		
<p><u>Q6</u>:- Give a brief comparison b/w RNA and DNA polymerase.</p> <p><u>Ans</u>: <u>DNA polymerase</u>:</p> <ul style="list-style-type: none">1: It used in DNA replication2: It synthesizes a double-stranded DNA molecule.3: It uses DNA nucleotides to synthesize a new strand.4: It active during S-phase of the interphase.5: It requires a primer to begin the replication process.6: It has a low errors rate.	<p><u>RNA polymerase</u>:</p> <ul style="list-style-type: none">1: It is used in transcription.2: It synthesizes a single-stranded RNA molecule.3: It uses RNA nucleotides to synthesize a new strand.4: It active during G₁ and G₂ phases of the interphase.5: It do not require a primer to begin the transcription process.6: It has a comparatively high errors rate.	
<p><u>Q7</u>:- Genetic codes are universal but not quite universal. Analyze this statement.</p> <p><u>Answers</u>: <u>Genetic code are universal</u> :- Genetic code is universal i.e. genetic code is identical in all organism.</p> <p><u>For example</u> :- codon - AGA specifies or work for - arginine in bacteria, in human and all other organisms whose genetic code has been studied.</p> <p><u>Genetic code are not quite universal</u> :- The study of -</p>		

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<p>Genetic code of mitochondrial-DNA show that genetic code is not that much universal.</p> <p>For example:- UGA-codon is normally a stop-codon but in mitochondria it works or code for Tryptophan amino-acid.</p> <p>Q8: - Summarize the structure of a eukaryotic gene. <u>Answer</u> :- see Question No-3. (write only structure of gene).</p> <p>Q9: - Define RNA Splicing. <u>Answer</u> :- See Question No-10 (write only last topic of Q-10)</p> <p>Q10: - what is the structure of lac Operon. <u>Answer</u> :- See Question No-13. (write on structure of lac-operon)</p>		
<p style="text-align: center;">Long Questions</p> <p>Q1: - Critically analyze the history of chromosome theory of inheritance <u>Answer</u>: see question No-4</p> <p>Q2: - Prove that an evidence of DNA as heredity material. <u>Answer</u>: see question No-5</p> <p>Q3: - Elaborate the work of Meselson & Stahl to justify the semi conservative replication as a correct model of replication. <u>Answer</u>: see question No-7</p> <p>Q4: - Describe the events of the process of DNA replication. <u>Answer</u>: see question No-8</p> <p>Q5: - Describe post transcriptional modification of mRNA. <u>Answer</u>: see question No-10</p> <p>Q6: - Explain the process of translation of mRNA into polypeptide. <u>Answer</u>: see question No-12</p> <p>Q7: - Discuss the regulation of gene expression with help of lac operon model. <u>Answer</u>: see question No-13</p> <p>Q8: - Describe cause, symptoms, and treatment of Down's syndrome. <u>Answer</u>: see question No-16 (write only 3- Down's syndrome)</p>		

