CHAPTER 3

OVERVIEW OF GENETICS

3.1 Introduction

Each of us is composed of hundred trillions \((10^{14})\) of cells and all of them are derived from a single diploid cell called zygote which is the product of union of male and female gametes during reproduction. Each cell has a nucleus which is a master cell organelle, within which coloured bodies called chromosomes are present. Each of these chromosomes contains very thin fibres a few centimetres long that play a major role in defining who we are, as human being, an animal, a microbe, a plant or any living organism. These important intracellular fibers are made up of DNA (Deoxyribose Nucleic Acid) which is regarded as a master molecule and controls all our characters. Every time a cell divides, its DNA is replicated and apportioned equally to two daughter cells. The DNA content of these cells—what we call the genome—is thereby conserved. This genome has a master set of genetic instructions, in fact a whole library of information that cells use to maintain the living state. Ultimately, all the activities of a cell depend on it. To know the DNA is therefore to know the cell and in a larger sense, to know the organism to which that cell belongs to.

Genetics, the study of heredity in general and of genes in particular deals with the molecular structure and function of genes, gene behaviour in context of a cell or organism (e.g. dominance and epigenetics), patterns of inheritance from parents to offspring, and gene distribution, variation and change in populations, such as through Genome-Wide Association Studies. Given that genes are universal to living organisms, Genetics can be applied to the study of all living systems, from viruses and bacteria, through plants and domestic animals, to humans (as in Medical genetics).

Although the influence of heredity has been recognized since pre-historic times, scientific understanding of inheritance is a fairly recent event. The science of Genetics began with the work of Gregor Johann Mendel, who studied the nature of inheritance in plants and discovered the fundamental laws of inheritance. Genetics has
grown at an explosive rate since 1900, when Mendel’s work was discovered by the scientific world. During the last half century, geneticists have determined the molecular structure of Mendel’s “factors” and the mechanisms by which they control the characteristics of an organism.

The relevance of Genetics to many aspects of human life and society, including health, behaviour, food production, forensics, and even politics, is chronicled almost daily in the news media.

Genetics has had and continues to have a profound impact in the field of medicine. Defective genes place humans at increased risk for developing many diseases. Treatment of inherited disorders by gene therapy has great potential but has not yet proven effective.

Genetics has had a huge impact on agriculture also. Today, both selective breeding and new molecular genetic approaches are being applied in the challenge to feed the world’s rapidly expanding human population.

The Human Genome Project is producing a wealth of new information about the Genetics of humans. If used wisely, the new information promises to enhance our quality of life.

3.2 Definitions

Genetics (from Ancient Greek genetikos, "genitive" and that from genesis, "origin"), a discipline of biology, is the science of genes, heredity, and variation in living organisms. (Griffiths et al., 2000)

Standard Dictionary of English Language defines Genetics as that portion of evolutionary science dealing with natural development uncomplicated by human interference; the experimental study of evolution, heredity and speciation of organisms. (“Genetics,” 1893,p. 1019)
Webster’s New International Dictionary defines Genetics as that branch of biology which individuals related by descent. The science of plant- and animal-breeding. The
inherited characteristics of an organism or group of organisms. (“Genetics,” 1996, p. 946)

**Dictionary of Genetics** defines Genetics as the study of inheritance patterns of specific traits. (Hotter, 2007, p.181)

**Encyclopedia Britannica** defines Genetics as the study of the way in which genes— the functional units of heritable material—operate and are transmitted from parents to offspring. (“Genetics,” 2010, p. 699)

**McGraw-Hill Encyclopedia of Science and Technology** defines Genetics as the science of biological inheritance, that is, the causes of the resemblances and differences among related individuals. (“Genetics,” 1997, p.773)

**Compton's Encyclopedia** defines Genetics as the branch of biology that deals with genes. (“Genetics,” 1986, p. 43a)

### 3.3 Origin and Early Theories

Through the ages men have speculated about heredity. In ancient Greece, for example, it was thought that the blood was in some way responsible for the transmission of hereditary traits, and the word “blood” is still often used to mean ancestry. Even before the beginning of written history men were aware of some of the ways in which heredity takes place. Domesticated animals and plants are proof of this. Domesticated horses, cattle, dogs, cultivated corn, wheat, cotton etc., differ greatly from their primitive, “wild” ancestors. They are the products of the art of ancient breeders for proper selection, well-controlled mating and careful choice of the selection of the best and promising offspring. The selected offspring should have greater qualitative and quantitative characters compared to the parents and meet the requirement of a breeder.

