**Human Genetics: Its Historical Background**

The science of Human genetics basically deals with the study of transmission and inheritance of characteristics, which may be in an individual, family, a race or population. From time immemorial, people are curious to know about the transmission of characteristics from parents to offspring. The history of human genetics cannot be studied separately from genetics, because most of the experimental and monumental works on genetics were carried out in non-humans rather than in human beings.

The foundation of modern genetics or human genetics was first laid down by Johann Gregor Mendel (1822-1884) in 1865 when he published his findings of his famous *Pea Plant Experiment*. His findings remain unnoticed for almost for 35 years and in1900, his findings were rediscovered independently by three European scientists, namely, Hugo de Vries, Carl Correns and Erich Von Tschermark. These findings have brought revolutionary and significant changes in the concept and ideas about the transmission of the characters from generation to the generation. But, it must be mentioned that even before Mendel, we can traced back some thought and theories related to human hereditary far back to the days of Greek Physician and thinker Hippocrates (460BC-375BC) who propounded the Brick and Mortar theory. Similarly Aristotle (384BC-322BC) had also put forwarded the idea that the characters are transmitted through semen and mother's menstrual blood, which mixed in the mother's womb. The Spanish physician Mercado in 1605 put forwarded the opinion that both parents contribute equally to the offspring. Similarly, Malphigi (1628-1694) propounded the ‘Preformation Theory’ according to which the whole organism is preformed in concrete shape in the ovum. In 1753 a French philosopher and scientist Maupertius for the first time published his work on the transmission of a dominant hereditary trait *Polydactylia* in four generationsin a Berlin family. In 1814 a British physician and surgeon Joseph Adams (1756-1818) published a book on human hereditary called *'A Treatise on the on the Supposed Hereditary Properties of Disease*'.

In the late 18th century, French naturalist Jean-Baptiste Lamarck (1744-1829) put forwarded his idea of inheritance through his theory of evolution. His theory was however severely criticised by evolutionary biologist August Weismann (1834-1914)who himself propounded the famous *Germplasm* theory. During the same period, another English scholar Sir Francis Galton (1822-1911) published a book 'Hereditary Genius' in 1869 where he tried to explain the behaviour of man on the basis of genetical differences. He was not only the first person to adopt *Twin method* to study the influence of heredity and environment on the phenotype of man but also considered as founder of the science E*ugenics and Biometric genetics*.

Towards the end of the 19th century, many research works and discoveries were made which triggered the development of genetics. In 1869, exactly three years after Mendel’s experiment, a Swiss physician Fredrich Miecher has successfully isolated the from the cell nuclei and led the foundation of molecular genetics. Some of the important landmark discoveries in the field of genetics during this period were *the detection of the chromosome by Karl Von Nageli Wilhelm in 1842* and later its identification as *chromosome in 1888 by German anatomist Von Waldeyer*, discovery of the *detail movement of the chromosome and separation of the sister chromatids in mitosis in 1879 by German physician Walther Flemming* (1843-1905) , *discovery of process of equal distribution of chromosomes to the daughter nuclei by Belgian biologist Edouard Van Beneden* (1846-1910)), and the works of *Oswald Avery, Colin Macleod and Maclyn McCarty in 1944 to established that substance responsible for transformation of genetic material was DNA*.

