5 SUMMARY

Over 800 individuals from three different ethnic population of Mongolian origin inhabiting North Bengal and adjoining areas were studied for G6PD deficiency. The incidence of erythrocytic G6PD deficiency varied from 2.91-8.12%. A relatively high frequency of the enzyme deficiency (8.12%) was found to be associated with the Rajbangsis living in the plains followed by the Kirati Complex residing in both plains and hills. The Hill Tribes inhabiting mostly the hills revealed a comparatively low frequency of enzyme deficiency (2.91%). The enzyme inactivity occurred at a comparatively high rate in the malaria free groups than in the malaria infected populations. The present observation not only strengthens the concept that G6PD deficiency owes its distribution to malaria, but also substantiates that the enzyme deficiency in human erythrocytes might confer resistence against malaria.

The prevalence of electrophoretic variants of the enzyme G6PD has also been investigated in the nondeficient (through qualitative test) subjects belonging to the Mongolian origin groups. The observation reveals a slow moving variant in the frequency of 0.62%. The Rajbangsis living in the plains depicted a relatively high frequency of the enzyme variant (1.10%). The Kirati Complex comprising of Rai, Limbu, Magar, Tamang and Gurung revealed 0.76%, while no variant could be detected in the Hill Tribes comprising of Bhutia, Lepcha and Sherpa. Enzymological characteristics in the nondeficient subjects reveals very mild red cell enzyme deficiency (67-77% of the normal B^+type). The electrophoretic mobility in phosphate and TEB buffers at pH 7.0 and 8.6 was noted as 90% and 80% respectively to that of normal B^+type. The variant enzyme shows an elevated rate of substrate analogues utilization and a normal thermostability. Similarly, the enzyme activity at different pH levels depicts a normal truncated type of pH optimum curve. Women representing a heterozygous condition with B^+type of enzyme and new slow moving type have also been observed in family study of the propositus. Enzymologic characteristics of the class 4 type (WHO Scientific Group 1967) and slow moving Indian variants did not show similarities with the present variant. From these observations it was considered to be a new variant and was designated as G6PD 'North Bengal.' The individuals

with the variant type did not have any history of drug-induced haemolytic anaemia and therefore did not suggest association with any clinical condition. The enzymologic characteristics of G6PD from the deficient subjects of Mongolian origin showed a low erythrocytic enzyme activity and were without haemolytic anaemia. Besides, the enzyme variant depicted an identical electrophoretic mobility, increased utilization rate of substrate analogues, low thermostability and a biphasic pH optima curve when compared to B^+ type. The composite picture of the characterization studies on G6PD from deficient subjects suggest identity with class 2 (WHO Scientific Group 1967) Mediterranean type of G6PD variant.