Neurodevelopmental Outcome after Hematopoietic Cell Transplantation in Inborn Errors of Metabolism: Current Considerations and Future Perspectives.

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Abstract

Inborn errors of metabolism (IEM) comprise an assorted group of inherited diseases, some of which are due to disordered lysosomal or peroxisomal function and some of which might be improved following hematopoietic cell transplantation (HCT). In these disorders the onset in infancy or early childhood is typically accompanied by rapid deterioration, resulting in early death in the more severe phenotypes. Timely diagnosis and immediate referral to an IEM specialist are essential steps in optimal management. Treatment recommendations are based on the diagnosis, its phenotype, rate of progression, prior extent of disease, family values, and expectations, and the risks and benefits associated with available therapies, including HCT. International collaborative efforts are of utmost importance in determining outcomes of therapy for these rare diseases, and have improved those outcomes significantly over the last decades. In this review, we will focus on the neurodevelopmental outcomes after HCT in IEM, providing an international perspective on progress, limitations, and future directions.

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