Chapter VII: SUMMARY AND CONCLUSION

Haemoglobinopathy has been described as a condition in which the production of normal adult haemoglobin (Hb A) is partly or wholly suppressed and is partly or wholly replaced by one or more haemoglobin variants which may include foetal type haemoglobin (Hb F).

Thalassaemia, either in homozygous form or in combination with Hb E appears to be the commonest form of haemoglobinopathy in Bengal.

In the present investigation, altogether 58 samples of blood had been studied which included samples from normal subjects, thalassaemia major, Hb E-thalassaemia, thalassaemia minor, nutritional anaemias and anaemias associated with primary tuberculosis, ascariasis and nephrotic syndrome. Five samples of cord blood from full term infants and three samples of haemoglobin solutions containing Hbs S, SC and CC were also included in the list.

Diagnosis of various cases of anaemias studied were done by routine clinical and pathological examinations. Diagnosis of haemoglobinopathies were confirmed on paper electrophoresis and by alkali denaturation test, apart from other routine clinical, pathological, biochemical and radiological examinations.

The patients suffering from haemoglobinopathies which included thalassaemia major and Hb E-thalassaemia were mostly children and presented
moderate to severe anaemia, enlargement of liver and spleen, and in some cases mongoloid facies. Peripheral blood showed hypochromic microcytic anaemia and presence of variable amounts of normoblasts. Radiological examination revealed generalised osteoporosis of the bones and in two cases typical sun-ray appearance of the skull bone were observed. Alkali denaturation test showed presence of variable percentage of foetal haemoglobin in all these cases and on paper electrophoresis presence of abnormal haemoglobins were detected.

The diagnosis of thalassaemia minor was confirmed by the presence of higher percentage of Hb A2 on paper electrophoresis.

Oxygen capacity was determined in all the cases of the present series, in samples of whole blood and their haemoglobin solution.

The majority of the samples of whole blood showed a reduction in the oxygen capacity than those of normal subjects in this series. The reduction of the capacity was maximum in cord blood. Amongst the cases of anaemias and haemoglobinopathies studied the maximum reduction of the capacity was registered by Hb E-thalassaemia. No relationship could be found out between the degree of reduction of oxygen combining capacity and the clinical features, and the contents of haemoglobin and foetal haemoglobin.

In haemoglobin solutions of all samples the oxygen capacity was increased. The value was higher than that of oxygen capacity of whole blood in normal subjects of the present series. The increase in the capacity in haemoglobin solution was maximum in case of thalassaemia major. Here also no relationship was observed between the degree of increase of oxygen capacity and the clinical features and the contents of haemoglobin and foetal haemoglobin.
The content of total lipids in haemoglobin solutions was found to be higher in samples of thalassaemia major and Hb E-thalassaemia.

To study the factors causing the inhibition of uptake of oxygen by haemoglobin in whole blood samples, attempts were made to assess the structural condition of red cell membrane or stroma of normal subjects and of patients suffering from thalassaemia major and Hb E-thalassaemia.

Estimation of total lipids of stroma revealed that the stroma of haemoglobinopathies contained greater amount of total lipids than that in normal subjects.

Estimation of hexosamine and nitrogen of the stroma which gave certain informations about the contents of mucopolysaccharides and protein respectively, revealed no significant change in these constituents.

The results of the present investigations also revealed that the red blood cells consisting wholly of abnormal haemoglobins (in the present series Hbs E & F) showed maximum reduction of the oxygen capacity in whole blood samples. In haemoglobin solutions, however, there was increase in the oxygen capacity, but the degree of increase in the oxygen capacity was less than that in haemoglobin solutions of samples from normal subjects, from thalassaemia major (consisting of Hbs A+F) and from other anaemias. The lipid content of the solutions of abnormal haemoglobins was also found to be higher than that in normal subjects. The difference in the increase of oxygen capacity in haemoglobin solutions may be partly attributed to the peculiar behaviour of a particular abnormal haemoglobin for the acceptance of oxygen and partly to the lipid content of the solution.

The high content of red cell membranes in haemoglobinopathies might bring about some sort of physiochemical change so as to inhibit the passage of oxygen into the cell to combine with haemoglobin.