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TO WHOM IT MAY CONCERN

This is to certify that the thesis entitled 'Cytogenetic Study of the Ethnic Population of North Bengal with special reference to Constitutive Heterochromatin Polymorphism' submitted for the award of the degree of Doctor of Philosophy of the University of North Bengal, is a record of research work carried out by Sri Rudra Narayan Agarwala, M.Sc. under my supervision. No part of this thesis has been submitted for any other degree or diploma. The help received from various sources has been duly acknowledged. There is nothing in his habits and character which may debar him from being admitted to the degree.

KB Datta 16.1.1984
(K.B.Datta)
Supervisor

Dedicated in memory

of my

Reverened 'Bhabhijee'

Late Urmila Devi Agarwala.

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Rudra N Agarwala
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PREFACE

Genetic polymorphism has been demonstrated in man for many characteristic features including blood groups, serum proteins, tissue enzymes and haemoglobins. The advent of chromosome banding techniques has opened a new horizon of cytogenetic research. The development and establishment of C-banding techniques have led to the discovery of class of chromosomal polymorphism involving constitutive heterochromatin which perhaps represents the most heteromorphic moiety of chromosome with great population and evolutionary stability. Moreover, the general correspondence between the loci of highly repititive DNA, constitutive heterochromatin and the C-banding on metaphase chromosomes alone warrants justification of studying C-band heteromorphism in man. Constitutive heterochromatin polymorphism involving chromosomes 1, 9 and 16 have been found to occur at a relatively high frequency in clinically normal population. Though the biological and clinical implications of C-heterochromatin polymorphism are poorly understood, yet its restriction to certain groups has been suggested. The incidence of C-band polymorphism varies in different populations and ethnic groups. Uptil now, the ethnic groups investigated represent selected sections of population inhabiting various countries. In view of the vastness of the Indian subcontinent as well as wide diversity of population, the lack of information about constitutive

heterochromatin polymorphism on Indian population is surprising.

The multitudeness of ethnic structure in North Bengal, an integral part of one of the north eastern states of India, provides unique opportunity for studying C-heterochromatin polymorphism. The objective of contemplating and understanding the present investigation has been manifold, one of which is to study and evaluate the variability of C-heterochromatic regions and to obtain an estimate of the frequency of C-band variants in the population of this area. The part I of the present dissertation has been devoted to a systematic study on the constitutive heterochromatin polymorphism in the normal population of Darjeeling - Himalayas, consisting of diverse ethnic groups and extending from the foothills to an altitude of 8500 ft and above. In view of repeated emphasis on the selective value of certain chromosomal polymorphisms in the adaptation of human populations to certain extreme environmental conditions, special emphasis has been given in the part 1 to a comparative analysis of C-band polymorphism in chromosomes 1, 9 and 16 of different racial groups for ascertaining selective value of chromosomal polymorphism in adaptation to varying environmental and geographical conditions.

Another facet of C-heterochrmatin polymorphism is the variability of C-band size categories. Studies on the possible occurrence of a differential distribution of C-band size categories in the different ethnic groups have been included in part 1. The reported variability on the incidence of C-band size categories in different populations have emphasized the need of a comparative study of the frequency of increased and decreased C-heterochromatin material in different ethnic populations. The part 1 considers as well the question of inter and intraracial differences in C-band variations in the ethnic groups each of which has been divided into several sub-groups mainly on the basis of anthropological origins.

In course of cytogenetic study of North Bengal population, some cases have been encountered with diverse inborn defects including various degrees of developmental and mental retardation. It is reduntant to emphasize the need for chromosomal studies of all such individuals with multiple congenital abnormalities and in particular the confirmation of all cases suspected of having a chromosomal aetiology from the clinical findings. The application of the newly discovered banding techniques have not only confirmed most of the previously establishmed syndromes but have also made possible precise identification of the

chromosomes involved in structural rearrangements. Establishment of the chromosomal basis of many clinical syndromes would not have been possible without the use of banding techniques. The part 11 of the thesis is concerned with the investigation on the possible association of chromosomal anomaly with the phenotypical abnormality in the subjects by application of Giemsa banding and fluorescent banding techniques.

Erythrocytic Glucose -6- Phosphate Dehydrogenase dificiency is one of the most prevalent and genetically informative hereditary defects of man. In view of suggested association of G-6-PD deficiency and distribution of malaria, as well as the hypothesis that the enzyme deficient individual offers resistance to malaria, it is worth investigating the prevalence of the deficiency in the different racial groups inhabiting the terrai and hill regions of North Bengal. The plains of North Bengal is a malaria endemic area and information is completely lacking on the incidence of G-6-PD deficiency among the population living in this part of India. The part 111 of the thesis deals with studies on the incidence of Glucose-6-Phosphate Dehydrogenase deficiency in the different ethnic groups of North Bengal population.