CHAPTER

1

Introduction

The science of genetics is the study of heredity which is the cause of similarities; and variation which is the cause of differences between individuals.

The science of heredity or genetics is the study of two contradictory aspects of nature : heredity and variation. The process of transmission of characters from one generation to next, either by gametes–sperms and ova–in sexual reproduction or by the asexual reproductive bodies in asexual reproduction, is called **inheritance** or **heredity**. Heredity is the cause of similarities between individuals. This is the reason that brothers and sisters with the same parents resemble each other and with their parents. **Variation** is the cause of differences between individuals. This is the reason that brothers and sisters who do resemble each other are still unique individuals. Thus, we have no trouble in recognizing the differences between sisters, for example, and even 'identical' twins are recognized as distinctive individuals by their parents and close friends. The science of genetics attempts to explain the mechanism and the basis for both similarities and differences between related individuals. It also tries to explain the phenomenon of evolution and cytodifferentiation.

The heredity and variations play an important role in the formation of new species (speciation). The biological science which deals with the mechanism of heredity and causes of variations in living beings (viruses, bacteria, plants and animals) is known as **genetics**. The word **genetics** was derived from the Greek root *gen* which means to become or to grow into and it was coined by **Bateson** in 1906 for the study of physiology of heredity and variations.

HISTORICAL

The history of most scientific disciplines including genetics are generally characterized by relatively long periods of stagnation punctuated by bursts of rapid progess. Most of these flurries of research are initiated by new technical developments. The science of genetics is a very young science in comparison to other biological sciences and its origin can be traced in the works of **Mendel** in the nineteenth century. But before Mendel's work men throughout the ages had some vague knowledge about genetics and more often have tried to explain the causes of heredity. About six thousand years ago men kept records of pedigrees of domestic animals such as horse and food plants as rice. The ideas or theories which have been forwarded from time to time to explain the phenomenon of inheritance can be categorized under the following headings : 1. Vapour and fluid theories; 2. Preformation theories; 3. Particulate theories.

1. Vapour and Fluid Theories

Early Greek philosophers such as **Pythagoras** (500 B.C.) proposed that every organ of animal body gives out some type of vapours. These vapours unite and form a new individual.

Hippocrates (400 B.C.) believed that the reproductive material is handed over from all parts of the body of an individual, so that the characters are directly handed over to the progeny.

Further, **Aristotle** (350.B.C.) thought that the semen of man has some "vitalizing" effect and he considered it as the highly purified blood. According to him the mother furnishes inert matter and the father gives the motion to the new life.

2. Preformation Theories

Leonardo da Vinci (1452–1519) proposed a theory that the male and the female parents contribute equally to the heredity of the offspring. **W. Harvey** (1578–1657) speculated that all animals arise from eggs and that semen only plays vitalizing role. **R. de Graaf** (1641–1673) observed that the progeny would have characteristics of father as well as of mother and, therefore, he proposed that both the parents should contribute to the heredity of progeny.

Malpighi (1673), the pioneer of preformationist school, concluded that development of any organism consisted simply of growth of preformed part. **A.V. Leeuwenhoek** in 1677 observed sperms of several animals (man, dog, rabbit and other mammals, frog, fish and insects) and also suggested their association with eggs. In 1679, **J. Swammerdam** studied development of insects and frog and suggested that development of an organism is a simple enlargement of a minute but preformed individual. The figure of **homunculus** (Fig. 1.1) or manikin, the miniature man in the sperm head, was published in 1695 by **Hartsoeker**. Such type of theories which advanced the concept of the presence of preformed embryo in the sex cells are known as **preformation theories**. Preformationists have, often, been divided into two schools: 1. **Ovists** who attached more importance to ova; they thought that "homunculus" was present in the ovum. 2. **Animalculists** or **spermatists** who attached more importance to sperm; they thought that a miniature but complete organism was present in the sperm.

