URL : https://news.amdi.usm.my/fullnews.php?id=eHJ3LzE5WTRvczhHLzNyZzV1WEl2QT09

AMDI Magazine

e-issn 2735-041X, Volume 5, Issue 2 (2024)

Expert Column

Thalassemia through time: The cure is here, prevention remains key

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PUBLISHED : 02 DECEMBER 2024

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Early Days but Still Relevant: Blood Transfusions and Iron Chelation Therapy

Thalassemia is the most common inherited blood disorder and is an important public health problem in Malaysia. Infants with thalassemia major experience health problems around 3 to 6 months of age due to severe anaemia, poor weight gain and enlargement of abdominal organs such as the liver and spleen. The treatment of thalassemia in the early days but still relevant today is regular blood transfusions and iron

chelation therapy. Chelation therapy prevents iron overload, a common side effect of frequent transfusions, while blood transfusions help control anaemia. Access to effective iron chelation therapy has significantly increased life expectancy, with some patients living into their 60s. This is a significant improvement, as previously, thalassaemia patients would often die in their teens due to heart failure.

Screening and Prevention

In 2004, Malaysia initiated a national Thalassaemia Prevention and Control Programme to decrease the incidence of severe thalassaemia and its associated complications. This programme involved mass public education campaigns, public awareness, and health education[1]. Concerned by the alarming statistics of one carrier in every 20 Malaysians, the Ministry of Health Malaysia initiated screening programs in 2008 with the goal of reducing the number of babies born with thalassemia[2]. The screening initiatives at the start targeted mainly the family members of thalassemia major patients (cascade screening) and antenatal women. The screening initiatives also included the option to screen before marriage. In 2016, Malaysia strengthened the screening program further by implementing a national policy on school-based thalassaemia screening programs, specifically targeting upper secondary adolescents aged 16 years old. These continuous efforts have overall seen a declining trend in affected births from 2015 onwards[3].

Introduction of Bone Marrow Transplantation: Curative

A bone marrow transplant (BMT) is a procedure that infuses healthy, blood-forming stem cells into a patient's bone marrow to replace one that is not producing enough healthy blood cells. BMT to date is considered the only well-established curative treatment for thalassemia. While it can potentially cure thalassemia, transplant-related complications such as severe infection, graft rejection, graft-versus-host disease and death could occur. This procedure is also not suitable for thalassemia patients who have had complications like iron overload. As of 2015, around 13 hospitals in Malaysia offer this procedure4. However, it is not widely accessible due to high costs, limited funding, and difficulty in finding fully matched stem cell donors. Many patients struggle to find a compatible sibling donor, and the availability of matched unrelated donors is scarce. The establishment of the Malaysian Stem Cell Registry in 2000 has aimed to increase the pool of available donors, but as of 2020, the number of registered donors remains relatively small[4]. We need to make more efforts to raise awareness and motivate volunteer donors.

Advances in Haploidentical Transplants and Gene Therapy

With the advancement of technologies, increased knowledge and experience in transplants, and the recognition of the challenges in finding fully-matched donors, the use of partially-matched donors (haplo-identical related donors) has increased. It also has the advantages of lower costs and outcomes have also greatly improved over the years with better steps taken at preventing graft-versus-host disease which is a

complication that develops when the donor stem cells start to become a threat and attack the recipient's body. All patients can now find a donor thanks to this alternative stem cell source, which expands the donor pool beyond siblings. This approach has significantly increased access to curative treatment for thalassemia in Malaysia. On the other hand, a lack of a proper regulatory framework has prevented local adoption of gene therapy, which focuses on correcting the genetic mutation responsible for thalassaemia by adding a functional gene to defective blood stem cells.

Conclusion

Treatment for thalassemia in Malaysia has advanced significantly, moving from basic transfusions and chelation therapy to advanced curative interventions such as bone marrow transplants. Despite these advancements, the most effective approaches still involve prevention through screening programs to identify carriers in the community and raising awareness and knowledge about the genetic inheritance of thalassaemia. It will take a long, continuous effort to further reduce the incidence of thalassaemia, while at the same time, the ever-growing medical innovations will make more treatment options available to those affected by the disease in the near future.

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