Coexistence of Gaucher Disease and High-Risk MDS: A Rare Case With Favorable Transplant Outcome

A Lal[1], C Anageldiyev[1], M khan[2], S Rath[3], S Naik[1]

1. Penn State Health Milton S Hershey Medical Center, Hershey, PA
2. George Washington University, DC,
3. All India Institute of Medical Sciences Bhubaneshwar, India

Introduction

Gaucher disease is rare lysosomal storage disorder, commonly associated with hematologic complications cytopenias, splenomegaly, and, sometimes, pathologic fractures. Although malignancy is extremely rare, when present, it most often involves hematologic cancers like multiple myeloma. We present a case of high-risk myelodysplastic syndrome (MDS) with complex cytogenetics in a patient with Gaucher disease, successfully managed with allogeneic stem cell transplantation.

Case Presentation

A 57-year-old female with long-standing Gaucher disease on enzyme replacement therapy presented with progressive pancytopenia. Her past medical history includes multiple orthopedic procedures, including bilateral hip and knee replacements, right ankle fusion, and left femoral plating. She had splenectomy at age of 5 and had long-standing chronic anemia and thrombocytopenia. Bone marrow biopsy revealed 4–5% blasts and a complex karyotype, including del(1)(p32p36.1), del(5)(q15q33), t(1;6)(q21;q23), and monosomy 7, consistent with diagnosis of MDS with elevated risk of transformation to acute myeloid leukemia (AML).

Due to absence of a matched donor, underwent haploidentical allogeneic stem cell transplantation, using fludarabine/cyclophosphamide (FluCy) conditioning regimen with post-transplant cyclophosphamide (PTCy), without total body irradiation.

Her pre-transplant bone marrow biopsy showed 4–5% blasts. Follow-up bone marrow biopsies at one and two years post-transplant showed no morphologic evidence of residual disease. At two years, donor chimerism analysis demonstrated 99.9% total and 99.86% T-cell chimerism. Enzyme levels normalized post-transplant, allowing for discontinuation of velaglucerase.

Discussion

This case highlights the rare association between Gaucher disease and MDS. The presence of complex cytogenetic, including monosomy 7, represent high-risk MDS, with a significant potential for transformation to acute leukemia. Factors such as prior splenectomy and chronic bone marrow stress may have contributed to clonal evolution. Despite multiple comorbidities, patient successfully underwent haploidentical stem cell transplantation, facilitated by early referral, a tailored reduced-intensity conditioning regimen, and vigilant post-transplant management. This case also emphasizes the importance of a multidisciplinary approach in managing patients with coexisting genetic and hematologic disorders.

Conclusion

Allogeneic stem cell transplantation represents viable and effective therapeutic option for patients with Gaucher disease and high-risk MDS. Haploidentical transplantation with a reduced-intensity conditioning regimen can achieve favorable outcomes, even in medically complex cases, when supported by comprehensive care and close monitoring.

SOHO2025