EDITORIAL

THALASSEMIA: STILL A CHALLENGE

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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 thalassaemia 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

1. Satwani H, Raza J, Alam M, Kidwai A. Endocrinal complications in thalassemias: Frequency and association with ferritin levels. Pak Ped J 2005; 29: 113-19.
2. Hahalis G, Alexopoulos D, Kremastinos DT, Zoumbos NC. Heart failure in beta thalassemia syndromes: a decade of progress. Am J Med 2005; 118: 957-67.
3. Pearson HA, Cohen AR, Giardina PJ, Kazazian HH. The changing profile of homozygous beta thalassemia: demography, ethnicity, and age distribution of current North American patients and changes in two decades. Pediatrics 1996; 97: 352-6.
4. Agarwal MB. Advances in management of thalassemia (Editorial). Indian Pediatrics 2004; 41: 989-92.
5. Cunningham MJ, Macklin EA, Neufeld EJ, Cohen AR. Complications of beta thalassemia major in North America. Blood 2004; 104: 34-39.
6. Aessopos A, Farmrkis D, Deftereos S, Tsironi M, Tassiopoulos S, Moyssakis I, et al. Thalassemia heart disease, a comparative evaluation of thalassemia major and thalassemia intermedia. Chest 2005; 127: 1523-30.
7. Borgne-Pignatti C, Cappellini MD, De Stefano P, Del Vecchio GC, Forni GL, Gamberini MR, et al. Survival and complications in thalassemia. Ann N Y Acad Sci 2005; 1054: 40-47.
8. Olivieri NF, Nathan DG, MacMillan JC, Wayne AS, Liu PP, McGee A, et al. Survival in medically treated patients with homozygous beta thalassemia. New Eng J Med 1994; 331: 574-78.
9. Chern JP, Su S, Lin KH, Chang SH, Lu MY, Jou ST, et al. Survival, mortality and complications in patients with beta thalassemia major in northern Taiwan. Pediatr Blood Cancer 2006; 45: 53-56.

10. Kremastinos DT, Flevari P, Spyropoulou M, Vrettou H, Tsiapras D, Stavropoulos-Giokos CG. Association of heart failure in homozygous beta thalassemia with the major histocompatibility complex. Circulation 1999; 100: 2074-78.

11. Shamshirsaz AA, Bekheirnia MR, Kamgar M, Pourzahedgilani N, Bouzari N, Habibzadeh M, et al. Metabolic and endocrinologic complications in beta thalassemia major: a multicenter study in Tehran. BMC Endocrine Disorders 2003; 3: 23-34.

12. De Sanctis V, Vullo C, Katz M, Wonke B, Tanas R, Bangi B. Gonadal function in patients with beta thalassemia major. J Clin Pathol 1988; 41: 133-37.

13. Mohammadian S, Bazrafshan HR, Sadeghi-Nejad A. Endocrine gland abnormalities in thalassemia major: a brief review. Pediatr Endocrinol Metab 2003; 16: 957-64.

14. Low Louis CK. Growth of children with beta thalassemia major. Indian J Pediatr 2005; 72: 159-64.

15. Perifanis V, Tziomalos K, Tsatra I, Karyda S, Patsiaoura K, Athanassiou-Metaxa M. Prevalence and severity of liver disease in patients with beta thalassemia major. A single-institution fifteen year experience (letter). Haematologica 2005; 90: 1136-38.

16. Prati D, Maggioni M, Milani S, Cerino M, Cianciulli P, Coggi G et al. Clinical and histological characterization of liver disease in patients with transfusion-dependent beta thalassemia. A multicenter study of 117 cases. Haematologica 2004; 89: 1179-86.

17. Zafeiriou DI, Economou M, Athanasiou-Metaxa M. Neurological complications in beta thalassemia. Brain Dev 2006; 28: 477-81.

18. Moorjani JD, Chithira I. Neurotic manifestations in adolescents with thalassemia major. Indian J Pediatr 2006; 73: 603-7.

19. Rahman M, Lodhi Y. Prospects and future of conservative management of beta thalassemia major in a developing country. Pak J Med Sci 2004; 20: 105-12.

20. Imran M. Hereditary anemias in North West Frontier Province of Pakistan. J Postgrad Med Inst Peshawar 1986; 1: 104-6.