


Subject: Anthropology

Production of Courseware

 **-Content for Post Graduate Courses****Paper No. : 01 Physical/ Biological Anthropology****Module : 21 Introduction to Human Genetics**

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Description of Module	
Subject Name	Anthropology
Paper Name	01 Physical/Biological Anthropology
Module Name/Title	Introduction to Human Genetics
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Contents of this unit

1. Introduction
2. Cell
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4. DNA structure
5. Function of DNA
 - a. Replication
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6. Chromosome and cell division
 - a. Mitosis
 - b. Meiosis

Learning Objectives

- To understand what is genetics
- To have a brief idea about the structure and function of cell
- To have a comprehensive knowledge of DNA structure and its function
- To know the basic concept of chromosome and cell division

1. Introduction

The question that often comes in our mind is why we are morphologically similar with our parents and different from members of other families? In the same way, why members of a population group share some resemblance with each other and differ from those of other population group? Or why such similarity and variation do exist among individuals? Is there 'something' that we inherit from our parent from the time of our birth? Or do we share that 'something' with the members of our population group that result in having similarities, morphologically? Yes, we do inherit 'something' from our parents that make us look alike with them. That 'something' comes to known as 'gene'. The word 'gene' was first coined by Wilhelm Johannsen (1857-1927) in early 1900s without knowing the biochemical properties but just because it is a nice small word to describe that 'something'. And it was Bateson who defined heredity as 'similarity due to relatedness'.

The basic nature of the gene was described by an Austrian monk, Gregor Mendel more than a century ago after a careful experiment conducted in common garden pea between 1856 and 1868. Summarizing his findings in two laws, the gene was recognized as a "particulate factor" that passes unchanged from parents to progeny. He describe that each particulate factor or gene exist in pairs in individual organisms. When an individual has two different particulate factors or genes, only one is expressed and said to be *dominant* to other, which is said to be *recessive*. During gamete formation, the paired particulate factors separate or segregate randomly with equal likelihood. This is known as *mendel's law of segregation*. The second law is known as *mendel's law of independent assortment*. In this law he describe that during gamete formation, segregating pairs of particulate factors assort independently of each other. That is, each paired gene freely separate sand do not influence the separation of other paired gene during gamete formation. These two laws explain the basic fundamental of inheritance. However, his groundbreaking research was discovered only in 1900 by two independent scientists – William Bateson and Hugo de Vries, 16 year after his death and 35 years after he published it.

After the discovery of Mendel's idea of inheritance in 1900, a whole new science called **genetics** (Bateson coined the term in 1908) emerged. So, genetics is that branch of science which deals with the study of inheritance of unit factor called gene. These genes are present inside the nucleus in the form of DNA in every cell.

1. Cell

Cell is the basic unit of every life. It is a microscopic organic body which carries genetic materials and other organelles required for cellular functions. All living organisms are made up of cells. Some are made up of a single cell and called as unicellular organism. Like bacteria and protozoan. This single cell performed all the activities of being a life. Others, including human are made up of multiple cells

and hence called as multi-cellular organism. Among these organisms different activities are performed by different specialized cells.

All the living organisms on earth can be divided into **prokaryote** and **eukaryotes**. Prokaryotes are those organisms with no major compartment within the cell to separate the genetic materials from other components of the cell. Bacteria, blue-green algae are some of the examples of prokaryotes. Prokaryotes are the earliest forms of life on earth appeared in the fossil record about 3.4 billion year ago (Kostianowsky, 2000).

On the other hand, eukaryotes are characterized by having nucleus that separates the genetic materials from the rest of the cell components. In the fossil record, eukaryotic cell do appears in about 1.5 billion year ago. Eukaryotic cells resemble to those found today evolved somewhere around 850 million year ago and multi-cellular organism made their first appearance only 600 million year ago (Kostianowsky, 2000).