— End —

Chapter-23	SS2	ETEA - MCQs															
Previous Entry Test MCQ's of Biology Chapter No -23: (Chromosomes and DNA)																	
2005 ETEA																	
<p>1. DNA and histones together form a bead like structure called:</p> <p>(a) Mesosome (b) Polysome (c) Nucleosome (d) Centrosome</p>		<p>(c) Opposite (d) None</p> <p>9. A true column of chromosomes number in Garden-pea, onion and tobacco is:</p> <table style="width: 100%; border-collapse: collapse;"> <thead> <tr> <th style="text-align: left;">Garden pea</th> <th style="text-align: left;">Onion</th> <th style="text-align: left;">Tobacco</th> </tr> </thead> <tbody> <tr> <td style="text-align: left;">(a). 13</td> <td style="text-align: left;">15</td> <td style="text-align: left;">17</td> </tr> <tr> <td style="text-align: left;">(b). 14</td> <td style="text-align: left;">16</td> <td style="text-align: left;">48</td> </tr> <tr> <td style="text-align: left;">(c). 15</td> <td style="text-align: left;">17</td> <td style="text-align: left;">49</td> </tr> <tr> <td style="text-align: left;">(d). 16</td> <td style="text-align: left;">18</td> <td style="text-align: left;">50</td> </tr> </tbody> </table>	Garden pea	Onion	Tobacco	(a). 13	15	17	(b). 14	16	48	(c). 15	17	49	(d). 16	18	50
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(a). 13	15	17															
(b). 14	16	48															
(c). 15	17	49															
(d). 16	18	50															
<p>2. In sickle cell Anemia only one glutamic- acid of normal hemoglobin is replaced by:</p> <p>(a) Valine acid (b) Alanine acid (c) Arginine acid (d) Methionine</p>		<p>10. Condensation of chromosomes reaches to its peak during early:</p> <p>(a) Prophase (b) Metaphase (c) Anaphase (d) Telophase</p>															
2007 ETEA																	
<p>3. The pneumococcal strain used by Griffith in his experiments was:</p> <p>(a) Lophotrichous (b) Amphitrichous (c) Atrichous (d) Monotrichous</p>		<p>11. In chromosomes, the material controlling heredity is:</p> <p>(a) Histone (b) RNA (c) DNA (d) All of them</p>															
<p>4. Which of these are carriers of genetic information:</p> <p>(a) tRNA (b) mRNA (c) DNA (d) Nucleotides</p>		<p>12. Which one among the following possesses a double-ringed structure:?</p> <p>(a) Cytosine (b) Adenine (c) Uracil (d) Thymine</p>															
2008 ETEA																	
<p>5. Anticodon of AUG will be:</p> <p>(a) TAC (b) UAC (c) AUG (d) CCA</p>		<p>13. Which one of the following disease is due to point mutation?</p> <p>(a) Down syndrome (b) Kline filter syndrome (c) Phenylketonuria (d) Turner syndrome</p>															
<p>6. The term BIVALENT means:</p> <p>(a) Two chromatids (b) Two chromosomes (c) Four chromatids (d) Four chromosomes</p>		<p>14. Increased products of RBC are called:</p> <p>(a) Leukemia (b) Polycythemia (c) Edema (d) Anemia</p>															
<p>7. If the sequence of the one strand of DNA is ATGCTC, the sequence of the other strand would be:</p> <p>(a) CACGTC (b) TAGCATG (c) TACGAG (d) GACGTG</p>		<p>15. An individual has an additional sex-chromosome which syndrome does it refer to</p> <p>(a) Down's syndrome (b) Turner's syndrome (c) Jacob's syndrome (d) Kline filter's syndrome</p>															
2009 ETEA																	
<p>8. Two parent's strands of DNA molecules are:</p> <p>(a) Parallel (b) Antiparallel</p>		<p>2010 ETEA</p>															
<p>16. The number of chromosomes of tobacco during replication plant are:</p>																	
2011 ETEA																	

Chapters-23	SS3	ETEP-MCQs
(a) 43 . (b) 29 (c) 46 (d) 48		
17. which sequence of nucleotides would bond with the D.N.A sequence TATGA:		chromosomes that an individual possesses is called as:
(a) AUAGA (b) ATACA (c) UAUGA (d) ATACT		a) Genotype b) Phenotype c) Karyotype d) Genome
18. The number of nitrogenous base common in both D.N.A and R.N.A are:		27. If the coding sequence on the DNA is AATTGCT, the sequence in the mRNA will be
(a) Two (b) Three (c) Five (d) Four		a) AAUUCGT b) UUAACGA c) TTAACGA d) UUUTCGT
2012 ETEA		28. Replication of DNA occurs during-
19. Replication of D.N.A occurs in:		a) Interphase b) Prophase c) Metaphase d) Anaphase
(a) Interphase (b) Prophase (c) Metaphase (d) Anaphase		29. A specific nucleotide sequence on DNA molecule to which RNA polymerase attach to initiate transcription of mRNA from a gene is called--
2013 ETEA		a) Polygene's b) Genome c) Promoter d) Pleiotropy
20. Replication progresses at a rate of about 50 base pairs per second in:		2015 ETEA
(a) Bacteria (b) Virus (c) Eukaryote (d) All of the above		30. In mitochondria UGA codon act to specify--
21. Avery, Macleod and McCarty repeated the Griffith experiment in the year:		a). arginine b). glutamic acid c). tryptophan d). valine
(a) 1869 (b) 1928 (c) 1944 (d) 1952		2016 ETEA
2014 ETEA		31. Stop codons are...
22. Both DNA and RNA are synthesized by the Process of----		a) UAA, UAG, UGA b) UGC, UCG, AAA c) UUG, UCG, UCA d) UAA, UGC, UCA
a) Transcription b) Replication c) Polymerization d) PCR		32. The mutation that occurs in an egg or sperm cell, or those that occur just after fertilization are called.....mutation.
23. Gene and chromosomes show parallel behavior except.		a) New b) De novo c) Drift d) Both A and B
a) Number b) Inheritance c) Heredity d) Composition		2017 ETEA
24. In eukaryotes, DNA replication proceeds at the rate of.		33. DNA polymerase adds nucleotide to the 3rd end of the primer so the direction of replication will be?
a) 50 base pairs per second b) 40 base pairs per second c) 30 base pairs per second d) 20 base pairs per second		(a) 5' to 3' (b) 3' to 5' (c) 3' end of the primer to 3' end of template stand (d) 3' end of template strand to the 3' end of the primer
25. Which of the following is enzyme lacking Disease---?		
a) PKU b) Alkaptonuria c) Anuria d) Diarea		
26. The particular array of		

