

# Emergence Of A New Integrated Discipline Of Medical Genetics And Biological Anthropology: A Super Specialty In Demand

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## Abstract

Anthropology is a common human science with its vast subject matter revolving around knowledge, applications and human surroundings in space and time. Metamorphosis in human life has enforced to widen the traditional vision (wisdom) and scope of particular discipline in terms of applications. In biological terms, heterosis is a process of coming together with another for vigor, virility, vitality and strength. Prudent heterosis means intra- and inter-disciplinary approaches to achieve a common goal. This paper over emphasizes the integral approaches of medical genetics and biological anthropology including human genetics in solving the emerging human health problems. These integrated approaches of medical genetics and biological anthropology will bring together unity in the discipline of Biomedical Anthropology and emerge as a super specialty in the medical and science world.

## INTRODUCTION

In a broader sense, anthropology is a common human science with its vast subject matter revolving around knowledge, applications and human surroundings in space and time. No other discipline of science is so vast with subject matter as that of anthropology. Today, the sea changes occurring in human life have enforced to widen the traditional vision (wisdom) and scope of particular discipline in terms of its applications and future course. In this context, heterosis is a process of coming together or joining hands with another for vigor, virility and vitality or strength. The prudent heterosis means intra- and inter-disciplinary approaches for achieving a common goal. This article, therefore, emphasizes the integral approaches of medical genetics and biological anthropology including human genetics in solving the human health problems in a variety of ways.

## SCIENCE OF GENETICS

Genetics (from the Greek *genno* = give birth) is the science of genes, heredity, and the variation of organisms. Heredity and variation form the basis of genetics. The knowledge of genetics in prehistoric times was applied by humans in the domestication and breeding of animals and plants<sup>2,3</sup>. Today, genetics provides an important tool in research for investigation of the function of a particular gene, e.g.,

analysis of genetic interaction. Within organisms, genetic information generally is carried on thread-like structures called, chromosomes where the chemical structure of particular deoxyribonucleic acid (DNA) molecules is represented. Genes encode the necessary information for synthesizing the amino acid sequences in proteins, which, in turn, play a large role in determining the final phenotype or physical manifestation of the gene in the organism. In diploid organisms, a dominant allele will mask the expression on one chromosome and that of a recessive allele on the other. The only way that a recessive allele can be expressed as a trait is, if the organism is homozygous (carries two copies of) the recessive allele. The interaction between alleles is often more complex than an explicit simple dominant-recessive relationship stated above.

Genetics determines much (but not all) of the appearance of organisms, including humans, and possibly how they act. Environmental differences and random factors also play a part. Monozygotic (identical) twins, a clone resulting from the early splitting of an embryo, have the same DNA, but show different personalities and fingerprints. Genetically, identical plants grown in colder climates incorporate shorter and less-saturated fatty acids to avoid stiffness. Molecular genetics builds upon the foundation of classical genetics but focuses on the structure and function of genes at a molecular

level. Molecular genetics employs the methods of both classical genetics (such as hybridization) and molecular biology. It is so called to differentiate it from other sub fields of genetics such as ecological genetics and population genetics. An important area within molecular genetics is the use of molecular information to determine the patterns of descent, and, therefore, the correct scientific classification of organisms: this is called molecular systematics. The study of inherited features, not strictly associated with changes in the DNA sequence, is called epigenetics.

Some take the view that life can be defined in molecular terms, as the set of strategies which RNA polynucleotides have used and continue to use to perpetuate themselves. This definition grows out of work on the origin of life, specifically the RNA hypothesis in the world.

Population genetics (quantitative and ecological) are all very closely related subfields and also build upon classical genetics (supplemented with modern molecular genetics). They are chiefly distinguished by a common theme of studying populations of organisms drawn from nature but differ somewhat in the choice of which aspect of the organism on which they focus. The foundational discipline is population genetics which studies the distribution of and change in allele frequencies of genes under the influence of the four evolutionary forces: natural selection, genetic drift, mutation and migration. It is the theory that attempts to explain such biological phenomena as adaptation and speciation. The related subfield of quantitative genetics, which builds on population genetics, aims to predict the response to selection, given data on the phenotype and relationships of individuals. A more recent development of quantitative genetics is the analysis of quantitative trait loci. Traits that are under the influence of a large number of genes are known as quantitative traits, and their mapping to a location on the chromosome requires accurate phenotypic, pedigree and marker data from a large number of related individuals.

Ecological genetics again builds upon the basic principles of population genetics but is more explicitly focused on ecological issues. While molecular genetics studies the structure and function of genes at a molecular level, ecological genetics focuses on wild populations of organisms, and attempts to collect data on the ecological aspects of individuals as well as molecular markers from those individuals. Population genetics is closely linked with the methods of genetic epidemiology. One method to study

gene-disease associations is using the principle of Mendelian randomization.

