# Preface

From the anthropological and population genetics perspective Indian subcontinent is most attractive because of its unique population structure, which is characterized by the division of its population into strictly defined hierarchical and endogamous castes, tribes and religious groups constituting the natural Mendelian units on which evolutionary forces operate. Because of the historical reasons Indian subcontinent is also endowed with enormous cultural, linguistic and ethnic diversity. Therefore, India is regarded as a rich natural laboratory and a gold mine for population genetics research. In order to gain systematic understanding of this diversity and complex population structure of India, Indian populations have been subjected to intense investigations since the beginning of 20$^{th}$ century. With few exceptions, the focus of most studies was in understanding biological and/or genetic composition of isolated castes and tribes with little emphasis on synthesis of population genetic or biological relationships at the regional and /or national levels. There have been attempts in recent decades to collate these enormous secondary data and reanalyze the same to derive patterns of regional, linguistic and ethic variation with reference to traditional genetic markers as well as quantitative biological variables like anthropometry and dermatoglyphics. However, with the advent of molecular genetic technology, particularly after the development of rapid screening techniques using PCR, sequencers etc. the focus has shifted to using molecular genetic markers in understanding the genetic variability and in unraveling the history and peopling of the Indian subcontinent. In the process, many anthropological hypotheses in vogue concerning the Indian social structure have also been tested. This volume on Trends in Molecular Anthropology is essentially a collection of 20 papers that attempt to bring out the current status of knowledge in the field of molecular anthropology of populations and diseases in general, albeit the focus had been on the Indian situation. The papers contributed to this volume are broadly grouped under two heads: i) Molecular Genetic Variation among the Populations and ii) Molecular Genetic Markers in Disease. Although the first 11 papers that deal with the patterns of molecular genetic variation primarily focus on Indian populations we have two papers on populations from outside the country, one dealing with the origins of the Yakut people, based on mtDNA genome, and another on genetic structure of the native populations from the Gran Chaco Region, South America, based on mtDNA, Y-Chromosome and autosomal markers.

The remaining 9 papers review the status of knowledge on the molecular genetic etiology of a number of complex genetic disorders like diabetic retinopathy, age-related macular degeneration, alcoholism, diabetes type II, recurrent pregnancy loss, obesity cardiovascular disease etc. One of the common features in all these papers, however, is the account on the status of research in these aspects on Indian populations and the prospects that the Indian populations can offer in dissecting genetic etiology of these disease conditions.

The first paper in the volume by Tripathy et al. provides a comprehensive and critical overview of the molecular anthropological studies and the chronological developments in these studies in India in the background of unique Indian population structure and the insights earlier provided by the classical genetic markers. In the concluding part of this paper the authors also present a synthetic view emerging on the origins, migrations and phylogenetic relationships of the Indian populations vis-à-vis others, based on different sets of molecular markers besides evaluating merits and drawback of these studies. The second paper is a critical analysis of Indian Y-chromosome data in comparison to East Asian data by Carvalho-Silva and Tyler-Smith who illustrate with examples the impact of caste system and population substructure on the male genetic variation. The subsequent two papers by Kumar et al and Chaubey et al are case studies demonstrating language shift by certain tribes of Central-Eastern and Northern India and test the models of language diffusion by demic/cultural diffusion using mtDNA and/or Y-chromosomal markers. The next four papers describe patterns of variation in different sets of molecular markers among the Indian populations. Singh et al. provide critical analysis of the APOC3 (SstI) variability among the Northwest Indian populations in comparison to other ethnic populations other parts of the world and suggest distinctness of Indian populations when compared to others. Tripathi et al. focus on the role of Alu elements in detecting population diversity with examples from around the world, supplemented by a review on Alu based phylogenetic studies on Indian populations and another paper by Veerraju et al. present analyses of the genetic

structure of the southern Indian tribes and their phylogenetic position in comparison to other Indian populations based on Alu markers. The paper by Maji et al. provide an overview of the frequency distribution of mtDNA macro haplogroup N with particular reference to the more predominant haplogroup R and its sub-haplogroup U in relation to the geographic, linguistic and ethnic heterogeneity of the populations. The next paper by Trivedi et al. is a comprehensive treatment on the origins and peopling of India by different linguistic groups, based on a large number of Y-Chromosomal markers, both STRS and SNPS, and nearly 90 population groups from different parts of India. Of the last two papers in this section, the one by Zlojutro et al. attempt critical analysis of mtDNA HVSI sequences of the Turkic populations in comparison to other Asians in order to trace the origins of Turkic speaking Yakut people of Northeastern Siberia; in the other paper Demarchi and Ministro present synthesis of the principal findings of the long term study on the genetic structure of native native populations of the Gran Chaco region in South America using an assortment of morphological (anthropometry and dermatoglyphics) and molecular markers (mtDNA HVS-I sequences and haplogroups, Y-chromosome markers and and nuclear STRs).

Of the papers in the 2nd section, while Pitchappan et al. talk about the implications of HLA genomic diversity in HIV pandemic in India the papers by Satagopan et al. and and Kaur et al. provide with a comprehensive review of the molecular genetic aspects in eye diseases, Diabetic Retinopathy and Age-Related Macular Degeneration, respectively. The paper by Govindaiah reviews the epidemic relationship between plasma homocysteine and MTHFR gene polymorphism in cardiovascular disease whereas Nayak et al. and Gupta et al. present comprehensive reviews on the molecular genetic aspects of alcoholism and diabetes type II, respectively. While Nirmala et al. provide an overview of the status of knowledge on the genetics of human obesity, the last two papers in the volume deal with the anthropological perspective on the molecular etiology of recurrent pregnancy loss ( Walia et al.) and Beta-Globin gene mutations and their linkage to β-haplotypes in India (Gupta et al.). Overall, the scientific papers in this volume provide broad overview of the molecular anthropology/genetic variation of both the normal and disease populations in general and particularly pertaining to the Indian subcontinent.

In the process of editing this special issue/volume our students were of great assistance and help. I particularly thank Drs. Vikrant Kumar and Vikal Tripathy, Mrs Aruna Meka and Miss Shilpi Dasgupta for their help. I also thank many of those who devoted time for reviewing the manuscripts.

There were three significant persons with whom I had interacted very closely during the formative stages of my carrier in anthropology. Of the three, while Prof. D.P. Mukherjee was responsible for initiating me into this discipline as a teacher during my M.Sc. course at the Sri Venkateswara University, Tirupati, and later as a Ph.D. supervisor, Prof. V.P. Chopra had been quite supportive, friendly and understanding as a host and took personal care of me during my tenure as a AvH Fellow at the University of Hamburg, Germany, so that our association was quite productive. Last but the most significant association was with Professor K.C. Malhotra with whom I spent more than 25 years, both as student and colleague. He has been a guide, philosopher and role model for many young anthropologists and for me this association had been reassuring and confidence boosting. He has the knack of transforming 'apparent problems into no problems'. The compassion and the level of understanding that he had shown towards me was memorable and probably unparalleled. As a token of gratitude to these associations I take this opportunity to dedicate this volume with great pleasure to Professors K.C. Malhotra, D.P. Mukherjee and V.P. Chopra.

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