

## CHAPTER 3

### ANTHROPOLOGY

#### Doctoral Theses

008. ANU BHAWANA

**Anthropological Approach to the Study of Health Seeking Behaviour : A Case Study of the Bihari Migrant Labourers in Delhi.**

Supervisor : Prof. Subhadra Mitra Channa

Th 22482

*Abstract*

Migration occurs in a variety of patterns and one of the significant ones is from rural to urban, where people shift their base in search of livelihood and better quality of life. Delhi being the capital of India provides an ideal example of glamour and glitz where people from the marginalized economic sections of society arrive from distant rural places with bunch of aspirations and have to face new challenges. The urge for a better quality of life is the first and foremost reason of this spatial shift. Their socio-cultural equation changes as a result of this demographic shift. Individual mobility in the form of migration has various future implications on the sociocultural differentials like gender, age, health, etc. The relatedness of migration and health is highly gendered in India. Like many developing countries in the world, India is also experiencing rapid internal migration in recent years which will have definite implications on migratory patterns, gender relations, caste-class system in India apart from the effects on health seeking behaviour of the migrant community as well as the rest of the population. People do myriad kind of works to earn their livelihood in industrial sectors and other manual labour works. Migration in a way affects an individual's identity, behaviour, lifestyle on the whole. This study is aimed at understanding the paradigm of socio-economic status of these daily wage labourers and the dynamism involved with their socio-cultural transition as a result of migration.

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009. BISWAS (Amitabh)

**Molecular Genetic Basis of Hypertrophic Cardiomyopathy.**

Supervisor : Prof. V R Rao

Th 22243

*Abstract*

HCM is one form of cardiomyopathy in which left ventricle is hypertrophied causing heart failure. In this study, a total of 59 patients were recruited along with their 123 family members and 102 clinically evaluated age and sex matched controls. Demographical, clinical, physiological and lifestyle information were taken along with 5ml intravenous fasting blood with written prior consent. DNA extracted from the blood were subjected to genetic analysis of 6 markers (25bp Del in MYBPC3, 5bp Del in TNNT2,

287bp I/D in ACE, T-786C in eNOS, E101K in ACTC1, 3bp Del in MYH7 and P77L in TNNT2). Sanger sequencing of hotspot region of the MYH7 gene (exon 23) of all the patients, unaffected family members along with controls and whole exome sequencing of a family were performed. From serum, lipid analysis was done. We found that sedentary lifestyle, waist hip ratio, total serum cholesterol and triglycerides along with 5bp Del in TNNT2 gene were found to be significantly associated with the disease condition ( $p < 0.05$ ; adjusted for age and sex). A novel (L926K) and two rare variants (E924K and C>T at codon 923) were identified in the hotspot region of MYH7 gene in three unrelated patients and predicted to be pathogenic by different in-silico prediction tools. Whole exome sequencing of a family revealed a rare variant in P2RX7 gene with stop gain function. In conclusion the significant results observed improve the understanding of genotype/phenotype relationships and potentially patient risk stratification and choice of treatment. Besides, offering counseling to the affected family members with risk genotype. Most significant findings of the study being, the discovery of potential new gene in Indian HCM population.

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Supervisor : Prof. P. C. Joshi

Th 22480

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Supervisor : Dr. P R Mondal

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Supervisor : Dr. R. P. Mitra  
Th 22483

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Supervisor : Dr. R P Mitra  
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Supervisors : Dr. K N Saraswathy and Prof. Manju Puri  
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018. MITRA (Siuli)  
**Polymorphisms of Paraoxonase 1 (PON1) Gene Among the Selected Populations of India.**  
Supervisor : Prof. Gautam K Kshatriya  
Th 22251