The science which grew out of the union of biochemistry and genetics is widely known as molecular biology. The term "genetics" is often widely conflated with the notion of genetic engineering, where the DNA of an organism is modified for some kind of practical end, but most research in genetics is aimed at understanding, functioning and explaining the effect of genes on phenotypes and in the role of genes in populations rather than genetic engineering.

A more recent development is the rise of genomics especially the functional genomics, which attempts the study of large-scale genetic patterns across the genome for (and in principle, all the DNA in) a given species. The field typically depends on the availability of whole genome sequences, computational tools and sequence profiling tool using bioinformatics approaches for analysis of large sets of data.

## **MEDICAL GENETICS**

Medical Genetics is the application of genetics to medicine<sup>4,5</sup>. Genetic medicine is a newer term for medical genetics. Medical genetics is a broad and varied field. It encompasses many different individual fields, including clinical genetics, biochemical genetics, cytogenetics, molecular genetics, the genetics of common diseases (such as neural tube defects, sickle cell disease, thalassemias, etc.), and genetic counseling<sup>6</sup>.

Each of the individual fields within medical genetics is a hybrid. Clinical genetics is a hybrid of clinical medicine with genetics. Biochemical genetics is a hybrid of biochemistry, mainly of amino acids and proteins, with genetics. Molecular genetics is a hybrid of the biochemistry of DNA and RNA with genetics. Cytogenetics is a hybrid of cytology and genetics; it involves the study of chromosomes under the microscope. Genetic counseling is a hybrid of genetics and nondirectional counseling.

The study of inheritance of human characteristics from parents to children is called human genetics. Inheritance in humans does not differ in any fundamental way from that in other organisms. The study of human heredity occupies a central position in genetics. Much of this interest stems from a basic desire to know who humans are and why they are as they are.

## **HUMAN GENETICS DIFFERS FROM MEDICAL**

## **GENETICS**

Human genetics differs from medical genetics in that human genetics may or may not apply to medicine, but medical genetics always applies to medicine. The study of Huntington disease (a progressive neurologic disease) is properly part of both human genetics and medical genetics, whereas the study of eye color (except in situations such as albinism) is part of human genetics but not medical genetics.

Similarly, for example, the aspects of etiology, pathology, symptomatology and clinical manifestations and treatment such as administration of medicines, repeated blood transfusion, stem cell or bone marrow transplantation in patients of sickle cell disease or thalassemia syndromes, and genetic counselling is a part of medical genetics; on the other hand, geographical distribution, prevalence/incidence, afflictions in communities or populations, mode of transmission, and evolutionary tendencies of these abnormalities fall within the gasket of human genetics; and prognosis, physical and physiological disabilities, morphological (appearance) distortions, mental retardation, nutrition, biological adaptation, and linked socio-cultural and psycho-economic factors constitute the subject of biological anthropology. All these overlapping components are intricately related to each other and determine the health and disease outcome.

Emphasizing the combination of Anthropology with the Health Sciences makes particular sense because there is an applied field called Medical Anthropology that is two-pronged. First, many physical anthropologists become involved in various aspects of medical research. Often this involves genetics, but can also range into other human biological areas. Second, medical anthropology also has a cultural side that specializes in the problem of understanding ideas about health and illness in other cultures. This is an applied anthropological area that keeps the problem of supplying health care across cultural boundaries in mind. Studying cultural anthropology helps potential health care deliverers understand some of the variety of ways people in other cultures conceive of illness, and helps them explain treatment in terms that such people can understand and accept. An understanding of illness questions in other cultures also entails a more holistic approach to well being that considers psychological and social conditions as well as the biomedical factors normally considered by doctors, nurses, and public health administrators/professionals<sup>7</sup>. Sometimes people are ill, not from diseases readily

recognized by Western medical science, but because their social relationships are maladjusted. In such cases referral to traditional curers may be better than just sending them home. As society becomes increasingly multicultural, that kind of anthropological understanding becomes more crucial to have in combination with biomedical knowledge. There are traditional curers (counseling or psycho-social therapy) among the Asian (Indian) tribal/nontribal communities in South East Asia, and that ideas about illness among people in those populations differ a great deal from those of the general public of western countries like that of United Kingdom, Australia, America, etc.