N. Grew in 1682 reported for the first time the reproductive parts of plants. **R. Camerarius** in 1694 described sexual reproduction in plants for the first time. He is also known to be first to produce a hybrid between two different plant species. In 1717, **Fairchild**

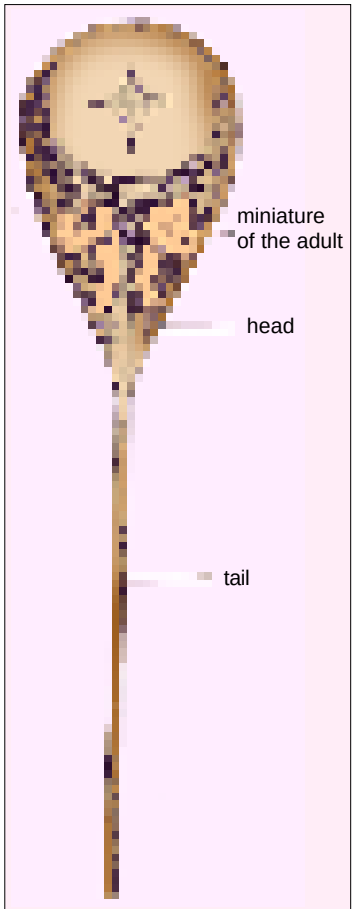


Fig. 1.1. Diagram of homunculus.

produced a hybrid having characteristics of both parents. This hybrid was called "**Fairchild's Sweet William**" or as "**Fairchild's mule**." This provides a means of artificial hybridization in plants. **J.G. Kolreuter** (1733–1806) obtained fertile hybrids from artificial crosses between two species of tobacco plants.

K.F. Wolff (1738–1794) finally refuted the preformation theory by proposing that neither egg nor sperm had a structure like homunculus but that the gametes contained undifferentiated living substance capable of forming the organized body after fertilization. Such an idea formed the very core of the theory of **epigenesis**. This theory suggested that many new organs and tissues which were originally absent, develop subsequently. However, Wolff believed that these tissues and organs developed *de novo* due to mysterious vital forces.

3. Particulate Theories

French biologist **Maupertuis** (1689–1759) has proposed that the body of each parent gives rise minute particles. In sexual reproduction, the particles of both individuals unite together to form a new individual. He thought that in certain cases the particles of the male parent might dominate on those of the female parent and produce the male individual. In the production of female individual the particles of female might dominate on particles of male. Thus, **maupertuis** proposed the concept of **biparental inheritance** by **elementary particles**. He studied the family pedigree of polydactyly and albinism in human beings.

The great biologist **Lamarck** (1744–1829) in 1809 proposed the phenomenon of "inheritance of acquired characters" among living organisms. But he failed to provide convincing evidences in support of his concepts.

In 1868 the well known naturalist **Charles Darwin** has given his famous **theory of pangenesis** which exclusively depends on the particulate theory. The central idea of pangenesis theory has been given first of all by **Hippocrates**. According to the pangenesis theory of Darwin each part of the animal body produces many minute particles known as **gemmules**. These gemmules are at first collected in the blood and later on are concentrated in the reproductive **organs**. When the animal reproduces into new individual, these gemmules pass on to it and it has blending of both parents. By this mechanism acquired characters would also be inherited because as the parts of the body changed so did the pangenes or gemmules they produced.

The theory of pangenesis was disapproved by **Galton** (1823–1911) and **Weismann** (1835–1934). **Weismann** in 1892 postulated the **theory of germplasm** to explain heredity. According to this theory the body of organisms contain two types of cells namely somatic cells and reproductive cells. The somatic cells form the body and its various organ systems, while the reproductive cells form sperm and ova. The somatic cells contain the **somatoplasm** and germinal or reproductive cells contain the **germplasm**. According to Weismann the germplasm can form somatoplasm but somatoplasm cannot form germplasm. Thus, the changes in the structure of somatic cells or somatoplasm which are caused by the environment (acquired characters) cannot influence the reproductive cells or germplasm. By cutting the tails of mice for many generations, Weismann always got tailed mice. So, by such experimental evidences he rejected the Lamarckism and pangenesis theory.


Gregor Mendel (1822-1884).

Though the particulate theory faced many problems in its beginning but its basic concept has formed the central core of the modern understanding of the genetics.

Augustinian Monk **Gregor Mendel** was the first investigator who laid the foundation of our modern concept of the particulate theory. He could understand the heredity problems more clearly than any one in the past, because his approach was simple, logical and scientific. By his famous experiments on pea plant he concluded that the inheritance is governed by certain factors which occur in the cells of each parent. He

thought that each parent has two such factors, while their sex cells (sperm or pollen and ovum or egg) have only one factor. However, he failed to explain the exact process by which these factors pass on the sex cells. **knight** (1799) and **Goss** (1824) conducted hybridization experiments on edible pea (*Pisum sativum*), but they failed to formulate any law of inheritance like the Mendel.