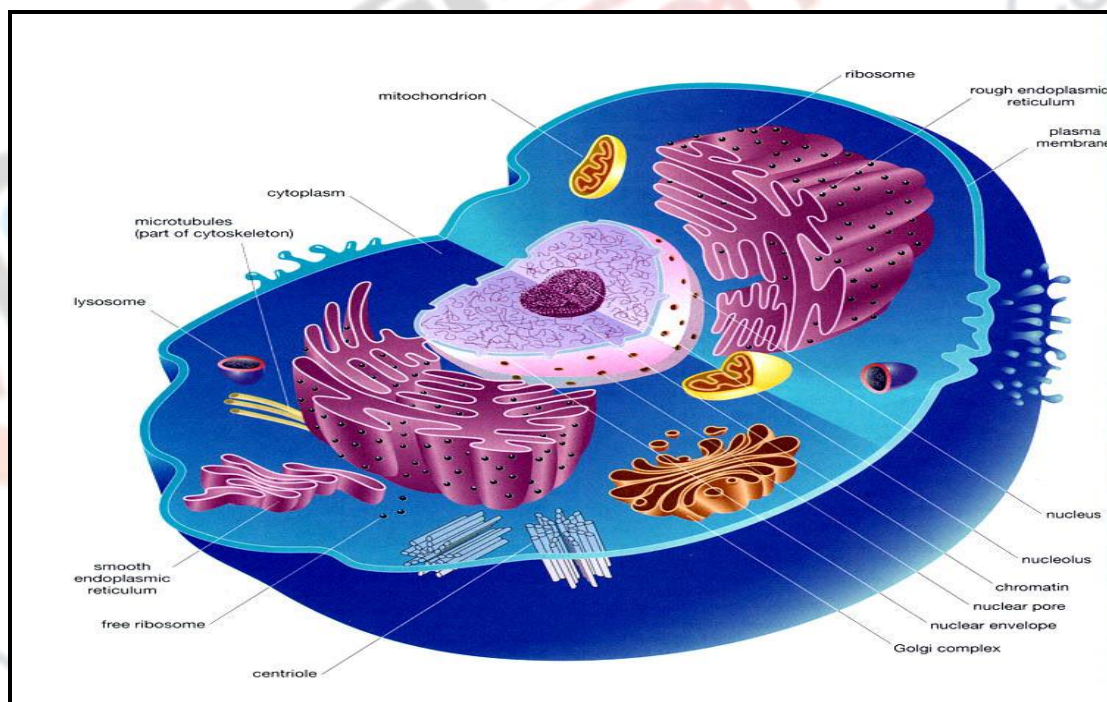


Fig1: a cell showing its components

(source: <http://www.thinglink.com/scene/522779055620096002>; accessed on 14th march 2015)

In multi-cellular organisms like human, different specialized cells perform different functions. For example, brain cell or neuron is structurally different from blood cell based on their different functions. However, cells in a complex multi-cellular organism can be divided into two types— **somatic** cell and

gametes cell. Somatic cells are body cells and do not directly involved in reproduction. Somatic cell again can be of different type base on different function. Whereas gametes are the sex cells which directly involved in reproduction. In human there are two types of gametes – *egg* (female gamete cell) and *sperm* (male gamete cell).

2. Cell anatomy

Generally eukaryotic cells share a similar structure. Every eukaryotic cell has a semi permeable membrane called **plasma membrane** that bound all the components of a cell together. Inside the cell all the organelles are float on a gel like substance called the **cytoplasm**. The most important and prominent structure in the cytoplasm is the **nucleus** which has its own thin envelope called **nuclear membrane**, separating its contents from the cytoplasm. Inside the nucleus contain two complex molecules – DNA and RNA. **DNA** or **deoxyribonucleic acid** is a double stranded complex molecule where genetic information is stored. **RNA** or **ribonucleic acid** is similar with DNA but have single strand. RNA is also found in cytoplasm. It is essential because it becomes the template for protein synthesis.

Another important organelle found in the cytoplasm is **mitochondrion** (*plural*, mitochondria). The main function of the mitochondria is energy production and hence is known as ‘power house’ of the cell. It is a capsule shaped structure and carry its own genetic material called mitochondrial DNA or mt DNA. Structurally and functionally, mt DNA is similar to that of nuclear DNA but differ in the pattern of organization. An mt DNA is an important tool for evolutionary studies. It follows the maternal line of inheritance, i.e. Inherited from mother to her progeny only. This is because father’s mt DNA doesn’t involve in zygote formation.

