

**INBORN ERRORS OF METABOLISM - II****THE ROLE OF HAEMATPOIETIC STEM CELL TRANSPLANTATION IN INBORN ERRORS OF METABOLISM****\*Revathi Raj****\*\*Ramya Uppuluri****\*\*Kavitha Ganesan**

**Abstract :** *Advances in the field of transplantation have resulted in dramatic cure for children with inborn errors of metabolism over the last three decades. Early diagnosis and optimal patient selection are the key to successful outcomes. The best outcomes have been seen in children with mucopolysaccharidoses who have been transplanted before the age of two years to prevent neurocognitive deficits. The counselling of these families involves many challenges including organ dysfunction during the procedure, the long time required to perceive improvement in the somatic features and the need for holistic follow up with cardiac, orthopaedic, neurology and ophthalmology colleagues. We present here, the data available with current studies on patient referral, timing of referral and the unique features in the care of these children.*

**Keywords:** *Hematopoietic stem cell transplantation, Inborn errors of metabolism, Lysosomal storage disorders, Gene therapy.*

**Points to Remember**

- *Hematopoietic stem cell transplantation (HSCT) for metabolic diseases involves inputs from a multidisciplinary team before, during and long after HSCT.*
- *The timing of HSCT must precede the onset of disease progression; HSCT halts progression but does not reverse existing disease related damages.*
- *HSCT can be performed using a matched sibling donor, a matched unrelated donor or a haploidentical parent donor.*
- *Myeloablative conditioning using treosulfan or targeted busulfan prevents rejection of the graft.*
- *Gene therapy using autologous stem cells offers the potential for better outcomes and lower toxicity in this unique group of children.*

**Acknowledgements**

We would like to acknowledge the little children with IEM from whom we had learnt and perfected the art of HSCT and the team involved in the care of children with IEM-Dr.Prasad Manne, Dr.Indira Jayakumar, Dr.Ravindra Prasad and Dr.Mahesh Narayanan.

**Bibliography**

1. Tan EY, Boelens JJ, Jones SA, Wynn RF, Hematopoietic Stem Cell Transplantation in Inborn Errors of Metabolism. *Front. Pediatr.* 2019 Oct 25;7:433.doi: 10.3389/fped.2019.00433. eCollection 2019.
2. Swaminathan VV, Meena S, Varla H, Chandar R, Jayakumar I, Ramakrishnan B, et al. Hematopoietic Stem Cell Transplantation for Children With Inborn Errors of Metabolism: Single Center Experience Over Two Decades. *Indian Pediatr.* 2022 Sep 15; 59(9):699-702.
3. Hivre M, Holkar SR, Vaishnav D, Surkar P, Chandankhede S, Vidhale D. Inborn Errors of Metabolism: A Comprehensive Clinical Review and Case-Based Analysis of Metabolic Disorders Across

---

\* Senior Consultant in Pediatric Hematology and Oncology

email : revaraj@yahoo.com

\*\* Consultant in Pediatric Hematology and Oncology, Apollo Speciality Hospitals, Chennai.

- Multiple Biochemical Pathways, Journal of chemical health risks, 2025; 15(4), 2168-2187.
4. Lum SH, Miller WP, Jones S, Poulton K, Ogden W, Lee H, et al. Changes in the incidence, patterns and outcomes of graft failure following hematopoietic stem cell transplantation for Hurler syndrome. *Bone Marrow Transplant.* (2017) 52:846-53. doi: 10.1038/bmt.2017.5
  5. Mahmood A, Raymond GV, Dubey P, Peters C, Moser HW. Survival analysis of haematopoietic cell transplantation for childhood cerebral X linked adrenoleukodystrophy: a comparison study. *Lancet Neurol.* 2007; 6 : 687-92
  6. Taylor M, Khan S, Stapleton M, Wang J, Chen J, Wynn R, et al. Hematopoietic stem cell transplantation for mucopolysaccharidoses: past, present, and future. *Biol Blood Marrow Transplant.* 2019; 25:e226-e46.
  7. Chiesa R, Wynn RF, Veys P. Haematopoietic stem cell transplantation in inborn errors of metabolism. *Curr Opin Hematol.* 2016; 23:530-35.
  8. Boelens JJ, Orchard PJ, Wynn RF. Transplantation in inborn errors of metabolism: current considerations and future perspectives. *Br J Haematol.* 2014; 167:293-303.