During 19th century and dawn of 20th century, the science of genetics have received solid support from landmark investigations in the field of cytology, embryology, biochemistry and genetics. **Von Baer** (1828) made discovery of the mammalian egg. **Pringsheim** (1855) first saw nuclear fusion in green algae (*Vaucheria*). Heredity transmission through the sperm and egg became known by 1860. **Ernst Haeckel**, noting that sperm consisted largely of nuclear material, postulated that the nucleus is responsible for heredity. **Oscar Hertwig** (1875) observed the entrance of the sperm into the sea urchin. He found nucleus to play an important role in hereditary mechanism. In 1884, **Hertwig** identified the hereditary substance with the chromatin of nucleus. **Strasburger** in 1875 discovered the chromosomes and he along with **Kolliker** and **Weismann** formulated the **nuclear theory of heredity**. **Flemming** (1882) investigated the process of mitosis.

Three plant breeders, namely **Hugo de vries** (Holland), **Karl Correns** (Germany) and **Erich Tschermak** (Austria), rediscovered the Mendel's laws in 1900. Each of them reached similar conclusions before they knew of Mendel's work. **Bateson** (1902) published a book "*The Principles of Heredity*." From 1902 to 1909 he introduced the terms **allelomorphs**, **homozygote**, **heterozygote**, **F1**, **F2** and **epistatic gene**. **Bateson** was the first to have Mendel's paper translated into English and the first to show that Mendel's theory also applied to animals. He coined the term **genetics** in 1905. In 1906, **Bateson** and **Punnett** reported first case of linkage in sweet pea, however, they failed to explain the phenomenon of linkage correctly. **R.C. Punnett** devised the **Punnett's square** for making gametic combinations theoretically. American cytologist, **Walter S. Sutton** in 1902 proposed the **chromosome theory of heredity** in his classic paper "*The Chromosomes in Heredity*," in which he postulated that the newly rediscovered Mendel's hereditary factors were physically located on chromosomes. This theory provided a mechanism of transmission to explain the behaviour of Mendel's factors and brought together two independent desciplines– the genetics and the cytology. Thus, the year 1903 is the year of birth of **cytogenetics**.

Archibald E. Garrod (1902) deciphered the inheritance pattern and metabolic nature of the human disease **alkaptonuria** (in which urine of patient turns dark to black upon exposure to air). In 1908, **Garrod** presented in a lecture nearly all the facts that we know today concerning this disease. He also postulated various enzymes involved in this metabolic error, but he could not identify them. In 1909 **Johannsen** formulated the **genotype-phenotype concept** to distinguish hereditary variations from environmental variations. According to him, the genotype of an individual represents the sum total of heredity, while phenotype of an individual represents the observable structural and functional properties which are produced by the interaction between genotype and environment. (In 1877, **Johannsen** coined the term **gene**). The hypothesis that "genes can change (mutate) to give rise to new genes (mutant genes)" was seriously tested, beginning in 1908 by American biologist **Thomas H. Morgan** and his young collaborators (Ph.D. students), such as, **Calvin B. Bridges**, **Hermann J. Muller** and **Alfred H. Sturtevant**. They worked on the fruit fly, *Drosophila melanogaster*. **(W.E. Castle** suggested the fruit fly to Morgan). In 1910, first white eye mutant was detected in *Drosophila* by this team of workers and it is first reported case of sex linkage.

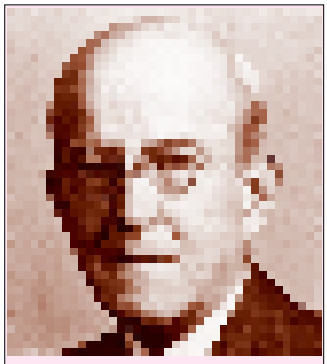
Sir A.E. Garrod, father of biochemical genetics.

T.H. Morgan (1866–1945) proposed in 1911 the **theory of linkage**. He turned the chromosome theory of inheritance into the concept of genes being located in a linear array on each chromosome. In


Thomas Hunt Morgan 1866-1945.

