Chapter 31

GENETICS -- Introduction, Brief History, Scope

Genetics *is the branch of biology that is concerned with the study of inherited variations*. It deals with origin of genetic variations and the expression of traits resulting from these variations. It is concerned with the transmission, expression and evolution of genes — *the molecules that control the function, development and ultimate appearance of individuals*. It is commonly referred to as **science of heredity**.

BRIEF HISTORY OF GENETICS

Our present understanding regarding heredity is the result of long history of thought and investigation. Two concepts provided the basis for thinking about heredity before 20th century.

1. Heredity within Species

For a long time people believed that animals are obtained by crossing widely different species, the phenomenon called **breeding**. For example, *Giraffe* was believed to be the result of a cross between a *camel* and a *leopard*. However, later on it was found that such crosses are not possible as the species had maintained themselves without a change since their creation.

2. Direct Transmission of Hereditary Material

Scientists observed variations among the members of a species and noticed that these are inherited to the offspring (children) from their parents. The logical question was that what is transmitted? Many theories were proposed to explain transmission of variations. These include:

i. Transmission of Gonos

Greeks believed that body parts of the parents were transmitted to the offspring directly. All the body parts contribute reproductive material known as **gonos** (seed). The information of each body part pass independent of each other and a new individual is formed after information from all parts of the body of the parents is transmitted.

ii. Darwin's Theory of Gemmules

Charles Darwin in 1868 proposed that all cells and tissues excrete microscopic granules called **gemmules**. These are transported by blood stream to the sex organs where these assemble into the gametes and are passed to the offspring upon fertilisation. They separate out to different parts of the body during development and guide the growth of these parts.

iii. Blending Theory

Another theory regarding transmission of hereditary characters assumed that male and female contributions are blended. According to this belief the hereditary material is in the form of liquid and the fluid of two parents blend. Therefore, offspring of these parents is a mixture of different parental characteristics. For example, it was observed that children of tall and short parents were intermediate to both parents in height at times. However, it was also frequently observed that children ... resemble either of the parents and there is no blending for many generations.

These two concepts led to a problem that if variations do not enter a species from outside and if variations within each species are blended in every generation, then all the members of a species should soon resemble one another exactly. But this does not happen because the individuals within most species differ widely from one another, and they differ in characteristics that are transmitted from generation to generation.

Hybridization Experiments

A solution to this problem was provided in the work of a German botanist **Josef Koelreuter**. He carried out first successful hybridization experiment of plant species in 1760. He crossed different species of *tobacco* and obtained fertile offspring. The hybrids differed in appearance from both of their parent strains. When hybrids were crossed, some of their offspring resembled hybrids, and a few resembled the original parent strains, i.e., gradparents. **Koelreuter's** ***work provided a clue about how heredity works. The traits he was studying were capable of being masked in one generation and reappear in the next generation.*** This pattern is not predicted by the theory of direct transmission. Also the traits did not blend and reappeared in the next generation in their original form.

Another important aspect of Koelreuter's hybridization experiments was that offspring of a single mating exhibited **alternative forms (alleles)** of the trait under study. Some offspring exhibit one alternative form of a trait, for example hairy leaves; and the other offspring from the same mating exhibit a different alternative form, for example smooth leaves. A modern geneticist may have explained it on the basis of law of segregation of gametes. This segregation of alternative forms of a trait helped Mendel to understand the nature of heredity.

Koelreuter's work was improved by many investigators and especially by farmers who were trying to improve varieties of agricultural plants. **T.A. Knight** (1790s) crossed varieties of **garden pea** (*Pisum sativum*) which were uniform from one generation to the next (now known as **true-breeding**). One of these varieties had purple flowers; the other, white flowers. All of the offspring had purple flowers. When hybrids were crossed, the offspring consitsted of some plants with purple flowers and few with white flowers. There is resemblance with Koelreuter's experiments in that a character from one parent was hidden in one generation and reappeared in the next.

From the above mentioned facts it can be concluded that:

a. one form of inherited trait is masked in one generation and reappear unchanged in the next generation;

b. alleles (forms of a trait) segregate among the offspring of a cross; and

c. some forms of a trait appear in more number than their alternative forms.

However, the work of these investigators failed to make an impact for over a century. The possible reasons being that the earlier workers did not maintain a numerical record of results that is crucial to the understanding of heredity. For example, it was noted that in pea plants some traits appear strongly than the others, but because the number of different classes was not recorded, therefore the results cannot be compared.

Particulate Nature of Hereditary Material --- Work of Galton

Francis Galton suggested that hereditary material is particulate in nature. According to him these particles must be handed down, that is why one parental trait disappears during hybridization and reappears again in a later generation.

Use of Biometry

Another importance of Galton work is that he used biometry in heredity experiments. He measured individuals for a variable characteristics such as height. He compared the measurement made for one generation with the next. But he was unable to explain that what happen when contrasting traits are brought together.

Role of Cytology

Genetics is related to **cytology**, ***the science of cellular structure and function***, therefore history of genetics can be traced to the discovery of cell. The cell was discovered by **Robert Hooke** (1665), and at that time the significant character of the cell was considered to be presence or absence of cell wall. The cellular structures were not considered necessary. **Robert Brown** (1931) noticed a constant element within the cell and named it as **nucleus**. Few years later **Schleiden** and **Schwann** proposed the **cell theory** that led to recognition of cell as unit of life. Cell theory also helped to recognise various disciplines of biology such as physiology, embryology, pathology, etc.