## **PHYSICAL OR BIOLOGICAL ANTHROPOLOGY**

Medical anthropology finds diverse interests and approaches among the anthropology faculty. Medical anthropology offers students important opportunities to integrate sociocultural and biomedical approaches. The research and teaching around health and illness cover diverse topics and specializations, including: ecological anthropology, semiotics and therapeutic processes, human molecular genetics, demographic process, disease and nutrition, international health policy and human rights, and the integration of biology, behavior, power, technologies in human reproduction, and evolutionary biology<sup>8</sup>. Students of medical anthropology are uniquely positioned to develop a robust knowledge of the diversity of anthropological methods needed to analyze health and illness, and may complement their anthropological training with a wide array of courses tailored to their individual needs, such as cultural psychology, behavioural adaptations, health policy and management, biostatistics, etc.

The knowledge that biological anthropologists gather on living populations falls into several overlapping categories of human genetics. Again, evolution and biosocial variation are underlying themes in studies that deal with nutrition, child growth, health in societies, the genetics of human populations, and adaptation (adjustment) to the environment<sup>9</sup>.

Human disease and its problems have both ecological and cultural bases, often tightly interconnected. Studies of the patterns of disease within human populations in different biological and social environments and across and within populations, represent one focus of the Biological Anthropology program. Research topics include human disease in diverse ecologies (including urban, agricultural, and foraging); the health implications of diet, including

studies of malnutrition, obesity, and medicinal plant use; evolutionary ecology of disease.<sup>1</sup>

Anthropologists demonstrate a particular capability in helping to solve human problems through building partnerships in research and problem solving; acknowledging the perspectives of all people involved; focusing on challenges and opportunities presented by biological variability, cultural diversity, ethnicity, gender, poverty and class; and addressing imbalances in resources, rights, and power. Such contributions are most effective in interdisciplinary settings where active and committed social scientists work in partnership with active and committed representatives of community, national, and international constituencies.

Medical anthropologists study health both cross-culturally and over evolutionary time scales. Anthropology contributes to our understanding of health by examining how they operate in diverse social, political, economic, cultural, and gender contexts across time and space.<sup>5</sup>

Biological anthropology is undergoing rapid and significant change as we enter the 21st century.<sup>5</sup> Biological anthropologists are developing broader interests beyond traditional themes in academic departments of anthropology, and finding new job opportunities in and outside of academia. Biological anthropologists can be found in medical schools, schools of public health, many companies producing pharmaceuticals and dietary items, and at major government research organizations such as the Indian Council of Medical Research (ICMR), the National Institutes of Health (NIH, USA), and the Centers for Disease Control and Prevention (CDC). Biological anthropology draws its students from a wide variety of disciplines that include the paramedical, nursing, behavioral sciences, natural sciences, social sciences and the humanities.

## **BIOMEDICAL ANTHROPOLOGY**

Anthropology of Disease or Biomedical anthropology is an emerging subdisciplinary area of biological anthropology,<sup>1, 5,10,11</sup>. It represents the interface between biomedicine and the behavioral and social sciences that shapes health status. As such, it does more than give lip service to integrating cross-disciplinary approaches. It represents an educational philosophy that has been recommended as part of a new and innovative graduate training initiative (Reshaping the Graduate Education of Scientists and Engineers, National Science Academy) implemented by the National Science

Foundation (NSF) all over the world.

Biomedical anthropology emphasizes biomedical, bio-behavioral and epidemiological approaches to understanding the transmission and dissemination of disease and the cellular and molecular mechanisms of pathogenesis and the dynamic interaction of biological and socio-cultural factors that shape health outcome.<sup>11, 12</sup>. The program includes rigorous field and wet laboratory-based research training.

There is a need by the discipline to provide necessary and appropriate training to supply a qualified Anthropological work force (holistic/integrative/multidisciplinary) for current and developing positions within the biomedical field. Anthropology can provide biomedicine with the best multidisciplinary-trained individuals who bridge the social, behavioral, biological and medical sciences. The National Academy of Sciences (NAS) and the National Science Foundation (NSF) have recently supported the reshaping of graduate education in the sciences. They strongly recommend preparing students for “an increasingly interdisciplinary, collaborative and global job market . . . and should not be viewed only as a byproduct of immersion in an intensive research experience. This program is in a unique position to make a major contribution to fulfill the needs of a changing world and a changing job market.<sup>13</sup>

Students with interest in cross-disciplinary, health-related professions will be drawn from Bachelors programs, in anthropology, biology, nursing, pre-medicine, public/international health programs, psychology and other sciences and social sciences. In addition, there is the expectation the international students; particularly those in the medical and public health professions from other countries will find this program of benefit. The list of target professions given below is not all-inclusive, but represents examples of the kinds of areas that Masters Degree recipients in Biomedical Anthropology may serve:

