ORAL Submission

HEMOGLOBINOPATHIES, PRIMARY IMMUNE DEFICIENCY DISEASES AND METABOLIC DISORDERS. (ORAL-652)

ADVANCING TOWARDS AN OPTIMAL CURE – HSCT EXPERIENCE IN INHERITED METABOLIC DISORDERS

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Aims & Objectives: Inborn errors of metabolism (IEM) comprise a large group of inherited diseases due to disordered lysosomal (mucopolysaccharidosis), peroxisomal (adrenoleukodystrophy), or mitochondrial function. Hematopoietic stem cell transplantation (HSCT) offers a therapeutic option, and we present the data on our case series.

Patients / Materials & Methods: We performed a retrospective study from January 2008 to February 2020 on children diagnosed with inherited metabolic disorders based on enzyme analysis or gene mutation. We documented HSCT details, including conditioning regimen, engraftment, and outcome.

Results: Twenty-two children (MPS - 12, Gauchers - 4, X- ALD 3, MLD - 2, Osteopetrosis-1) underwent HSCT. The donor was a matched family donor in 5 , a mismatched family donor in 1, matched unrelated donor in 9, and a haploidentical family donor in 8 children. All children received myeloablative conditioning consisting of fludarabine with treosulfan in 17/22, busulfan in 4/22, and melphalan in 1/22 children. Peripheral blood stem cell was the predominant stem cell source, and three children received a cord unit. Engraftment occurred in 20 /21 children (90%), and one child died of diffuse alveolar hemorrhage before engraftment. Two children had secondary graft failure. We documented viral reactivation in ten children (45%). The incidence of acute graft versus host disease (GVHD) was high at 59% (13/22) with skin involvement in 7 children (32%), gut involvement in 5 children (23%), and liver in one child. Two children have survived with extensive chronic and musculoskeletal GVHD. Two of the three children with X-ALD suffered a progression of their disease and died. One child with Gauchers disease transplanted post-splenectomy died a year after HSCT due to septic shock.

Discussion & Conclusion: The overall survival after HSCT for children with IEM was 69% in our cohort. With increasing awareness and early diagnosis, optimal outcomes are feasible with HSCT in low and middle-income countries with no access to lifelong enzyme replacement therapy or gene therapy

Disclosure of Interest: None Declared

Keywords: HSCT, inherited metabolic disorders, Outcome