undergo polymerization under low oxygen tension causing change in shape of the REC from biconvex 4:11iSc teielorisated sickle like structure.

c. Pherrylikkonuirla• inborn error of metabolism inherited as autosornal recessive trait. The affected individual lacks an enzyme that converts the amino acids phenylalanine to tyrosine that results into mental retardation.

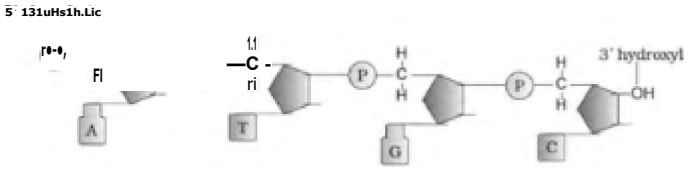
Chromosomal Clisorders-Failure of segregator' of chromatids during tell division results iri loss or gain of chromosome called aneuplaidy. The failure of cytokirbesis leads to two sets of chromosome called polyploldy,

a. Denim[P]s Syndrome. is due to presence of additional topyr of the chreimotorne number 21. The affected individual is short Mtlirecl with small rounded head, furrowed tongue and partially opened rrbOUthi. Mental Clew ell5prnent 'FS retarded.

h. ~~Klinefleter's Syndrome- clue to presence of an additional copy of X-chrorrbosorne MY).~~ Such persons have owerall masculine development. They are sterile.

C. Turner's Syndrome – canted due to the a bserket of one of the X chromoszome. 45 with XIB, such females are Eterile *as* ovaries are rudimentary. They lack secondary sexual characters.

Cytosine is common for both DNA and ANA and Thymine is present in DNA. IJ radii is present In RNA at the place of Thyrnine.



nitrogenous base i5 linked to pentose sugar with N-glycosFdic. linkage to form to form a nucleoside_ When phosphate group is linked 5'90H of a nucleoside through phosphoester linkage nucleotide is formed. Two nucleotides are linked through 3'-5' phosphodiester linkage to form di nucleotide. More nucleotide joins together to form polynucleotide •

In RNA, nuckoticle residue has additional —C11.1 group present at T•position In nb•se and wadi Is found at the place of Thymine.

Double 1 lebi. Model for Struttura of DNA - James Watson and Frantis Crick. based on X ray diffraction data prcwiucecl by Wilkin and Rosalind proposed this model of DNA_

The sailent features of this model a re-

- DNA Is made of two pohnucleoticie chains In which backbone Is made up of sugar- phosphate and bases projected inside iL
- Two chains have anti-parallel polarity_
- The bases in two strands are paired through H-bonds. Adenirbe and Thy rnine forms double hydrogen bond and Guanine and Cytosine forms triple hydrogen bonds,
- The bases in two strands are paired through Fl-bonds. Adenine and Thymine forms double hydrogen bond and Guanine and forms triple hydrogen bonds
- Two chains are coiled in right handed fashion. The pitch of helix is 3A nm and roughly 10 by in each to m_
- The plane of one base pair stacks over the other in double helix to confer stabillty.

Pay king of DNA helix-

- in prokaryotes, well defined nucleus is absent and negatively charged DNA Is combined with 50me positively charged proteins called nucleolds.
- In eukaryrotes, histones, positively charged protein organized to form S rnbletuleg unit called histone votomer. Negatively charged DNA is wrapped around the Nacre orkomer to form nucleosome. $ingla nucleosome contains about 200 base pairs. Chromatin Is the repeating unit of nucleosome.

Reolkolon of DNA

Watson and Crick suggested that *two* strands of DRIP, separate from each other and act as template For synthesis of new complementary strands. After the completion of replication each DNA molecule would have one parental Arid one newly synthesised strand, this method is lled semitontervative replication_

Messe!son and stahl's shows experiment!l evidence of serniconservative replication. Using radioartikee Nitrogen

Replication of DNA require Enzyme DNA poiymerase that catalyse the poLvmerisation in one strand 5'43' only, So. replication **in one stand** is continuouA and othier strand it is distontinuouA due to the synthesized **fragments that are joined together by enzyme DNA ligase.**

replimenmer

trapargiptIock **tranrimElksa**
m1R2⅛A **protein.**

Centred docilsM.