Another complex structure look like a folded sheet called **endoplasmic reticulum** (er) is found in the cytoplasm. The main function of er is to increase surface area within the cell for metabolic reaction to take place. **Ribosome** is another organelle within the cytoplasm which main function is protein synthesis. Ribosome which are attached to the er is called as attached ribosome and those found floating in the cytoplasm as free ribosome.

3. DNA structure

DNA is the hereditary material contain inside the nucleus. DNA carries the inherited (from parents) genetic information required for life. DNA directs all the cellular functions of an organism through proteins. So, in order to understand the mechanism of genetics, it is necessary to know the structure and function of DNA molecule.

The exact physical and chemical properties of DNA molecule were successfully discovered by two young scientists - James Watson and Francis Crick in 1953. Watson and Crick developed their model of DNA base on some of the properties of DNA which has already established by other scientists. In the

late 1940s and early 1950s, biochemist Erwin Chargaff and his colleagues have discovered that DNA consists of four different nitrogenous bases and these bases are distributed in a unique pattern. In a given sample of DNA, the amount of adenine is similar to the amount of thymine and amount of cytosine to that of guanine. Another two scientists, Rosalind Franklin and Maurice Wilkins were also successfully able to produce an x-ray crystallography of DNA molecule clearly showing that the structure is helical. Discovery of DNA molecular structure have altered our understanding of biological and evolutionary mechanism and completely revolutionized the field of biology and medicine. For their famous discovery, Watson, Crick and Wilkins were awarded nobel prize in 1962 but Rosalind Franklin died four years before the award at the age of 37 year because of cancer.