## **PUBLIC/COMMUNITY HEALTH**

- International Health/Health Development
- Epidemiology
- Forensic Anthropology/Molecular Identification/Paternity Issues
- Molecular Anthropology/Biology
- Ethnomedicine/Natural Products Research

- Administration/Research/Health Care Delivery in
  - Government Agencies
  - Non-Government Organizations
  - Private Volunteer Organizations
  - Relief Organizations
  - Hospitals
  - Medicine/Health Care Delivery
  - Health Care Development/World Bank Advisors
  - Grants Officers
  - Research Coordinators
  - Human Subjects/Biomedical Ethics Advisors

## CONCLUSION

Biomedical anthropology is disease-centered and its investigations inquire into the impact of the disease (or condition) upon biological processes of individuals or the populations to which they belong. The prudent heterosis between biological or physical anthropology (including human genetics) and medical genetics has already been emphasized. This includes the integral approaches of physical (biological) anthropologist with interests in human biology, human genetics, molecular medicine, human growth, development and nutrition, human ecology, and human being as physical entity, and of a medical anthropologist whose interests are in the areas of health behavior, medical care (intervention) systems, health planning, psychosomatic illness including mental health, and correlation of demographic variables. The contention is that a biological anthropological approach is best suited when it emphasizes the biological basis of health and disease, while at the same time actively incorporating and understanding the socio-cultural nature of the sickness process in the society. Thus, integrated approaches of medical genetics and biological anthropology including human genetics will bring together not only the unity in diversity but emerging Biomedical Anthropology as a super specialty in the medical and science world.

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## References

1. Balvir RS. Biomedical anthropology in the service of mankind in the new millennium: Are we ready? In: Bhasin MK, Malik SL, editors. *Anthropology: Trends and Applications*. New Delhi: Kamala-Raj Enterprises, 2002. p.141-147.
2. Balvir RS. An upsurge of biotechnology in India: A commitment towards human health and disease. *Indian J Multidisciplinary Res.*2005; 1:153-64.
3. Balvir RS. Medical biotechnology in human health and disease in North-Eastern India: A commitment towards the betterment of society. In: Basu Debashis, Kulirani B Francis, Datta Ray B, editors. *Agriculture, Food Security, Nutrition and Health in North East India*. New Delhi: Mittal Publications, 2007. p.333-46.
4. Balvir RS. Medical genetics in clinical practice in India. *Curr Med Trends.* 1999; 3: 567-72.
5. Balvir RS. Biomedical anthropology in contemporary tribal society of India. In: Behera, Deepak Kumar, Pfeffer, Georg, editors. *Contemporary Society: Tribal Studies (Tribal Situation in India)*, Vol.6. New Delhi: Concept Publishing Company, 2005. p.292-301.
6. Balvir RS. Human genetics in community health practice in India: An urgent need of action. In: Sharma K, Pathak RK, Mehta S, Talwar I, editors. *Genes, Environment and Health: Anthropological Perspectives*. New Delhi: Serials Publications, 2007. p. 171-86.
7. Balvir RS. Medical genetics in public health administration in India: A handicap of bureaucracy, bias and corruption. *Health Administrator (Theme: Health of the Educational Systems)*, 2005; 17: 101-9.
8. Balvir RS. Health and morbidity pattern in the tea garden labour communities of North East India. In: Sengupta Sarthak, editor. *The Tea Labourers of North East India*. New Delhi: Mittal Publications, 2006. p.223-35.
9. Balvir RS. Dimensions of rural tribal health, nutritional status of Kondh tribe and tribal welfare in Orissa: A biotechnological approach. *Proceedings of the UGC Sponsored National Conference on Human Health and Nutrition: A Biotechnological*

Approach (Lead Lecture), Thane, December 12-13, 2004. p. 47-57.

10. Balgir RS. Biomedical anthropological approaches in North Eastern India: A synthesis of the tea garden population of Assam. In: Sengupta Sarthak, editor. Peoples of North East India: Anthropological Perspective. New Delhi: Gyan Publishing House. 1996. p. 91-104.

11. Balgir RS. Biomedical anthropology and health status of the tribal Muslim Gujjars of Himachal Pradesh, India. In: Sengupta Sarthak, editor.

Health, Healers and Healing: Studies in Medical Anthropology, Chapter I. Dibrugarh: National Library Publishers. 1999. p.1-19.

12. Johnston FE, Low SM. Biomedical anthropology: An emerging synthesis in Anthropology. Yearbook of Physical Anthropology, 1984; 27: 215-27.

13. Balgir RS. Academic curricula of Anthropology need overhauling: An observation. Man in India, 2002; 82: 407-12.

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