

HEMATOPOIETIC STEM CELL TRANSPLANTATION WITH REDUCED TOXICITY CONDITIONING REGIMEN IN MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME (MNGIE)

Gülcihan Ozek¹, Serap Aksoylar¹, Sema Kalkan Uçar¹, Ebru Canda¹, Mediha Akcan¹, Ozgur Carti¹, Zuhul Siviş¹, Yesim Oymak¹, Havva Yazıcı¹, Bridget Bax², Fatma Derya Bulut¹, Merve Yoldaş Çelik¹, Fehime Erdem¹, Mahmut Çoker¹, and Savaş Kansoy¹

¹University of Michigan Department of Pediatrics

²St George's University of London Molecular and Clinical Sciences Research Institute

January 10, 2023

Abstract

Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE) is a rare autosomal recessive disorder due to mutations in the *TYMP* gene. Clinical findings are characterized by neurologic manifestations and severe gastrointestinal dysfunction. The syndrome is usually fatal, the most effective treatment appears to be hematopoietic stem cell transplantation (HSCT). Aim: In this retrospective study, we evaluated HSCT that was performed using a reduced toxicity myeloablative conditioning regimen in patients with MNGIE at our center. Results: A total of 6 allogeneic transplant procedures were performed in 4 patients. Three patients' donors had fully matched donors, and one patients' donor was haploidentical. Treosulfan-based myeloablative conditioning regimen was applied in 5 of 6 transplants. Bone marrow was used as a stem cell source. One patient is being followed up in the 4th year of posttransplant with full chimeric and without Graft versus host disease (GVHD). One patient died of acute stage IV GIS GVHD. Two patients underwent second transplantation due to engraftment failure, one of which was the patient who had a haploidentical transplant. Conclusions: Treosulfan-based regimen is well tolerated. Engraftment failure with this conditioning regimen can be a significant problem. We share our haploidentical transplant experience, which will be the first reported case in the literature.

Hosted file

MNGIE.docx available at <https://authorea.com/users/469887/articles/618279-hematopoietic-stem-cell-transplantation-with-reduced-toxicity-conditioning-regimen-in-mitochondrial-neurogastrointestinal-encephalopathy-syndrome-mngie>

Hosted file

MNGIE-TABLE 1.docx available at <https://authorea.com/users/469887/articles/618279-hematopoietic-stem-cell-transplantation-with-reduced-toxicity-conditioning-regimen-in-mitochondrial-neurogastrointestinal-encephalopathy-syndrome-mngie>

Hosted file

MNGIE TABLE 2.docx available at <https://authorea.com/users/469887/articles/618279-hematopoietic-stem-cell-transplantation-with-reduced-toxicity-conditioning-regimen-in-mitochondrial-neurogastrointestinal-encephalopathy-syndrome-mngie>