1926, his book '*The Theory of the Gene*' was published and he got Nobel prize in 1934. Cytological basis of **crossing over** was first described by the Belgian cytologist **F.A. Janssens** in 1911. **H.J. Muller** and **L.J. Stadler** independently discovered that X-rays induce mutations. **H.J. Muller** got Nobel Prize in 1946 for the discovery of the induction of mutation in *Drosophila* by X-rays. In 1916, **Bridges** made discovery of the phenomenon of **non-disjunction** in *Drosophila*. In 1921, he proposed the **genic balance** mechanism of sex determination in *Drosophila*.

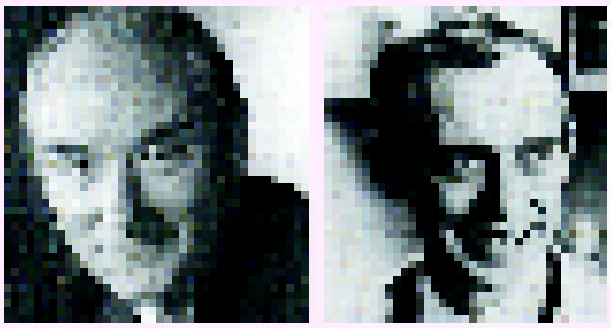
B.O. Dodge in the late 1920's and early 1930's first determined the genetics of *Neurospora*. **T.M. Jenkins** (1924) reported a case of **cytoplasmic inheritance**, called **Iojap striping** in maize. **Barbara McClintock** and **Harriet Creighton** working at Cornell University, USA, with the corn plant, *Zea mays*, devised an elegant demonstration of chromosome breakage and rejoining during crossing over. In 1937, **Richard Goldschmidt** stimulated exploratory questions on the chemical nature of gene (of *Drosophila*). In the 1940's, two significant discoveries were made concerning the chemical nature of the gene: 1. **Oswald Avery**, **C.M. MacLeod** and **M. McCarthy** (1944) were able to establish by experiments with pneumonia-causing (virulent) bacteria that *genes were composed of a specific type of nucleic acid, called* **deoxyribonucleic acid (DNA)**, *and not proteins.* 2. While studying the biochemical basis for the eye colour in *Drosophila*, **George Beadle** and **E.L. Tatum** were able to show that the lack of brown colour in various mutants was due to a defect in one step in the biosynthesis of the brown pigment. They proposed the **one-gene one-enzyme hypothesis** which suggested that the action of each gene is through the synthesis of a protein (enzyme) which in turn catalyzes a single chemical reaction. They proved this hypothesis through the use of multitude of mutants in the fungus, *Neurospora* (in 1941). In most cases, each mutation was due to a change in a single gene. Thus, they initiated the branch of **biochemical genetics**. The term **molecular biology** was first used in 1945 by **William Astbury**, who was referring to the study of the chemical and physical structure of biological macromolecules.. **Joshua Laderberg**


Oswald Avery 1877-1955.


Max Delbruck 1906-1981.

(1946) first demonstrated the phenomenon of recombination in the bacteria *E.coli*. **Beadle**, **Tatum** and **Laderberg** got Noble prize in 1958. Recombination in phage was first demonstrated in 1948 by **Max Delbruck** and **Mary Delbruck**. The chemistry of DNA and RNA has been worked out by **A. Kornberg** and **S. Ochoa** ; both got Noble Prize in 1959.

Prior to discovery of the chemical structure of the genetic material, the 'gene' was an abstract, indivisible unit of heredity (comparable to old concept of the indivisible atom). This period in history is referred to as **classical** or **formal genetics**. The word "formal" pertains to the extrinsic aspect of something as distinguished from its substance or material. The era of **molecular genetics** followed the discovery of DNA structure (*i.e.*,1953) when the fundamental unit of heredity was determined to be DNA nucleotide and the 'gene' was found to consist of an aggregate of nucleotides. In 1953, one of the most significant twentieth-century discoveries in biology was made by **James watson** and **Francis Crick**. Their paper published in the British Journal *Nature* in which they proposed the molecular structure of DNA, *i.e.*, the molecular composition of the gene. **Watson**, **Crick** and **Wilkins** got Nobel Prize in 1962 for the discovery of double helix model of DNA which opened the new vistas in the genetical world.



Francis Crick and James Watson.

