soho2023
ELEVENTH ANNUAL MEETING

Allogenic Stem Cell Transplantation for ADA2 deficiency

ZEYNEP TUĞBA GÜVEN[1], ELİF SUYANI[1]
[1]Adana City Hospital, Department of Hematology, Adana, Turkey

Add your logo here

INTRODUCTION

Adenosine deaminase 2 (ADA2) deficiency is an autoinflammatory disease that develops due to mutations in the cat eye syndrome chromosome region candidate 1 (CECR1) gene, called ADA2.



The disease has a broad clinical spectrum, including vasculopathy, relapsing fever, immunodeficiency, and bone marrow dysfunction. Hematopoietic stem cell transplantation (HSCT) can be curative if a suitable donor is available. Here we describe an 18-year-old case of ADA2 deficiency who underwent allogeneic stem cell transplantation.

CASE REPORT

A 15-year-old female patient was evaluated for a history of recurrent infections since the age of 10. The patient had anemia and low total leukocyte count with decreased neutrophils. Bone marrow evaluation was hypercellular. Bone marrow karyotyping was normal. He was treated with intermittent injections of filgrastim; however, no adequate response was received. Steroid therapy was given for resistant cytopenias. however, severe steroid-related side effects were observed. Evaluation of the immune system revealed low B cell counts and low IgG. Immunoglobulin replacement was initiated. homozygous mutation in CECR1 (c.G506A) was found by exome sequencing.

A diagnosis of immunodeficiency due to ADA2 deficiency was made. Anti-TNF treatment was started in the patient who developed livedo reticularis, but no response was obtained. Due to her severe disease, allogeneic hematopoietic stem cell transplantation (aHSCT) was planned. The patient underwent a myeloablative conditioning with busulfan, fludarabine and anti-thymocite globulin.

Graft versus host disease (GVHD) prophylaxis consisted of, short methotrexate, cyclosporine (from day 0 to day +28) than replaced by tacrolimus (from day +29). Post-transplant, she had neutrophil and platelet engraftment on days +11 and +12 respectively. Complete donor chimerism was detected on day +33. She completely resolved the clinical signs of ADA2 deficiency in the patient who was approaching the 1st year of transplantation.

CONCLUSIONS

HSCT is an effective treatment option for treatment of ADA2 deficiency.

CONTACT INFORMATION

drztkarabulutguven@gmail.com