Chapter-8-23	554	ETEA - MCQs																																																
34. The experiments by Hershey and Chase helped confirm that DNA was the hereditary material on the basis of the finding that:	(a) 1 (b) 2 (c) 4 (d) 6	2019 ETEA																																																
(a) Radioactive phage were found in the pellet (b) Radioactive phage were found in the supernatant (c) Radioactive sulphur was found inside the cell (d) Radioactive phosphorus was found in the cell																																																		
35. How many nucleotides are 12 mRNA codons?		43. DNA polymerase III works always in:																																																
(a) 12 (b) 24 (c) 36 (d) 48		(a) 5' – 2' direction (b) 5' – 3' direction (c) 3' – 5' direction (d) 2' – 5' direction																																																
36. Which of the following is a non-sense codon?		44. Particular amino acid and tRNA molecule binds together by the action of an enzyme named:																																																
(a) UGA (b) UAU (c) CAU (d) GUA		(a) tRNA synthase (b) Amino tRNA synthase (c) tRNA ligase (d) Aminoacyl tRNA synthase																																																
37. If a disorder is not present in a child family but the fetus itself is infected before birth, it is known as?		45. The ribosomes responsible for protein synthesis are present in the cell:																																																
(a) Somatic mutation (b) Hereditary mutation (c) Germ line mutation (d) De novo mutation		(a) Floating in the cytosol (b) Localized in the nucleus (c) Bound to rough endoplasmic reticulum (d) Both a and c																																																
38. What will happen if a nucleotide is deleted from a gene having 9 nucleotides in its transcriptional unit?		46. _____ enzyme need a primer for the initiation of its function:																																																
(a) Change in phenotype (b) No change in phenotype (c) Synthesis of 3 amino acids (d) Synthesis of 4 amino acids		(a) RNA polymerase (b) DNA polymerase (c) Primase (d) Ligase																																																
2018 ETEA		47. The following histone proteins form a nucleosome complex except:																																																
39. In protein synthesis the initiator tRNA carrying amino acid methionine lands on which site of ribosome:		(a) H1 (b) H2A (c) H2B (d) H3																																																
(a) E site (b) P site (c) A site (d) C site		2020 ETEA																																																
40. Polyploidy is more common in:		48. At what phase the DNA content of a cell is doubled?																																																
(a) Plants (b) Animals (c) Bacteria (d) Virus		a. Prophase b. Interphase c. Anaphase d. Telophase																																																
41. Male having Down's syndrome have sex chromosomes:		Key:-																																																
(a) XXY (b) XY (c) XYY (d) XYYY		<table border="1"> <tr><td>1.c</td><td>2.a</td><td>3.c</td><td>4.b</td><td>5.b</td><td>6.b</td></tr> <tr><td>7.c</td><td>8.b</td><td>9.b</td><td>10.b</td><td>11.c</td><td>12.b</td></tr> <tr><td>13.c</td><td>14.b</td><td>15.d</td><td>16.d</td><td>17.d</td><td>18.b</td></tr> <tr><td>19.a</td><td>20.c</td><td>21.c</td><td>22.c</td><td>23.a</td><td>24.a</td></tr> <tr><td>25.a</td><td>26.a</td><td>27.b</td><td>28.a</td><td>29.c</td><td>30.c</td></tr> <tr><td>31.a</td><td>32.d</td><td>33.a</td><td>34.d</td><td>35.c</td><td>36.a</td></tr> <tr><td>37.d</td><td>38.a</td><td>39.b</td><td>40.d</td><td>41.b</td><td>42.d</td></tr> <tr><td>43.b</td><td>44.d</td><td>45.d</td><td>46.a</td><td>47.a</td><td>48.b</td></tr> </table>	1.c	2.a	3.c	4.b	5.b	6.b	7.c	8.b	9.b	10.b	11.c	12.b	13.c	14.b	15.d	16.d	17.d	18.b	19.a	20.c	21.c	22.c	23.a	24.a	25.a	26.a	27.b	28.a	29.c	30.c	31.a	32.d	33.a	34.d	35.c	36.a	37.d	38.a	39.b	40.d	41.b	42.d	43.b	44.d	45.d	46.a	47.a	48.b
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43.b	44.d	45.d	46.a	47.a	48.b																																													
42. Amino acid leucine is coded by how many codons:																																																		

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