Seymour Benzer performed extensive investigations on the genetics of T_4 bacteriophage of *E. coli* and in 1955, was able to define the gene in terms of function (**cistron**), recombination (**recon**) and mutation (**muton**) and to place an accurate molecular size estimate on the conceptual gene components. In simple terms, Benzer demonstrated that the linear array of genes on chromosomes as shown by Morgan, was extended down to the molecule of DNA making up the chromosome. In 1961, **Francois Jacob** and **Jacques Monod** provided genetic evidence for a method of gene regulation in bacteria, now called the **operon**. In 1965, **Jacob**, **Monod** and **Lwoff** were awarded Nobel Prize for their contribution to microbial genetics. **Gaulian** and **Kornberg** isolated, purified and utilized **DNA polymerase** of *E. coli*. **R.W. Holley** got Nobel Prize (1968) for the discovery of base sequence of tRNA. **Holley** died in 1993. During 1961–1968, the genetic code of DNA was solved by **M.W. Nirenberg**, **J.H. Matthaei**, **p.Leder**, and **H.G. Khorana**. They synthesized small RNA molecules (mRNAs) of known composition and observed which amino acid was incorporated into protein in a cell-free protein synthesizing system. In 1968 **Nirenberg** and **Khorana** discovered the complicated DNA code known as **genetic code**. Both scientists along with **Holly** received the Nobel Prize in 1968.

N.L. Dhawan and **R.L. Paliwal** (1964) studied the cytoplasmic inheritance in maize. The term **transposons** (*i.e.*, jumping genes) is used in 1974 by **R.W. Hedges** and **A.E. Jacob** of Hammersmith Hospital in London, for a DNA segment or genetic element which could move from one molecule to another and carried resistance for antibiotic ampicillin in the bacterial cells. These transposons, however, were originally discovered in maize plant by **Barbara McClintock** by the name **controlling elements** in 1956. During the late 1970's, the science of genetics entered a new era dominated by the use of **recombinant DNA technology** or **genetic engineering** to produce novel life forms not found in nature. Through this technology, it has been possible to transfer genes from mammals into bacteria, causing the microbes to become tiny factories for making (in relatively large quantities) proteins of great economic significance such as **hormones** (insulin, growth hormones) and **interferon** (lymphocyte proteins that prevent replication of a wide variety of viruses). These proteins are produced in such small quantities in humans that the cost of their extraction and purification from tissues has been very expensive, thus, restricting their medical use in **prophylaxis** (prevention) and **therapeutics** (treatment) of disease. By genetic engineering, it has become possible to produce various **blood clotting factors**, **complement proteins** (part of the immune system) and other substance for the improvement of genetic deficiency diseases (**euphenics**) other current fields of genetic research are oncogenes (cancer), antibody diversity (immunogenetics), homeotic mutation and behaviour.

Joshua Lederberg

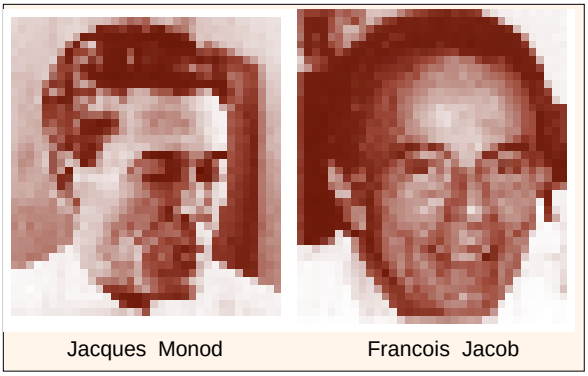
SCOPE OF GENETICS

Geneticists study all aspects of genes. The study of the mode of gene transmission from generation to generation is broadly called **transmission genetics**; the study of structure and function of the gene forms the **molecular biology**, and the study of behaviour of genes in populations is called **population genetics**. These three major subdivisions of genetics are arbitrary and there is considerable overlapping. It is the knowledge of how genes act and how they are transmitted down through the generations that has unified biology; previously, specific set of biological phenomena had each been

assigned to separate disciplines. An understanding of how genes act is now essential prerequisite for such biological fields of study as development, cytology, physiology and morphology. An understanding of gene transmission is a fundamental aspect of areas such as ecology, evolution and taxonomy. Further unification has resulted from the discovery that the basic chemistry of gene structure and function is very similar across the entire spectrum of life on the earth. Thus, not so long ago, biology was divided into many camps that rarely communicated with each other; today, however, every biologist must be a bit of a geneticist, because the findings and techniques of genetics are being applied and used in all fields. In fact, genetics contributed the modern prototype for all of the biology. It provides a unifying thread for the previously diverse fields of biology.

