CHAPTER 5

Summary and Conclusion
5.1 Review of Hemoglobinopathies and thalassemias

Thomas Cooley was the first to observe thalassemia in red blood cell in human being in 1925. Basak and Ozcelik in 1963 was able to demonstrate in imbalance in production of globin chains and the production of an inadequate number of red cells. Aulehla and Scholz was depict that the defect may affect the a, b, g or d chain, or may affect some combination of the b, g, and d chain. In 1978 Deidda showed that abnormal globin chains can produce adverse effects on the red cell and lead to destruction of the red cell in the marrow that is he called ineffective erythropoiesis. Oner in 1982 proved that some but not all, hemoglobinopathies and thalassemias are hemolytic anemias. These nosologic concepts are summarized by the Venn diagram below.

5.2 Review of geographical Distribution of Hemoglobin Disorders including Thalassemia

Thalassemia is a global problem. The two major form of thalassemias i.e. a and b-thalassemia are the most common inherited single-gene disorders in the world with the highest prevalence in areas where malaria was or still is endemic. The burden of this disorder in many regions is of such a magnitude that it represents a major public health concern. In some endemic countries in the Mediterranean region,
longestablished control programs have achieved 80-100% prevention of newly affected births. In Iran, it is estimated that about 8,000 pregnancies are at risk each year. Thalassemias foci of origination and prevalence are the Mediterranean, Asia, and Africa, spreading through Europe, the Americas and Australia due to population migration. In the eastern Mediterranean region, the prevalence of heterozygous b-thalassemia varies from about 3.5 – 4.5 % in the Gaza strip and Jordan to 5.5 % in Pakistan.

Thalassemia disorder is found almost all Indian states. The highest frequency of b-thalassemia trait was found in Gujarat, followed by Punjab, Tamil Nadu, South India and its prevalence rate was found to be highest in Central India i.e. 2.4 % to 4.4 %. (Average frequency). The frequency of b-thalassemia was found in Maharashtra were near about 1.9 %. The frequency of b-thalassemia trait in Western Maharashtra reported as 7.0 % is in between that of Punjab and Tamil Nadu. In Amravati region, out of total sample examined showed the prevalence of thalassemia trait is near about 0.85%

5.3 Consanguineous

The high rate of consanguinity increases the inbreeding coefficient, and the frequency of genetic disorders in populations. This affects health, psychological state and economic status of many unfortunate families, and it has implications on the whole population. I thought level of education was a key causative element. But to my disappointment, it is not. Clearly, The roots of this problem go deeper. And regardless of the reasons behind it, there has to be an effort at least in the medical field and population genetics to evaluate the situation. There is a lack of research and information concerning the problem. I wasn’t able to find the frequencies of...
endemic genetic disease in the area like thalassemia. Besides, genetic screening has to become both available and mandatory for affected families. Finally, premarital genetic counseling could be of great importance in helping young couples understand the high risk associated with consanguineous marriages. Some thalassemia trait from present study also showing consanguinity.

5.4 Haematological analysis

5.4.1 NESTROFT

The NESTROFT test is very cheap, cost effective and easy to perform. The stock solution once made can be kept well in a stoppered bottle. At one time one can perform 40-50 tests in a hour. Thus, NESTROFT seems to be valuable as a screening test in our country with low socio-economic status. A practical approach would be to perform NESTROFT in high-risk community for detection of heterozygotes of β-thalassemia.

5.4.2 Peripheral Blood Smear Examination

Peripheral blood smear examination showed different size and shape of R.B.Cs including schistocyte, acanthocyte, spur cells, tear drop cell and burr cells. Microcytosis also depict. Red blood cell count showed lower values in 57% of patients studied. The haemoglobin level was found to be below normal. The haematocrit value was also found to be below normal (i.e. below 35 %). However the white blood cell count was consistently elevated in the population afflicted with thalassemia. The foetal haemoglobin percentage was found to be quite high in majority of the patients studied (mean = 5.6 %). The higher average level of foetal haemoglobin could contribute to the generally less severe thalassemic disease in population studied.
5.5 Biochemical analysis:

The bilirubin levels were found to be quite high indicating that the population under study might be suffering from haemolytic blood disease. The patients also suffered from intra-and extrahepatic biliary tract obstruction, intravascular and extravascular hemolysis, physiologic neonatal jaundice.

The serum urea and creatinine levels were found to be in the normal range (22.04 mg/dl) thus indicating that renal failure was a rare occurrence in population studied. The serum alkaline phosphatase activity showed high values (136.25 IU/L) This may be due to congestion or obstruction of the biliary tract, which may occur within liver, the ducts leading from the liver to the gallbladder, or the duct leading from the gallbladder through the pancreas that empty into the duodenum. More precisely due to hepatobiliary disorder seen in thalassemic patients.

5.6 Ethnobiology:

The percentage of ABO blood groups found in the thalassemic diseased population under study were A= 8.3 %, B= 51.3 %, O=22.4 %, AB= 2.5 %, Rh +ve = 95.3 %, Rh –ve = 4.7 %. These blood group percentages match with the blood group percentages exhibited by the normal population.

Thalassemia disease was found to be more common in males than in females, the male : female ratio being 71:29 in the population studied.

Positive history of consanguinity also seen in 12 patients affected with thalassemia. 5 New married couple with thalassemia minor condition was also detected. So in-breeding depression still working on population of Amravati.

Sindhi population was found to be mostly affected population in Amravati region. This population was also under the category of high risk population because
the frequency of thalassemic trait was found to be near about 0.85%. Besides this, the Sindhi community some other community like Navboudhas, Hindus and Muslims also showing the frequency of b-thalassemia was near about 0.67%.

71% patients from pediatric group needed blood transfusion early in their life. On an average every patients suffering with b-thalassemia needed blood transfusion of 8 times in a year.

Inheritance pattern of thalassemia showed Mendelian pattern of inheritance in present study. There is an urgent need for making the people aware of this lethal malady. Health education is an important component of the preventive genetic programmes. With improving environmental and socio-economic conditions, better public health care and medical facilities, effective malarial prophylaxis and better nutrition, children suffering from thalassaemia and haemoglobinopathy can be better managed and rehabilitated in India. This requires proper health education and adequate sensitization to the individual, family or community to accept these preventive remedial measures. Thus, thalassaemia and haemoglobinopathy, which are prevalent throughout India are heritable, treatable, curable and preventable disorders. We must make efforts to eliminate these from our society. In this way, we may be able to alleviate and ameliorate the sufferings of the affected masses in our country. High cost of treatment, repeated blood transfusion and chelation therapy, and economic burden on family resources, all suggest that prevention is better than cure. Thus a joint venture of antenatal and inductive screening seems to be the most fruitful strategy for haemoglobinopathy in India.