Over the centuries more and more knowledge has been acquired about the control of heredity for practical purposes. However, scientists remained baffled about the actual process of trait transmission till 1990 before Mendel proposed his famous laws on inheritance. (“Genetics,” 1986, p.43a)
The modern science of Genetics, which seeks to understand the process of inheritance, only began with the work of Gregor J. Mendel, a German-Czech Augustinian monk and scientist in the mid-19th century. His breeding experiments with garden peas, *Pisum sativum* led him to formulate the basic laws of heredity. Although he did not know the physical basis for heredity, Mendel observed that organisms inherit traits via discrete units of inheritance, which are now called “genes” but referred as “factors” by Mendel.

In his paper "Experiments on Plant Hybridization", presented in 1865 to the Society for Research in Nature in Brünn, Mendel traced the inheritance patterns of seven selected traits in pea plants and described them mathematically. The importance of Mendel's work did not gain wide understanding until the 1890s, after his death, when other scientists working on similar problems rediscovered his research. Walter Sutton of U.S. has proposed that chromosomes-major component of the cell nucleus-were the site of Mendel’s hereditary factors. The infant science of Genetics then flowered rapidly. By 1902 William Bateson, a proponent of Mendel's work, coined the word Genetics in 1905.

Bateson popularized the usage of the word Genetics to describe the study of inheritance in his inaugural address to the Third International Conference on Plant Hybridization in London, England, in 1906. The Hardy-Weinberg law, which established the mathematical basis for studying heredity in populations, was independently by English mathematician Godfrey H. Hardy and the German physician Wilhelm Weinberg in 1908 (Tamarin, 2001).

After the rediscovery of Mendel's work, scientists tried to determine which molecules in the cell were responsible for inheritance. In 1911, Thomas Hunt Morgan American geneticist argued that genes are on chromosomes, based on observations of a sex-linked white eye mutation in fruit flies (*Drosophila melanogaster*). In 1913, his student Alfred Sturtevant used the phenomenon of “genetic linkage” to show that genes are arranged linearly on the chromosome and move to the gamets together
during gametogenesis, which has disproved the Mendel’s law of independent assortment.

Although genes were known to exist on chromosomes, chromosomes are composed of both proteins, DNA and RNA—scientists did not know which of these is responsible for inheritance. In 1928, Frederick Griffith discovered the phenomenon of transformation in bacteria: dead bacteria could transfer genetic material to "transform" other still-living bacteria. During the 1940s George W. Beadle and Edward L. Tatum of United States demonstrated that genes exert their influence by directing the production of enzymes, proteins that facilitate chemical reactions in the cell. Sixteen years later, in 1944, Oswald Theodore Avery, Colin McLeod and Maclyn McCarty identified the molecule responsible for transformation as DNA. The Hershey-Chase experiment in 1952 also showed that DNA (rather than protein) is the genetic material of the viruses that infect bacteria, providing further evidence that DNA is the molecule responsible for inheritance (Tamarin, 2001).

The application of Physics and Chemistry to biological problems led to the development of molecular biology. Molecular biology is particularly concerned with the flow and consequences of biological information at the level of genes and proteins. The discovery of the DNA double helix made clear that the genes are functionally defined parts of DNA molecules and that there must be a way for cells to make use of their DNA genes in order to make proteins.

James D. Watson and Francis Crick along with x-ray crystallographer Maurice Wilkins determined the molecular structure of DNA in 1953 using the x-ray crystallographic analysis of the individual components of DNA. They were subsequently awarded with the Nobel Prize in medicine. This structure showed that genetic information exists in the sequence of nucleotides on each strand of DNA. In the following years, scientists tried to understand how DNA controls the process of protein production. With this molecular understanding of unit of inheritance, an explosion in the field of Molecular biology became possible and laid a foundation stone for the study of Molecular genetics.
When Watson and Crick produced their double helix model of DNA, it was known that most of the specialized features of the many different life forms on Earth are made possible by proteins. Structurally, proteins are long chains of amino acid subunits. In some way, the genetic molecule, DNA, had to contain instructions for how to make the thousands of proteins found in cells. From the DNA double helix model, it was clear that there must be some correspondence between the linear sequences of nucleotides in DNA molecules to the linear sequences of amino acids in proteins. The details of how sequences of DNA instruct cells to make specific proteins were worked out by molecular biologists during the period from 1953 to 1965. Francis Crick played an integral role in both the theory and analysis of the experiments that led to an improved understanding of the genetic code.