IMPORTANCE OF GENETICS

The cultural evolution of human beings is strongly influenced by knowledge of hereditary phenomena of early man. Civilizations itself become possible when nomadic tribes learned to domesticate plants and animals. Long before biology existed as a scientific discipline, people selected grains with higher yields and greater vigour and animals with better fur, meat or milk. They also were mystified about the inheritance of desirable and undesirable traits in the human population. Despite this long-standing concern with heredity and the practice of selective breeding, it was not until the discovery of Mendel's laws that we were able to explain the actual basis for inheritance.



Jacques Monod Francois Jacob

Like other disciplines of science, the genetical insight has produced new challenges as well as solutions to some human problems. For example, early in this century a new wheat strain called **Marquis** was developed in Canada. This high-quality strain is resistant to disease; furthermore, it matures two weeks earlier than other commercially used strains—a very important factor where the growing season is short. The introduction of Marquis strain of wheat had opened up millions of square kilometres of fertile soil to cultivation in such northern countries as Canada, Sweden and the USSR. Likewise, **IR26** strain of rice was developed by geneticists in 1973; it has a wide range of desirable characteristics such as resistance to several viral and fungal diseases and protection from insects such as green leaf hopper, brown hopper and stem borer. In addition to improving crop varieties, geneticists have learned to change the genetic systems of insects to reduce their fertility. This technique has provided an important new tool in the age-old struggle to keep insects away of human crops and habitations.

In recent years, such successes led to the concept of "**Green Revolution**". Using sophisticated breeding techniques based on new knowledge about genes, geneticists created high-yielding varieties of dwarf wheat and rice. Large-scale planting of these crops around the world did provide new food supplies, but new problems quickly became apparent. These specialized crops require extensive cultivation and costly fertilizers. The use of the new high-yield varieties produced diversed social and economical problems for the poor/developing countries where they are most needed. Furthermore, the spread of **monoculture** (the extensive dependence on a single plant variety) left vast areas at the mercy of some newly introduced or newly evolved form of pathogen (*e.g.*, plant disease) or insect pest. With the huge population of humans on earth, our dependence on high yield varieties of crop plants and domestic animals has become increasingly clear. In fact, the stability of human society depends on the ability of geneticists to juggle the inherited traits that shape life forms, keeping a jump ahead of the destructive parasites and predators (Fig. 1.2).

In recent years, advances in **biotechnology** have led to the creation of special genetically engineered strains of bacteria and fungi that carry specific genes from unrelated organisms such as

humans. (**Note**: Biotechnology means use of living organisms or processes to modify or make products and to improve plants or animals, see **Peter Funk**, 1995). As already has been mentioned under the head of historical, these microbes produce such useful compounds as insulin, human growth hormone and the antiviral (or anticancer) agent–the interferon.

Further, the most exciting and alarming application of genetic knowledge is to the human species itself. Genetic discoveries have had major effects on medicine. One can now diagnose hereditary or genetic disease before or soon after birth, and in some cases we can provide secondary treatments. Using **family pedigree analysis**, a genetic counsellor can give prospective parents the information they need to make intelligent decisions about the risks of genetic disease in their offspring. Some refined techniques as **amniocentesis** and **fetoscopy** provide information about possible genetic disease at early stages of pregnancy. A battery of post natal chemical tests can detect problems in the newborn infants, so that some corrective methods can be applied immediately to lessen the impact of many genetic diseases.

However, our new ability to recognize genetic disease poses an important moral dilemma. An estimated 5 per cent of our population survives with severe physical or mental genetic defects. This percentage probably will increase with extended exposure to various environmental factors and paradoxically, with improved medical technology. For example, of those patients admitted to paediatric hospitals in North America, 30 per cent estimated to have genetic diseases. This will certainly increase great financial burden on human society.

Lastly, knowledge of genetic mechanisms has made us aware of some new dangers as well. Primarily, some geneticists fear that there may be an accidental release from some laboratory of an artificial pathogen that has never existed on this planet before and that pathogen may cause havoc. Some geneticists even fear that increased exposure to chemical food additives and to vast array of chemicals in other commercial products may be changing the human genetic makeup in a very undesirable and haphazard manner. This type of random genetic changes can also be caused by such environmental agents as fallout from nuclear weapons, radioactive contamination from nuclear reactors, and radiation from various X-ray machines. These agents may be contributing to inherited disease, but they almost certainly are contributing to the incidence of cancer (which is a genetic disease of the somatic or body cells). Anyhow in modern era the genetics has revolutionised the agriculture, horticulture, animal husbandry and many other branches of science. The science of genetics has proved worthy in removing many faulty concepts of man about the inheritance.