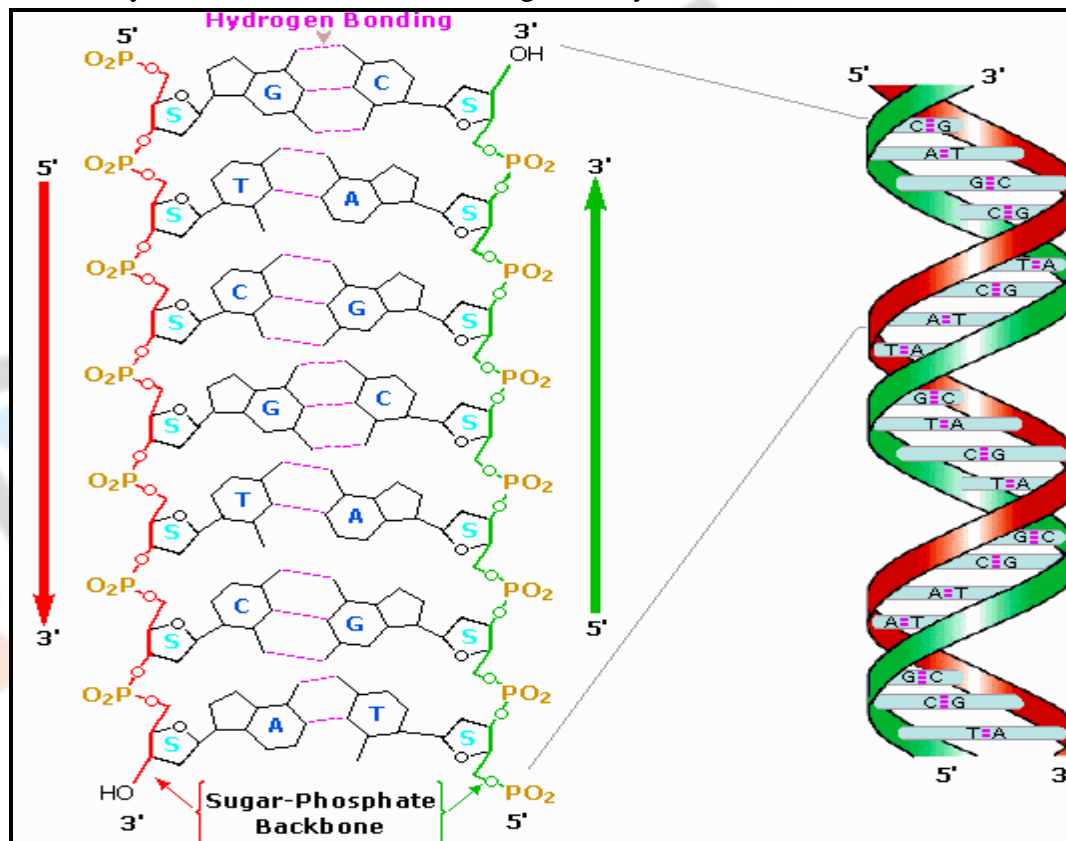


Fig 2: structure of DNA molecule

(source: <https://www2.chemistry.msu.edu/faculty/reusch/virttxtjml/nucacids.htm>; accessed on 14th march 2015)

The structure of DNA molecule is double strands forming a double helix resembling a ladder twisted around its central axis. Each strand is made up of a chain of **nucleotides**. Nucleotide is the basic unit of DNA (and RNA) molecule. Each nucleotide is again made up of three components – a sugar (deoxyribose), a phosphate unit and a nitrogenous base. There are four different types of nitrogenous bases belong to two classes – *purines* and *pyrimidines*. The purines are *adenine* (A) and *guanine* (G);

the pyrimidines are *cytosine* (C) and *thymine* (T). Based on these four different nitrogenous bases, there are four nucleotides which can be represented by the initial letter of each base – A – G – C – T.

In a nucleotide each phosphate is bond to a sugar molecule and each sugar molecule bond with a nitrogenous base. The nitrogenous base of a strand forms hydrogen bond with the nitrogenous base of the complimentary strand thereby resulting double stranded DNA molecular structure. The bonding between the nitrogenous bases followed a fix pattern and it is always A – T and C – G as it is possible to form two hydrogen bonds between a – t and three hydrogen bonds between c – g. In other word, the combination is between a purine and a pyrimidine. Purines are larger molecules than the pyrimidine and therefore this combination is necessary to maintain a constant distance between the two strands. For example, if a strand of DNA has a sequence of nucleotides as attcgcaagc, the corresponding strand will have TAAGCGTTCG sequence of nucleotides. So, if we consider the analogy of a ladder the side of the ladder is represented by the chain of sugar and phosphate and the rungs are represented by the nitrogenous bases and bonds forming between them.

RNA is similar to DNA except that –a) it is single stranded unlike DNA's double strands. B) deoxyribose is replaced by ribose as the sugar in nucleotide. C) pyrimidine base thymine is replaced by another pyrimidine base *uracil* (u) and bonds with adenine.

4. Function of DNA

The genetic information to build up an organism is transmitted and store as a sequences of nucleotides called DNA from parents to progeny. These complete set of genetic information for any organism is called **genome**. Genome itself does not perform any active role in the functioning of organism rather it carries the information for producing proteins in the appropriate time and place. So, all the cells of an organism should carry the exact genetic information. In order to achieve this, DNA should be able to replicate itself and also should be able to decode the information for protein synthesis.

a. Replication

Every multi-cellular organism starts from a single celled zygote. This single celled zygote was divided billion times to form a complex organism. So, when a mother cell divided into two daughter cells an exact copy of genetic material much be transmitted to each daughter cell. The structure of DNA molecule suggests the mechanism for its replication. Replication of DNA starts with the separation of double strands into two separate strands by breaking the weak hydrogen bonds between the nucleotide. Each of these separate strand acts as a template for the formation of a new complementary strand of DNA by assembling nucleotides. Every step of DNA replication is mediated by a particular enzyme. After completion, two copies of mother DNA molecule are formed, each having one original strand and one newly form strand. These two DNA molecules are than pass on to two daughter cells during cell division.

