EDITORIAL

THALASSEMIA MAJOR – A MAJOR CHALLENGE IN PAKISTAN

β-Thalassemia major is an autosomal recessive haemoglobinopathy being one of the most frequent genetic disorders worldwide[1]. In Pakistan, there was very little awareness about the occurrence of this disease till late 60`s and early 70`s when first reports of this condition started appearing in the literature. In fact, the initial reports of patients being diagnosed as Thalassemia major came from AFIP and other cities in Pakistan mainly Karachi[2,3]. However, as the awareness about the problem increased among pediatricians and hematologists, the diagnostic services improved and an increasing number of children were recognized to be suffering from β-thalassemia major. Now we know, it as one of the major genetic problems affecting our children since Pakistan is in the thalassemia belt on the world map[4]. Annually around 5000-9000 babies are born with β-thalassemia major, the gene frequency being 5-7% and there are an estimated 9.8 million carriers in the country[5]. The World Health Organization (WHO) has designated control of haemoglobinopathy particularly β-thalassemia major in the developing countries as one of the major priorities. In Pakistan, this is a major pediatric health challenge as these children are dependent upon regular blood transfusion and chelation for survival. The estimated requirement for treating a birth cohort for one year is 90,000 units of blood and desferrioxamine; an iron chelator worth 22 million dollars[6]. This entails considerable burden upon our transfusion services and pediatric care. Patients from the lower socio-economic strata consequently suffer from early complications of the disease leading to mortality. Those who survive, continue to be a heavy burden upon the health services as well as remaining chronically dependent upon the family for long term care. The introduction of prenatal diagnosis and genetic counseling for these families has opened avenues for planning, prevention of births with thalassemia major .

The introduction of bone transplantation as a mode of treatment through opening of new bone marrow transplantation (BMT) centers in Pakistan provided distressed families a light at the end of the tunnel despite considerable cost of such treatment. However in Pakistan the key to control of β-thalassemia major remains developing a comprehensive program for prevention. Keeping in view the experience and success of such programs in Cyprus, Sardinia and Iran; screening of the targeted population, genetic counseling, prenatal diagnosis and the option of terminating pregnancies with affected fetuses remain the main strategies for prevention[7]. The option of pre marital counseling regarding forbidding marriage between two carriers of the gene for thalassemia remains another option. Currently there are around 217 mutations for β-thalassemia major, out of which about 20 mutations account for 90% of the β-globin gene in the world and the frequency of unique mutations for each population and ethnic group remains distinct for that population[8]. Pakistan being a resource constrained country cannot afford to persist with bearing the exorbitant cost of curative and maintenance services for the increasing population of children with thalassemia major. The viable long term answer remains to adopt measures for preventing the birth of thalassemia major patients through prenatal diagnosis[9], genetic counseling[10] and educating the population about the importance of planning marriages of young adults after screening for β-thalassemia gene[11] and forbidding the marriages of carriers through creating awareness among the population and proper legislation in due course of time[12].

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