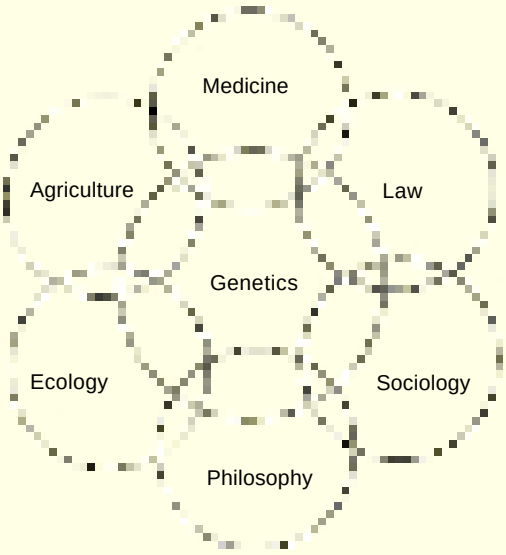


Fig. 1.2. Impact of genetics on different interacting areas of human ventures (after Suzuki *et al.*, 1986).

BRANCHES OF GENETICS

In recent years, the science of genetics has proliferated into numerous distinctive subdisciplines. Some of the significant branches of genetics are the following :

1. **Plant genetics.** The genetics of plants.
2. **Animal genetics.** The genetics of animals.
3. **Human genetics.** It involves the study of heredity of human traits, human disorders, betterment and correction of human disorders.

4. Microbial genetics. It deals with the genetics of microorganisms (*viz.*, viruses, bacteria, unicellular plants and animals).

5. Fungal genetics or mycogenetics. The genetics of fungi.

6. Viral genetics. Genetics of virus.

7. Drosophila genetics. Genetics of fruit fly, *Drosophila* sp.

8. Mendelian genetics. It involves study of heredity of both qualitative (monogenic) and quantitative (polygenic) traits and the influence of environment on their expressions.

9. Quantitative genetics. It involves the study of heredity of quantitative traits such as height, weight and IQ in human beings and milk production in cattle.

10. Morganian genetics. It includes study of recombination (crossing over) in all kinds of organisms such as higher plants, animals, fungi, bacteria and viruses. It also involves the preparation of linkage maps of chromosomes.

11. Non-Mendelian genetics. It involves a study of the role of cytoplasm and its organelles (particularly chloroplasts and mitochondria) in heredity.

12. Mutations. They involve study of heredity of both chromosomal changes (structural and numerical) and also gene mutation.

13. Cytogenetics. It provides the cytological explanations of different genetical principles.

14. Molecular genetics. It includes the study of structure and function of gene and regulation of its activity.

15. Transmission genetics. It includes the study of mode of gene transmission from generation to generation. The kind of studies that Mendel performed are now included in the discipline of transmission genetics.

16. Clinical genetics. Genetics involved in the detection of causes of diseases such as haemophilia, colour blindness, diabetes, phenylketonuria.

17. Immunogenetics. It deals with genetics of production of different types of antibodies; the diversity of antibodies has been found to be under control of genetic regulation.

18. Behavioural genetics. It involves the interaction of genes with the environment to produce a particular pattern of behaviour. In *Drosophila* many behaviour genes have been identified, *e.g.*, mutants described as sluggish, non-climbing, flightless, easily shocked, etc., and genes regulating sexual behaviour. In primates including humans, it has been found that IQ (intelligence quotient) is governed by genetics (parentage), environment (adopted parents) and developmental stage (age) of an individual.

19. Forward genetics and reverse genetics. During the last decade, the term **reverse genetics** has been used for physical mapping and isolation of genes whose protein products are unknown. The term **forward genetics** has been used for genes which are mapped on the basis of phenotype (or gene product or protein), using the technique of classical genetics. However, recently in 1991, the term reverse genetics has been redefined by **Paul Berg** (Nobel Laureate). According to him, the term reverse genetics should be restricted to those studies, where we start the study with a DNA segment with unknown phenotypic effect, introduce this DNA (without any alteration or other modification) into a plant or an animal and then follow the phenotypic effect.