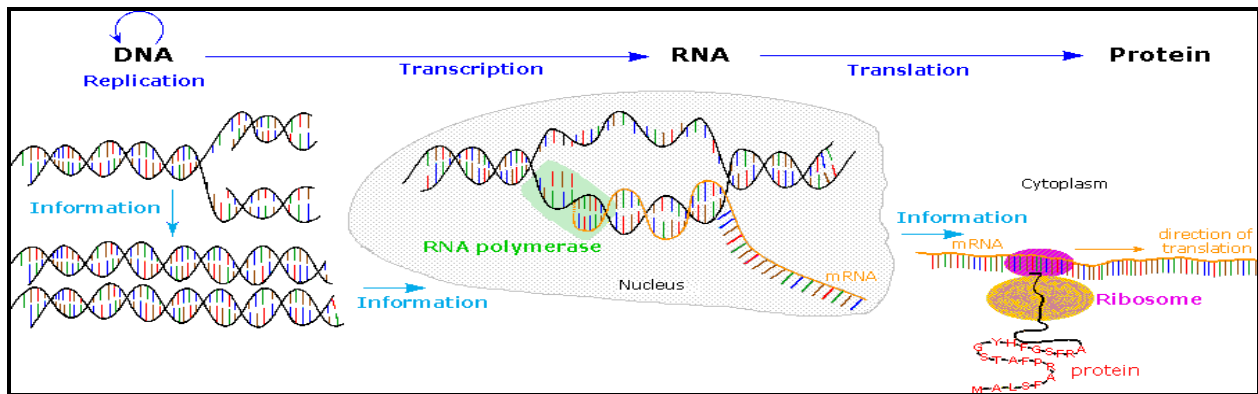
b. Protein synthesis

The cellular function of an organism is executed by various proteins. For example, *hemoglobin* found in red blood cells binds the oxygen molecule and carry to cells throughout the body. Similarly, *insulin* (protein) helps in metabolism of sugar molecules in the body. The production of these proteins in different time and place is possible only by following the genetic information stored in the DNA through the process called protein synthesis. Proteins are complex molecules made up of smaller molecules called **amino acids**. Amino acids have the ability to form long chains by bonding to each other. There are twenty amino acids; of which nine are not able to synthesis in the body and have to be obtained from diet. These are known as essential amino acids. Remaining eleven non-essential amino acids are produced inside the body (laidlaw and koppel, 1987). These amino acids line up in a particular sequence to produce a specific protein.

By now we know that nucleotide sequence of DNA carry the code for protein synthesis and protein is made up of chain of amino acids. So, the sequence of amino acids is ultimately determined by the sequence of the nucleotides. In DNA, a triplet of nucleotides or group of three nucleotides specifies an amino acid. For example, if the sequence of nucleotide is CGA then it specifies the amino acid *alanine* (see table 1). Similarly, a sequence of TTA GGA CTT GTC CTA AGA will give a chain of amino acids – *asparagines-proline-glutamic acid-glutamine-aspartic acid-serine*. A typical protein may be formed by a chain of about 200 amino acids.

The process of protein synthesis involved two steps – *transcription* and *translation*. Transcription takes place inside the nucleus. In this step, the nucleotide sequence coded for the particular protein is copied in the form of RNA called messenger RNA (mRNA). This mRNA exactly copies the nucleotide sequence of the DNA and become the temple for assembling the corresponding amino acids. The second step, translation takes place in the cytoplasm. So, the mRNA comes out of the nucleus through the nuclear membrane into the cytoplasm.

In the cytoplasm mRNA is bind at the ribosome and its information is translated in groups of three mRNA nucleotide called *codons*. Each mtDNA codon specifies one amino acid like a triple do in DNA. Another RNA called transfer RNA (tRNA) helps in forming amino acids chain in the ribosome. Each tRNA molecule binds to one specific amino acid. These tRNA molecules take the amino acid that matches the codon being translated to the ribosome. These amino acids join together in the sequence dictated by the sequence of mRNA codons. In this way amino acids form the required chain to be a protein.