*Abstract*

Genetic variation in paraoxonase activity and PON1 polymorphisms among populations across continental regions and among Indian populations has been previously documented. The underlying genomic processes that have shaped up the polymorphic variation of PON1 in different populations were not investigated. To comprehend genetic diversity, nature and extent of linkage disequilibrium among Indian populations, eight polymorphic sites of PON1 gene were examined among ten tribal and non-tribal populations. The data obtained was compared with diversity at the loci reported across world populations to find out the extent of genetic differentiation among PON1 loci. Allele and genotype frequency distribution at rs662 showed significant differences among the ten study groups. Some of the study groups were not in Hardy-Weinberg equilibrium at the rs662 locus carrying the Q192R polymorphism while no deviation was observed in the L55M polymorphism occurring in rs854560 locus. Global analysis mirrored the findings with an uneven distribution at Q192R and a homogenous high L allele frequency at L55M. No specific trend was observed across the promoter region. LD analysis revealed high recombination between the Q192R and L55M polymorphisms and was equal to or close to unity within the promoter region. Q192R was also found to deviate from the expectations of selective neutrality and limited fluctuations were detected at different loci in the  $F_{ST}$  and AMOVA analysis. Findings on geographic structure in Q192R were concurrent with the heterogeneous allele frequency distribution in most regions and a localized homogenous distribution in Europe at the loci. Although PON1 activity has been recommended as the preferred diagnostic marker for studying disease risk, studying population differences of PON1 polymorphisms will be important to understand genomic processes that have shaped up diversity in this gene and their role in the selective sweep that has occurred in the chromosomal region carrying the gene.

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Supervisor : Prof. P C Joshi  
Th 22249

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Supervisor : Dr. P. R. Mondal  
Th 22478

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021. SINGH (Piyoosh Kumar)  
**Molecular Anthropological Study of Depression and Suicide in IDU Mishmi Tribe of Arunachal Pradesh.**

Supervisor : Prof. V. R. Rao  
Th 22476

*Abstract*

Thesis work titled “Molecular Anthropological study of Depression and Suicide in Idu Mishmi Tribe of Arunachal Pradesh” performed in Tibeto-Burman Idu Mishmi tribe of Dibang valley and Lower Dibang Valley district of Arunachal Pradesh. The work was based on psycho-socio-genetic analysis of depression and suicide in an isolated tribal population of North East India. The field study was conducted from July 2011 to February 2014 that involved more than 200 days of field work. Study validate the events of suicide through deep analysis of depression and other associated psychiatric traits like other anxiety syndrome, panic disorder, eating disorder, hopelessness, alcoholism, aggression, impulsivity and lack of social support in high risk Idu Mishmi Tribe of Arunachal Pradesh. Study includes analysis of demographic (household composition, age, sex, surname, nata ayucha (Gotra), education, occupation, marital status, consanguinity, age at marriage, food habits etc.), socio-economic, psychological/psychiatric, anthropometric (height, weight, Biceps, Triceps, Suprailiac, Abdomen, sub scapular), Blood Group, Blood Glucose and genetic factors of Idu Mishmi. In psychosocial estimation well validated widely used tools were employed. Genetic analysis embraces already implicated candidate polymorphisms of depression and suicide behaviour, that includes genetic polymorphisms of plasticity gene (BDNF (Val66Met)), dopamine rate limiting gene (COMT(Val158Met)), Serotonin transporter gene ((5-HTTLPR), (5-HTT (STin2)), serotonin rate limiting gene (MAOA (uVNTR)). Suicide attempt, as assessed with Columbia Suicide Severity Rating Scale and face to face interview, was found in 18.38% individuals and with female preponderance. In categories of composite behavior, current suicide ideation (41.30%), and current suicide ideation with lifetime suicide attempt (32.61%) predominates in suicidality category. The key findings of the study implied that risk of suicide attempt increases significantly with the presence of psychosocial traits of depression, impulsivity, childhood trauma and lack of social support in genetically vulnerable person of 5-HTTLPR (SS) and MAOA (H) activity genotype.

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1. Introduction. 2. Rationale and objectives of the study. 3. Literature review. 4. Material and method. 5. Result. 6. Discussion. 7. Conclusion. References and annexures.

