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PRESIDENT'S PAGE

Thalassemia: Looking Ahead

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The 8th of May every year marks the International Thalassemia Day. As we embrace this year's theme: "Together for Thalassemia: Uniting Communities, Prioritising Patients," it serves as a poignant reminder to combat this genetic blood disorder with involvement and support of all stakeholders including treating pediatricians and physicians, patients and their families, communities, governmental and non-governmental organizations (NGOs).

With an age-standardized prevalence rate (ASPR) and age-standardized incidence rate (ASIR) in 2021 of 18.28 (95% UI 15.29–22.02) and 1.93 (95% UI 1.51–2.49) per 100,000 persons, respectively, thalassemia continues to account for a significant disease burden globally [1]. Although the global ASPR and ASMR have declined significantly since 1990, there has been an increase in the prevalence of thalassemia among the elderly. While this offers hope of increasing longevity in these patients, it also warns against the significant health challenges that need to be addressed as the disease progresses due to evolving complications. While younger children with transfusion-dependent thalassemia (TDT) typically present with symptoms due to anemia within the first two years of life and need regular blood transfusions, the adolescent and young thalassemia cohorts encounter several complications. These primarily include complications due to systemic iron overload such as poor growth, cardiac and hepatic dysfunction, and endocrinal disturbances like hypothyroidism, hypoparathyroidism, hypogonadism and osteopenia/osteoporosis; complications due to extramedullary hematopoiesis like splenomegaly and facial changes; and transfusion transmitted infections like hepatitis B and C.

Looking at the scenario in India, thalassemia continues to be a major public health concern with an estimated burden of 100,000 patients, although in the absence of National Registries, the exact figures are not available [2]. Over the last two decades, India has made a significant progress in the management of thalassemia. Regular transfusions and iron chelation using oral medications like deferasirox or deferiprone or parenteral deferoxamine are available at several thalassemia centers across the country, albeit the availability of NAAT (nucleic acid amplification test)-tested packed red cells and leucodepleted blood are marred by significant regional disparities. The National Health Mission has sponsored thalassemia management, including prevention and control activities in several states, fostering better patient outcomes. In 2017, Coal India Corporate Social Responsibility (CSR)-funded Hematopoietic Stem Cell Transplantation (HSCT) program was launched in a few apex centers in India to provide a onetime cure opportunity for children with hemoglobinopathies like thalassemia and sickle cell disease who have a matched family donor. This initiative was targeted to provide financial assistance to patients by providing a package cost up to Rs. 10 lakhs per HSCT. Since then, the access to HSCT has increased in India with several centers now offering promising results. Luspatercept was granted approval for treating beta-thalassemia by the Central Drugs Standard and Control Organization in December 2022 and it has shown good results in decreasing the transfusion frequency. Although the US Food and Drug Administration (FDA) granted approval to gene therapy for beta-thalassemia in 2023, the huge financial implications continue to keep this cure out of reach for the majority [3].

Considering our country's typical situation with marked regional disparities in the availability of thalassemia care services, affordable healthcare models for thalassemia management are the way forward. Spreading awareness for compulsory prenatal and carrier screening and facilities for genetic counseling, along with access to cost-effective screening and diagnostic tests, can strengthen the prevention of thalassemia. Efforts are needed to implement newborn screening for thalassemia across the country. Researchers need to focus on optimizing the existing therapies, while simultaneously improving HSCT techniques and exploring genetic modifications.

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Meanwhile, government policies including benefits offered under the Right to Persons with Disabilities Act (2016) continue to offer hope for a better quality of life to these patients with opportunities for better education and jobs and some financial assistance. With increasing life expectancy, developing uniform "Transition of Care Guidelines" will ensure continued care to these aging patients [4].

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