

PREFACE

Genetic polymorphism has been demonstrated in man for many characteristic features including blood groups, serum proteins, tissue enzyme and haemoglobin. The development and establishment of chromosome banding techniques have led to the discovery of a 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 repetitive DNA, constitutive heterochromatin and C-banding on metaphase chromosomes alone warrants justification of studying C-band heteromorphism in man. Polymorphism of human constitutive heterochromatin, especially in the secondary constriction of chromosomes 1, 9 and 16 has been found to occur at a relatively high frequency. Although such heteromorphisms were initially reported in some clinical material, later works have demonstrated that heteromorphisms occur with equal frequency even within phenotypically normal controls. Though the biological and clinical implications of human chromosome polymorphism are poorly understood, yet it has been suggested that

these heteromorphisms are restricted to certain groups. Investigations carried out here and elsewhere have demonstrated that the incidence of C-band polymorphism varies in different population and ethnic groups. Observations made in this laboratory have revealed that ethnic groups of Mongolian origin exhibit, in general, a relatively high frequency of C-band polymorphism involving chromosomes 1, 9 and 16.

The multitudeness of ethnic structure in North Bengal, an integral part of the North Eastern states of India, provides a unique opportunity for studying C-heterochromatin polymorphism. The objective of the present investigation has been manifold, one of which is to study intensively and to evaluate the variability of C-heterochromatin regions and to obtain an accurate estimate of the frequency of C-band variants in the Mongolian population of this area. The Part I of the present dissertation has been devoted to a systematic study on constitutive heterochromatin polymorphism in the Mongolian population of North Bengal and adjoining areas, constituting of diverse ethnic groups and extending from the foot hills to an altitude of 8500 feet and above. In view of repeated emphasis on the selective value of certain polymorphism in adaptation of human population to certain extreme environmental conditions, special emphasis has been contemplated for a

comparative analysis of C-band polymorphism in different racial groups of Mongolian origin for ascertaining selective value of chromosomal polymorphism in adaptation to varying environmental and geographical conditions.

Erythrocytic glucose-6-phosphate dehydrogenase is the key enzyme in the pentose-phosphate pathway of glucose metabolism in man. Deficiency of the erythrocytic enzyme G6PD, an inborn error of metabolism, is one of the most prevalent, genetically and clinically informative hereditary defects of man. In view of the suggested association of G6PD deficiency and distribution of malaria as well as the hypothesis that the enzyme deficient individuals offer resistance to falciparum malaria, it is worth investigating the prevalence of the deficiency in the different racial groups, inhabiting terai and hill regions of North Bengal. The plains of North Bengal is a malaria endemic area and information is almost lacking on the incidence of G6PD deficiency among the ethnic groups living in this part of India. The Part II of the thesis is concerned with the investigations on the incidence of G6PD deficiency in the different ethnic groups of Mongolian origin inhabiting North Bengal and adjoining areas.

Human G6PD shows great genetic heterogeneity. About 300 genetically determined variants of G6PD molecule have been described from all over the world. The frequency of this variant among various populations differs according to race and geography. These variants differ from normal and from

each other in one or more physical and kinetic properties. No attempts have yet been made to characterize G6PD variants in the ethnic populations of North Bengal. It is of great value to examine the characteristic of the enzyme for many purposes. A systematic study on the incidence of G6PD deficiency and characterization of G6PD variants in the ethnic population of North Bengal in general and high altitude dwellers in particular would be useful not only for clinical and genetical reasons but also for tracing relatedness and origin of different ethnic groups. The Part II of the dissertation also covers the investigation on the red cell enzyme variant in 3 Mongolian racial groups following starch gel-electrophoresis.