REVISION QUESTIONS

1. Explain the following :
   Germplasm theory; pangenesis theory; preformation theory; particulate theory; and variation.
2. Why the study of genetics is important for human society?
3. Give a brief account of the scope and importance of genetics, outlining the newer areas of study in this subject.
4. Describe the historic growth of genetics in last two decades.
5. Enumerate and define various branches of genetics.

CHAPTER

2

Genetical Terminology

Albinos illustrate pleiotropy.

Like other sciences, the science of genetics has its specific terminology which minimizes the chances of confusion, inconvenience and unnecessary repetition of full sentences. We are giving here certain most common terms which are used more frequently in genetics.

Acquired character. The alteration in the morphology or physiology of an organism in response to its ecological factors (environment) is known as acquired character. Acquired characters are usually not heritable.

Albinism. Absence of colour in skin, hair and eyes or absence of chloroplast in a plant; an inherited trait.

Albino. The animal without pigmentation in skin, hairs and eyes is called albino.

Allele (Allelomorph). One of two or more forms that can exist at a single gene locus, distinguished by their differing effects on the phenotype. Alleles are genes controlling the same characteristic (*e.g.* hair colour) but producing different effects (*e.g.* black or red), and occupying corresponding positions on homologous chromosomes.

Amniocentesis. Puncture of the uterine wall with a needle for the purpose of obtaining amniotic fluid, which can be analyzed to determine whether the foetus has a genetic abnormality. Amniotic fluid contains sloughed foetal cells.

Aneuploidy. Karyotypic abnormality in which a specific chromosome(s) is present in too many or too few copies.

Animal breeding. The practical application of genetic analysis for development of pure-breeding lines of domestic animals suited to human purposes.

Autosome. The chromosomes which are not associated with sex are known as autosomes. Except the sex chromosomes (X) and (Y) other chromosomes are the autosomes.

Amniocentesis.

Back cross. The cross of a progeny individual with its parents is known as back cross.

Barr body. A densely staining mass that represents an X-chromosome inactivated by dosage compensation.

Bead theory. The disproved hypothesis that genes are arranged on the chromosome like beads on a necklace, indivisible into smaller units of mutation and recombination.

Bivalent. A pair of synapsed homologous chromosomes is known as a bivalent.

Blending inheritance. A discredited model of inheritance suggesting that the characteristics of individual result from the smooth blending of fluid-like influences from its parents.

Carrier. A heterozygous individual. An individual who possesses a mutant allele but does not express it in the phenotype because of a dominant allelic partner; thus, an individual of genotype *Aa* is a carrier of *a* if there is complete dominance of *A* on *a*.

Chiasma (plural **chiasmata**). A cross-shaped structure commonly observed between nonsister chromatids during meiosis; the site of crossing-over.

Chromatin. A DNA, RNA, histone and non-histone protein containing thread-like coiled structure of interphase nucleus is called chromatin.

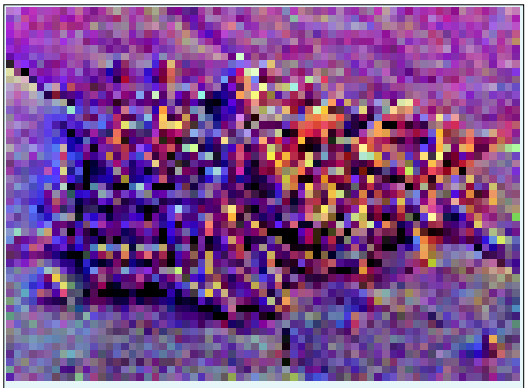
Chromosome. The nucleoprotein structure which are generally more or less rod-like during nuclear division. The genes are arranged on the chromosomes in a linear fashion. Each species has a characteristic number of chromosomes. Chromosomes play most important role in inheritance.

Cis arrangement. Linkage of dominants of two or more pairs of alleles on one chromosome and the recessive on the homologous chromosome.

Cistron. A term equated with the term gene. It is a region of DNA that encodes a single polypeptide (or functional RNA molecule such as tRNA or rRNA).

Clone. A group of genetically identical cells or individuals derived by asexual division from a common ancestor.

Cloning. Asexual production of a line of cells or organisms or segments of DNA genetically identical to the original.



Cells of yeast multiply rapidly by budding, producing vast clones of genetically identical cells.