(source: <https://www2.chemistry.msu.edu/faculty/reusch/virtualexperiments.htm>; accessed on 14th march 2015)

Fig 3: DNA replication and protein synthesis

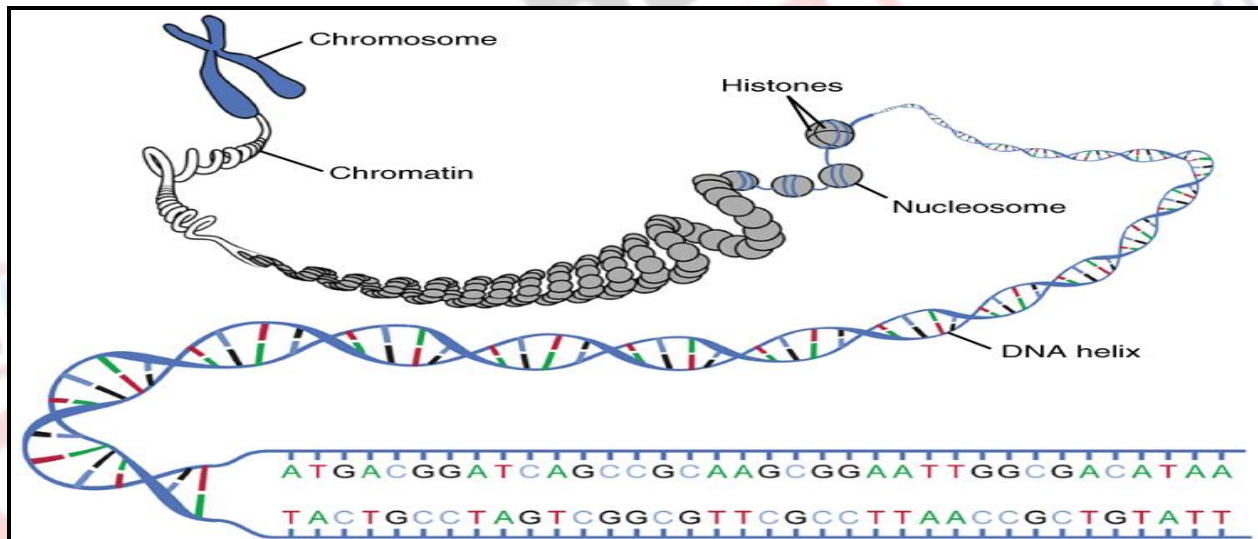
Table 1: list of amino acid with genetic codes

Amino acid	DNA triplets	MRNA codons
Alanine	CGA, CGG, CGT, CGC	GCU, GCC, GCA, GCG
Arginine	GCA, GCG, GCT, GCC, TCT, TCC	CGU, CGC, CGA, CGG, AGA, AGG
Asparagine	TTA, TTG	AAU, AAC
Aspartic acid	CTA, CTG	GAU, GAC
Cysteine	ACA, ACG	UGU, UGC
Glutamine	GTT, GTC	CAA, CAG
Glutamic acid	CTT, CTC	GAA, GAG
Glycine	CCA, CCG, CCT, CCC	GGU, GGC, GGA, GGG
Histidine	GTA, GTG	CAU, CAC
Isoleucine	TAA, TAG, TAT	AUU, AUC, AUA
Leucine	AAT, AAC, GAA, GAG, GAT, GAC	UUA, UUG, CUU, CUC, CUA, CUG
Lysine	TTT, TTC	AAA, AAG
Methionine (initial codon)	TAC	AUG
Phenylalanine	AAA, AAG	UUU, UUC
Proline	GGA, GGG, GGT, GGC	CCU, CCC, CCA, CCG
Serine	AGA, AGG, AGT, AGC, TCA, TCG	UCU, UCC, UCA, UCG, AGU, AGC
Threonine	TGA, TGG, TGT, TGC	ACU, ACC, ACA, ACG
Tryptophan	ACC	UGG
Tyrosine	ATA, ATG	UAU, UAC
Valine	CAA, CAG, CAT, CAC	GUU, GUC, GUA, GUG
Termination codons	ATT, ATC, ACT	UAA, UAG, UGA