Transcription is the process of copying genetic information from one strand of DNA Into RNA. In transcription **only one segment** of ~~**1314A and only One strerid is copied In RNA The Adenosine forms**~~ be pair **with** until ins.Lead **of Thyrni ne.**

Transcription of DNA includes a promoter, the structural gene and a terminator. The strands that has polarity **3' act as template and called template strand and other strand is called coding strand.**

- Promoter is located at 5 end and that Wild the enzyme RNA polymerase to start transcription. The terminator Is located at I'end of coding. stra mei and usually defines the end of transcription_
- Exons are those sequences that appear in (nature and pro-cesseci RNA. Exons are interrupted by introns. Introns do not appear in mature and processed RNA.

:or In bacteria there are three types of RNA's· rnRINA, t-RNA and r-RNA. All these are needed for synthesis of protein in the cells_

- The m·RNA provide the template.. t-RNA brings the amino acids and read the genetic code, the r-RNA play grurtural and catalytic role during translation.
- The primary transcript contains bath exon and intron and is non-functional. It undergoes the process of spilcirti in which iritrons are removed and exonsare joined in a defined order.
- The hnHNA heterogeneous nuclear RUN' undergo additional processing called as capping and tailing_ In calving Ire unusual nucleotide iirietlhiel guanosine triohosphate) to the 5'end of hnRNA_ In tailing a denylatelate is added at Tend in a tern plate at independent manner_

Genetic Code is the relationship of amino acids sequence in a polypeptide and nucleotide/base sequence in ruRNA, It directs the sequence of amino acids during synthesis of proteins.

Salient features of Genetic Code are-

i. The code is triplet. 61 cordons code for amino acids and 3 codons do not code for any an acids called stop cciclons..

ii. Codon is unam higuous and specific, code for one amino acid_

Lii. The code is degenerate. Some amino acids are coded by more than one codon.

iv, The codon Is read In rrifiNA lna contiguous fashion witthout any punctuation.

11. The codon is nearly universal. AUG has dual functions. It codes for rnethionine and also act as initiator co-don.

Mutations and Genetic code

4' A change of single base pair (point mutation) In the chain of Beta globin chain that results in the change of amino add residue glutamate to valine. These results into diseased condition called Sickle cell anaemia

- Insertion and deletion of three or its multiple bases insert or delete one or multiple codons hence one or morE amino atidS and reading frame remain unaltered from that point onwards. Such mutations are called frame-shift insertion or deletion mutations..

The t-RNA called as adaptor molecules. It has an anticodon loop that has bases complementary to code and also has an amino acid acceptor to which amino acid binds_ t-R NA is specific for each amino. acids_ It also contain another specific t-RNA referred as initiator t-FINA. The secondary structure of t-RNA is depicted as clover-leaf_ In actual structure, the t•RNA is a compact molecule which looklIke inverted L

- Translation is the process of polymerisation of amino acids to form a polypeptide_ The order and sequence of amino acids are defined by the sequence of bases in the mRNA. Amino acids are Joined bv peptide bonds_ It involved following steps-
  .r) Charging of t-P N.A.
  c Formation of peptide bonds between two charged tRNA_
- The start colon is AUG. An m R NA has Kirne additional sequence that are not translated called untr,3nsiated region
- For initiation ribosome binds to mRNA at the start codon. Ribosomes moves from codon to cotton along mRNA for elongation of protein chain_ At the end release factors binds to the stop oodon, termi mating the translation and release of polypeptide form ribosome.

The Lac Oberon

- Lac operon consists of one regulatory gene (1[ ] and three structural genes [10,z and all. Gene i code for the repressor of the lac operon. The z .ge-ne code for beta·galaciodase. that is responsible for hydrolysis of disaccharide. lactose Into monomeric units,. galactose and 8:lucose. Gene v code for perry ease, which increases permeability of the cell. Gene a encode for transacetylase.
- Lactose is the substrate for enryme beta-galactosidase and it regulates switching an and off of the operon, so It is called Inducer.
- Re.gulation of Lac operon is done by repressor referred as negative regulation. Operation of Lac operon is also under the control of positive regulation.

Human (enome Project was launched In 199:1 to find out the complete DNA sequence of human genorne using genetic engineering technique to isolate and done the DNA segment for determining DNA seq uence.

DNA finzerpfiriling is Bi very quick way to compare the DNA sequence of any two individual. It includes identifying difiepehres in some specific region in DNA sequence called a5 repetitive DNA because in this region, a small stretch of DNA is repeated many tirnes.

Depending upon the base composition, length of segment and number of repetitive units satellite DNA is classified Into maw). categories.

Polmorphism in DNA sequence Is the basis for genetic mapping of human genuine as well as fingerprinting_

- The technique of fingerprinting was Initially developed by Alec Jeffrey. He used a satellite DNA as probe to so high polymorphism was called Variable Number of Tendon Repeats O MR).