022. TALWAR (Seerat)  
**One Carbon Metabolism and Recurrent Miscarriages : A Genetic and Epigenetic Study From Delhi**  
Supervisors : Dr. K N Saraswathy and Prof. Manju Puri  
Th 22244

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023. THAKUR (Sunil Kumar)  
**Anthropogenetic Study Among High and Low Altitude Gaddis of Himachal Pradesh.**  
Supervisors : Prof. P. K. Ghosh and Dr. Benrithung Murry  
Th 22477

*Abstract*

Adaptation to variation depends upon the adaptive ability (Darwinian fitness) of organism which is either habitat specific (in the individual) or environment specific (in population). Human adaptations to high altitude involve the changes in physiology, morphology, anatomy and genetic to combating with the harsh environment of high altitude. It is evident that populations at high altitude have different mechanisms of adaptation to high altitude. Considering the effects and population specific responses (genetic, physiological, morphological) to high altitude, the present study was an attempt to understand high altitude adaptations/ maladaptations holistically comprising of morphology, physiology, biochemistry, genetics along with lifestyle variables among Gaddis (a tribal population of western Himalayas, Himachal Pradesh, northern India) Comparatively, high prevalence of smoking, alcoholism, stress, non-veg diet, less physical activity, TC, LDL, general and abdominal obesity was observed in high altitude than low altitude Gaddis. Hypertension, MetS and obstructive pattern of spirometry were also more prevalent among high altitude compare to low altitude Gaddis. For abnormal spirometry pattern at high altitude, reduction in high and low BMI and increase in FBS are adaptive and high TC is maladaptive. For metabolic syndrome, reduction in low BMI and TC alongwith MTHFR gene polymorphism were adaptive and high BMI, high TG, and high WC were maladaptive. Similarly, ACTN3 gene polymorphism was adaptive and increased BMI, abdominal obesity, TC and eNOS gene polymorphism were maladaptive for hypertension at high altitude. On the other hand, increased FBS was adaptive and increased WHR was maladaptive for pre-hypertension at high altitude. The final outcome of the present study showed holistic interaction of lifestyle variable, genetic markers and high altitude environment which shape-up the morphological and biochemical changes. These changes, whether adaptive or maladaptive, in turn shape up the different physiological processes (for the present study spirometry pattern, MetS and Blood pressure).

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024. TRINAYANI BORDOLOI

**Reproductive Performances and Complications : Association with Body Structure and Cardiovascular Functions Among the Women of Assam, North East India.**

Supervisor : Prof. A K Kapoor

Th 22246

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025. VERMA (Deepali)

**Insulin Resistance Among Adolescents of Delhi - Insight From Individual and Familial Risk Factors.**

Supervisor : Prof. Satwanti Kapoor

Th 22245

*Abstract*

The resistance to insulin-mediated glucose disposal has been recognized as prominent pathophysiological factor leading to diabetes mellitus and other cardio-metabolic disease. Adolescence period is marked by significant alteration in biological processes and engagement in behaviour disadvantageous to health. This emphasis on the need to look across an individual's or cohort's life experiences or across generations for clues to current patterns of health and disease, whilst recognising that both past and present experiences are shaped by the wider social, economic and cultural context. The present study was conducted to determine the risk factors associated with insulin resistance among adolescents (n=200) of Delhi which included- early life experience (birth weight), individual lifestyle pattern (physical activity, calorie intake and obesity), family environment (family history and cardio-metabolic profile) and genes (ACE and TCF7L2). Insulin resistance was estimated by homeostasis model (HOMA). Insulin resistance was found to be related with higher body fat among normal weight adolescents while central adiposity among overweight adolescents. A significant proportion of them were sedentary, however the potential effect of more time spent in sedentary activities on insulin resistance was apparent among overweight rather than normal weight adolescents. Insulin resistance was associated with higher cardiovascular risk among these adolescents. Familial clustering of cardio-metabolic risk predisposed adolescents to adiposity but no association was elucidated with respect to insulin resistance. ACE gene was found to be significantly associated with adiposity while TCF7L2 gene (rs7903146) with insulin resistance with gender difference in expression. Since the risk of insulin resistance among adolescent was observed at a lower degree of obesity, it emphasizes on revision of American Diabetes Association guidelines which recommend screening of obese children and adolescents for type 2 diabetes. The study signifies consideration of optimal period of time spent in physical activity along the spectrum of obesity.

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