Source: laidlaw and kopple, 1987

5. Chromosome and cell division

DNA, inside the nucleus exists as dispersed and uncoiled strands called **chromatin**. During cell division this chromatin become condense, coiled and tightly wound into a discrete structure called **chromosome**. Each chromosome has distinct shape and size which is partly determined by the position of **centromere**, a condensed and constricted region on the chromosome. The number of chromosome is fixed for every species; say, every human has 46 or 23 pairs of chromosome. Each somatic cell has two copies of each chromosome or *diploid* number of chromosome inherited one each from mother and father. But gamete cell has *haploid* number of chromosome (half the number of chromosome). The gene or the fragment of DNA coded for a protein is distributed across the chromosome. The location of a specific gene on a chromosome is called *locus*. Since somatic cell has two copies of chromosome, each gene is also exists in two copies, one at each copy of chromosome or locus. These alterative variant of a same gene is called **alleles**.



(source: http://cnx.org/resources/4e8553539bc5b4ae8654d3a621b80b76/0321_DNA_macrostructure.jpg; accessed on 14th march 2015)

Fig 4: chromosome and DNA

A. mitosis

Mitosis cell division occurs in somatic cell when it replicated to form two daughter cells. It is required for growth of the organism, healing of wounds or whenever new cells are required to replace the old one. The life cycle of a cell can be divided into different stages. When the cell is not involved in any cell division, it is called *inter-phase*.

The first stage of mitosis is called *prophase* characterized by disappearance of nuclear membrane; condense of chromatin to form chromosome composed of two identical sister *chromatids*. This is

followed by *metaphase* where the chromosomes are aligned at the center of the cell. Then the sister chromatids are split apart and migrated to opposite ends of the cell in *anaphase*. The final stage is called *telophase*, where the cytoplasm split forming two completely separate daughter cells.

Not every somatic cell undergoes mitosis. Red blood cell, brain and nerve cell (neurons) and liver cells usually do not reproduce once it is fully developed. Red cells are continuously produced inside the bone marrow.

B. meiosis

Meiosis has two divisions forming four daughter cells with haploid number of chromosomes unlike forming two daughter cells with diploid number of chromosomes in mitosis. It occurs in the specialized diploid cell in testes of male and ovaries of female. These haploid gametes produced through meiosis are necessary because when a sperm and egg are fertilized to form a zygote, the original diploid number of chromosomes is restored.

The first meiotic prophase is similar to the prophase of mitosis but with some critical differences like pairing of partner chromosomes forming four chromatids or tetrads. Pairing of chromosomes is an important phenomenon because exchange of genetic information called **crossing over** takes place. After crossing over, the cell begins to divide. The members of paired chromosomes separate and migrate to opposite ends of the cell forming two daughter cells but not identical to each other or to the mother cell. Each cell contains only a member of paired chromosome or haploid. The second meiotic division executes in the same way as mitosis occurred, resulting in four daughter haploid cells – two from each of the two daughter cells of the first meiotic division.

Summary

- The word 'gene' was first coined by Wilhelm Johannsen (1857-1927) in early 1900s without knowing the biochemical properties but just because it is a nice small word to describe that 'something'.
- After the discovery of Mendel's idea of inheritance in 1900, a whole new science called **genetics** emerged.
- Genetics is that branch of science which deals with the study of inheritance of unit factor called gene.
- Cell is the basic unit of every life.
- Cells in a complex multi-cellular organism can be divided into two types – **somatic** cell and **gametes** cell.

- Somatic cells are body cells and do not directly involved in reproduction. Somatic cell again can be of different type base on different function. Whereas gametes are the sex cells which directly involved in reproduction.
- DNA is the hereditary material contain inside the nucleus.
- The exact physical and chemical properties of DNA molecule were successfully discovered by two young scientists - James Watson and Francis Crick in 1953.
- The genetic information to build up an organism is transmitted and stored as sequences of nucleotides called DNA from parents to progeny. These complete set of genetic information for any organism is called **genome**.
- Mitosis cell division occurs in somatic cell when it replicated to form two daughter cells. It is required for growth of the organism, healing of wounds or whenever new cells are required to replace the old one.
- Meiosis has two division forming four daughter cells with haploid number of chromosome unlike forming two daughter cells with diploid number of chromosome in mitosis. It occurs in the specialized diploid cell in testes of male and ovaries of female.