Beta thalassemia is the most common single gene disorder in Pakistan with a gene frequency of 5-8% and about 8-10 million carriers in the country.1 It has become a worldwide clinical problem due to increasing immigration of ethnic groups with high prevalence of thalassemia.2,3 Over the past three decades, regular blood transfusions and iron chelation has dramatically improved the quality of life and transformed thalassemia from a rapidly fatal disease in early childhood to a chronic disease compatible with prolonged life.1,4 Today life expectancy varies between 25-55 years, depending on the compliance with medical treatment.4 Despite increased life expectancy, complications keep arising. These relate to inadequate transfusions, transfusion-related infections, allosensitization, iron-overload related cardiac, endocrine and liver disturbances and toxicities of iron chelators.4,5 Many of these problems are strongly age dependent.5

Heart disease is the most important complication and the main determinant of survival. It is responsible for more than half of the deaths.6,7 It may take the form of cardiomyopathy, pulmonary hypertension, heart failure, arrhythmias, pericarditis and myocarditis.6,7,8,9 Although iron overload is the main cause but other factors; genetic, immune or infective, may also be important.10

Endocrine complications include diabetes mellitus, hypothyroidism, hypoparathyroidism, hypogonadism and delayed puberty.11,12 These complications have contributed little to morbidity and mortality in the past; however as a result of increased longevity, these have become more common and contribute significantly to the morbidity in these patients.13 Endocrine complications along with osteoporosis, trace elements deficiency and other metabolic disturbances also lead to growth failure and short stature.11,14

Less significant complications include hepatic involvement,15,16 neurological complications,17 and psychological manifestations.18

In short, beta thalassemia has a spectrum of varied manifestations and complications. The importance of thalassemia in Pakistan is masked by infections and nutritional deficiencies.19,20 We need to develop preventive strategies like pre-marital screening, genetic counseling and ante-natal diagnosis. A part from early diagnosis, there is a need to increase the therapeutic facilities like blood transfusions, chelation therapy and bone marrow transplant.

It is really a great challenge and needs an organized plan for action.

REFERENCES