Discovery of Chromosomes

A dye named **aniline** was discovered in 1870. It helped in staining the cell and its components in a better way. At about the same time **condenser** and **oil immmersion lens** were developed that helped scientists to observe cellular structures in detail. The most significant cellular structures observed during this period were **chromosomes**. A number of biologists, e. g., **Flemming**, **Strausberger**, **Hertwig** and **Fol**, and **Weismann** observed division and duplication of chromosomes and cell divisions, the **mitosis** and **meiosis**. Botanists and Zoologists reached conclusions independently that **chromosomes are the physical basis of heredity** because only these are distributed precisely during cell division. This fact was supported by another finding that the size of the nucleus is same in gametes although the cytoplasmic contents may vary.

Chromosome Theory of Heredity

By 1880, there were strong suggestions that chromosomes are the carriers of heredity material. This was named **chromosome theory of heredity** and the chief proponents of this theory were **Hertwig**, **Kolliker**, **Weismann** and **Strausberger**. Weismann formulated the **continutity of the germ plasm** and concluded that a special kind of cell division during which the number of chromosomes is reduced to one-half is the basis of genetic continuity. This process is now known as **meiosis**, but the details of it were worked out after 1900.

Mendel and Inheritance

The year 1900 is very important in the history of genetics as the work of Gregor Mendel was rediscovered. Mendel has actually presented his work in his paper read at Brunn, Czechoslovakia, about thirty five years earlier. This was before any detailed observation had been made on the cell nucleus and chromosomes. Therefore, his work was not given importance at that time. But later on work of **DeVries, Correns** and **Von Tschermak** confirmed Mendel's results working with plant material and era of **Mendelian genetics** started. *Mendel's findings fit quite well with the particulate nature of individual chromosomes that separated during cell division.*

Identification of Chromosomes as Carriers of Genes

The constancy of chromosomes helped explain the constancy of heredity; and the different possible combinations of chromosomes during meiosis helped explain some of the variability of heredity. This led to the identification of chromosomes as carriers of genetic material by **Sutton** (1876-1916), **Boveri** and others.

The beginning of the twentieth century saw the basic groundwork of genetics embodied in the idea that an actual heredity material existed, that it was a particulate nature composed of **genes carried on chromosomes**, and that its behaviour in transmission from one generation to another could be predicted.

SCOPE OF GENETICS

The scope of topics included in the field of genetics is enormous. Studies have involved viruses, bacteria, and a wide variety of plants and animals and have spanned all levels of biological organisation, from molecules to populations. However, the science of genetics has been used in the areas of agriculture and medicine mainly.

I. Agriculture and Genetics

Although the cultivation of plants had begun long before, however the rediscovery of Mendel's work spurred scientists to apply genetic principles to the process. In plants four major categories of improvements have been possible:

a. more effiecient energy utilization during photosynthesis, resulting in more vigorous growth and increased yields;

b. increased resistance to natural predators and pests, including insects and disease-causing microorganisms;

c. production of hybrids exhibiting a combination of superior traits derived from two different strains or even two different; and

d. selection of genetic variants with increased protein value or an increased content of limiting amino acids which must be present in the human diet.

These improvements have resulted in a tremendous increase in yield and nutrient value in crops such as *barley*, *beans*, *corn*, *oats*, *rice*, *rye*, and *wheat*. For example, in Mexico, where *corn* is the staple crop, a significant increase in protein content and yield has occurred. Similarly an effort has also been made to improve the growth of *Mexican wheat*. Favourable genes from wheat strains found in various parts of world have been incorporated creating a superior variety which is now grown in many underdeveloped countries.

II. Animals and Genetics

Applied research in genetics has developed superior breeds of animals. Enormous increases in usable meat supplies produced per unit of food intake have occurred. For example, selective breeding has produced chickens which grow faster, produce more high quality meat per chicken, and lay greater numbers of larger eggs. In larger animals such as cows, the use of artificial insemination has been particularly important. The sperm of a sinlge male with superior genetic traits may now be used to fertilize thousands of females located in all parts of the world.

III. Medicines and Gentics

Advances in genetics resulted in development of medicines. Numerous disorder in humans have discovered to result from either a single mutation or a specific chromosomal abnormality. For example, the genetic basis of **sickle-cell anemia**, **erythroblastosis fetalis**, **hemophilia**, **Down syndrome**, and countless **metabolic disorders** in *humans* have been discovered. Recognition of genetic basis of these disorders has provided direction for the development of treatment and preventive measures. For example, **genetic counselling** provides parents with objective information upon which they can make rational decisions. It is estimated that every childbearing couple stands an approximate three percent risk of having a child with some form of genetic anomaly.

Applied research in genetics has provided other medical benefits. Increased knowledge in **immunogenetics** has made possible compatible blood transfusions as well as organ transplants. The discovery of tissue bound antigens has led to the important concepts of **histocompatibility** and **tissue typing**. With the development of immunosuppresive drugs, transplant operation involving human organs, including the heart, liver, and lungs are increasing annually.

Recombinant DNA Technology

Recombinant DNA technology is also an important part of applied genetics. By cloning human genes for such medically important molecules such as **insulin** and **interferon**, bacteria can serve source of mass production of these gene products. Recombinant DNA techniques will also play an increasing and essential role in human genetic engineering, which involves the direct manipulation of the genetic material (**gene therapy**). It involves replacement of defective genes in patient's body by copies of healthy genes.

The recombinant DNA techniques have also been applied and used for improvement of plants. Genes for herbicide and insect resistance have been introduced into the plants. Efforts are being made to introduce genes into the cultivated grasses that enable these to form root nodules. This would make plants free from nitrogen fertilisers.