An important discovery was the chain-termination DNA sequencing in 1977 by Frederick Sanger. This technology allowed scientists to read the nucleotide sequence of a DNA molecule. In 1983, Kary Banks Mullis developed the polymerase chain reaction, providing a quick way to isolate and amplify a specific section of a DNA from a mixture. Single copy of a gene can be reproducible into million copies in in-vitro conditions within three to four hours and can be subsequently used to produce genetically modified organisms through gene transfer technology (recombinant DNA technology).

DNA fingerprinting, also called DNA profiling or DNA typing, is a method of isolating and making images of sequences of DNA (deoxyribonucleic acid). The technique was developed in 1984 by the British geneticist Alec Jeffrey, after he noticed the existence of certain sequences of DNA (called minisatellites) that do not contribute to the function of a gene (non-coding) but are repeated within the gene and in other genes of a DNA sample. Jeffrey also determined that each organism has a unique pattern of these minisatellites (VNTR-Variable Number Tandem Repeats), the only exception being multiple individuals from a single zygote (e.g., identical twins). An early use of DNA fingerprinting was in legal disputes, notably to help solve crimes and determine paternity. Today the technique has been refined through the use of
more-specific and more-sensitive probes and better blotting membranes. It has been recognized that DNA fingerprinting, similar to other DNA-analysis techniques, is limited by the quality of the sample obtained. Data generated through this technique may be used in *Personal Identification, Paternity and Maternity analysis*, Diagnosis and Cures for Inherited Diseases, Criminal identification and forensics, registration of Intellectual Property Rights for any biological materials etc. Through the pooled efforts of the **Human Genome Project** and the parallel private effort by ** Celera Genomics**, these and other methods culminated in the sequencing of the **human genome** in 2003(Snustad & Simmons, 2012). In spite of the Human Genome Project, there are so many challenges yet to be solved.

**Future Challenges in Genetics**

- What lies in the future?
- What will the twenty-first century, the century of genomics, bring?
- Will geneticists a hundred years from now speak of a complete cure for cancer, heart disease, and mental illness?
- Will we have cure for autoimmune diseases such as diabetes and arthritis?
- Will aging be slowed or even prevented?
- Will we have a complete understanding of the process of development and a concurrent elimination of birth defects and developmental problems?
- Will Genetics put an end to world hunger?
- How will we live, and what will be the quality of our lives?

These are the challenges posed to genetists who will learn and contribute the solutions to these questions as time progresses.

**3.4 Branches of Genetics**

**Behavioural genetics** is the field of study that examines the role of Genetics in animal (including human) behaviour. Often associated with the "nature versus nurture" debate, Behavioural genetics is highly interdisciplinary, involving contributions from **Biology, Genetics, Ethology, Psychology** and **Statistics**.
Behavioural geneticists study the inheritance of behavioural traits. In humans, this information is often gathered through the use of the twin study or adoption study. In animal studies, breeding, transgenesis, and gene knockout techniques are common; Psychiatric genetics is a closely related field.

Classical genetics consists of the technique and methodologies of Genetics that predate the advent of molecular biology. A key discovery of Classical genetics in eukaryotes was genetic linkage. The observation that some genes do not segregate independently at meiosis broke the laws of Mendelian inheritance, and provided science with a way to map characteristics to a location on the chromosomes. Linkage maps are still used today, especially in breeding for plant improvement.

Developmental biology is the study of the process by which organisms grow and develop. Modern developmental biology studies the genetic control of cell growth, differentiation and which is the process that gives rise to tissues, organs and anatomy.

