HUMAN GENETICS

RAMSADAY COLLEGE

LOAKNATH GHOSH

Human genetics is the scientific study of human heredity. In other words, the study of human heredity occupies a central position in human genetics. Inheritance in humans does not differ in any fundamental way from that in other organisms. An understanding of human heredity is of critical importance in the prediction, diagnosis, and treatment of diseases that have a genetic component. The quest to determine the genetic basis of human health has given rise to the field of medical genetics. In general, medicine has given focus and purpose to human genetics.

Human genetics today comprises a number of overlapping fields, including:

- **Classical or formal genetics** -- the study of the transmission of single genes within families and the analysis of more complex types of inheritance.
- **Clinical genetics** -- the diagnosis, prognosis and, in some cases, the treatment of genetic diseases.
- **Genetic counseling** -- an important area within clinical genetics involving the diagnosis, risk assessment, and interpersonal communication.
- **Cancer genetics** -- the study of genetic factors in inherited and sporadic cancer.
- **Cytogenetics** -- the study of chromosomes in health and disease.
- **Biochemical genetics** -- the biochemistry of nucleic acids and proteins including enzymes.
- **Pharmacogenetics** -- how genes govern the absorption, metabolism and disposal of drugs and untoward reactions to them.
- **Molecular genetics** -- the molecular study of genetics including particularly DNA and RNA.
- **Immunogenetics** -- the genetics of the immune system including blood groups, HLA, and the immunoglobulins.
- **Behavioral genetics** -- the study of genetic factors in behavior in health and disease including mental retardation and mental illness.
- **Population genetics** -- the study of genes within populations including gene frequencies, the gene pool, and evolution.
- **Reproductive genetics** -- the genetics of reproduction including genes and chromosomes in germ cells and the early embryo.

- **Developmental genetics** -- the genetics of normal and abnormal development including congenital malformations (birth defects).
- **Ecogenetics** -- the interaction of genetics with the environment.

- **Forensic genetics** -- the application of genetic knowledge, including DNA, to legal matters (William, C. et al.).

AIM AND APPLICATION OF HUMAN GENETICS IN ANTHROPOLOGY

This intimate relationship between genetics and anthropology was first characterized in 1973, in a volume entitled "*Methods and Theories of Anthropological Genetics".*

Scientific interest in the relationship between race and human biological variation has intensified recently with advances in genomics. There is hope that an increased knowledge of the human DNA sequence and the discovery of DNA sequence variations within and among individuals will provide definitive answers to the long-standing questions about the biological aspects, and indeed the biological validity, of the idea of race.

Genetic diversity is measured from DNA sequence differences between alleles. There are many methods for estimating genetic diversity.

However, all of the methods reveal three major features that typify a unique pattern of human genetic diversity. The three basic properties of human genetic variation are seen in patterns of *nucleotide diversity.*

1. The first feature is that the amount of diversity at the DNA level is only a fraction of what would be expected for a species that consists of billions of members (example- humans have low diversity, is apparent when comparing humans and chimpanzees).
2. The second feature is that the genetic diversity in people living outside of sub-Saharan Africa is mostly a subset of the genetic diversity in populations within sub-Saharan Africa.
3. The third feature is that, at most genetic loci, a variant allele that is common in one human population in the entire species (example- human genetic variations are illustrated by DNA sequences from widely dispersed populations in Africa, Asia, and Europe).

The word *race* should be used carefully because different meanings have been affixed to it in scientific, social, and historical contexts. Population geneticists typically define *race* as a group of individuals in a species showing closer genetic relationships within the group than to members of other such groups (Example-The genetic variation found between Europeans, Asians and African population).

Health researchers are actively debating the value of race and ethnicity in the diagnosis and treatment of chronic diseases such as diabetes, high blood pressure, and cancers. It is well known that chronic diseases are unevenly distributed in the general population. Depending on the disease, some groups are more or less prone than others.

The architecture of human genetic variation is ultimately explained by the evolutionary history of our species and best understood in that context (Example-The molecular revolution and how DNA markers can provide insight into the processes of evolution).

The field of anthropological genetics utilizes a comparative approach on small, isolated populations and topics such as human variation, evolutionary theory, reconstruction of the human Diaspora (out-of-Africa), genetic epidemiology, and forensic sciences.

Anthropological geneticists have been successful in mapping quantitative trait loci involved in biological pathways of diseases such as diabetes mellitus, cancers, obesity, osteoporosis, and coronary heart disease.

The prominent role of anthropological genetics used in legal interest (using classic genetic markers and molecular methods).

In population studies, genetic markers have been defined as “discrete, segregating genetic traits which can be used to characterize populations by virtue of their presence, absence, or high frequency in some populations and low frequencies in other.