Conference Abstract

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Management of Beta Thalassaemia Major in Pakistan: Past, Present, and Future

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Past Treatments: In Pakistan, beta thalassaemia major, a severe inherited blood disorder, has long posed a significant public health challenge due to its high prevalence. As there is a high rate of family marriages in the region. Historically, the management of beta thalassaemia major was limited to supportive care. Blood transfusions, introduced in the mid-20th century, became the primary means of treatment. Patients with beta thalassaemia major require regular transfusions to maintain adequate haemoglobin levels, allowing for normal growth and development. However, the repeated transfusions led to iron overload, which could damage organs such as the heart, liver, and endocrine glands.

Iron chelation therapy was introduced in the 1970s to address the complications of iron overload. The use of deferoxamine, an iron-chelating agent, became a standard treatment. Though effective, the treatment was burdensome, requiring daily injections and being prohibitively expensive for many families in Pakistan. Access to advanced medical care and therapies was limited, especially in rural areas, where 70% of the population resides, resulting in suboptimal outcomes for majority of patients.

Present Treatments: Over time, there has been significant progress in the treatment and management of beta thalassaemia major in Pakistan. Due to the more widespread role played by the NGOs in providing treatment and awareness. The use of Hb F inducing medicines is also being used successfully in very few centres. Today blood transfusions and iron chelation therapy remain the backbone of treatment, as newer, oral iron chelators, such as deferiprone and deferasirox, are available. These oral medications have improved patients' quality of life by offering more convenient alternatives to deferoxamine's daily infusions. However, due to high cost this treatment is not affordable by the majority of the patients. Thus, iron over-load is still a very major issue leading to high morbidity and mortality.

Awareness about thalassaemia has grown, and screening programs, often spearheaded by NGOs and govermental health advocacy groups, have been implemented in some regions of the country. These programs focus on targeted family screening and also, premarital screening to reduce the incidence of the disease. However, widespread awareness campaigns and premarital screening are still lacking, leading to ongoing high birth rates of children with beta thalassaemia. In more advanced centers, hematopoietic stem cell transplantation (HSCT), or bone marrow transplantation, offers the only curative treatment. However, the procedure is costly for the patients, and finding a compatible donor remains a significant challenge in Pakistan. Despite these challenges, some centers in larger cities such as Karachi, Lahore, and Islamabad offer the procedure, although access is limited due to financial constraints and the availability of suitable healthcare infrastructure.

Future Treatments: Looking ahead, the future treatment of beta thalassaemia major in Pakistan is promising, particularly with the emerging knowledge of newer and more cost-effective Hb F inducing medicines. The global advancement and development of new pharmacological treatments approved by FDA and gene therapies, including gene editing technologies, such as CRISPR, are being explored and could offer a definitive cure for beta thalassaemia by correcting the genetic mutations responsible for the disease. But due to very high cost, the patients in Pakistan and other developing countries will not have access to these for quite some time. The most promising future management of thalassaemia patients in our region is to explore the knowledge and establish a national guideline, which should also include safe and cost-effective use of Pharmacological haemoglobin-F

inducing medicines. This would eliminate the need for lifelong transfusions and chelation therapy in most of the patients. Several successful clinical studies from China India and Pakistan are published, showing good clinical response to Thalidomide alone and with Hydroxyurea combined. This has resulted in marked improvement in the quality of lives of Beta Thalassaemia major patients. This will also lead to more availability of the donated blood for saving lives of the other patients. Thus, improving the overall Health care system in Pakistan. To further reduce the incidence of thalassaemia, there should be a growing push for the implementation of nationwide mandatory premarital screening programs, along with targeted family screening. If widely adopted, such programs could help significantly reduce the number of children born with the condition.

Conclusion: While significant strides have been made in the management of beta thalassaemia major in Pakistan, challenges such as limited access to advanced treatments and high treatment costs persist. Future innovations in gene therapy, pharmacological advances, and nationwide screening programs offer hope for a brighter future for individuals living with this condition. The continued effort to improve healthcare infrastructure and expand access to these therapies will be essential for managing the disease effectively in the coming years.

Disclosure

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