Conservation genetics is an interdisciplinary science that aims to apply genetic methods to the conservation and restoration of biodiversity. Researchers involved in Conservation genetics come from a variety of fields including Population genetics, molecular ecology, biology, evolutionary biology, and systematics. Genetic diversity is one of the three fundamental levels of biodiversity, so it is directly important in conservation of biodiversity, though genetic factors are also important in the conservation of species and ecosystem diversity. Conservation of genetic variability is important to the overall health of populations because decreased genetic variability leads to increased levels of inbreeding, and reduced fitness.

Ecological genetics is the study of Genetics in natural populations. Research in this field is on traits of ecological significance—that is, traits related to fitness, which affect an organism's survival and reproduction. Examples might be: flowering time, drought tolerance, polymorphism, mimicry, avoidance of attacks by predators.
Evolutionary genetics, the modern evolutionary synthesis is a union of ideas from several biological specialties which provides a widely accepted account of evolution. It is also referred to as the new synthesis, the modern synthesis, the evolutionary synthesis, millennium synthesis and the Neo-Darwinian synthesis.

Genetic engineering, also called genetic modification, is the direct human manipulation of an organism's genome using modern DNA technology. It involves the introduction of foreign DNA or synthetic genes into the organism of interest. The introduction of new DNA does not require the use of classical genetic methods. However traditional breeding methods are typically used for the propagation of recombinant organisms.

Genetics of Intelligence is the study of the heritability of IQ investigates the relative importance of Genetics and environment for phenotypic variation in intelligence quotient (IQ) in a population. "Heritability", in this sense, "refers to the genetic contribution to variance within a population and in a specific environment". There has been significant controversy in the academic community about the heritability of IQ ever since research began in the 19th century.

Genomics is a discipline in Genetics concerned with the study of the genomes of organisms. The field includes efforts to determine the entire DNA sequence of organisms and fine-scale genetic mapping. The field also includes studies on intra-genomic phenomena such as heterosis, epistasis, pleiotropy and other interactions between loci and alleles within the genome.

Human genetics is the study of inheritance as it occurs in human beings. Human genetics encompasses a variety of overlapping fields including: Classical genetics, Cytogenetics, Molecular genetics, Biochemical genetics, Genomics, Population genetics, Developmental genetics, Clinical genetics, and Genetic counselling.

Medical genetics is the speciality of medicine that involves the diagnosis and management of hereditary disorders. Medical genetics differs from Human genetics, in that Human genetics is a field of scientific research that may or may not
apply to medicine, but Medical genetics refers to the application of Genetics to medical care.

**Microbial genetics** is a subject area within [microbiology](https://en.wikipedia.org/wiki/Microbiology) and [genetic engineering](https://en.wikipedia.org/wiki/Genetic_engineering). It studies the Genetics of very small (micro) organisms. This involves the study of the genotype of microbial species and also the expression system in the form of phenotypes. It also involves the study of genetic processes taking place in these microorganisms i.e., recombination, transformation, conjugation etc.


**Psychiatric genetics**, a branch of [behavioural neurogenetics](https://en.wikipedia.org/wiki/Behavioral_neurogenetics), studies the role of Genetics in psychological conditions such as [alcoholism](https://en.wikipedia.org/wiki/Alcoholism), [schizophrenia](https://en.wikipedia.org/wiki/Schizophrenia), [bipolar disorder](https://en.wikipedia.org/wiki/Bipolar_disorder), and [autism](https://en.wikipedia.org/wiki/Autism). The basic principle behind Psychiatric genetics is that genetic [polymorphisms](https://en.wikipedia.org/wiki/Polymorphism), as indicated by linkage to e.g. a [single nucleotide polymorphism](https://en.wikipedia.org/wiki/SNP) (SNP), are part of the etiology of psychiatric disorders.

**Quantitative genetics** is the study of continuously measured traits (such as height, weight, colour etc.) and their mechanism of inheritance. It can be an extension of simple [Mendelian inheritance](https://en.wikipedia.org/wiki/Mendelian_inheritance) in that the combined effects of one or more genes and the environments in which they are expressed give rise to continuous distributions of phenotypic values. Inheritance pattern of Quantitative traits do not follow the Mendelian pattern.

Out of these branches of Genetics, 10 branches have been selected for the present study based on their priority.
References