The discovery of ABO blood group system by the Austrian physician Karl Landsteiner in 1901 was one of the most significant discoveries in the beginning of the 20th century. Some of the landmark developments in the field of human genetics during this time were the *demonstration of inheritance pattern of a human genetic disorder Alkaptonuria* by English physician A. Garrod in 1901, *discovery of the role of chromosome in hereditary* by German scientist Theodor Boveri (1862-1915) in 1902, *revelation of the mechanism of the division of chromosome in reduction division (meiosis)* by Walter Sutton in 1903 and *coining of the term ‘genetics’ by William Bateson* in 1905. During this period, we witnessed the growth of the concept of ‘Neo-Darwinism ’through assimilation of the principles of Mendelian inheritance with that of Darwinian concept of natural selection by R.A. Fisher, J.B.S. Haldane and Sewall Wright. This synthesis of the Mendelism and Darwinism marked the foundation of a new branch of genetics called ‘population genetics’. The discovery of Hardy-Weinberg principle in 1908 independently by G.H. Hardy and W.Weinberg was a major advancement in population genetics. In 1910 Thomas Hunt Morgan (1866-1945) discovered that genes are located in the chromosomes like beads in a string and are the bearer of hereditary information. He also established human gene maps for haemophilia and colour blindness on the X chromosome. In 1911 Alfred Henry Sturtevant (1891-1970) created the first genetic map of chromosome. In 1910-11 when Ludwik Hirzfield and E.Von Dungen discovered the inheritance pattern of the ABO blood group.

From the mid of the 20th century, advancement in human genetics becomes more rapid. The relationship between proteins and the genes was revealed by George W. Beadle (1903-1989) and Edward L. Tatum (1909-1975) in 1940. Following this discovery, Fred Sanger (1918-2013) made remarkable contribution by constructing the sequences of amino acids in insulin, a protein secreted by the pancreas. The ground breaking discovery in the genetic research took place in 1953 when American biologist James Watson and English physicist Francis Crick unrevealed the double helix structure of DNA and laid the groundwork of molecular genetics. One of the major developments that took place in the study of human genetics was the correct estimation of the number of chromosome as 46 in man by J.H.Tjio and A.Levan in 1956. Prior to this discovery, for prolonged 30 years, it was believed that the number of chromosomes was 48. The discovery of the causes of sickle cell anaemia by Linneaus Pauling in 1949 and that of Down syndrome in 1959 by Jerome Lejeune was also an important discovery in the field of medical genetics.

In 1961, Francis Jacob and Jacques Monod demonstrated that the products of certain genes regulated the expression of other genes. Since the mid -seventies, studies in genetics undergone a revolution on the use of sophisticated technology to isolate and identify specific fragments of DNA. The first step towards mapping human chromosome was occurred in 1968 when Rojer Donahue and others discovered the location of Duffy blood group locus in the human chromosome 1. The advancement of technology in the field of cytogenetics and biochemical genetics during this period has made it possible to study human chromosome particularly with the help of Branding techniques developed by Caspersson in 1969 and G Branding Pattern technique by Maximo Drets and Margery Shaw.

A new field called genetic engineering was emerged with the invent of recombinant DNA technology. The first ever “recombinant DNA” was created by Dr. Paul Berg in 1971 followed by Stanley Cohen and Herbert W. Boyer in 1972. In the same year Walter Fiers, along with his team for the first time sequences the gene of Bacteriophage MS2. Following these developments, in 1977 Richard J. Roberts and Phillip Sharp discovered that genes can be split up into segments. These developments in genetic sequencing had marked a turning point in molecular genetics.The Sanger group in 1977 developed techniques to sequence, or decodes, DNA.. These developments have led to the first International Human Gene Mapping school workshop in New Haven at Yale University in June 1973 where geneticist have able to mapped 100 genes. In 1999, for the first time the genetic code of an entire human chromosome 22 was deciphered by Researchers of the Sanger Centre near Cambridge, University of Oklahoma,; Washington University and Keio University of Japan

In the last six and half decades rapid technological developments in the field of genetics have revolutionised the study of human genetics. The progression of DNA sequencing technology led to the launch of the Human Genome Project in 1990 and its completion in 2003. The HGP was launched worldwide to determine the whole sequence of chemical base pairs of human DNA and of identifying and mapping all the genes of the human genome.

Another milestone in the history of human genetics was the launching of UK10K project by UK in 2010 with an aim to uncover the genes that are involved in causing diseases by studying the genetic code of 10000 populations. The science of human genetics has reached a new height after completion of HGP. Many developments in genetic and cellular level have been taking place that could yield enormous positive implication in the years to come.